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GENOMICS IN NURSING PRACTICE IN AUSTRALIA: A CRITICAL REALIST CASE STUDY

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Dedication

I dedicate this thesis to the memory of my father, Eric Worsley Wright,

or, as we knew him, '*Eric the Wonderful*'.

Statement of Original Authorship

The work contained in this thesis has not been previously submitted to meet requirements for an award at this or any other higher education institution. To the best of my knowledge and belief, the thesis contains no material previously published or written by another person except where due reference is made.

Signature:

Date: 11th December 2018

Statement on the Contribution of Others

Nature of Assistance/ Contribution	Contribution	Names, Titles and Affiliations of Co-contributors
Intellectual support	The supervisory team provided assistance with proposal writing and data analysis within the remit of their roles, as described in the policy document, <i>The Role of the Advisory Panel in Providing Regular Guidance and Support to Research Students</i>	Professor Melanie Birks, Discipline Head, Nursing and Midwifery, James Cook University Professor Jane Mills, Adjunct Professor, Nursing and Midwifery, James Cook University Dr Lin Zhao, Senior Lecturer, Nursing, RMIT University
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Abstract

Genomic research continues to ‘change the landscape’ of healthcare worldwide (Camak, 2016, p.86). Genomics is beginning to reshape healthcare delivery by changing the way we prevent, diagnose, treat and monitor illness, providing the opportunity to offer more precise and tailored treatments. As genomic developments change healthcare, so too are they changing the nursing profession. This revolution has led to a new responsibility for all nurses to be knowledgeable of genomics and incorporate genomics into nursing practice. Research addressing the integration of genomics into nursing practice in Australia is limited. The aim of this study was to determine how nurses engage with genomics in nursing practice in this country.

Case study research was used to achieve the research aim. A case study is ‘an empirical inquiry that investigates a contemporary phenomenon (the ‘case’) in depth and within the real-world context’ (Yin, 2014, p. 16). A single holistic case study design drawing on the works of Robert Yin (2014) was conducted. This case study was underpinned by a critical realist philosophy. Critical realism is concerned with the nature and knowability of the social world and social phenomena (Schiller, 2016), making it a suitable framework to guide an exploration of Australian nurses’ engagement with genomics.

Data were collected via a cross-sectional survey of Australian registered nurses and midwives in 2016, and via semi-structured interviews with registered nurses working in oncology departments within a regional Australian hospital in 2018. Key case findings were generated using thematic analysis, and grouped into three categories: Point of learning (education), Point of reference (professional expectations) and Point of care (clinical practice). These three categories were used as a framework to describe the case, and presented in relation to the key tenets of critical realism - (i) the primacy of ontology, (ii) the stratified character of the real-world (reality) and the search for generative mechanisms, and (iii) the interplay between social structures and human agency (Bhaskar, 1975/2008, 1979/1998, 2011).

The case indicated that Australian nurses have limited engagement with genomics at the point of learning, point of reference and point of care. Nurses’ inadequacy at each of these points is sequential, meaning that if nurses are not knowledgeable about genomics and are unclear about professional expectations, they cannot be expected to adequately integrate genomics into their

practice. The critical realist philosophy underpinning the case led to consideration of the way point of learning, point of reference and point of care form the context for nursing practice. How nurses respond to this context determines the extent to which they are able to transform education, policy and practice.

Australian nurses' limited engagement with genomics has consequences for the nurse, the patient and the wider nursing profession. This limited engagement must be addressed. It is recommended that (i) genomics be embedded throughout the nursing curricula with healthcare applications made clear to the learner (point of education), (ii) nursing policy articulates the alignment between the NMBA's Standards for Practice and genomic competencies (point of reference), and (iii) nurses incorporate genomics knowledge and skills into practice (point of care). The 'genomic revolution' (Jenkins et al., 2005, p.98) will require further development of Australia's capacity, capability and infrastructure if these are to support the integration of genomic information and technology into the national health system (Australian Health Ministers' Advisory Council, 2017b). As the largest component of the Australian health workforce, nursing cannot ignore the opportunity before us.

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List of Abbreviations

AGHA	Australian Genomics Health Alliance
AHPRA	Australian Health Practitioner Regulation Agency
DNA	Deoxyribonucleic Acid
ELSI	Ethical, Legal and Social Implications
G2NA	Global Genomics Nursing Alliance
GNCI	Genomic Nursing Concept Inventory
HER2	Human Epidermal Growth Factor Receptor 2 Protein
HGP	Human Genome Project
HREC	Human Research Ethics Committee
ISONG	International Society of Nurses in Genetics
NMBA	Nursing and Midwifery Board of Australia
QGHA	Queensland Genomics Health Alliance
UK	United Kingdom
US	United States

Terminology

Competency

Competency refers to an observable, measurable, performance-based outcome that indicates the achievement of a particular knowledge component, and application or demonstration of a psychomotor behaviour or skill.

Genetics

Genetics is the study of individual genes and their influence on single gene disorders.

Genetic counselling

Genetic counselling is a communication process that aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions.

Genetic literacy

Genetic literacy refers to knowledge sufficient to develop genetic and genomic competency.

Genomics

Genomics is the study of all the genes in the human genome together or as a subset, including their interactions with each other and the environment, and the influence of other psychosocial and cultural factors.

Genetics/genomics nursing

Genetics/genomics nursing refers to the protection, promotion and optimisation of health and abilities, prevention of illness and injury, and alleviation of suffering through the diagnosis of human response and advocacy in the care of the genetic and genomic health of individuals, families, communities and populations.

Genetics nursing practice

Genetics nursing practice refers to providing client-centred nursing care, education or research based on understanding the underlying genomics of individuals, families, communities or populations affected by, or at risk for, a disease or condition with a genetic component.

Scope of practice

Scope of practice refers to the area in which nurses are educated, competent to perform and permitted to perform by law. The actual scope of practice is influenced by the context in which the nurse practises, the health needs of people, the level of competence and confidence of the nurse, and the policy requirements of the service provider.

Standards for practice

Standards for practice in this document refer to the expectations of registered nurse practice. They inform the education standards for registered nurses, regulation of nurses and determination of nurses' capability for practice, and guide consumers, employers and other stakeholders about what to reasonably expect from a registered nurse, regardless of the area of nursing practice or years of nursing experience. They replace the previous *National Competency Standards for the Registered Nurse*.

Prologue

Genomics is an unusual area of interest for a nursing academic in Australia—I know this based on experience. Nurses frequently ask each other about their nursing specialty or area of interest, and I always answer by saying *genomics*. The responses I receive from colleagues range from genuine enthusiasm to borderline hostility. However, in the middle are colleagues who respond with polite scepticism, evidenced by a genuine desire to know ‘what has that got to do with nursing?’. This polite scepticism can be seen in wider academia. Three manuscripts were submitted for publication over the course of this dissertation. The editors’ tepid responses to the manuscripts are reflective of the nursing professions’ scepticism of genomics. As one reviewer stated in relation to one manuscript, ‘This is a well-written paper on a topic I have extensive reservations about’. The reviewer’s reservations were clearly directed at the topic, as opposed to the manuscript itself. The reviewer also stated that ‘I have never used genomics in practice and am unlikely to. Furthermore, I can confidently say I don’t need genomic literacy for my practice area’. I believe this is a ‘case in point’ for the ‘case’ of genomics in nursing practice in Australia, yet I do not hold the reviewer accountable for this short-sightedness. Rather, I argue that this response reflects the lack of awareness of the wider nursing profession. I would like to see this awareness change. Nursing academics in the United States and United Kingdom are leading the way in nursing and the genomic revolution. I would like to see Australia follow suit with our own nuanced approach to incorporating genomics into nursing education and practice. Thus far, I have been met with great support from my colleagues and even greater support from my doctoral advisors, albeit with frequent good-natured taunts from some—yes, Professor Birks, that means you. Many of those who know me will, at some point, have asked the ‘million dollar’ question: ‘what has that got to do with nursing?’. You will all be pleased to know that I have reduced my answer from a one-hour lecture to one sentence: ‘Genomic information will ultimately pervade all of health care’ (Jenkins, Grady, & Collins, 2005, p. 98). Nurses are healthcare professionals—get on board.

Chapter 1: Introduction

1.1 Introduction

The study described in this thesis explores Australian nurses' engagement with genomics in practice and contributes to an understanding of the genomic knowledge and skills employed by nurses, nurses' perceptions of the relevance of genomics in nursing practice, and nurses' experience of using genomics in day-to-day nursing practice. This chapter introduces the study and serves as a precursor to the detailed exploration in the chapters that follow. The chapter will introduce key terminology and discuss the impetus for the study, including the author's personal perspective on the topic. Context to the study will be provided through reference to the Human Genome Project (HGP) and its translation into genomic healthcare, as well as genomics in nursing and nursing education. The aim and objectives will be presented, followed by the case design and underlying theoretical framework used in this research. Moreover, the significance of the study will be addressed, demonstrating its anticipated contribution to the literature.

1.2 Terminology

The terms 'genetics' and 'genomics' are frequently used interchangeably in the literature; however, they are distinct terms. Genetics refers to 'the study of individual genes and their impact on relatively rare single gene disorders', whereas genomics is 'the study of all the genes in the human genome together, including their interactions with each other, the environment, and the influence of other psychosocial and cultural factors' (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, pp. 8–9). Genetics is perhaps the more familiar term; however, genomics has a 'broader and more ambitious reach' (Guttmacher & Collins, 2002, p. 1512). The term 'genomics' will largely be used throughout this thesis to refer to both the study of single genes (genetics) and the study of an individual's entire genetic makeup (genome), along with how it interacts with environmental or non-genetic factors. Exceptions to this rule will occur when it is necessary to differentiate between the two terms, where the more familiar term 'genetics' is more appropriate, or in reference to the literature, where a particular term is considered more consistent with the author's intentions. The terms 'genomics' and 'genomic knowledge' used in this document refer to the data, information and learnings derived through genomic research and technologies used for testing, analysing and

furthering the discovery of genomic knowledge. This terminology was selected to reflect that used by the Australian Government's *National Health Genomics Policy Framework 2018–2021* (Australian Health Ministers' Advisory Council, 2017b).

1.3 Call to the Question

The author developed an interest in genetics during her time working as an associate genetic counsellor with a government genetic health service. Genetic counsellors have specialised education in genetics and counselling. In Australia, prospective genetic counsellors are required to complete a clinical Master of Genetic Counselling, after which an individual can seek employment as an associate genetic counsellor and apply for board eligibility from the Human Genetics Society of Australasia.

Genetic counselling can be defined as 'a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions' (Resta et al., 2006, p.80). As a registered nurse and an associate genetic counsellor, the author came to appreciate the role of genomic knowledge and skills in healthcare. The genetic counselling process integrates the following:

- interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
- education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research
- counselling to promote informed choices in view of risk assessment, family goals, ethics and religious values
- support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (Resta et al., 2006).

These skills or 'functions' (interpret, educate, counsel and support) are not restricted to genetic counselling—they have a place in all healthcare professions in various capacities. Genetic counselling and nursing are distinct professions with separate scopes and standards of practice. However, as with many healthcare professions, some crossover of knowledge and skills is inevitable. Nurses interpret, educate, counsel and support as part of their nursing care delivery.

These skills can be tailored to nursing practice, just as they are tailored to other healthcare professionals.

Since acknowledging the crossover between genetic counselling and nursing, the question was raised in the author's mind about what Australian nurses know about genomics and how Australian nurses are using genomics. Having worked as a registered nurse after working as a genetic counsellor, the author's impression was that nurses know little about genomics and are not adequately using genomics in practice. It appears that the opportunity to interpret, educate, counsel and support individuals with respect to genomic matters is not being seized. The author acknowledges the inherent bias that comes with seeing one profession (nursing) through the eyes of another profession (genetic counselling); however, this research study was conducted from a nursing perspective, and sought to provide information that can benefit the nursing profession. Any potential bias on the part of the researcher was managed through early articulation of her assumptions and counterbalanced by the supervisory team.

Although anecdotal evidence indicates that genomics is present in nursing practice in Australia, a comprehensive investigation has not been undertaken. Thus, nurses' engagement with genomics in practice in Australia remains unclear. As such, this project sought to determine how Australian nurses engage with genomics and create a picture of Australian nurses' engagement with genomics through summarising the genomic knowledge and skills of nurses, and how these are used in the delivery of nursing care.

1.4 Genomics

1.4.1 Human Genome Project

The greatest advances in genomics are derived from the HGP. The HGP was an international research initiative with the aim of sequencing the entire human genome (National Human Genome Research Institute, 2016). The HGP was launched in 1990 with the intent to be completed within 15 years. In 2000, a 'working draft' of the human genome was announced and published in the journal *Nature* in February 2001 (90% of the sequence of the genome's three billion base-pairs). The full sequence was completed and published in April 2003—two years ahead of schedule. This sequence represents the broad architecture of all human genomes that scaffolds current and future work aiming to characterise individual sequence variation. Surprise findings in the human genome sequence included: (i) the relatively small number of

human genes (as few as 30,000); (ii) the complex architecture of human proteins compared with their homologs in other species, such as *C. elegans* (roundworms) or *Drosophila* (fruit flies); and (iii) the role of repeat sequences of deoxyribonucleic acid (DNA) (National Human Genome Research Institute, 2016).

Advances in DNA technologies have accelerated the sequencing process. These advances include improvements in the methods used to decipher the DNA base-pair sequences, improvements in the computing facilities required for data management and improvements in the analytical instruments. This rapid evolution of next-generation DNA sequencing technologies has reduced the cost of sequencing a human genome (Rehm, 2017). The estimated cost for generating that initial ‘draft’ human genome sequence is ~\$300 million worldwide, and for advancing the ‘draft’ human genome sequence to the ‘finished’ sequence is ~\$150 million worldwide (National Human Genome Research Institute, 2016). Based on the data collected from National Human Genome Research Institute–funded genome-sequencing groups, the cost to generate a high-quality ‘draft’ human genome sequence had fallen to ~\$14 million by 2006, and fallen to below \$1,500 by late 2015 (National Human Genome Research Institute, 2016). These advances and subsequent cost reductions have rendered clinical genomics a viable option for healthcare.

Alongside the HGP ran the Ethical, Legal and Social Implications (ELSI) program. The ELSI program was founded in 1990 as an integral part of the HGP, designed to identify and address issues raised by genomic research that would affect individuals, families and society (National Institutes of Health, 2018). The ELSI program focused on the possible consequences of genomic research in four main areas: (i) privacy and fairness in the use of genetic information; (ii) the integration of new genetic technologies into practice; (iii) ethical issues surrounding genetic research with people; and (iv) education of healthcare professionals, policy makers, students and the public about genetics and the complex issues associated with genomic research.

The findings of the HGP have made their way into the public domain, and genomics has an increasing presence within this domain. There are frequent references to genetics and genomics with respect to healthcare in the media and other publications that are freely available to the public. There are new terms, concepts and issues appearing in the media every day, such as stem cells, cloning, biobanks and ‘saviour siblings’, to name a few. Each of these concepts has

a place in healthcare, and the public may well turn to healthcare professionals, such as nurses, for further information.

Genetic testing is also more accessible to the public. For example, companies such as ‘23 and me™’ are offering personalised genome screening. Personalised genome screening ensures that risk assessment of disease, health screening and promotion, and disease treatments are tailored to the individual’s genetic profile (Garcia, Greco, & Loescher, 2011). This direct-to-consumer testing is now available, and is allowing the public to learn about their genetic makeup (Cashion, 2009). The success of these companies indicates that the public are aware of genetic tests and their application to healthcare. Increased public awareness will mean that healthcare professionals are required to accommodate patients’ questions about genomics and the way that genomics affects their healthcare (Rogers, Lizer, Doughty, Hayden, & Klein, 2017).

1.4.2 Genomics in Healthcare

The completion of the HGP in 2003 ushered in a new era in healthcare—an era in which health professionals increasingly use genetics/genomics technology and information to improve the health of those in their care. Genomic applications in healthcare continue to increase, transforming healthcare delivery in ways that have the potential to increase quality and safety, decrease costs and improve health outcomes (Calzone et al., 2018b; Williams, Feero, Leonard, & Coleman, 2017). Table 1.1 presents some examples of the ways genomics can benefit healthcare.

Table 1.1: Examples of Ways Genomics Can Benefit Healthcare

Benefit	Description	Example
Improve health outcomes	Prevent unnecessary medical procedures based on an individual's genetic status	A person who does not have a genetic predisposition to bowel cancer can avoid unnecessary colonoscopies, even if they are at 'population risk' for bowel cancer.
	Select medical procedure and/or treatments based on an individual's genetic status	Imatinib mesylate (Gleevec®) targets the BCR-ABL fusion protein, which is made from pieces of two genes that are joined together in some leukaemia cells and promote the growth of leukemic cells.
Increase quality and safety	Preventing or minimising adverse reactions to medications	Warfarin has a narrow therapeutic index. Small changes in plasma levels may result in concentration-dependent adverse drug reactions or therapeutic failure. Therefore, warfarin dose is tailored to each person according to the person's response (measured as international normalised ratio) and the condition being treated. The <i>VKORC1</i> and <i>CYP2C9</i> genotypes are the most important known genetic determinants of warfarin dosing.
	Varied responses to medications	Genetic influences are responsible for metabolism of opioids through the cytochrome p450 system (CYP), including <i>CYP2D6</i> , <i>CYP3A</i> and <i>CYP2B6</i> . These genetic influences can enhance or reduce a patient's response to opioids.
Decrease costs	Prevent or minimise illness, leading to reduced healthcare costs	Decreasing the frequency of adverse drug effects and increasing the probability of successful therapy will likely lower the cost of healthcare. For example, identification of patients with homozygous familial hypercholesterolaemia via genetic testing and prophylactic therapy (predominantly statins) can reduce patient deaths and major adverse cardiovascular events.

Genomic information and technologies can now be used at any stage of the healthcare continuum to determine disease risk, predisposition, diagnosis and prognosis, and the selection and prioritisation of therapeutic options (Kirk, Campalani, et al., 2011, p. 6), thereby leading to improved healthcare outcomes. Table 1.2 provides examples of genomics being used across the healthcare continuum, from prevention to diagnosis, treatment and prognosis.

Table 1.2: Genomics across the Healthcare Continuum

Stage in Healthcare Continuum	Example
Prevention	A young woman is concerned about her risk of breast cancer after a significant family history of breast and ovarian cancer. Her mother was recently diagnosed with an invasive ductal carcinoma; she underwent genetic testing and was found to have a <i>BRCA1</i> gene mutation. The young woman had predictive genetic testing, and was also found to have the <i>BRCA1</i> gene mutation. Rather than have a prophylactic mastectomy, the young woman elected to have frequent breast screening to detect any early changes in the breast tissue that may indicate breast cancer.
Diagnosis and prognosis	Prenatal diagnostic testing is offered to couples with an increased risk of having a baby with a genetic or chromosomal disorder. Prenatal diagnostic testing is used to detect changes in a foetus' genes or chromosomes that may indicate a genetic disease or disorder. A tissue sample for prenatal testing can be obtained through amniocentesis or chorionic villus sampling.
Treatment	Tamoxifen® is used to treat breast cancer that is hormone receptor positive (HER2 ⁺). An example of such a differentially expressed target is the human epidermal growth factor receptor 2 protein (HER2). HER2 is expressed at high levels on the surface of some cancer cells. Several targeted therapies are directed at HER2, including Herceptin®, which is approved to treat certain breast and stomach cancers that overexpress HER2.

1.4.3 Precision Medicine

The terms ‘precision medicine’ and ‘personalised medicine’ are frequently used interchangeably. ‘Personalised medicine’ was the original term used to denote individualised care; however, according to the National Research Council (National Institutes of Health, 2015), there was concern that the word ‘personalised’ could be misinterpreted to imply that treatments and preventions are being developed uniquely for each individual. This is not the case. ‘Precision medicine’ includes the concept of personalised medicine at a more exact level through advances in science and technology, such as genetics and genomics sequencing (Ashley, 2016; Hammer, 2016). In ‘precision medicine’, the aim is to identify which approaches will be effective for which patients based on genetic, environmental and lifestyle factors. ‘Precision medicine’ is now the preferred term. Precision medicine has made visible the healthcare benefits of genomics. Treatments are no longer solely based on anatomic site and tumour histology. Instead, new targeted therapies allow drug selection based on mutations present in an individual tumour, and are often associated with effective therapies (Ewing, 2014). An example of targeted therapy is the use of trastuzumab (Herceptin™) in HER2-amplified breast cancer (Ewing, 2014). The use of Herceptin™ in HER2-amplified breast cancer is one of the earliest examples of personalised treatment, and has led to improved patient outcomes.

A nursing workforce knowledgeable in genetics and genomics is vital in this era of personalised and precision healthcare (Rogers et al., 2017). Precision medicine brings with it advanced testing, care and treatment. The communication, support and advocacy for patients associated with personalised and precision medicine mean that nurses face new challenges: ‘Nurses must have adequate preparation and knowledge of the ongoing evidence to care for patients using personalized strategies’ (Vorderstrasse, Hammer, & Dungan, 2014).

1.4.4 Genomic Healthcare in Australia

Australia acknowledges that genomics plays a key role in health. The Australian Institute of Health and Welfare (2018a) argues that health is a state of wellbeing that reflects the complex interactions of a person’s genetics, lifestyle and environment. They state that:

Australia’s health system currently faces many challenges. These include demographic changes and the demand for health services; coordinated management of chronic conditions; greater availability and access to health data; and advances in medical research, science and technology (such as genetic testing). (Australian Institute of Health and Welfare, 2018a, p. 38)

Further, advances in medical science have seen a growth in genetic testing services in Australia (Australian Institute of Health and Welfare, 2018a). Implementation of genomic discoveries into healthcare optimally includes evaluation of outcomes for recipients of care, providers, payers and healthcare systems (Williams et al., 2017). Genomics has the potential to reshape clinical practice and fundamentally change the way we prevent, diagnose, treat and monitor illness, providing the opportunity to offer more precise and tailored treatments. The ability to respond to this change is dependent on further developing Australia’s capacity, capability and infrastructure needed to support integration of genomic technology into the national health system (particularly with regard to clinical utility, workforce, education, data security and sharing, quality and accreditation of nursing programs, cost-effectiveness and research) (Australian Health Ministers’ Advisory Council, 2017b). Achieving and maximising health outcomes for all Australians requires a collaborative and coordinated approach at all levels of government and across stakeholders. The *National Health Genomics Policy Framework 2018–2021* presents a shared commitment to leveraging the benefits of genomics in the health system for all Australians (Australian Health Ministers’ Advisory Council, 2017b). The vision is to ‘help Australians live longer and better by integrating genomics into the health system through

taking coordinated action across agreed strategic priority areas' (Australian Health Ministers' Advisory Council, 2017b, p. i).

Developing Australia's capacity, capability and infrastructure needed to support integration of genomic technology into the national health system comes at a cost. Australia's healthcare spending is sizable. Australia spent nearly \$181 billion on health in 2016 to 2017 (Australian Institute of Health and Welfare, 2018b). The real growth (adjusted for inflation) in health spending of 4.7% in 2016 to 2017 was 1.6 percentage points higher than the average over the past five years (3.1%) (Australian Institute of Health and Welfare, 2018b). The exact amount spent on genomic healthcare is difficult to determine; however, there has been funding clearly directed at genomics. The Australian Government (2018b) will invest \$500 million over 10 years in an Australian Genomics Mission to help save or transform the lives of more than 200,000 Australians through research into better testing, diagnosis and treatment. The \$500 million Australian Genomics Health Futures Mission is the centrepiece of the government's \$1.3 billion National Health and Medical Industry Growth Plan, announced in the 2018 to 2019 federal budget. The first genomics project will be Mackenzie's Mission, with \$20 million being provided for a preconception screening trial for rare and debilitating birth disorders, including spinal muscular atrophy, fragile X syndrome and cystic fibrosis (Australian Government, 2018a).

Genomics in healthcare is also being addressed at the state level. The Queensland Genomics Health Alliance (QGHA) aims to improve the health of Queenslanders by delivering genomic medicine (Queensland Genomics Health Alliance, 2018). The QGHA seeks to understand how genomics can improve healthcare and health outcomes throughout the communities of Queensland. The QGHA will drive collaboration between the state's health system and research and academic communities. Queensland Health published *Queensland Advancing Health Research 2026* (Queensland Health, 2018), which is concerned with supporting, integrating and expanding the conduct and translation of research in the health system. *Queensland Advancing Health Research 2026* is designed to guide Queensland Health's research investment decisions and actions to achieve a vision of healthier Queenslanders through research-informed healthcare. *Queensland Advancing Health Research 2026* seeks to support the QGHA and other national initiatives to pioneer the introduction of genomics into healthcare and ensure that the state is a leading contributor to this technological advance (Queensland Health, 2018).

New Australian genomic organisations have been created to foster the integration of genomics into healthcare. The Australian Genomics Health Alliance (AGHA), frequently referred to as 'Australian Genomics', is a national research collaboration of clinicians, researchers and diagnostic geneticists working together to provide evidence for the equitable, effective and sustainable delivery of genomic medicine in healthcare. Australian Genomics unites 80 organisations, including the clinical and diagnostic genetics services across the nation, along with major research and academic institutions. Australian Genomics operates on many levels, including state-based genomics initiatives, state and federal government, and international policy.

1.5 Genomics in Nursing

1.5.1 Genomics in Nursing Practice

Genetic knowledge has traditionally been viewed as useful, but not necessary to nursing practice (Calzone et al., 2010)—this view is changing. Completion of the HGP led to the expansion of genetics and genomics nursing practice (Kerber & Ledbetter, 2017), and new scientific developments come with new nursing considerations (Cheek, Bashore, & Brazeau, 2015; Cheek & Howington, 2017). Nurses must be able to respond to the clues and cues that could affect the prevention, early detection or treatment of common conditions, such as cancer or heart disease (Skirton, 2017). The ability to respond to patients' presentation will require nurses to be aware of genetic influences on health and disease.

Nurses are involved across the healthcare continuum, and subsequently need to be well informed if they are to use genetics and genomic technologies in their clinical practice. Genomics has started to pervade healthcare across all stages of life, from preconception to adult medicine (Rehm, 2017). For people to benefit from widespread genetic/genomic discoveries by the HGP, nurses must be:

competent to obtain comprehensive family histories, identify family members at risk for developing a genomic influenced condition and for genomic influenced drug reactions, help people make informed decisions about and understand the results of their genetic/genomic tests and therapies, and refer at-risk people to appropriate healthcare professionals and agencies for specialized care. (Calzone et al., 2010, p. 27)

Table 1.3 provides exemplars of these skills. These nursing skills are applicable to all nurses, not just those working in specialist areas. Genetics is no longer confined to rare and single gene disorders. It is now known that genetics contributes to numerous common conditions and diseases, such as diabetes, cardiovascular disease and cancer (Cashion, 2009). This new ‘layer’ to disease means that nurses will be caring for patients with genetic diseases and disorders on a daily basis; thus, nurses require adequate genomic knowledge and skills to deliver appropriate care.

Table 1.3: Examples of Ways Nurses Can Use Genomics in Practice

Skill	Example
Competent to obtain comprehensive family histories	The nurse is able to collect a three-generation family history, noting information relevant to the individual’s condition, such as age at diagnosis and age at death in a family with bowel cancer
Identify family members at risk for developing a genomic-influenced condition and for genomic-influenced drug reactions	The nurse is able to identify an at-risk individual by identifying ‘red flags’, such as type of cancer, cancer across multiple generations and early age at diagnosis
Help people make informed decisions about and understand the results of their genetic/genomic tests and therapies	The nurse is able to help individuals make informed decisions about genetic testing, such as the advantages of genetic testing and the potential disadvantages of genetic testing, such as discrimination
Refer at-risk people to appropriate healthcare professionals and agencies for specialised care	The nurse is familiar with the local genetic services and refers at-risk individuals to these services

Collegiality in the genomics healthcare community is strengthening. The International Society of Nurses in Genetics (ISONG) (2018) is a global nursing specialty organisation dedicated to fostering the scientific and professional growth of nurses in human genetics and genomics worldwide. ISONG fosters work in genomic healthcare, education, research and scholarship. The mission of ISONG is to serve both the nursing profession and public. ISONG (2018):

fosters and advocates for the scientific and professional development of its members and the nursing community, in the discovery, interpretation, application, and management of genomic information, for the promotion of the public’s health and wellbeing. ISONG advocates for public understanding of genomic health and use of genomic information. (para.2)

Similarly, the Global Genomics Nursing Alliance (G2NA) was formed in January 2017 to ‘accelerate the integration of genomics across everyday nursing practice’ (para.2). The G2NA is not focused on the genetic specialist, but on the general nursing community. The G2NA aims to share genomic resources among the international nursing community, with the intent that

this genomic knowledge mobilization will increase resource accessibility and decrease duplication of efforts via leadership, collaboration and sharing. Overall, the G2NA aims to ‘increase nursing capacity to integrate genomics into practice through supporting improvements in genomic literacy critical to adoption in practice’ (Calzone, Kirk, et al. 2018, p. 54).

1.5.2 Genomics in Nursing Education

Since the publication of Brantl and Esslinger’s (1962) seminal work, various individuals, consensus panels and organisations have promoted genomics education in nursing. In recent years, there have been various publications addressing the integration of genetics and genomics into nursing curricula (Daack-Hirsch, Dieter, & Quinn Griffin, 2011; Garcia et al., 2011). In particular, publications addressing faculty readiness have appeared frequently in the literature (Jenkins & Calzone, 2012; Read & Ward, 2016; Williams et al., 2011). All publications continue the call for the integration of genetics/genomics into nursing education, yet these frequent calls for the integration of genomics into nursing curricula are largely going unanswered. Genomics is still not fully integrated into nursing education (Kirk, Calzone, Arimori, & Tonkin, 2011).

1.5.3 Genomics in Nursing Education in Australia

In 2017, the author conducted a desktop analysis of pre-registration nursing and midwifery curricula in Australian universities to determine the extent to which genomic information is included in pre-registration nursing and midwifery programs in Australian universities. The purpose of this survey was to obtain a snapshot of current genomics education in nursing in Australia, while also informing and confirming the larger body of work undertaken in this thesis.

The author accessed the public websites of 34 universities, colleges and institutions, each offering a nursing or nursing and midwifery course. Genetics or genomics appeared in one or more subjects at 15 universities. The words ‘genetics’ and ‘genomics’ did not appear in any subject titles, yet were present in the aim and/or synopsis of 10 subjects, and in the learning outcomes or specific content of 16 subjects. In cases where genetics was present in a subject (aim, synopsis, learning outcomes or specific content), it was generally related to anatomy and pathophysiology. The findings prompted the author to conduct a survey with Australian universities.

The author surveyed Australian universities seeking information about their respective nursing curricula. Heads of schools at Australian universities, colleges and institutions offering a nursing or nursing and midwifery course in Australia were contacted via email to inform them of the study and request the contact details of the program coordinator or other suitable university representative. These program/subject coordinators or other appropriate representatives were subsequently invited to complete a short survey. The survey contained 10 questions and was expected to take 20 minutes to complete.

The curriculum survey requested information about: (i) the type or category of genomic information included in the curriculum, (ii) the subject/s in which the genomic information was provided and (iii) the amount of time allocated to teaching genomics. The content requested was based on the four categories outlined in the Genomic Nursing Concept Inventory (GNCI[®]): human genome basics, mutations, inheritance patterns and healthcare applications. For each category, respondents were asked if their university's curricula included information on that topic; in which subject the topic was included; and, for each subject listed, the amount of time spent teaching that topic. An example question is: 'Does the nursing and midwifery curriculum at your university include information on "human genome basics"?' The respondents were given the option to select 'yes', 'no' or 'unsure'. Examples of each topic were provided. Examples of information related to 'human genome basics' include genome composition/organisation, homo/heterozygosity, gene function and expression, genotype–phenotype association and genome homogeneity.

Responses were received from 16 university representatives; however, because of a functional error in the survey, multiple responses were received from some university representatives, meaning that only 13 universities were represented in the survey. The survey findings are presented in Table 1.4. The findings indicated that all topics—human genome basics, mutations and inheritance patterns—were addressed. Minimal data on timing were collected; however, these data indicated that limited time was devoted to teaching genomic content.

Table 1.4: Summary of Findings for Curriculum Survey

Question	Yes (n)	No (n)	Unsure (n)	Total (n)	Time Range
Does the nursing and midwifery curriculum at your university include information on the topic ‘human genome basics’?	9 (60.00%)	2 (13.33%)	4 (26.67%)	15	30 mins to 4 hrs
Does the nursing and midwifery curriculum at your university include information on the topic ‘mutations’?	6 (42.86%)	4 (28.57%)	4 (28.57%)	14	30 mins
Does the nursing and midwifery curriculum at your university include information on the topic ‘inheritance patterns’?	8 (61.54%)	3 (23.08%)	2 (15.38%)	13	15 mins to 2 hrs
Does the nursing and midwifery curriculum at your university include information on the topic ‘genomic healthcare applications’?	6 (46.15%)	4 (30.77%)	3 (23.08%)	13	30 mins to 4 hrs

The findings of the desktop analysis and curriculum survey combined to form the broader curriculum audit. The findings for the curriculum audit reflect the current international literature, which states that genomics is still not adequately integrated into nursing education.

1.6 Study Details

1.6.1 Significance

Genomics has the potential to transform healthcare delivery by increasing quality and safety, decreasing costs and improving health outcomes (Alexander, 2017; Calzone, Jenkins, et al., 2018; McCormick & Calzone, 2016). However, to reach this potential, nurses must engage with genomics. This study sought to determine how Australian nurses engage with genomics, and produce findings that can be used to improve the delivery of genomically informed nursing care, and direct genomic education for nurses at the pre- and post-registration levels. Given that Australian literature in this area is limited, this study contributes greatly to the existing research, especially in the Australian context.

1.6.2 Research Question

This study’s research question asked:

How are nurses engaging with genomics in nursing practice in Australia?

1.6.3 Research Aim

The aim of this research study was to determine how nurses engage with genomics in nursing practice in Australia.

1.6.4 Research Objective

The objective of the study was to document instances of nurses' engagement with genomics (genomic knowledge, skills and/or technologies) in their nursing practice in Australia. The researcher sought to gain a deeper understanding of:

1. the genomic knowledge and skills employed by nurses in nursing practice
2. nurses' perceptions of the role of genomics in nursing practice
3. nurses' experience of using genomics in daily nursing practice in terms of patient care and/or communication within the healthcare team
4. the barriers and enablers to nurses applying genomics in nursing practice.

1.6.5 Research Design

There were an extensive number of research designs available to the researcher. Research design selection depends on the researcher's philosophical assumptions about the nature of reality (ontology), how the researcher knows what is known (epistemology), the inclusion of the researcher's values (axiology), the nature in which the research emerges (methodology) and the researcher's writing structure (rhetorical) (Creswell, Hanson, Clarke, & Morales, 2007). This research employed case study research. Originally viewed as a 'soft' form of research (Yin, 2014), case study research has now been adopted in several disciplines and is becoming increasingly popular in nursing and midwifery research. This study employed a single holistic case study, as outlined by Yin (2014), to explore genomics in nursing practice. Critical realism provided the philosophical framework for the study.

Case study research is not assigned to a fixed ontological, epistemological or methodological position. This flexibility lends case study research a degree of 'philosophical versatility' (Harrison, Birks, Franklin, & Mills, 2017) that can accommodate critical realism as an underlying philosophy. Critical realism combines a realist ontology with a relativist epistemology in subscribing to a form of 'robust relativism' (McEvoy & Richards, 2003, p. 411). Therefore, this critical realist case study allowed the researcher to explore the

contemporary phenomenon of genomics in nursing practice within the real-world context of the Australian healthcare system using a ‘critical realist’ lens.

1.7 Thesis Structure

This thesis is presented in the standard doctoral format. The thesis contains eight chapters, as follows. ‘Chapter 1: Introduction’ provides an introduction to the thesis. ‘Chapter 2: Literature Review’ provides a more extensive background to the study and presents a publication that summarises the literature and provides context for the current study. ‘Chapter 3: Critical Realism’ outlines critical realism as the philosophical lens through which this research was viewed and conducted. ‘Chapter 4: Research Design’ presents the combined case study methodology and methods. ‘Chapter 5: Findings—Genomic Literacy of Registered Nurses and Midwives in Australia’ and ‘Chapter 6: Findings—Genomics in Oncology Nursing Practice’ present the publications associated with the individual studies. ‘Chapter 7: Discussion’ outlines the full ‘case’ findings and provides a description of the case of Australian nurses engaging with genomics. ‘Chapter 8: Conclusion’ concludes the thesis by discussing the contribution of this work to existing knowledge, the implications of the findings, the limitations of the research and recommendations for further study.

1.8 Summary

This chapter has provided an introduction to this study by explaining the theory, aims and objectives of the study, and providing the context by describing the impetus and significance of this work. The study design and underlying philosophical framework were stated, and will be discussed at length later in this thesis. The following chapter will provide a detailed context for the research and reinforce the importance of genomics in nursing practice, while discussing nurses’ current genomic literacy and competency.

Chapter 2: Literature Review

2.1 Introduction

In recent years, there has been an increase in literature addressing nurses' preparedness to deliver genomic healthcare. Chapter 1 provided an outline of genomics as it relates to healthcare. This chapter will provide a detailed description of genomic literacy and competency, including the relationship between the terms 'genomic literacy' and 'genomic competency'. It will outline the genomic literacy and competency of nurses internationally, as described in the literature. The chapter will also present exemplar genomic competency documents intended to guide nurses in applying genomics in practice. This chapter also contains a published article addressing nurses' competence in genomics. Previous literature addressing genomic literacy and competency has concluded that nurses are not demonstrating the competencies required to provide comprehensive genomic healthcare. This literature review provides an update on the existing literature and serves to further justify similar research in the Australian context.

2.2 Terminology: Genomic Literacy and Genomic Competency

To provide genomic healthcare, nurses must first 'learn the language of genetics' (Cashion, 2009, p. 535). This language can be thought of as 'genetic/genomic literacy'. Genomic literacy for nurses is appropriately defined as 'knowledge sufficient to develop genetic and genomic competency', as outlined in the *Essentials of Genetic and Genomic Nursing* competency document (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). The terms 'genomic literacy' and 'genomic competency' are frequently used interchangeably; however, they are distinct terms. In general, literacy is more closely aligned with knowledge, while competency infers the ability to apply that knowledge (Ward, 2011). Genomic literacy is necessary, yet insufficient for genomic competence (Ward, 2011). Thus, for nurses, genomic literacy requires knowledge sufficient to complete the activities that make up those competencies. The delivery of genomic healthcare does not require nurses to have detailed knowledge of genetic mechanisms. However, it does require an understanding of genetic and genomic terminology, and a solid grasp of the underlying concepts of genome science (Ward, 2011).

2.3 Genomic Literacy and Competency Documents in Nursing and Midwifery Practice

In accordance with the increasing importance of genomics in healthcare, competency documents have been developed by lead researchers in the United States (US) and United Kingdom (UK). In the US in 2006, a consensus panel on genetics in nursing published the *Essentials of Genetics and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators*, with a second edition published in 2009 (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). In 2011, these genetic and genomic competencies were reviewed and tailored to graduate nurses, and published as *Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees* (Greco, Tinley, & Seibert, 2011). Similarly, in the UK in 2003, the Genomics Policy Unit at the University of Glamorgan and the Medical Genetics Service for Wales at the University Hospital of Wales prepared *Fit for Practice in the Genetics Era: A Competence-based Education Framework for Nurses, Midwives and Health Visitors*, which proposed seven competency statements (University of Glamorgan and University Hospital of Wales, 2003). In 2010, the *Fit for Practice in the Genetics Era* framework was reviewed, and an eighth competency statement was added. Earlier in 2001, the National Coalition for Health Professional Education in Genetics (2007) published the *Core Competencies in Genetics for Health Professionals*, with a second and third edition published in 2005 and 2007, respectively. In 2008, the European Society of Human Genetics published the *Core Competences in Genetics for Health Professionals in Europe* (European Society of Human Genetics, 2008), which recommended core competences for generalist health professionals or those specialising in a field other than genetics, as well as core competences for specialist genetics health professionals. These competency documents are designed to guide nurses, midwives and other healthcare professionals in the application of their professional skills and responsibilities.

2.4 Genomic Literacy and Competency of Nurses Internationally

Several studies have been conducted internationally to assess nurses' and midwives' genomic knowledge and competence. Most studies have been conducted in the US. This is consistent with the findings of a bibliometric review by Anderson and Monsen (2014), which indicated that almost three-quarters of the literature about this topic were published in the US. Other studies have been conducted in the UK, Turkey, Italy, Jordan, Japan and Canada. Systematic

reviews have appeared in recent years to summarise the nursing and midwifery literature related to genetic literacy and competency.

Two literature reviews were published in 2012, each addressing the genomic literacy and competency of nurses. Godino and Skirton (2012) published a review article titled ‘A Systematic Review of Nurses’ Knowledge of Genetics’ in the *Journal of Nursing Education and Practice*. The aims and objectives of this review were to examine the available evidence on nurses’ genetics knowledge. The search retrieved 137 papers, with six eligible for inclusion. The findings indicated that both perceived and actual knowledge of genetics was poor, and that the amount of genetics education delivered to nurses in these studies was low overall. The same year, Skirton, O’Connor, and Humphreys (2012) published a review article titled ‘Nurses’ Competence in Genetics: A Mixed Method Systematic Review’ in the *Journal of Advanced Nursing*. The aim of the systematic review was to ascertain the extent to which nurses are achieving the core competences in genetics appropriate for nursing practice. The search retrieved 269 papers, with 13 eligible for inclusion. The findings indicated that there is limited evidence on this topic; however, the available evidence suggests that nurses are not demonstrating the competences needed to offer holistic healthcare to people with genetic conditions.

An earlier review article, ‘Genetic Competence of Midwives in the UK and Japan’ was published by Skirton, Murakami, Tsujino, Kutsunugi, and Turale (2010). The review was undertaken to determine the extent to which midwives were achieving the genetic competences prescribed for their practice. Unlike the aforementioned reviews, this review had stricter inclusion criteria. Given that the review concerned midwifery practice in Japan and the UK, the authors elected to include only studies with data collected from those countries. The search retrieved 111 papers, with eight eligible for inclusion. The findings indicated that midwives were not achieving the competences and were not confident in their genetics knowledge, nor were women being supported to make informed decisions regarding antenatal screening.

These literature reviews produced largely similar findings—that genetic literacy and competency is limited, and nurses and midwives are not confident in using genetics in practice. There is a general call for more work and research regarding competency achievement in practice, as well as changes to nursing and midwifery curricula and further continuing education to ensure that nurses and midwives are able to provide competent genetic care. Many years have passed since these systematic reviews were published, and it is unclear whether

there has been an increase in nurses’ genetic literacy and competency, or improved attitudes towards genomics.

2.5 Literature Review

Chapter no.	Details of publication on which the chapter is based	Nature and extent of the intellectual input of each author, including the candidate
2	Wright, H., Zhao, L., Birks, M., & Mills, J. (2018). Nurses’ competence in genetics: An integrative review. <i>Nursing & Health Sciences</i> , 20(2), 142-153. doi:10.1111/nhs.12401	Wright conducted the literature review. Wright and Zhao individually assessed the papers using the STROBE or CASP-Qualitative Research Checklist. Wright wrote the first draft of the manuscript which was revised with editorial input from Zhao, Birks and Mills.

The author conducted an integrative literature review to see what progress, if any, has been made towards achieving the core genomic competencies appropriate for clinical practice. The author replicated (where possible) the review methodology used by Skirton et al. (2012). The findings of the integrative review can be grouped into three themes: (i) genomic knowledge and use—nurses have poor genomic knowledge and competency; (ii) perceived relevance to practice—most nurses believe genomics is important to their practice; and (iii) genomic education—genomics is not adequately addressed in nursing curricula. Overall, nurses were shown to have poor genomic knowledge and/or competency, yet there was consensus that most nurses believe genomics is important to their practice. The review indicated that, in the past five years, nurses and midwives have made minimal progress towards achieving the core genomic competencies appropriate for clinical practice. This integrative review, together with other international reviews, indicates limited engagement with genomics.

2.6 Theoretical Statement

A theoretical statement provides a ‘blueprint’ for the study (Yin, 2014, p. 38). A theoretical statement addresses the research questions, propositions, units of analysis, logic connecting data to propositions, and criteria for interpreting the findings. This research study was centred on the theory that Australian nurses do not adequately engage with genomics because education, policy and practice do not support engagement. This theory was used as the starting point for the study, and will be revisited at the conclusion of the thesis to determine if and to what extent it proves correct.

REVIEW ARTICLE

Nurses' competence in genetics: An integrative review

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Abstract

The aim of this integrative review was to update a mixed method systematic review by Skirton, O'Connor, and Humphreys (2012) that reported on nurses' levels of competence in using genetics in clinical practice. Three electronic databases were searched using selected key words. Research studies published in English between January 2011 and September 2017 reporting levels of nurse competence in genetics or genomics were eligible for inclusion. The selected studies were subjected to thematic analysis. Three main themes were identified: (i) genomic knowledge and utilization, (ii) perceived relevance to practice, and (iii) genomic education. While the reviewed papers produced varied findings, many nurses were shown to have poor genomic knowledge and/or competency, and yet there was a consensus that most nurses believe genomics is important to their practice. The present review indicated that in the past 5 years nurses have made minimal progress toward achieving the core genomic competencies appropriate for clinical practice.

KEYWORDS

Competency, family history, genetics, genomics, nursing, pedigree

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2.7 Summary

This chapter has provided context for this study by outlining genomic literacy and competency, and presenting a current literature review of genomic competency in nursing practice. The integrative literature review indicated that, in the past five years, nurses have made minimal progress towards achieving the core genomic competencies appropriate for clinical practice. It is unclear whether Australian nurses have similar genomic literacy and competency levels to their international counterparts, which warrants further investigation. To investigate the full picture of Australian nurses' engagement with genomics, the author conducted a case study using a critical realist framework. Critical realism and the framework it provides for the study will be discussed in the following chapter. Critical realism in relation to case study research will be further addressed in Chapter 4.

Chapter 3: Critical Realism

3.1 Introduction

Critical realism provided the theoretical framework for this study. Philosophers of critical realism are concerned with the nature and knowability of the social world and social phenomena (Schiller, 2016), and it is because of this intent that it was considered suitable to use in an exploration of Australian nurses' engagement with genomics. In this chapter, critical realism will be situated in relation to social science, and a justification of the use of critical realism in this study will be provided. A brief overview of philosophy is presented, including empiricism, positivism and postpositivism (constructivism and interpretivism), before a discussion of the development of Roy Bhaskar's philosophy of critical realism. The fundamentals of critical realism are outlined, including the definition, central tenets, scope and methodology, and usefulness to nursing and nursing research.

3.2 Philosophy in Qualitative Research

3.2.1 Philosophy and Philosophical Assumptions

Philosophy is derived from the Greek words '*philo*' (love) and '*sophia*' (knowledge), and literally means 'the love of knowledge' (Brinkmann, 2018, p. 1). Philosophy is concerned with human beings' fundamental questions—questions that are theoretical (e.g., what is the nature of reality?) and practical (e.g., how should we act?) (Brinkmann, 2018, p. 8). Birks (2014) defined philosophy as 'a view of the world encompassing the questions and mechanisms for finding answers that inform that view' (p. 18). Creswell and Poth (2018) provided a similar description of philosophy as 'the use of abstract ideas and beliefs that inform our research' (p. 16).

According to Creswell and Poth (2018, p. 14), there are four philosophical assumptions to be explored by researchers: their beliefs about ontology, epistemology, axiology and methodology. Ontology is concerned with the nature of reality and existence, while epistemology considers the nature of knowledge and what can be known. Axiology focuses on values, while methodology considers the research approach, with methods describing the practical means by which data are collected and analysed (O'Reilly & Kiyimba, 2015). Ontology and epistemology are perhaps the most philosophically valuable. The term 'ontology'

derives from the Greek terms '*logos*' (study) and '*ontos*' (being) (Brinkmann, 2018, p. 9), making ontology the study of being (Birks, 2014, p. 21). Ontology is concerned with the nature of reality and its characteristics (Creswell & Poth, 2018; Ormston, Spencer, Barnard, & Snape, 2014) and essentially determines whether we believe reality exists separate from human practices (Braun & Clarke, 2013). Epistemology is concerned with knowledge and the means by which we gain knowledge of this reality (Petty, Thomson, & Stew, 2012), or 'how knowledge is known' (Creswell & Poth, 2018, p. 21). Ontology and epistemology 'each demarcates what can and cannot count as meaningful knowledge and informs our methodology and the process of producing knowledge' (Braun & Clarke, 2013, p. 26).

3.2.2 Positivism, Postpositivism and the 'Realist Turn'

Paradigms are a 'basic set of beliefs that guide action' (Guba, 1990, p. 17) and 'frameworks that represent a shared way of thinking in respect of how we view the world and we generate knowledge from the perspective' (Birks, 2014, p. 18). Therefore, a paradigm refers to the way researchers position themselves when conducting research based on a specific philosophical, ontological and epistemological perspective (Nagy, Mills, Waters, & Birks, 2010). The major research paradigms that influence qualitative research are positivism, postpositivism, postmodernism, critical theory, constructivism and the participatory paradigm (Lincoln, Lynham, & Guba, 2011).

For early researchers, knowledge was considered empirical, in that experience is the foundation of knowledge (Paley, 2008a). In a practical sense, empirical knowledge comes from observation (Cruickshank, 2012) and, since empirical knowledge is able to be observed by others, it stands that anything that cannot be observed, directly or indirectly through instruments, ultimately cannot exist (Mingers, 2006). Positivism is closely aligned with empiricism (Braun & Clarke, 2013). The nature of empiricist ideas embedded in positivist thought means that positivism can be considered a variant of empiricism (Paley, 2008b). Early examples of positivist thought in research can be seen in the works of René Descartes (1596 – 1650) and his focus on objectivity and evidence in the search for truth; David Hume (1711–1776) as the founder of empiricism; and Auguste Comte (1798–1857), who is considered the founder of positivism (Ormston et al., 2014). Positivism 'asserts the existence of a single reality that is there to be discovered' (Birks, 2014, p. 20)—simply stated, 'the things we experience are things that exist' (Wainwright, 1997, p. 1263). Therefore, a positivist stance is that knowledge is produced through the senses, based on observation of reality (a single reality)

that can be known accurately (Ormston et al., 2014). Over time, positivism has attracted criticism—the most common being that positivism excludes various other sources of understanding of the world (such as human experiences, reasoning or interpretation) and that it addresses the nature of a social world devoid of context (Fox, 2008).

As a result of this criticism, positivism has been largely overlooked by researchers in favour of postpositivism. Postpositivism ‘rejects the concept of a measurable reality that exists in isolation of the observer’ (Birks, 2014, p. 20). Instead, postpositivism states that knowledge of the world is produced through testing propositions, where hypotheses about causal relationships are derived from scientific theories and then evaluated against observations, where reality can be known, yet only approximately (Ormston et al., 2014). The postpositivist turn was initiated by philosophers such as Immanuel Kant (1724–1804), who argued that there are ways of knowing the world other than through direct observation (Ormston et al., 2014). Kant believed that perception relates to a human interpretation of what the senses tell us, resulting in knowledge—a position that led to *interpretivism*. Interpretivists acknowledge interpretation as well as observation as they seek to understand the social world (Ormston et al., 2014). The related movement of *constructionism* posits that knowledge is actively ‘constructed’ by human beings, rather than being passively received by them (Ormston et al., 2014). Constructionism questions the idea that knowledge is an objective reflection of reality, and instead posits that our ways of knowing the world are linked to the social world in which we live (Braun & Clarke, 2013). The terms ‘interpretivism’ and ‘constructivism’ are frequently used synonymously in paradigmatic discussions. Table 3.1 outlines the three key research paradigms described above and their differences with regard to ontology, epistemology and methodology.

Table 3.1: Research Paradigms in Respect of Ontology, Epistemology and Methodology

Issue	Positivism	Postpositivism	Constructivism
Ontology	Naïve realism—‘real’ reality, but apprehensible	Critical realism—‘real’ reality, but only imperfectly and probabilistically apprehensible	Relativism—local and specific co-constructed realities
Epistemology	Dualist/objectivist Findings true	Modified dualist/objectivist Findings probably true	Transactional/subjectivist Co-created findings
Methodology	Experimental/manipulative Verification of hypothesis Chiefly quantitative methods	Modified experimental/ manipulative Falsification of hypotheses May include qualitative methods	Hermeneutical/dialectical

Source: adapted from Lincoln et al. (2011, pp. 97–128).

Realism offers an alternative position to the dominant positivist, postpositivist and constructivist paradigms. Bhaskar (2011) described realism as a theory in which the objects of scientific enquiry exist and act (for the most part) independently of scientists and their activity. Simply stated, the ‘features’ that form our world are not essentially visible (Wainwright, 1997). In this way, realism shifts the emphasis from epistemology to ontology (Wainwright, 1997, p. 1264), thereby rendering realism a theory of *being*, not of knowledge or truth (Bhaskar, 2011, p. 13). Realism provided a new set of perspectives on society (and nature) and how to understand them thereby making it ‘a philosophy of and for the whole of the natural and social science’ (Sayer, 1992, p. xi). It is from realism that critical realism developed, for those who decided to follow the ‘realist turn’ away from positivism and constructivism (Gorski, 2013, p. 659) and focus on a more integrated and holistic view of the world. In the philosophy of science, tradition has depended on an implicit ontology of empirical realism; however, it became Bhaskar’s (2011) intent to show that ‘only a realism fully consistent with the principle (or definition) of realism—*transcendental realism*—can sustain the intelligibility of the experimental and theoretic work of science’ (p. 13).

3.3 Roy Bhaskar and the Development of Critical Realism

The positivist/constructivist dichotomy resulted in the paradigm wars of the 1980s (Denzin & Lincoln, 2011). Postpositivists, constructivists and critical theorists retaliated against the dominant positivist research culture of the time (Mills & Birks, 2014). To overcome the impasse presented by the dichotomy of traditional philosophy, and to encompass developments in modern thought concerning the nature of the social world, philosophers and social theorists—such as Roy Bhaskar, Margaret Archer, Mervyn Hartwig, Tony Lawson, Alan

Norrie and Andrew Sayer—began to develop the philosophy of critical realism (Walker, 2017). However, British philosopher Roy Bhaskar (1944–2014) is credited with the creation of critical realism, and rendered it ‘a coherent philosophical language’ (Danermark, 2002, p. 4).

There are two key movements in the development of critical realism: *transcendental realism* and *critical naturalism*. Bhaskar published three seminal books that together became the basis for basic (or original) critical realism: *A Realist Theory of Science* (1975), *The Possibility of Naturalism* (1979) and *Scientific Realism and Human Emancipation* (1986). The first two publications, *A Realist Theory of Science* (1975) and *The Possibility of Naturalism* (1979), align with the two key movements in critical realism: transcendental realism and critical naturalism, respectively. Bhaskar’s transcendental realism was the first movement in critical realism. Transcendental realism regards the objects of knowledge as the structures and mechanisms that generate phenomena (Bhaskar, 1998b, p. 19). Bhaskar (1998b) stated that ‘these objects are neither phenomena (empiricism) nor human constructs imposed upon the phenomena (idealism) but real structures which endure and operate independently of our knowledge, our experience and the conditions which allow us to access them’ (p. 19). Bhaskar published his first book, *A Realist Theory of Science*, in 1975. In *A Realist Theory of Science*, Bhaskar (1975/2008) sought to develop a ‘systematic realist account of science’ (p. 8) that he believed would provide a comprehensive alternative to the positivism. Originally, two strands of criticism were directed towards the positivist view of science—first by writers such as Thomas Kuhn and Karl Popper, who emphasised the social character of science, and second by writers such as Mary Hesse and Rom Harre, who focused on the stratification of science (Bhaskar, 1975/2008). Bhaskar’s (1975/2008) *A Realist Theory of Science* presents:

novel and stunning resolutions of problems generated by classical empiricism and rationalism, and the newer philosophy of Science, problems such as that of induction and that of reconciling the relativity of scientific knowledge as a social process with realism about its objects. (p. ix)

Critical naturalism was the second movement in critical realism. Critical naturalism acknowledges the significant difference between natural and social structures and the forms of their appropriate science (Bhaskar, 1979/1998). Bhaskar published his second book, *The Possibility of Naturalism*, in 1979, presenting critical naturalism. Bhaskar’s third book, *Scientific Realism and Human Emancipation*, published in 1986, encouraged the combination of ethical naturalism and ideology-critique, in what became known as the theory of explanatory critique (Bhaskar 1986/2009)—a theory implicit in *Possibility of Naturalism* (Bhaskar, 1998a).

The term ‘critical realism’ is an amalgam of the phrases ‘transcendental realism’ and ‘critical naturalism’ (Bhaskar, 2016, p. 10), with Bhaskar (1978) combining them to describe the interface between the natural and social worlds. Bhaskar (2016) stated that critical realism’s ‘credentials as a *realism* were obvious’ (p. 10). The *critical* in critical realism is less clear. The term ‘critical’ reflects Kant’s use of the word as a synonym for ‘transcendental’ (Bhaskar, 2016, p. 10). Using *critical* rather than *transcendental* infers that the philosophy is critical—not just of other philosophies, but of scientific practices, common beliefs and the structures or circumstances underlying them (Bhaskar, 2011, p. 190). Bhaskar’s work extends beyond ‘basic’ or ‘original critical realism’ (transcendental realism or critical naturalism) to other forms, such as ‘dialectical critical realism’ and ‘transcendental dialectical critical realism’ (or ‘meta-Reality’) (Gorski, 2013). This research study is underscored by Bhaskar’s *original* critical realism—referred to simply as ‘critical realism’. Thus, this will be discussed at length in the following section.

3.4 Defining Critical Realism

Defining critical realism is challenging. Critical realists draw on many authors meaning there is no one unitary framework, set of beliefs, methodology or dogma that unites critical realists as a whole (Archer et al., 2016). Broadly, critical realism seeks to ‘investigate and identify relationships and non-relationships, respectively, between what we experience, what actually happens, and the underlying mechanisms that produce the events in the world’ (Danermark, 2002, p. 21). For the purposes of this study, the operational definition of critical realism is as follows:

Critical realism states that an (objective) world exists independently of people’s perceptions, language or imagination; and that part of that world consists of subjective interpretations which influence the ways in which it is perceived and experienced. (O’Mahoney & Vincent, 2014, p. 2)

In this definition, critical realism answers the fundamental question as to whether a world exists independent of human consciousness, by stating that, yes, there exists both an external world independent of human consciousness, and a dimension that includes our socially determined knowledge about reality (Danermark, 2002). The goal of a research study underpinned by critical realism is to synthesise, from the available ideas and relevant data, an account of what is occurring in key social mechanisms and processes (Ackroyd & Karlsson, 2014). Realism (reality is independent of human ways of knowing about it) and relativism (reality is dependent

on human interpretation) are at opposing ends of the ontological continuum (Braun & Clarke, 2013). However, critical realism best represents the relationship between ontology and epistemology in that it combines a realist ontology with a relativist epistemology by subscribing to a form of “‘robust’ relativism’ (McEvoy & Richards, 2003, p. 411).

3.5 Key Tenets of Critical Realism

Critical realism has three key tenets: (i) the primacy of ontology, (ii) the stratified character of the real-world (reality) and the search for generative mechanisms and (iii) the interplay between social structures and human agency (Bhaskar, 1975/2008, 1979/1998, 2011).

3.5.1 The Primacy of Ontology

Critical realism is primarily concerned with ontology. Ontology refers to ‘what is’ or ‘what exists’ (Schiller, 2016). Bhaskar (1978) argued that the fundamental question in the philosophy of science is ‘what properties do societies and people possess that might make them possible objects for knowledge?’, and it is this ontological question—not the epistemological question of how knowledge is possible—that must serve as the starting point for a philosophy of reality (Danermark, 2002). As stated earlier, Bhaskar (1998b, p. 27) argued that positivism is an ‘epistemic fallacy’—that is, the reduction of ontology to epistemology, or the limitation of ‘reality’ to what can be empirically known (e.g., through scientific experiments) (Fletcher, 2017). The same critique applies to constructivism, where researchers view reality as entirely constructed through and within human knowledge or discourse. Despite the seeming opposition between the constructivist and positivist perspectives, each reduces reality to human knowledge, whether that knowledge acts as a lens or container for reality. Critical realism advocates for the primacy of ontology (Joseph, 2014) and enquiry into the nature of things. Ontological realism asserts that much of reality exists and operates independently of our awareness or knowledge of it. Thus, our human perceptions of the world (epistemology) cannot be synonymous with the world’s objective state (ontology) (Bhaskar, 1998b).

3.5.2 Reality is Stratified

Bhaskar (1998b) interpreted reality as existing at three different layers of knowledge or ontological ‘domains’—the *empirical*, *actual* and *real*—essentially providing an ‘ontological map’. Bhaskar argued that underlying structures, powers and processes must act together under certain circumstances to influence observable events, and that these underlying phenomena are

as real as the observable effects and outcomes they cause. Experiences, events and mechanisms constitute three overlapping domains or reality, present in the *real*, *actual* and *empirical* domains, as summarised in Table 3.2 (Bhaskar, 1998b, p. 41). According to Bhaskar (1998b), events must occur independently of the experiences in which they are apprehended, just as structures and mechanisms are distinct from the experiences in which they are apprehended. If ontology is based on experience—as in the empirical world—the three domains of reality are collapsed into one (Bhaskar, 1998b)

Table 3.2: Real, Actual and Empirical Ontological Domains

Domain	Description	Experiences	Events	Mechanisms
Empirical	Fallible human perceptions and experiences, including science	✓		
Actual	Events and actions that are more likely to be observed	✓	✓	
Real	Underlying powers, tendencies and structures that cause events in the actual domain	✓	✓	✓

Source: adapted from Bhaskar (1998b, p.41).

The empirical domain refers to what we experience (Danermark, 2002) and comprises only human perceptions and experiences (Clark, Lissel, & Davis, 2008). Humans come to know empirical information through direct and indirect experiences, which in turn are a result of the interaction of generative mechanisms in the real domain (Schiller, 2016). For critical realists, human perceptions and speculations in the empirical domain are considered fallible representations of the real domain (Clark et al., 2008; Schiller, 2016).

The actual domain is the level at which events occur, whether we experience them or not—essentially stating that what happens is not the same as what is observed (Danermark, 2002). In the actual domain, humans are able to actually experience a portion of those events that have been caused by the complex interaction of the generative mechanisms (Clark et al., 2008). Mechanisms sometimes generate an event, and, when they are experienced, they become an empirical fact. However, an event is considered to have occurred whether or not it is experienced or perceived by a human being (Schiller, 2016). Essentially, what occurs in the world is not equivalent to only that which is observed by humans (Schiller, 2016).

The real domain contains both the structures (objects) and the mechanisms that generate phenomena (McEvoy & Richards, 2003). The real domain is independent of the thought, awareness and even existence of human beings (Schiller, 2016). This is the level beneath the

level of events (actual) and the level of our empirical world (empiricism)—a deeper dimension where those mechanisms make the events occur. As Fletcher (2017) stated, ‘human knowledge captures only a small part of a deeper and vaster reality’ (p. 182). This is the ‘point of departure’ for the natural sciences (Danermark, 2002, p. 53).

Critical realism provides this stratified ontology to distinguish between the different layers of knowledge and to understand reality as it exists in the actual and real domains (Danermark, 2002). Bhaskar (1978) believed the empirical world allows for the ‘epistemic fallacy’ by reducing the three ontological domains to one—that is, it reduces what ‘is’ to what we can ‘know’. Bhaskar (1998) used the term ‘generative mechanism’ to refer to the causal powers or tendencies of ways of acting of structured things. Causality is defined as ‘the power to bring about change’ (Hartwig, 2007, p. 57). Causal analysis seeks to explain why what occurs actually does occur (Danermark, 2002). These causes can be natural or social, and can be activated to produce particular outcomes that may, or may not, be consciously experienced or known by human beings (Schiller, 2016; Walsh & Evans, 2014). In critical realism, causation itself is viewed as being generative. Understanding the powers of objects and the conditions that generate mechanisms to operate and produce events is key in critical realism (Bhaskar, 2011).

Critical realists acknowledge that generative mechanisms are not directly observable, yet maintain that they are real and identifiable through their effects (McEvoy & Richards, 2003, p. 412). In this way, unobservable structures are real on the grounds that their effects can be experienced or observed (Walsh & Evans, 2014). Generative mechanisms can be understood through, and exist within, phenomena at the empirical level (domain), and contribute to our understanding of the actual domain (Fletcher, 2017). Similarly, it is possible to partially discern and understand the real domain, with the opposite being true that it may *not* be possible to access these objects and mechanisms nor observe every aspect of them (Schiller, 2016). Regardless, these real structures and mechanisms generate phenomena (McEvoy & Richards, 2003; Schiller, 2016). In Bhaskar’s view, the objective of science is to produce knowledge about the generative mechanisms and structures that combine to produce phenomena (Schiller, 2016), and it is these phenomena that are available for scientific investigation (Fletcher, 2017).

For social scientists, phenomena occur in ‘open systems’, rather than the artificially controlled ‘closed systems’ provided by laboratory experiments. Critical realism treats reality as an open system where multiple mechanisms operate simultaneously, and the everyday events that

humans observe may or may not be activated under certain conditions (Souza, 2014). Events in this open system are ‘a product of many factors coming together in certain combinations and given the right circumstances or context to causally generate new events’ (Clark et al., 2008). Only a fully ‘closed system’ could yield universally valid patterns of interplay between the causal events associated with real objects and mechanisms, which would generate truly ‘law-like regularities’ in the actual world (Bhaskar, 1998a). In contrast, most social settings are far from being closed systems; rather, they are highly complex, and the actualisation of generative mechanisms is subsequently dependent on the variable conditions presented by the social setting (Bhaskar, 1998b; McEvoy & Richards, 2003).

This concept of a stratified ontology differentiates the critical realist paradigm from ontologies of other philosophical frameworks. Many other paradigms only engage with, and seek to recognise, the actual or empirical domains of the world, and fail to consider an independent reality (Schiller, 2016). A study underpinned by critical realism results in an understanding that scientific claims are attempts to clarify the various circumstances or contexts under which a particular event is likely to occur, or a particular explanation is likely to be valid (Bhaskar, 1998a, 1998b).

3.5.3 Interplay between Structure and Agency

There are two approaches to the analyses of social phenomena in the social sciences: structural and agential approaches. *Structural* approaches emphasise the social worlds and organisations within which individuals are embedded. Alternatively, *agential* approaches place greater emphasis on the way that human agents respond to their surroundings, based on the meanings they give to things or events (McEvoy and Richards, 2003). Thus, unlike natural structures, social structures do not exist independently of the activities they govern, nor are they independent of agents’ conception of their activities (Joseph, 2014). Social structures cannot be reduced to individuals; however, they are a prerequisite for human action; thus, in this way, they enable action, yet at the same time set limits on what actions are possible (Danermark, 2002). Therefore, the question is ‘how much freedom actors possess and to what degree society constrains their behaviour’ (Houston, 2014, p.2). This represents an enduring debate in the social sciences about the relative importance of individual (‘agency’) factors (such as beliefs, attitudes and personal meanings) and contextual (‘structural’) factors (such as social norms, culture, geography and environment) (Clark et al., 2008). Critical realists attempt to ‘bridge the gap’ by emphasising the interdependence of structure and agency. They acknowledge that

social structures provide the resources necessary for individuals to act, and place limits on individual behaviour. However, human agents are also able to transform social structures by responding to their circumstances (Connelly, 2000; McEvoy & Richards, 2003). As Bhaskar states ‘actors shape their social worlds but, in turn, are constrained by social structures embedded in the fabric of social life’ (Houston, 2014, p. 2).

3.6 Criticisms of Critical Realism

Positivists and interpretivists have each presented their critiques of this paradigm of thought. Positivists argue that critical realists risk bias because the application of values in any given situation is a judgement call (Hammersley, 2009). Critical realists respond by saying that these values underpin all research endeavours, whether acknowledged or not; thus, an informed judgement call is warranted. Interpretivists are suspicious about the existence of a layered ontology, since any reality is provisional and contestable, and our knowledge of it is partial and subjective. Critical realists respond by saying that the deeper layers of ontology are real because their effects are real, and it is the responsibility of researchers to seek them out.

Another criticism of critical realism is that what denotes the ‘critical’ in *critical realism* is not always clear. Although Bhaskar detailed his progression through the evolving conception of ‘critical’ philosophy, there is no clear explanation of what ‘critical’ philosophy actually is, nor explanation of its place in critical realism. Little (2013) asked the question: what is ‘critical’ about critical realism? Little (2013) commented:

He [Bhaskar] is a careful and explicit philosopher in much of his writing; but on the subject of ‘critical’ method, he is surprisingly elliptical. And to me, this suggests that the import of Bhaskar’s system is more on the side of ‘realism’ than its ‘critical’ methodology.

As Hammersley (2009, p. 1) suggested, there is an expectation that all research be critical. However, the phrase ‘critical realism’ and the notion of ‘critical social science’ generally extend beyond this generic expectation. Bhaskar (2016, p. 10) argued that his use of the term ‘critical’ reflects Kant’s use of the word as a synonym for ‘transcendental’. Preference for the term critical, rather than transcendental, indicates that the philosophy is critical not just of other philosophies, but of scientific practices, common beliefs and the structures or circumstances underlying them (Bhaskar, 2011).

3.7 Bringing Critical Realism to Nursing Practice

Critical realism has been adopted by many disciplines (Williams, Rycroft-Malone, & Burton, 2017), yet nursing research studies have not used it widely to date, perhaps because the legacy of the paradigm wars—the ‘qualitative’ (constructivist) and ‘quantitative’ (positivist) dichotomy—remains an issue. Wainwright (1997, p. 1262) argued that adopting a dualistic approach ignores the option of realism, and that the philosophy of the human and social sciences, and therefore of nursing, is better viewed as a *triad* of paradigms: positivist, constructivist and realist. Critical realism presents an alternative approach to research and particularly nursing research because it has the potential to ‘frame, identify and understand those complex phenomena that comprise the social science world’ (Schiller 2016, p.88). Contemporary nursing practice is embedded within complex social situations. A realist methodology recognises the ‘complex nature of programs or interventions and focuses on explaining what is working under specific conditions or contexts’ (Williams et al., 2017, p. 2) and provides for a more inclusive picture of the reality of our world. Most importantly, critical realist studies can provide new insights into the complexity of nursing practice and healthcare, and the influence of different factors on this work (Williams et al., 2017).

Critical realism supports a wide range of research methodologies and methods as a means to explore and understand events and experiences (Schiller, 2016); however, as a philosophy, it is far from directive. Authors of treatises on critical realism provide limited guidance regarding which precise methods—including methods of data collection, coding and analysis—are best suited to applied critical realist research (Fletcher, 2017), which jeopardises the application of critical realism (Fletcher, 2017). This is a problem, as Bhaskar (2014) himself stated, because ‘if critical realism is to be “serious”, it must be applicable’ (p. v). A literature review by Fletcher (2017) indicated that critical realist literature usually falls into one of two categories—(i) high-level philosophy of science and theory or (ii) reports on empirical research meant to explain social problems or inform policy—with neither providing a detailed description of the methods used. This leaves aspiring critical realist qualitative researchers without methodological guidelines for the deployment of methods (Morse, Barrett, Mayan, Olson, & Spiers, 2002).

3.8 Why the Researcher Chose Critical Realism

An important consideration in any study is understanding a researcher's choice to use a particular paradigm to frame his or her research (Schiller, 2016). This personal perspective was captured by Lysaght (2011), who stated that:

A researcher's choice of framework is not arbitrary but reflects important personal beliefs and understandings about the nature of knowledge, how it exists (in the metaphysical sense) in relation to the observer, and the possible roles to be adopted, and tools to be employed consequently, by the researcher in his/her work. (p. 572)

The current researcher chose critical realism as the theoretical framework to underpin this research study because it appealed to her worldview. The researcher is not alone in this, with critical realism appealing to a wide audience, as it relates to how many of us think about the world (O'Mahoney & Vincent, 2014; Williams, Rycroft-Malone, & Burton, 2016). Critical realism states that an (objective) world exists independently of people's perceptions, language or imagination, and that part of that world consists of subjective interpretations that influence the ways it is perceived and experienced (O'Mahoney & Vincent, 2014, p. 2). The premise of a subjective view of an objective world appeals to the current researcher. Critical realism 'takes the middle ground' because it does not reduce the world to unknowable chaos or a positivistic universal order, nor does it place objective truth value on the perspectives of human beings or remove the influence and importance of human perspectives (Clark et al., 2008). Instead, as stated previously, critical realism combines a realist ontology with a relativist epistemology to create a form of "'robust" relativism' (McEvoy & Richards, 2003, p. 411). Critical realism is useful for unpacking and understanding complex social phenomena (Cruickshank, 2012; Schiller, 2016), such as the engagement of Australian nurses with genomics, and this potential led the researcher to choose critical realism for this study.

3.9 Summary

This chapter has reviewed the general approaches to social science, the path to realism and the subsequent development of Roy Bhaskar's critical realism. The fundamentals of critical realism were outlined, including the definitions, goals, key tenets and methodology. Moreover, critical realism's usefulness in nursing research was explored. This discussion of critical realist ontology (what exists) and epistemology (how we can come to know about it) will be followed

by methodology—the means of acquiring this knowledge—in the next chapter. The next chapter will introduce case study as the chosen methodology for this research study.

Chapter 4: Research Design

4.1 Introduction

Case study designs, which include particular combinations of philosophy, methodology and methods, allow for the comprehensive study of complex issues in context (Anthony & Jack, 2009), making them ideal for nursing and social science in general. This chapter will discuss the development of case study research, including the history of case study research, the contribution of key case study researchers and the different definitions that each of these researchers developed based on their particular philosophical and methodological orientations. The methods of case definition, data collection and analysis of datasets as applied in this study will be discussed. Strategies to ensure the quality of a case study will be outlined, with the application of these strategies in this study reserved for the conclusion chapter. The research design used in this research study was influenced by the work of several lead case study researchers; however, it drew mainly on the methodology and methods proposed by Robert Yin (2014).

4.2 Overview of Case Study Research

4.2.1 History

Early case studies were included in ethnographies of individuals and cultures conducted in the discipline of anthropology in the 1900s (Merriam, 2009; Simons, 2009; Stewart, 2014). Quantitative methods dominated case study designs from the 1940s to 1970s, with qualitative case studies considered unfashionable (Johansson, 2003). During these years, case study was restricted to being a method within a quantitative study, or a descriptive research study investigating a selected phenomenon (Harrison et al., 2017; Merriam, 2009). In the 1960s, there was renewed interest in qualitative methodologies (Anthony & Jack, 2009), and case study re-emerged as a means to study complex issues in context. By the 1980s, researchers were writing about case study as a methodology (Merriam, Tisdell, & Ebscohost, 2016). Robert Stake, Robert Yin and Sharan Merriam were the main protagonists, with their individual philosophical leanings framing how they thought about case study as a methodology. Stake (1995) favoured a relativist-constructivist/interpretivist approach, Yin (2014) favoured a realist-postpositivist approach, and Merriam (2009) favoured a more pragmatic/constructivist approach.

In recent times, nursing practice has become more complex and, as such, case study designs offer a useful way to explore and understand these complexities (Anthony & Jack, 2009; Harrison & Mills, 2016; Rosenberg & Yates, 2007). Case study enhances our understanding of the complex contextual, cultural and behavioural factors affecting practice (Atchan, Davis, & Foureur, 2016; Stake, 1995; Yin, 2014) and allows the holistic nature of nursing care to be addressed (Heale & Twycross, 2018; Sandelowski, 1996). However, nurses have not always embraced case study research. This reluctance may be due to criticisms of case study methodologies and the perceived shortcomings of qualitative research in general. Case study has been plagued by a lack of clarity, being labelled as a research design, research methodology, research method, research strategy, data collection method and teaching technique (Anthony & Jack, 2009). This lack of clarity may have dissuaded many nurses from using case study research because of a perceived lack of credibility and rigour.

4.2.2 What is Case Study?

In its simplest terms, a case study allows for the comprehensive study of complex issues in context (Anthony & Jack, 2009). The case study researcher has the opportunity to explore, describe or explain the case of interest, and develop a context-derived, in-depth, holistic knowledge and understanding about ‘real-life’ events (Luck, Jackson, & Usher, 2006). Using a case study, the researcher is able to ‘see something in its completeness’ (Thomas, 2011, p. 23) and, in this way, is able to ‘get close to reality’ (Flyvbjerg, 2006; Thomas, 2016). However, the varied definitions and descriptions of case study research have led to what one academic described as a ‘definitional morass’ (Gerring, 2007, p. 17). Confusion is escalated by the term ‘case study’ being used to refer to ‘both the unit of study (the “case”) and the product of this type of investigation’ (Anthony & Jack, 2009). The most commonly used definitions come from the works of Stake, Yin and Merriam. Their definitions and focus are compared in Table 4.1. These definitions share some similarities. There is a common denominator between the case study definitions in that each includes a ‘case’, which is the object of study, and that the ‘case’ should be a complex functioning unit, be contemporary and be investigated in its natural context using several methods of data collection and analysis (Johansson, 2003). The differences presented by the definitions relate to the focus of the case study—be it exploration, process or product.

Table 4.1: Comparison of Case Study Definitions and Emphasis

Author	Definition	Case Study Defined by...	Focus/Emphasis
Stake	'the study of the particularity and complexity of a single case, coming to understand its activity within important circumstances' (Stake, 1995, p. xi)	Object of the case	Emphasis on inductive exploration, discovery and holistic analysis that is presented in thick descriptions of the case
Yin	'An empirical inquiry that investigates a contemporary phenomenon (the "case") in depth and within its real-world context' (Yin, 2014, p. 16)	Methods and techniques	Emphasis on the scope, process and methodological characteristics of case study research, emphasising the nature of enquiry as empirical, and the importance of context to the case
Merriam	'intensive, holistic description and analysis of a single unit or bounded system' (Merriam, 1998, p. 1998)	Case characteristics	Emphasis on defining and understanding the case through the products of enquiry

Yin (2014, p. 16) provided a twofold definition of case study. The first part of the definition, as included in Table 4.1, details the scope of a case study:

A case study is an empirical inquiry that investigates a contemporary phenomenon (the 'case') in depth and within the real-world context, especially when the boundaries between phenomenon and context may not be clearly evident (Yin, 2014, p. 16)

The second part of the definition outlines the features of a case study:

A case study inquiry:

- copes with the technically distinctive situation in which there will be many more variables of interest than data points, and as one result
- relies on multiple sources of evidence, with data needing to converge in a triangulating fashion, and as another result
- benefits from the prior development of theoretical propositions to guide data collection and analysis (Yin, 2014, p.17).

The twofold definition indicates the way case study research addresses design logic, data collection and analytical techniques (Yin, 2014). While the definition remains relatively unchanged from Yin's first edition in 1984, over time, Yin has articulated previously implicit concepts, these being: (i) in-depth enquiry, (ii) presenting the phenomenon being studied as the 'case', (iii) the triangulation of evidence and (iv) having more variables of interest than data points.

According to (Yin, 2014) selecting case study as a research design will depend on the type of research question; the extent of control a researcher has over actual behavioural events; and the degree of focus on contemporary events, rather than entirely historical events. This view is clearly underpinned by Yin’s postpositivist/realist methodological position, which flows through into the way that methods of data collection and analysis are deployed.

4.2.3 Philosophy in Case Study Research

Case study researchers must demonstrate coherence between their philosophical position and research design (Taylor & Thomas-Gregory, 2015). Case study is not assigned to a fixed ontological, epistemological or methodological position, but instead can be oriented on the continuum from a realist/positivist perspective through to a relativist/interpretivist perspective. This flexibility lends case study a degree of ‘philosophical versatility’ (Harrison et al., 2017). This versatility can be viewed as an advantage because it allows the researcher to accommodate his or her worldview. An analysis of the philosophical and methodological positions of lead case study researchers is presented in Table 4.2.

Table 4.2: Ontological, Epistemological and Methodological Positions of Lead Case Study Researchers

Author	Philosophical Stance	Ontology	Epistemology	Methodology	Methods
Stake	Interpretivism	Relativism/constructivism	Transactional/subjectivist created findings	Hermeneutical/dialectical	Interviews and observations are the preferred and dominant data collection method
Yin	Postpositivism	Critical realism	Modified dualist/objectivist	Falsification of hypothesis	Documentation, archival records, interviews, direct observations, participant observation and physical artefacts
Merriam	Pragmatism	Relativism/constructivism	Transactional/subjectivist created findings	Hermeneutical/dialectical	Interviews and observations

Stake (1995) argued that relativism/constructivism orients qualitative case study research because ‘most contemporary qualitative researchers hold that knowledge is constructed rather than discovered’ (p. 99). Similarly, Merriam (1998) stated that relativism/constructivism orients qualitative case study research because ‘the key philosophical assumption upon which all types of qualitative research are based is the view that reality is constructed by individuals interacting with their social worlds’ (p. 9).

Yin (2014) largely subscribed to a critical realist perspective—assuming the existence of a single reality that is independent of any observer. However, Yin (2014) also argued that case study can accommodate a relativist perspective—acknowledging that reality is dependent on the observer. The postpositivist researcher uses science to understand reality, while accepting that all measurement is imperfect, and, for this reason, favours multiple methods to triangulate data analysis to understand reality as close as possible to the ‘truth’ (Lincoln et al., 2011). Yin’s postpositivist stance is evident in his methodology and multiple methods of data collection, all of which align well with the researcher’s position in the current study.

4.3 Case Study Research Design

4.3.1 Approaches to Case Study Research

The way a case study is characterised is in many ways dependent on the parameters applied by the individual researcher. Most case studies are identified by their approach, and there are several approaches to case study research that need to be considered. These approaches are not necessarily exclusive, since an individual case study can fall into more than one category (Taylor & Thomas-Gregory, 2015). Stake (1995) characterised case studies as intrinsic, instrumental or collective; Yin (2014) characterised them as descriptive, exploratory or explanatory; and Merriam (1998) characterised them as descriptive, interpretative or evaluative. Table 4.3 provides a brief description of each approach.

Table 4.3: Different Approaches to Case Study Research

Approach	Author	Description
Intrinsic	Stake, Thomas	When a researcher undertakes an investigation out of interest for its own merits
Instrumental	Stake, Thomas	When a researcher undertakes the research for a specific reason
Collective	Stake	Studying several cases within the same project
Descriptive	Merriam, Yin	Enables the researcher to describe the phenomenon (the 'case') in its real-world context
Exploratory	Yin, Thomas	Enables the researcher to understand more about a particular problem or situation, and identify the research questions or procedures to be used in a subsequent research study
Explanatory	Yin, Thomas	Enables the researcher to explain something about the phenomenon under investigation, and explain how or why some condition came to be
Interpretative	Merriam	Assumes an in-depth understanding and immersion in the environment
Evaluative	Merriam, Thomas	Aims to enable the researcher to determine whether something has worked or is working

Source: adapted from Taylor and Thomas-Gregory (2015), Yin (2014) and Thomas (2016, pp. 113, 120).

4.3.2 Case Selection

Case selection is an important case study method. Case studies are focused on cases; however, what constitutes a case needs to be clearly delineated. A case is a 'spatially and temporally defined entity created by researchers via a process' (Ragin, 1992, p. 217), frequently referred to as 'casing'. According to Ragin (1992, p. 217), casing is a 'research tactic', whereby researchers 'concoct' cases as a way to manage complexity. Researchers do not so much find cases as they define, delimit and declare them. In this way, casing creates discrete objects for case study by designating them as cases (Sandelowski, 2011).

The case is the object of the study and is commonly referred to as the 'unit of analysis'. The decision regarding what constitutes the 'unit' or case to be studied is made by the researcher (Stewart, 2014), who must define and bound the case (Yin, 2014). Defining the case is necessary to determine what is to be studied. The 'classic' case study usually focuses on an individual person, small groups, communities, events (Yin, 2014, p. 31), a program, an institution or a specific policy (Merriam et al., 2016, p. 38).

Once the case is defined, it must be clarified or 'bound' (Yin, 2014, p. 33). Bounding the case applies a frame to focus the research process on the object of the study and manage contextual variables, allowing the researcher to 'fence in' what is to be studied (Merriam et al., 2016, p.

38). The bounded context is crucial to case study research, so that users of the research can determine whether the findings are relevant to their particular context (Yin, 2014).

4.3.3 Case Design

A research design is the logical sequence that connects the data to the initial research question and then to its conclusions (Yin, 2014), or, as Yin (2014) colloquially describes it, a ‘logical plan for getting from here to there’ (p. 28). Many research methods come with a clear research design framework; however, in case study research, there is no ‘standard catalog’ of research designs (Yin, 2014, p. 27). Yin (2014, p. 29) suggested that there are five common components of case study design: (i) the research question/s, (ii) the propositions (if any), (iii) the potential unit/s of analysis, (iv) the logic linking the planned data to the propositions and (v) the criteria for interpreting the findings. The first three components lead the study design towards the data that are to be collected, and the last two components lead the study design towards methods of analysis relevant to the data that have been collected.

4.3.4 Specific Types of Case Design

Yin (2014) described case study design based on the number of cases (single versus multiple) and the units of analysis within each case (holistic versus embedded). Yin (2014) proposed four basic types of case study designs: (i) Type 1—single-case holistic design, (ii) Type 2—single-case embedded design, (iii) Type 3—multiple-case holistic design and (iv) Type 4—multiple-case embedded design.

4.3.4.1 Single or Multiple

A single case occurs when there is just that—a single ‘case’. Yin (2014) described case study design based on the number of cases (single versus multiple) and proposed five rationales for using a single-case design: critical, unusual, common, revelatory and longitudinal. Yin argued that a single-case study is analogous to a single experiment. Multiple-case study occurs when the same study contains more than a single case. Using a multiple-case study allows for a more in-depth understanding of the cases as a unit (Heale & Twycross, 2018). Evidence arising from multiple-case studies is considered more compelling than single-case research (Yin, 2014). However multiple-case study can require extensive resources and time that place it beyond reach of many researchers (Yin, 2014).

Multiple-case designs are generally preferred by case study researchers. Multiple-case research allows for a more in-depth understanding of the cases as a unit, through comparison of the similarities and differences of the individual cases (Heale & Twycross, 2018). Multiple-case studies frequently produce more reliable evidence than single-case research, and, according to Yin (2014), contribute to construct validity, thereby increasing research quality.

4.3.4.2 Holistic or Embedded

A case study in which the research question is concerned with the global nature of the case is a holistic case. The holistic case study has limitations; however, holistic case study can be conducted at an abstract level, without the operational detail frequently seen in multiple-case studies. Using a single-case holistic design results in the researcher ‘put[ting] all their eggs in one basket’ (Yin, 2014). It is also possible that the nature of a single-case holistic study may change as the study evolves, rendering the research design inappropriate for the research questions (Yin, 2014). Alternatively a single-case study may involve units of analysis at more than one level. These are embedded subunits that can be incorporated into either a single- or multiple-case design, depending on the definition of the case. Embedded designs confer an analytic advantage to the researcher because of the multiplicity of data sources used. However, embedded case designs also have limitations because they can result in the researcher focusing on the subunits and failing to consider the larger unit of analysis (Yin, 2014) that describes the case as a whole.

4.3.5 Data Sources and Data Collection

Data in case studies are frequently qualitative in nature (Heale & Twycross, 2018). The most common *qualitative* methods used in case study research are interviews, observations and documents (Houghton, Casey, & Smyth, 2017; Simons, 2009; Stake, 1995; Swanborn, 2010), with other qualitative methods including critical incidents, open letters, narratives, video analyses, photographs, log entries and artefacts (Simons, 2009). Although qualitative methods are more common, *quantitative* methods can also be used in case study research (Heale & Twycross, 2018). Quantitative methods can include surveys, examinations results and questionnaires (Simons, 2009; Taylor & Thomas-Gregory, 2015). Yin (2014) proposed six common sources of evidence: documents, archival records, interviews, direct observations, participant observations and physical artefacts—a list that includes the three most frequently used methods in case study research. Survey method is not one of Yin’s nominated six sources

of evidence; however, survey remains an acknowledged quantitative source of data (Simons, 2009; Taylor & Thomas-Gregory, 2015).

Yin (2014) proposed four principles of data collection: (i) using multiple sources of evidence, (ii) creating a case study database, (iii) maintaining a chain of evidence and (iv) exercising care when using electronic sources. Perhaps the most significant of these principles, and certainly the most ubiquitous in terms of case study methodology, is using multiple sources of evidence. Irrespective of the individual methods used, multiple sources of evidence are critical to producing a high-quality case study (Houghton et al., 2017). Incorporating multiple sources of evidence is far more crucial to case study research than other research designs (Yin, 2014). Triangulating from multiple sources of evidence (data) enables the researcher to capture the complexities of phenomena, thereby enabling a more complete description of the case (Houghton et al., 2017; Walshe, Caress, Chew-Graham, & Todd, 2004), as well as enhancing rigour (Houghton, Casey, Shaw, & Murphy, 2013). However, using multiple data sources has its challenges. First, the large and potentially overwhelming amounts of data mean that researchers can sometimes become ‘lost’ (Baxter & Jack, 2008, p. 554). Second, it can be difficult to bring the data together during analysis for reporting (Baxter & Jack, 2008). To overcome these challenges in this study, the researcher maintained a chain of evidence and created a case study database. These are strategies proposed by Yin (2014) to increase construct validity and reliability, respectively, and will be discussed further in Chapter 8 in reference to quality and rigour.

4.3.6 Data Analysis and Interpretation

Data analysis and interpretation receive the least attention in the case study literature. Data analysis and interpretation are distinct terms. Data analysis allows the researcher to ‘make sense’ of the data to produce findings and an overall understanding of the case, whereas interpretation refers to the insight derived from a more holistic and intuitive view of the data (Simons, 2009, p. 117) in relation to context. The difference between analysis and interpretation is subtle in that quantitative data analysis produces findings; however, the interpretation of these findings by the author is subjective, with ‘meaning’ created by considering the findings in the context of the literature. Thus, data analysis and interpretation are not discrete processes, and occur in an interactive and iterative manner (Simons, 2009).

Yin (2014) proposed four general analytic strategies to support a postpositivist position while managing the interplay between analysis and interpretation: (i) relying on theoretical

propositions, (ii) working the data from the ‘ground up’, (iii) developing a case description and (iv) examining plausible rival explanations. The first strategy of ‘relying on theoretical propositions’ requires the researcher to follow the theoretical propositions that led to the case study. In contrast, the second strategy of ‘working the data from the “ground up”’ is an inductive strategy. It ignores theoretical propositions and, instead, the researcher finds patterns or concepts within the data. The third strategy of ‘developing a case description’ involves organising the case study according to a descriptive framework. The fourth strategy of ‘examining plausible rival explanations’ operates in combination with the previous three strategies. It allows the researcher to collect evidence about possible other influences on the case.

In addition to the four analytic strategies, Yin (2014) proposed five individual analytic techniques for case studies: (i) pattern matching, (ii) explanation building, (iii) time-series analysis, (iv) logic models and (v) cross-case synthesis. The first technique of pattern matching compares an empirically based pattern with a predicted pattern. The second technique of explanation building is a type of pattern matching; however, in this instance, the aim is to analyse the case study data by building an explanation about the case. The third technique of time-series analysis searches for a match between the empirical trend and either the significant or rival trend articulated prior to the study. The fourth technique of logic models uses a complex chain of occurrences or events over an extended period. The final technique of cross-case synthesis is only applicable in multiple-case studies.

4.4 Adopted Case Study Design

4.4.1 Case Summary

A single, holistic, exploratory case study underpinned by the philosophical position of critical realism was used to explore how nurses engage with genomics in Australia. Critical realism was selected to provide the philosophical underpinnings of this research study, as it allowed the case study design to be grounded in separate ontological and epistemological assumptions. Critical realism combines a *realist* ontology with a *relativist* epistemology by subscribing to a form of ‘robust relativism’ (McEvoy & Richards, 2003, p. 411). This ‘robust relativism’ makes critical realism a suitable philosophy for case study research. The author was able to explore the obdurate reality (concrete structures, such as curriculum, policy and practice requirements) that shapes the relativist experience of nurses engaging with genomics in their practice.

In this case study, the phenomena of interest was genomics in nursing practice, while the case was genomics in nursing practice in Australia, and the context for the case study was the Australian healthcare setting. Participants were Australian registered nurses and midwives. Engagement in this case study referred to nurses’ attitudes, understanding, knowledge and application of genomics in their nursing practice. Using the term ‘engagement’ enabled a more comprehensive and holistic exploration of all aspects involved in the ways nurses might use genomics in nursing practice. The following section will outline the case study design used in this research study. The key components of the case study are presented in Table 4.4.

Table 4.4: Key Components of Case Study Design

Key Component	Description
Theoretical statement	This research study investigated the theory that Australian nurses do not fully engage with genomics because education, policy and practice do not support this engagement
Research aim	Determine how nurses engage with genomics in nursing practice in Australia
Issues	Australians nurses’ genomic knowledge and competency is poor Australian nurses are not engaging with genomics
Research question	How are nurses engaging with genomics in nursing practice in Australia?
Phenomenon	Genomics in nursing practice
Concrete manifestations	Examples of nurses applying genomic information and skills in the provision of nursing care
Propositions	The genomic literacy and competency of Australian registered nurses and midwives is low Australian nurses are not adequately incorporating genomics into nursing education, policy or practice
Unit of analysis	Registered nurses and midwives in Australia
Data collection	Cross-sectional survey and semi-structured interviews
Data analysis	Descriptive and inferential statistics, and thematic analysis

4.4.2 Case Justification

Genomics in nursing practice is a complex phenomenon. Engaging with genomics in practice involves possessing genomic knowledge and skills, as well as the insight to see why, when and how to use genomics in practice (Calzone et al., 2013). The clinical application of genetic and genomic knowledge has major implications for the entire nursing profession, regardless of academic preparation, role or practice setting (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). The literature addressing Australian nurses’ engagement with genomics

in Australia is limited. Thus, this research is necessary to determine nurses' engagement with genomics in Australia and provide insight into how engagement can be improved.

4.4.3 Type of Case Study Selected and Reasons for This Choice

The current researcher chose to undertake case study research because it allows for the investigation of a 'contemporary phenomenon (the "case") in depth and within its real-world context' (Yin, 2014, p. 16). More simply stated, a case study allows for the comprehensive study of complex issues in context (Anthony and Jack, 2009). Therefore, case study was the ideal design for the comprehensive study of genomics in nursing practice within the Australian healthcare setting. The premise for the case study was that viewing the subject from several angles would enable the researcher to 'close in' on 'why' and 'how' (Thomas, 2011, p. 40) genomics is applied in nursing practice. The researcher required no control over behavioural events (registered nurses' actions). Finally, and most importantly, genomics in nursing practice is a contemporary event, in the 'genomic era' of healthcare.

4.4.4 Key Components

4.4.4.1 Theoretical Statement

Case study research design embodies a 'theory' of what is being studied (Yin, 2014). The theoretical statement addresses the research questions, propositions, units of analysis, logic connecting data to propositions, and criteria for interpreting the findings—Yin's five components of research design—thereby essentially providing a 'blueprint' for the study (Yin, 2014, p. 38). Yin's (2014) theory-first approach to case study is evident in the second part of his definition of case study, where he contended that case study enquiry benefits from the prior development of theoretical propositions to guide data collection and analysis. Yin advised case study researchers to construct a preliminary theory related to the topic of the study. Construction of theory prior to the study distinguishes case study from grounded theory and ethnography. This research study was centred on the theory that *Australian nurses do not adequately engage with genomics because education, policy and practice do not support engagement*. Yin (2014) also advocated for anticipating rival explanations, so they can be 'captured' during data collection. In this research study, the rival theory was that *Australian nurses do not adequately engage with genomics because of lack of professional interest or perceived relevance to practice*.

4.4.4.2 Research Aim

The aim of this research study was to determine how nurses engage with genomics in nursing practice in Australia.

4.4.4.3 Issues

A literature review is typically conducted prior to the study. This may include a review of the literature, grey literature, media, reports and more. The literature review determines what is already known about the case. It provides a basic understanding of the case and informs the development of research questions (Heale & Twycross, 2018). Issues about the problem being investigated are identified in the literature. These issues are transformed into the research questions or propositions (Harrison & Mills, 2016). Together, they reflect the purpose of the study.

It is evident in the literature that the ‘genomic era’ of healthcare is upon us, yet international studies have shown that nurses’ genomic knowledge and competency is poor (Skirton et al., 2012). Nurses’ limited engagement with genomics in other countries indicates that there is potential for limited engagement with genomics by the Australian nursing profession. The issues identified for this research study were as follows: (i) *the genomic literacy of nurses and midwives is low* and (ii) *nurses and midwives are not adequately incorporating genomics into their nursing practice*. Although preliminary analyses indicate that genomics is present in nursing practice in Australia, a comprehensive investigation has not been undertaken; therefore, nurses’ engagement with genomics in practice in Australia remains unclear.

4.4.4.4 Research Question, Phenomenon and Concrete Manifestations

The form of the research questions—whether a ‘who’, ‘what’, ‘where’, ‘how’ or ‘why’ question—indicates the most relevant methods of data collection and analysis. ‘How’ and ‘why’ questions lend themselves best to case study research (Yin, 2014, p. 29). Research questions primarily focus on answering queries related to ‘what is’ and ‘what has happened’, or explaining the ‘how and why’ of a situation (Yin, 2014). The research question is centred on the phenomena of interest. This must be a real-life phenomenon that has a concrete manifestation (Yin, 2014). The research question for this study was: *How are nurses engaging with genomics in nursing practice in Australia?* In this case study, the phenomenon was

‘genomics in nursing practice’ and the concrete manifestations were examples of nurses applying genomic knowledge and skills in the provision of nursing care.

4.4.4.5 Propositions

The development of theoretical propositions is recommended during the design phase of a case study, as they can provide a ‘blueprint’ for the study (Yin, 2014, p. 38). The ‘how’ and ‘why’ of a research question may lead to the case study design; however, questions alone will not provide a clear direction of the case that should be studied. Propositions direct attention to the phenomenon that should be examined within the scope of the study, and direct attention towards potential evidence to be collected and analysed (Yin, 2014). As Yin (2014) argued, without clear research questions and propositions, the researcher ‘may be inclined to cover everything’ (p. 31). This research study had two key propositions that supported the research question: (i) *genomics has a limited presence in the nursing profession in Australia* and (ii) *Australian nurses are not adequately incorporating genomics into nursing education, policy or practice*.

4.4.4.6 Units of Analysis

Defining the case is necessary to determine what is to be studied. According to Yin (2014, p. 31), the ‘classic’ case study usually focuses on an individual person, while a case can encompass small groups, communities and events. Once the case is defined, it must be clarified or ‘bound’, allowing the researcher to ‘fence in’ what is to be studied (Merriam et al., 2016, p. 38). The unit of analysis or ‘case’ in this research study was *genomics in nursing practice* with the context of the *Australian healthcare setting*. Oncology nurses were selected as a subunit because genomics has a higher presence in the oncology literature, thereby suggesting that these nurses’ engagement with genomics may be higher than other specialties or generalist nurses.

4.4.4.7 Logic Connecting Data to Propositions

Linking data to propositions guides the choice of data collection and analysis methods (Yin, 2014). The following section outlines the methods of data collection and analysis that were chosen to address each proposition. In this study, two propositions were developed, based on the research question: *How are nurses engaging with genomics in nursing practice in Australia?* The first proposition—that Australian nurses have a limited genomic literacy—required a method of data collection and analysis that would result in findings generalisable to

the profession. The second proposition—that Australian nurses are not adequately incorporating genomics into nursing education, policy or practice—focused on the lived experience of clinicians. As such, this required a method of data collection and analysis that provided an opportunity for the researcher to ask questions of Australian nurses. Each proposition and the associated data collection method and analytical technique will be discussed in the next section.

4.4.4.8 Proposition 1: Australian Nurses Have Limited Genomic Literacy

4.4.4.8.1 Recruitment

Recruitment for the survey was undertaken in a variety of ways to maximise responses. Initially this recruitment took the form of advertisements containing the survey link in the Australian College of Nursing (ACN) newsletter, along with distribution of the survey link by chapters of the Australian Nursing and Midwifery Federation (ANMF) to its membership. These strategies were supplemented by the researcher's attendance at the ACN National Nursing Forum, where participants could either complete the survey online on an iPad provided, or a paper-based version at the university booth. A flyer with a QR code linking to the online survey (Appendix 1) was also distributed at this event to enable participants to easily access the survey at a time convenient to them.

These recruitment strategies were intended to reach the broader population of registered nurses and midwives in Australia. This population numbered 284,245 in 2015 (AIHW, 2016). G*Power 3TM analysis determined that 232 was the minimum sample size required for the survey to demonstrate statistical significance. A systematic review conducted by Fan and Yan (2010) reported that surveys sponsored by credible agencies about topics of high interest to the population and of limited length are associated with higher response rates. In recognition that the topic area may not be of high interest to the contemporary nursing population, strategies to maximize response rates were employed, including: sending multiple reminders (or repeated invitations); offering the survey in multiple formats; and providing incentives for participation (Sauermann & Roach, 2013). As recruitment was initially slow, approval from the university's Human Research Ethics Committee was obtained in September 2016 to offer an incentive to participants to enter a draw to win a \$200 gift card for completion of the survey (Appendix 2). The data collection period was also extended to December 2016, by which time sufficient participant numbers were achieved.

4.4.4.8.2 Data Collection

A cross-sectional survey was conducted to determine the genomic literacy of Australian registered nurses and midwives. Survey data were collected in electronic form using Qualtrics™. Demographic information was collected in addition to the main genomic literacy survey. The demographic items were based on key variables listed in surveys conducted by the Australian Institute of Health and Welfare, Australian Bureau of Statistics and Australian Health Practitioner Regulation Agency (AHPRA), which also align with the demographics collected in similar genomic literacy studies (McCabe, Ward, & Ricciardi, 2016; Read & Ward, 2016; Ward, Haberman, & Barbosa-Leiker, 2014). The alignment of the demographic questions with those of these sources allowed for comparative work. Demographic characteristics were re-categorised into smaller subgroups for the purpose of data analysis and reporting.

Genomic literacy data were collected using the GNCI®. The GNCI® assesses genomic knowledge across four topical categories (human genome basics, mutations, inheritance patterns and genomic healthcare applications) and 18 concepts. Although designed for nursing practice, the genetic and genomic concepts that informed the GNCI® are sufficiently relevant to midwifery practice to justify its use in assessing the genomic literacy of midwives. Permission was obtained from the author of the GNCI® to use the instrument in Australia on the condition that the instrument was not reproduced (Appendix 3). The psychometric properties of the GNCI® were reported by McCabe et al. (2016), the authors of a recent study conducted in the US, who used this tool to assess genomic knowledge among practising nurses. Internal consistency (Cronbach's α) was calculated as $\alpha = 0.76$ (McCabe et al., 2016), and the total scale difficulty (overall percentage of correct items) was 44.2% correct responses (McCabe et al., 2016), which was within the target scale difficulty (measured as the per cent of correct responses) set at 40% to 55% during the instrument's development (Ward et al., 2014). The GNCI® was subjected to initial content validity testing via an independent expert panel (a genetics specialist, registered nurse and registered midwife) prior to distribution. The panel reviewed the GNCI® for content validity for the Australian context. Minor amendments were recommended and subsequently made to the GNCI®.

4.4.4.8.3 Data Analysis

Descriptive and inferential statistics were used to analyse the survey data. The numerical data were collated and analysed using IBM SPSS Statistics 24. Three hundred and ninety-eight

survey responses were recorded. More than 100 respondents did not complete the GNCI[®] scale, with the missing data points shown to be random. Completed surveys (those with 30 or more questions completed) were included in the final analysis. Expectation maximisation function was used to replace the missing values, allowing a total value to be calculated. A total of 253 ($n = 253$) respondents were used in the final statistical analysis, equating to a 64% completion rate. Although 253 respondents were used in the GNCI[®] analysis, not all 253 respondents answered all demographic items in the survey. Missing demographic values were found to be random; therefore, they were retained in the final analysis. Given that this was an online study advertised by social media networks and electronic mailing lists, it was not possible to calculate the number of surveys issued or the resultant response rate.

Descriptive statistics were used to determine general trends in the demographic data. The correct responses (via percentage) were provided for each GNCI[®] item (question), and the mean correct responses (via percentage) per concept and category were calculated using the item data. The GNCI[®] scores were dichotomised into high and low groups using the median GNCI[®] score (13) as the point of separation. The relevance to practice and perceived knowledge reported by nurses and midwives were reported separately. Chi-square (χ^2) analyses were used to determine if there was a significant difference in genomic knowledge based on demographics.

4.4.4.9 Proposition 2: Australian Nurses Are Not Adequately Incorporating Genomics into Nursing Education, Policy or Practice

4.4.4.9.1 Recruitment

Negotiating access to a case study site is the first step in case study research (Stewart, 2014). In the current study, a 'gatekeeper' was selected to assist the researcher to collect the necessary permissions to conduct the research. It is important that the researcher establish an open and transparent relationship with 'gatekeepers' (Simons, 2009).

Participants were recruited at the hospital unit level through printed flyers (Appendix 4), a short presentation (or 'in-service') and an email invitation to the nurse unit manager with the request that it be forwarded on to nursing staff. Nurses who were interested in participating in the study were asked to contact the principle investigator via the contact details provided in the study presentation and/or printed materials. Once contacted, the principle investigator arranged a time to speak with the potential participant, either in person or via telephone, to answer any

questions he or she may have about the study. If the potential participant declined to participate, no further action was taken. If the potential participant agreed to be involved in the study, he or she was required to read the participant information sheet and complete the consent form (Appendices 5 and 6 respectively). To ensure consent was informed, this consent form was reviewed with the potential participant before he or she completed the form.

4.4.4.9.2 Data Collection

Determining the sample size in qualitative research is challenging because guidelines for determining sample sizes are scarce. Sample size in qualitative interviews is debated in the literature, with various recommendations on the size and composition of sample participants (Beitin, 2012). Research once required a clearly defined, predetermined number of participants. However, there has been a recent trend to view sample size as fluid and emerging throughout the research design. This has led many researchers to focus on the research process as informing the ultimate number of participants (Beitin, 2012). Purposive sample size typically relies on the concept of 'saturation' or the point at which no new information or themes are observed in the data (Guest, Bunce, & Johnson, 2006). Studies have shown that a small sample size is sufficient in qualitative research. In this study, it was anticipated that no more than 12 interviews would be required to reach saturation. Nine ($n = 9$) participants were interviewed. Participant demographics are presented in Table 4.5.

Table 4.5: Demographics of Interview Participant Group

Participant Characteristic	Number (n)
Total participants	9
Gender	
Female	8
Male	1
Setting	
In-patient oncology unit	1
Out-patient oncology unit	8
Age range	
18–24 years	1
25–34 years	3
35–44 years	1
45–54 years	2
55–64 years	2
65–74 years	0
75+ years	
Year registered	
Prior to 2000	4
2000–2010	2
After 2010	3
Years working in oncology	
5 years or less	4
6–15 years	3
16–20 years	2
20+ years	0
Postgraduate qualifications	
Postgraduate qualifications in oncology (completed or currently enrolled)	4
Postgraduate qualifications or specialist training in genetics/genomics	0

Semi-structured interviews were conducted to determine how Australian nurses are incorporating genomics into nursing education, policy and practice. Interviews are commonly used in case study research (Yin, 2014, p. 110). Interviewing participants has been defined as a ‘conversational practice where knowledge is produced through the interaction between an interviewer and an interviewee or a group of interviewees’ (Brinkmann, 2008, p. 470). The conversation provided participants with the opportunity to talk about their experiences and perspectives, and allowed the researcher to capture their language and concepts relevant to the topic (Rubin & Rubin, 2012). Semi-structured interviews are the dominant form of interview

processes (Braun & Clarke, 2013). In semi-structured interviews, the researcher uses an interview guide with predetermined, yet open-ended, questions that are used to guide the conversation with the participant. This gives the researcher more control over the topics of the interview, yet there is no fixed range of responses to each question (Ayres, 2008a), thus providing scope for the participant to raise issues that may not have been anticipated by the researcher (Braun & Clarke, 2013).

In this research study, the semi-structured interview guide used was based on the cross-sectional survey, as well as the literature review. The interview guide included questions that were typically brief, simple and open (Appendix 7). Opening questions were used to start a dialogue with the participant—for example, ‘Can you tell me about a time when you saw or heard another registered nurse use genomics in their practice?’. A closing or ‘clean-up’ question was used to give participants an opportunity to raise any issues not covered in the interview—for example, ‘Are there any questions or comments that you would like to make about genomics in nursing practice?’. Each interview question was carefully worded and sequenced so that the questions flowed in topic-based sections (Braun & Clarke, 2013). Interview prompts and probes were added to the interview guide to encourage participants to expand on their answers (Ayres, 2008b; Braun & Clarke, 2013)—for example, ‘Probe: Can you give me an example of how genomic information can be used in practice?’. Non-directive examples were occasionally provided to participants if they were unclear about the ‘type’ of answer required. The interview guide was piloted and subsequently re-drafted with selected questions reworded and the question sequence altered.

Interviews require intense focus by the researcher, making them emotionally draining (Braun & Clarke, 2013); thus, only one interview per day was conducted. Interviews were conducted in the participants’ own time, so all interviews were scheduled at a time convenient to the participant. All interviews were conducted on the hospital grounds to ensure the safety of the researcher (Braun & Clarke, 2013). The researcher was permitted to use small rooms within the oncology units, with the familiar surroundings making participants comfortable. The rooms were quiet with minimal background noise to minimise distractions and ensure optimal audio recording. It was expected that the interviews would take approximately 30 to 60 minutes, with most interviews falling in this range.

Interviews were digitally recorded and professionally transcribed prior to analysis. Digital recordings capture the language and concepts that participants use to discuss their experiences

and perspectives (Braun & Clarke, 2013). Two recording devices were used in case one recording device failed or produced a poor-quality recording. All participants consented to have their interview recorded.

4.4.4.9.3 Data Analysis and Interpretation

Yin's (2014) analytic techniques—pattern matching, explanation building, time-series analysis, logic models and cross-case synthesis—all represent ways of linking data to propositions. The actual analysis requires the researcher to combine the case study data as a direct reflection of the study propositions (Yin, 2014). Yin's (2014) general analytic theory of 'working the data from the "ground up"' (p. 137) sees the researcher reviewing or 'playing' with the data and noting a pattern or useful concept, which may lead the researcher down a particular analytic path. Yin's (2014, p. 143) individual analytic technique 'pattern matching' compares an empirically based pattern with a predicted pattern. The general analytic strategy of 'working on data from the "ground up"' and individual analytical technique of 'pattern matching' are, at their simplest level, inductive strategies that allow the researcher to find patterns or concepts within the data, similar to inductive coding and thematic analysis techniques. In this light, the terminology may be different, yet the premise is the same. Therefore, in the current study, inductive coding and thematic analysis were used as an alternate analytic strategy to those offered by Yin. The qualitative traditions are known for their more holistic nature, and frequently promote holism, simultaneous data collection and analysis, insights resulting from case comparison, emphasis on an inductive and exploratory approach, the generation of central concepts, the alignment of theory and concepts, and the abstraction reached by 'bottom-up' analysis (Swanborn, 2010).

Thematic analysis is a data reduction and analysis strategy by which qualitative data are segmented, categorised, summarised and reconstructed in a way that captures the important concepts within the dataset (Ayres, 2008b). Thematic analysis seeks uniqueness, commonalities/similarities and patterns, and, in this way, is similar to Yin's pattern matching technique. Coding was used to identify themes in the data. A code in qualitative research is 'a word or short phrase that symbolically assigns a summative, salient, essence-capturing, and/or evocative attributes for a portion of language-based or visual data' (Saldana, 2016, p. 4). It is a researcher-generated construct that 'translates' data by attributing meaning to each individual datum for later purposes of analysis (Saldana, 2016). Coding is not just labelling—it is *linking* (Saldana, 2016, p. 9). Linking implies a successive nature, where coding leads the researcher

from the data to the idea, and from the idea to all the data pertaining to that idea (Richards & Morse, 2013).

The researcher used each of the stages of coding and analysis described by Braun and Clarke (2013, p. 202): (i) transcription (for interviews only), (ii) reading and familiarisation, (iii) coding, (iv) searching for themes, (v) reviewing themes, (vi) defining and naming themes and (vii) writing. Saldana (2016) stated that coding is a cyclical act, and argued for first- and second-cycle coding. First-cycle coding refers to the processes that occur during the initial coding. Second-cycle coding is an advanced way of re-organising and re-analysing data coded through first-cycle methods. In the current study, the author originally assigned 46 codes on the first cycle of coding. These codes were reduced to 23 codes in a second-round cycle. The codes were originally grouped into four themes: *genomic knowledge, application, relevance and education*. These themes were reviewed and re-categorised into two main themes—*genomic literacy* and *relevance to practice*—with *genomic literacy* having three subthemes: *knowledge of genomics, application of genomics* and *genomics education*. Demographics were coded separately.

The program NVivo was selected for use in this study. Computer-assisted software is useful for assisting researchers to organise their intellectual work and to bring together identified categories of data for easy comparison (Julien, 2008). These programs also offer tools to define categories, annotate text, write memos and calculate frequencies of categories and codes. NVivo can enhance the rigour of the research by providing a comprehensive trail of decisions made during data collection and analysis (Houghton et al., 2017; Silverman, 2013). This is achieved by the researcher writing and recording notes about decisions within the NVivo program. In the current study, these notes were reviewed and updated by the researcher throughout the process of analysis, and assisted in the integration of findings from the quantitative and qualitative studies, and the interpretation of meaning both individually and in the triangulation of findings.

4.4.4.10 Interpreting the Case Findings

Interpreting the findings is the final key component of a case study design, as outlined by Yin (2014). In quantitative research, statistical estimates serve as the criteria for interpreting research findings—for example, $p < 0.5$ indicates statistical significance (Yin, 2014, p. 36). However, because case study research can also include qualitative findings, other criteria for interpretation are also required. Yin (2014) suggested addressing rival explanations as a criterion for interpreting case study findings. The idea is that the more rival explanations that have been addressed and rejected, the stronger the study findings. Yin listed several types of rival explanations. In this study, the author selected the ‘rival theory’, which was defined by Yin (2014) as follows: ‘a theory different from the original theory explains the results better’ (p. 141). The original theory and rival theory were as follows:

Theory (theoretical statement): Australian nurses do not adequately engage with genomics because education, policy and practice do not support engagement.

Rival theory: Australian nurses do not adequately engage with genomics because of a lack of professional interest or perceived relevance to practice.

Yin (2014) argued for anticipating rival explanations so they can be ‘captured’ during data collection. In this research study, relevance to practice was explored to achieve this goal and to provide for the researcher’s interpretation of findings to identify that which could be judged closer to reality.

In practical terms, interpretation of the study findings occurs at the individual data collection (source) level and the ‘case’ level. The findings from the cross-sectional survey and semi-structured interviews were interpreted in isolation (and presented in individual manuscripts), and then combined and interpreted together using thematic analysis. Thematic analysis is enhanced by triangulation. Triangulation enhances credibility by using multiple data sources. The two main purposes of triangulation are to confirm data and to ensure data are ‘complete’ (Walshe et al., 2004). Combined data sources allow for in-depth insight and completeness in the cases and their context (Houghton et al., 2017). These individual findings were used to develop a description of the case of genomics in nursing practice in Australia.

4.5 Data Management

Data management for this research project was in accordance with the James Cook University *Code for the Responsible Conduct of Research*, adapted from the *Australian Code for the Responsible Conduct of Research*. The principle investigator ensured that all raw data were stored in accordance with the *Code for the Responsible Conduct of Research*. Electronic data are stored on the principle investigator's password-protected computer, with data backed up on an external hard-drive to safeguard against accidental data loss. The external hard-drive is housed in a separate location away from the university in a secure location. For the duration of the study, and upon completion of the study, raw data will be stored in the principal investigator's office in the Nursing Sciences building at James Cook University, Townsville Campus, in a locked filing cabinet. Data will be held for a minimum of five years from the end of the year of publication of the last refereed publication or other form of public release to an audience outside of the university that is based on the data.

This was a low/negligible risk qualitative study; thus, a Data and Safety Monitoring Committee was not required. The principle investigator was responsible for data and safety monitoring during the project. The research team met frequently during the project to review study conduct. It was agreed that the project would be discontinued if the research team believed data and safety had been sufficiently compromised.

4.6 Safety Considerations/Patient Safety

The protection of research participants takes precedence above all other considerations in a study. However, given the nature of this study, the likelihood of adverse events or serious adverse events was negligible. Adverse events were likely limited to minor discomfort or feelings of embarrassment or incompetence if the participant believed his or her genomic literacy to be limited. It was extremely unlikely that interviews would trigger painful memories or experiences for the participants, where they could become distressed. Any adverse events were to be recorded by the principle investigator and reported to the research team and nurse unit manager within 24 hours. Action taken would depend on the nature of the adverse event. No adverse incidents occurred during the course of the study.

4.7 Ethics Approval

This research involved two separate research studies, each with a separate ethical approval. The survey research study titled ‘Genomic Literacy of Australian Registered Nurses and Midwives: A Cross-sectional Survey’ was granted approval by James Cook University Human Research Ethics Committee (HREC) (H6587). The interview research study titled ‘Genomics in Oncology Nursing Practice’ was granted approval by Townsville Hospital and Health Service HREC (HREC/17/QTHS/241 and SSA/17/QTHS/247).

4.8 Summary

This chapter has provided an overview of case study research. It has addressed the history of case study research, and the contribution of lead case study researchers and their nuanced versions of case study design. It has also outlined the key components of case study research, including case selection, data collection and data analysis. The specific design used in this study was presented, with each step in the research process described separately. The following chapters will present the findings of the individual data collection methods and the combined case findings.

Chapter 5: Findings—Genomic Literacy of Registered Nurses and Midwives in Australia

5.1 Introduction

International studies have indicated that the genomic literacy of registered nurses and midwives is limited. To date, no comparable studies have been conducted in Australia. A brief summary of genomic literacy will be provided as an adjunct to the work presented in Chapter 2, as well as acknowledging the call for a genomically literate workforce by key government institutions and Australian organisations. This chapter also contains an article accepted for publication (in press) addressing the genomic literacy of Australian registered nurses and midwives. This article summarises Australian registered nurses' and midwives' genomic literacy, as well as their perceived knowledge and attitude towards genomics in nursing and midwifery practice.

5.2 Genomic Literacy

The case has been made in preceding chapters that registered nurses and midwives require a degree of genomic literacy if they are to adequately communicate with other healthcare professionals provide optimal care to patients, their families and the community. The term 'genetic literacy' was defined in Chapter 2 as 'knowledge sufficient to develop genetic and genomic competency', as outlined in the *Essentials of Genetic and Genomic Nursing* competency document (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). In short, nurses must 'learn the language of genetics' (Cashion, 2009). A review of the literature indicated that several studies assessing nurses' and midwives' genomic knowledge and competence have been conducted internationally. Most studies have been conducted in the US, with other studies conducted in the UK, Turkey, Italy, Jordan, Japan and Canada. Regardless of country of origin, studies exploring nurses' and midwives' genomic literacy produced largely similar findings—that nurses' knowledge of genomics is limited. This assertion is confirmed by the systematic literature reviews appearing in the literature in recent years, including the integrative literature review published by the author and presented in Chapter 2. The absence of Australian findings represents a significant gap in the nursing literature.

5.3 Genomics in Australia

There have been many calls to prepare the workforce to deliver genomic care. In 2018, the Australian Government by way of the Australian Health Ministers' Advisory Council (2017b) published the *National Health Genomics Policy Framework 2018–2021*, reflecting a shared commitment to 'leveraging the benefits of genomics in the health system for all Australians' (p. i). A priority of the *National Health Genomics Policy Framework 2018–2021* is to build a skilled workforce that is literate in genomics through increasing capacity and capability in genomics and bioinformatics (Australian Health Ministers' Advisory Council, 2017b, p. 7). The document is generic in its reference to the 'health workforce' and 'health professionals', clearly indicating that it is not simply designed for use by medical and/or genetic specialists. As the largest components of the health workforce, nursing and midwifery fall well within the remit of this document.

Australian organisations are also being formed with the intent to improve the genomic literacy of the Australian health workforce. The AGHA—frequently referred to as Australian Genomics—is a national research collaboration of clinicians, researchers and diagnostic geneticists working together to provide evidence for the equitable, effective and sustainable delivery of genomic medicine in healthcare. The AGHA believes that the successful implementation of genomics in healthcare will depend on the availability of a workforce able to deliver genomic medicine; thus, it has developed the 'Genomics Workforce, Education and Ethics' research program to apply quantitative and qualitative methods to identify the current landscape with respect to workforce education and training, patient understanding and ethics. As with the *National Health Genomics Policy Framework 2018–2021*, the AGHA is for clinicians, researchers and diagnostic geneticists. Nursing and midwifery again falls within the remit of the 'clinical' workforce referred to in this document.

The *National Health Genomics Policy Framework 2018–2021* shares a similar aim to that proposed by the authors of the seminal document, *Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators*, which aims to prepare the nursing workforce to deliver competent genetic- and genomic-focused nursing care (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 7). No comparable Australian genetic/genomic nursing competency documents were located in the conduct of this present study. The AHPRA and Nursing and Midwifery Board of Australia (NMBA) have made no similar recommendations nor produced any publications advocating for a genomically literate

nursing and midwifery workforce. With no Australian-developed genomic competencies, Australian nurses and midwives must rely on international documents and resources to inform their practice. Therefore, despite the Australian Government and genomic organisations, such as the Australian Genomic Health Alliance, acknowledging the need for a genomic workforce, it seems that key Australian nursing and midwifery organisations and governing bodies are not.

5.4 Assessing Genomic Literacy: Genomic Nursing Concept Inventory (GNCI[®])

Several studies assessing nurses' and midwives' genomic knowledge and competence have been conducted internationally; however, no such studies have been conducted in Australia. Thus, the genomic literacy of Australian nurses and midwives is unknown. As such, the current author sought to assess the genomic literacy of Australian nurses and midwives as part of a wider study examining nurses' engagement with genomics in practice. The GNCI[®] was selected as the instrument to assess genomic literacy in this study. The GNCI[®] was developed by Assistant Professor Linda Ward (2011) as a Doctor of Philosophy project in 2011. The GNCI[®] is a 'scale to measure understanding of the genetic/genomic concepts most critical to nursing practice' (Ward et al., 2014, p. 511). The 'concepts most critical to nursing practice' are designed to measure nurses' knowledge of key concepts underlying the *Essentials of Genetic and Genomic Nursing: Competencies, Curricula, Guidelines and Outcome Indicators* document (Ward, 2011). These essential competencies were developed by an independent panel of nurse leaders from clinical, research and academic settings, with the intent of identifying the minimum standards necessary to prepare the nursing workforce to deliver competent genetic- and genomic-focused nursing care, and guide nurses in the application of their professional skills and responsibilities (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). The GNCI[®] was selected for use in this study because of its alignment with this iconic genomic nursing document.

5.5 Genomic Literacy of Registered Nurses and Midwives in Australia: Cross-sectional Survey Findings

This cross-sectional survey was conducted in 2015 with the aim of measuring the genomic literacy of Australian nurses. The details of this phase of the research were published in full as a journal article, which is presented as the findings in the following section of this chapter. The findings of the survey also form a major component of the case study presented in Chapter 7.

Chapter no.	Details of publication on which the chapter is based	Nature and extent of the intellectual input of each author, including the candidate
5	Wright, H., Zhao, L., Birks, M., & Mills, J. (in press). Genomic literacy of registered nurses and midwives in Australia: A cross-sectional survey. <i>Journal of Nursing Scholarship</i> . Manuscript ID is JNU-03-18-101.	Wright designed the study. Wright collected the data and performed the data analysis with assistance from Zhao. Wright developed the figures and tables with assistance from Zhao. Wright wrote the first draft of the paper which was revised with editorial input from Zhao, Birks and Mills.

The author conducted a study to measure the genomic literacy of Australian registered nurses and midwives through assessing participants' understandings of the genomic concepts most critical to nursing and midwifery practice, as well as their perceived knowledge and attitudes towards genomics in nursing and midwifery practice. The author conducted a cross-sectional survey of Australian registered nurses and midwives using the GNCI[®]—a 31-question multiple-choice survey instrument (*see Appendices 1-3*). Descriptive and inferential statistical techniques were used to calculate the total GNCI[®] score and scores on individual subcategories, as well as the relationships between demographic variables and GNCI[®] scores. The findings indicated that the genomic literacy of registered nurses and midwives in Australia is low. It is expected that the findings from this study will serve as a catalyst to improve the genomic literacy of the Australian nursing and midwifery workforce, thereby allowing for improved health outcomes for individuals and the wider Australian public.

Genomic Literacy of Registered Nurses and Midwives in Australia: A Cross-Sectional Survey

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Key words

genetics, Australian, genomics, knowledge, midwife, registered nurse

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Abstract

Purpose: Registered nurses and midwives require a degree of genomic literacy if they are to adequately communicate with other healthcare professionals and provide optimal care to patients, their families, and the community. Several studies have been conducted internationally to assess the genomic literacy of nurses; however, the genomic literacy of Australian registered nurses and midwives has not been investigated. The aim of this study was to measure the genomic literacy of Australian registered nurses and midwives through assessing participants' understandings of genomic concepts most critical to nursing and midwifery practice, as well as their perceived knowledge and attitude towards genomics in nursing and midwifery practice.

Design: Cross-sectional survey of Australian registered nurses and midwives using the Genomic Nursing Concept Inventory (GNCI[®]), a 31 multiple-choice question survey instrument. Participants were recruited via two key Australian nursing and midwifery organizations over an 8-month period in 2016.

Methods: Descriptive and inferential statistical techniques were used to calculate the total GNCI[®] score and scores on individual subcategories, as well as relationships between demographic variables and GNCI[®] scores.

Findings: Most respondents worked as clinicians (71.4%) in a hospital or hospital-based setting (61.8%). Most registered nurses (80.5%) and midwives (97.2%) reported that genetics was relevant to clinical practice; however, over 80% of registered nurses and midwives believed their knowledge of genetics was poor or average. Genomic knowledge was assessed using the GNCI[®]. Scores ranged from 3 to 29 (out of a possible 31), with a mean score of 13.3 (*SD* 4.559) based on 253 (*N* = 253) respondents, indicating that genomic literacy is low. There was a significant difference between genomic knowledge scores and education and training level (*p* = .036).

Conclusions: The genomic literacy of registered nurses and midwives in Australia is low. More must be done to ensure Australian registered nurses and midwives have an adequate level of genomic literacy to provide optimal care to patients, their families, and the community.

Clinical Relevance: Modern medicine requires a healthcare workforce that is literate in genomics. Findings from this study may serve as the catalyst to improve the genomic literacy of the Australian nursing and midwifery workforce, allowing for improved health outcomes for individuals and the wider Australian public.

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Table S1. Demographic Background of Participants

	<i>N</i> (%)
Age	
20-34 years	45 (17.8)
35-44 years	43 (17.0)
45-54 years	86 (34.0)
55-64 years	66 (26.1)
65+ years	13 (5.1)
Gender	
Female	229 (91.6)
Male	21 (8.4)
Registration type	
Registered nurse	216 (85.7)
Registered midwife	36 (14.3)
Nursing registration year	
<1970	5 (2.0)
1970-1989	98 (38.7)
1990-2009	94 (37.2)
>2009	47 (18.6)
Highest qualification in nursing	
Hospital / vocational training	22 (8.7)
Bachelor degree	88 (34.8)
Graduate Certificate /diploma	66 (26.1)
Master and doctorate degree	77 (30.4)
Primary practice setting	
Public health service	186 (73.8)
Private health service	66 (26.2)
State	
New South Wales	41 (16.2)
Victoria	39 (15.4)
Queensland	123 (48.6)
South Australia	25 (9.9)
Western Australia	9 (3.6)
Tasmania	6 (2.4)
Northern Territory	4 (1.6)
Australian Capital Territory	4 (1.6)
Geographical region	
Major cities of Australia	121 (48.0)
Inner regional Australia	65 (25.8)
Outer regional Australia	53 (21.0)
Remote Australia	4 (1.6)
Very remote Australia	3 (1.2)
Migratory	2 (0.8)
Unsure	4 (1.6)
Principal role	
Clinician	180 (71.4)
Administrator	14 (5.6)
Educator	34 (13.5)
Researcher	10(4.0)
Others	14 (5.6)
Primary work setting	

Hospital based	155 (61.8)
Non-hospital based care	53 (21.1)
External organisations	6 (2.4)
Education institutes	19 (7.6)
Other	18 (7.2)

Table S2. Performance in GNCI[®]

Topical category (4)	Concept (18)	Item (31)	% of correct responses per item	Mean % of correct responses per concept	Mean % of correct responses per topical category
Genome basics (12 items)	Genome composition and organization	2	38.7%	32.8%	28.7%
		4	27.7%		
		5	39.1%		
		8	25.7%		
	Homozygosity and heterozygosity	13	15.4%	21.2%	
		29	26.9%		
	Gene function	1	27.7%	26.8%	
		6	17.8%		
		9	34.8%		
	Gene expression	11	22.9%	22.9%	
	Human genome homogeneity	3	37.9%	37.9%	
	Genotype-phenotype association	7	30.4%	30.4%	
Mutations (3 items)	Mutations and disease	19	35.2%	48.3%	50.3%
		21	61.3%		
	Germline and somatic mutations	18	52.2%		
Inheritance (8 items)	Dominance	10	42.3%	42.3%	46.9%
	Autosomal inheritance	24	28.9%	28.9%	
		30	48.2%	53.8%	
	Autosomal recessive	15	61.3%	63.1%	
		16	64.8%		
	X-linked	17	48.6%	48.6%	
Multifactorial	25	44.7%	44.7%		
Genomic healthcare (8 items)	Family history	23	23.3%	49.2%	48.9%
		26	75.1%		
	Pharmacogenomics	12	90.5%	65%	
		27	43.1%		
		28	61.3%		
	Cancer genetics	20	21.3%	21.3%	
	Genetic testing	14	81%	60.1%	
22		39.1%			

Table S3. Demographic Characteristics of Participants With GNCI® Outcome

Characteristics	GNCI® total score ≥13	<i>p</i>
	<i>N</i> (%)	
Gender		.184
Female	118 (51.5)	
Male	14 (66.7)	
Primary practice setting		.232
Private	39 (59.1)	
Public	94 (50.5)	
Education/training level		.036
Bachelor degree or below	50 (45.5)	
Graduate Certificate or above	84 (58.7)	
Principle role		.576
Clinician	93 (51.7)	
Non-clinician	40 (55.6)	
Registration year		.071
≤1989	61 (59.2)	
≥1990	67 (47.5)	
Profession type		.279
Registered nurse	111 (51.4)	
Registered midwife	22 (61.1)	
Registered nurse speciality area		.379
Non-medical/surgical	75 (55.6)	
Medical/surgical	58 (50.0)	

Table S4. Perceived Relevance and Knowledge of Genomics Relevant to Nursing and Midwifery Practice by Profession Type

Question	Profession	Undecided	Not relevant	Somewhat relevant	Moderately relevant	Very relevant	Extremely relevant
How relevant do you believe genetics/genomics is to nursing and/or midwifery practice?	Registered nurse (N=216)	15.7%	3.7%	20.8%	22.7%	25.9%	11.1%
	Registered midwife (N=36)	0%	2.8%	13.9%	36.1%	36.1%	11.1%
	Profession	Unsure	Poor/limited	Average/fair	Good	Very good	Excellent
How would you rate your knowledge of genetics/genomics as it relates to nursing and/or midwifery practice?	Registered nurse (N=216)	4.2%	45.4%	35.2%	13.0%	1.9%	0.5%
	Registered midwife (N=36)	0%	19.4%	69.4%	8.3%	2.8%	0%

5.6 Summary

This chapter has presented the findings of a cross-sectional survey designed to measure the genomic literacy of Australian registered nurses and midwives. The survey indicated that the genomic literacy of Australian registered nurses and midwives is limited, despite nurses' and midwives' perception that genomics is relevant to nursing and midwifery practice. Although these findings provided a measure for genomic literacy, the nuances around nurses applying genomics in practice were not captured. To explore the ways in which nurses apply genomics in clinical practice, the author conducted semi-structured interviews with oncology nurses—a demographic selected for its increased prevalence of genomics. The findings of the semi-structured interviews will be presented in the following chapter.

Chapter 6: Findings—Genomics in Oncology Nursing Practice

6.1 Introduction

Genomics is becoming an everyday component of cancer care (Beamer, Linder, Wu, & Eggert, 2013), thereby creating the need for oncology nurses to learn about genomics and integrate genomic knowledge and skills into their specialty practice (Aiello-Laws, 2013). This chapter provides a description of genomics in the area of oncology nursing care. The chapter commences with a summary of advances in science and technology that are changing oncology practice, discusses genomic applications in oncology, and outlines genomic literacy as required by oncology nurses. The chapter also contains a manuscript (under review) describing how genomics is applied in oncology nursing practice in a regional hospital in Queensland, Australia. The manuscript will provide insight into how registered nurses apply genomics in oncology-based settings in Australia, summarising the genomic knowledge and skills of registered nurses, and the ways this knowledge and these skills are used in the delivery of patient care.

6.2 Cancer in Australia

Cancer is a major cause of illness in Australia. Cancer is responsible for the largest number of years of healthy life lost in Australia through premature death or disability, and most of the total cancer burden is due to premature death (Australian Institute of Health and Welfare, 2017). In 2017, it is estimated that 134,174 new cases of cancer will be diagnosed, and 47,753 people will die from cancer (Australian Institute of Health and Welfare, 2017). These figures indicate the burden of cancer in Australia, and support the need for genomically informed oncology nursing care. Cancer Australia is the Australian Government's national cancer control agency. Cancer Australia (2014, p. 290) works to translate evidence to inform policies and programs in cancer control, and to promote evidence-based practice to health professionals across Australia, each in an effort to minimise the effects of cancer. Funding for such initiatives is secured. The Australian Government will invest \$500 million over 10 years in an 'Australian Genomics Mission' to help save or transform the lives of more than 200,000 Australians through research into better testing, diagnosis and treatment (Australian Government, 2018a).

6.3 Genomics in Oncology Nursing Practice in Australia

Developments in science and technology are changing cancer care. New discoveries in sequencing, genetic molecular markers, genetic mutations and variants, genomic sequencing, risk-reduction methods and targeted therapies are together enhancing clinical practice (Boucher, Habin, & Underhill, 2014; Calzone et al., 2013; Flória-Santos et al., 2013). Clinical applications of cancer genetics and genomics now include identifying patient risk through assessing family history, directing screening and surveillance guidelines, facilitating genetic testing and counselling services, applying risk-reduction methods, creating treatment guidelines, administering and monitoring targeted therapies, and prognosis (Aiello-Laws, 2013; Boucher et al., 2014). Meeting the challenges that will accompany the increased burden of cancer will require oncology nurses to have a sound understanding of genetics and genomics (Boucher et al., 2014).

As the frontline caregivers of people diagnosed with cancer (Beamer et al., 2013), oncology nurses must understand the influence of genomics on cancer prevention, screening, diagnosis, treatment and survivorship—factors that are crucial to these health professionals providing quality cancer care. This position is reiterated by Cancer Australia’s (2014) statement recommending the ‘systematic implementation of evidence-based strategies for prevention, screening, early detection, diagnosis, treatment, supportive care, follow-up care, palliation and end-of-life care’ (p. 5). The literature indicates that genomics has an increased presence in oncology, in comparison with other nursing areas and specialties (with the exception of midwifery). The increased presence of genomics in oncology nursing led the researcher to undertake targeted research with nurses working in this specialist area.

6.4 Genomic in Oncology Nursing Practice: Interview Findings

Interviews were conducted in 2018 with the aim of exploring how genomics was applied in oncology nursing practice in a regional hospital in Queensland, Australia. The details of the study and the findings are summarised in the following section and reported in full in the manuscript that follows. While this publication presents the outcomes of this phase of the research as a stand-alone study, these findings also form a significant component of the larger case study presented in Chapter 7.

Chapter no.	Details of publication(s) on which chapter is based	Nature and extent of the intellectual input of each author, including the candidate
6	Wright, H., Zhao, L., Birks, M., & Mills, J. (under review). Genomics in oncology nursing practice in Australia. <i>Nurse Education Today</i> . Manuscript ID NET_2018_608.	Wright designed the study. Wright collected the data and performed the coding and data analysis with assistance from Birks. Wright wrote the first draft of the paper which was revised with editorial input from Zhao, Birks and Mills.

Though relevant to all areas of healthcare, genomics has a higher presence in oncology. The study described in this paper explored how genomics was applied in oncology nursing practice in a regional hospital in Queensland, Australia. Semi-structured interviews were undertaken with registered nurses working in oncology departments within a regional Queensland hospital in 2018 (*see Appendices 4-7*). This paper describes the four key themes that were identified: (i) genomic knowledge, (ii) applying genomics in practice, (iii) genomics relevance to practice and (iv) genomics education. As can be seen from this article, most participants believed their genomic knowledge was poor or average. Interestingly, while the participants believed that genomics is relevant to oncology nursing practice, many were unclear about how genomics can be applied other than in ‘targeted treatments’, and were not actively using genomic knowledge with any regularity, beyond obtaining a family history. The findings of this phase of the broader study indicate that oncology nurses are not sufficiently incorporating genomics into their practice.

Manuscript Details

Manuscript number	NET_2018_608
Title	Genomics in oncology nursing practice in Australia
Article type	Research Paper

Abstract

Purpose There is a new responsibility for all nurses to be knowledgeable of genomics and incorporate genomics into nursing practice. This study describes how genomics is applied in oncology nursing practice in a regional hospital in Queensland, Australia. **Methodological approach** Semi-structured interviews were conducted and analysed using thematic data analysis. **Participants and setting** Registered nurses working in oncology departments within a regional hospital in Queensland, Australia. **Findings** Nine semi-structured interviews were conducted. Three key themes were identified: (1) adequacy of knowledge for practice, (2) relevance of knowledge to practice, and (3) applying knowledge in practice. Most participants believe their genomic knowledge is poor or average. While most participants believe genomics is relevant to practice, many are not clear about how genomics can be applied other than 'targeted treatments', and are not actively using genomics with any regularity beyond taking a family history. **Conclusion** Most nurses have limited genomic knowledge and are applying genomics in practice in a reduced capacity that falls below current expectations for competent oncology care. **Implications for nursing** The findings of the study can be used to improve the utilisation of genomics in oncology nursing practice, which in turn may lead to better quality healthcare for patients and their families.

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Research Data Related to this Submission

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GENOMICS IN ONCOLOGY NURSING PRACTICE IN AUSTRALIA

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Ethical Approval

This project received ethical approval (HREC Reference number: HREC/17/QTHS/241)

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Title

Genomics in oncology nursing practice in Australia

Abstract

Purpose

There is a new responsibility for all nurses to be knowledgeable of genomics and incorporate genomics into nursing practice. This study describes how genomics is applied in oncology nursing practice in a regional hospital in Queensland, Australia.

Methodological approach

Semi-structured interviews were conducted and analysed using thematic data analysis.

Participants and setting

Registered nurses working in oncology departments within a regional hospital in Queensland, Australia.

Findings

Nine semi-structured interviews were conducted. Three key themes were identified: (1) adequacy of knowledge for practice, (2) relevance of knowledge to practice, and (3) applying knowledge in practice. Most participants believe their genomic knowledge is poor or average. While most participants believe genomics is relevant to practice, many are not clear about how genomics can be applied other than ‘targeted treatments’, and are not actively using genomics with any regularity beyond taking a family history.

Conclusion

Most nurses have limited genomic knowledge and are applying genomics in practice in a reduced capacity that falls below current expectations for competent oncology care.

Implications for nursing

The findings of the study can be used to improve the utilisation of genomics in oncology nursing practice, which in turn may lead to better quality healthcare for patients and their families.

Key words

Genetics, genomics, oncology, competence, relevance

Introduction

Advances in genetic and genomic science and technology are changing the way healthcare, including nursing care, is delivered (Alexander, 2017). Genomics informs clinical decision making by identifying risks for and manifestations of disease, the development of new therapies, and responses to interventions such as medications (Calzone, Kirk, Tonkin, Badzek, et al., 2018). The use of genomic information across the healthcare continuum impacts the care provided by all clinicians, especially nurses (Calzone, Jenkins, Culp, Bonham, & Badzek, 2013).

As the largest professional group in healthcare in Australia (Australian Institute of Health and Welfare, 2016b), nurses are well placed to incorporate genomic information into healthcare. Genomic healthcare requires a prepared workforce (Calzone, Kirk, Tonkin, Badzek, et al., 2018). However, the literature shows limited progress in the integration of genomics by the nursing workforce (Calzone et al., 2012). Most nurses remain inadequately prepared to translate genomic information into personalized healthcare (Calzone et al., 2013). Genomics has the potential to transform healthcare delivery by increasing quality and safety, decreasing costs and improving health outcomes (Alexander, 2017; Calzone, Kirk, Tonkin, Laurie Badzek, et al., 2018; McCormick & Calzone, 2016).

Genetic and genomic information is now being utilized in most healthcare settings (Bancroft, 2013; Seven, Akyüz, Elbüken, Skirton, & Öztürk, 2015). Though relevant to all areas of healthcare, genomics has a higher presence in oncology. Oncology nurses led the translation of genetics and genomics into clinical nursing practice (Eggert, 2017). Since that time genomics has begun to ‘revolutionize oncology care’ (Jenkins, 2011, p. 64) with genomics now central to the care of most oncology patients (Santos et al., 2013).

This paper will provide insight into how registered nurses apply genomics in oncology-based settings in Australia. The study findings will be used to create a ‘picture’ of genomics in oncology nursing practice in Australia, and how this knowledge and these skills are used in the delivery of patient care. This information may be used to improve the delivery of genomically-informed healthcare by registered nurses, and direct genomic education at the pre- and post-registration level.

Background

The terms ‘genetics’ and ‘genomics’ are well defined in the literature. Genetics can be defined as “the study of individual genes and their impact on relatively rare single gene disorders” and genomics as “the study of all the genes in the human genome together, including their interactions with each other, the environment, and the influence of other psychosocial and cultural factors” (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, pp. 8-9). Genomics is central to the care of most patients with cancer (Santos et al., 2013). Genomics is currently integrated into cancer care in terms of

aetiology; cancer risk assessment and genetic testing; genomics of tumour profiling, pharmacogenomics, and targeted cancer therapy; and cancer management (Jenkins, 2011). Oncology nurses are the ‘frontline caregivers’ of persons diagnosed with cancer (Beamer, Linder, Wu, & Eggert, 2013). Understanding the influence of genomics in cancer prevention, screening, diagnosis, treatment, and survivorship is necessary for nurses to provide optimal nursing care (Santos et al., 2013). Oncology nurses must include genomics into their nursing practice to provide competent, evidence-based care and to potentially improve patient outcomes (Jenkins, 2011). Competent care in oncology includes “taking a family health history, constructing a pedigree, identifying red flags for those at increased risk, providing appropriate patient education and counselling, referring patients to a genetics professional when appropriate, supporting patients through the informed decision-making and consent processes, and advocating for competent genetics services for patients” (Aiello, 2015, p. 204). Genomics adds a new and changing dimension to patient care (Eggert, 2017) that oncology nurses must be ready to adopt.

Cancer is a major cause of illness in Australia. In 2017, it is estimated that 134,174 new cases of cancer (excluding basal and squamous cell carcinoma of the skin) will be diagnosed in Australia, an average of 367 diagnoses each day (Australian Institute of Health and Welfare, 2017). In 2017, it is estimated that 47,753 people will die from cancer in Australia, an average of 131 deaths each day (Australian Institute of Health and Welfare, 2017). These figures reflect the magnitude and needs of cancer care in Australia, and support the need for genomically informed oncology nursing care.

Research addressing the integration of genomics into oncology nursing practice in Australia is limited. As such, it is unclear how Australian registered nurses are applying genomics in oncology nursing practice. This study is part of a larger research study in which the authors seek to gain a deeper understanding of (i) the genomic knowledge and/or skills employed by nurses; (ii) nurses’ perceptions of the relevance of genomics in nursing practice; and (iii) nurses’ experience of using genomics in ‘day to day’ nursing practice in terms of patient care and/or communication within the healthcare team.

Methods

Aim

This study will determine how genomics is applied in oncology nursing practice in a regional hospital in Australia.

Design

This study employed semi-structured interviews with registered nurses working in oncology-based units within a regional hospital in Queensland, Australia. The study was conducted over a six week period from January – February 2018.

Sample and setting

To be eligible for the study, participants had to be a nurse currently registered with the Nursing and Midwifery Board of Australia in Division 1 of the register. Nurses with dual registration (registered nurse/registered midwife) were eligible for inclusion however their role was limited to that of nursing for the purpose of the study. The nurses also had to work in oncology-based unit/s at the hospital. Oncology-based units included 'inpatient' and 'outpatient' services such as the oncology ward, ambulatory oncology unit, haematology and bone marrow therapy unit, and radiation oncology unit. Palliative care units were included in the study as they were deemed to provide significant oncology-based nursing care to their patients however no participants from this specialty volunteered to participate. Nurses caring for an oncology patient outside an oncology-based unit were not eligible for inclusion in the study. Participants were recruited using flyers, a short presentation (or 'in-service') delivered by the researcher and an email invitation to the Nurse Unit Managers with the request that it be forwarded on to nursing staff. Flyers were also placed at various locations around the hospital. Nurses interested in participating in the study were asked to contact the principal investigator via the contact details provided.

Data collection

Data collection commenced following approval from the Human Research Ethics Committees of the Health Service and University. Participants were interviewed by the lead author at a time and location (within the hospital grounds) suitable to the participant. Interviews were conducted in the participants own time and were generally 30-60 minutes duration. An interview guide was used. This interview guide contained a framework that allowed the researcher to pursue alternate questions and to allocate more time to some questions than to others. The interviews were recorded and transcribed by a professional transcription service. Nine interviews were required to reach data 'saturation' or the point at which no new information or themes are observed in the data (Guest, Bunce, & Johnson, 2006). Data analysis was performed concurrently with data collection (interviews).

Data analysis

The researchers used the stages of coding and analysis described (Braun & Clarke, 2013, pp. 202-203), these are (1) transcription (for interviews only), (2) reading and familiarisation, (3) coding, (4) searching for themes, (5) reviewing themes, (6) defining and naming themes, and (7) writing. Individual coding was informed by using first and second cycle coding methods (Saldana, 2016). The lead author was responsible for initial coding and theme generation, with the resulting themes reviewed by the research team. The computer-assisted qualitative data analysis software program NVivo™ was used to assist with thematic data analysis.

Findings

Demographics

Nine registered nurses participated in this research. Eight participants were female and one male. One participant worked in the oncology ward and the remaining participants worked in other oncology related units including oncology day unit, radiation oncology, and clinical trials unit. Five participants were aged 44 years or younger and four participants were aged 45 years or older. Year registered ranged from 1986-2017 with years working in an oncology setting (full time and part time inclusive) ranging from one year to 19 years. Three participants had post-graduate qualifications in oncology nursing and one participant was currently enrolled in same. No participants had post-graduate qualifications and/or specialist training in genetics or genomics.

Themes

Three key themes were identified: (1) adequacy of knowledge for practice, (2) relevance of knowledge to practice, and (3) applying knowledge in practice.

Theme 1: Adequacy of knowledge for practice

Participants were asked to rate their genomic knowledge. Most participants reported their genomic knowledge as poor or average with only a few participants reporting their genomic knowledge as good. No participants reported their genomic knowledge as very good or excellent.

Despite reporting low genomic knowledge levels, several respondents believed that their current knowledge of genomics was sufficient. One participant stated their genomic knowledge was “[adequate] for day to day practice” (P3). Even amongst those that believed they had adequate genomic knowledge, there was the suggestion that it could be better, as one participant reported “[adequate] for our current context, yes. But...I think it could be a lot better” (P2). Other participants indicated that their genomic knowledge and/or skills was less than sufficient or “... probably not as adequate as what it could be” (P1). Regardless of the adequacy of their genomic knowledge, many participants acknowledged that patient care could be improved through enhanced genomic knowledge. Statements such as “I think my practice would be improved with a deeper knowledge of genetics and genomics” (P2) and “when diagnosing tumours and things like that, knowing what it’s positive to and what it’s not, so that we’re able to target it better and we’re able to provide better outcomes for our patients” (P1) suggest that participants were aware of the impact that improved genomic knowledge could have on patient care.

Few participants reported genomics as being a part of their nursing education. For these participants, genomics was presented as biology. No participants reported learning about genomics beyond the basic biological concepts and/or terms. As one participant stated “I’m sure we did something in the nursing

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science and biology section of it [undergraduate nursing degree]” (P5). Other participants stated that the genomic content was basic saying that they “*did not learn anything other than basic chromosome stuff*” (P9). One participant indicated that they may have learnt about genomics without acknowledging it at the time, for example “*I probably have learnt a bit about it, and I probably know some more stuff about it, but I don’t know how to harness that and link that as being genetics and genomics...*” (P4). No participants reported learning about genomics in terms of its value within the healthcare continuum.

Most participants were interested in learning more about genomics. These participants were more interested in informal education than formal post-graduate programs. ‘In-services’ and workshops were the most commonly requested mode for learning. Several participants indicated they would be willing to attend education sessions in their own time. Attending education sessions can be challenging. Participants reported several barriers to further learning such as lack of personal time, finances, and availability during workhours. As one participant stated “*...sometimes the logistics of getting there [to education sessions] doesn’t work out*” (P4). Participants wanted to learn about genomics that was relevant to their practice and delivered at an appropriate level. As one participant stated “*there’s no point learning things that you don’t see. We want things that we can relate to the patients...*” (P6) and “*tell us about it all at a level we understand*” (P6). In terms of genomics improving their practice, one participant stated “*if we had some deeper knowledge about that, we might understand more so the implications for people...*” (P2).

Theme 2: Relevance of knowledge to practice

Most participants stated that genomics was relevant or very relevant to nursing practice. One participant said “*I think it’s very relevant to practice. We are moving towards individualized medicine and care. So, that’s quite reliant on our understanding of people’s genetic profile, and as a whole, the genomics. So, I think it’s very relevant*” (P2). A few participants believed genomics was not relevant to nursing practice with two participants stating that “*in terms of day to day nursing I think genetics doesn’t even cross my mind*” (P2) and “*I’m more worried about dealing with the problems a patient has got that are physical...*” (P3). Similar responses were seen in regard to importance of genomics in nursing practice. As one participant said “*if it’s relevant, it’s got to be important*” (P2). However, the importance of genomics may be undervalued, as indicated by one participant who said “*...as nurses, we probably don’t appreciate the importance of genetics and genomics...*” (P2).

Several participants view genomics as the responsibility of the doctor, nurse specialist or senior nurses. The responsibility to use genomics appears to align with participants’ views towards how genomics is used in healthcare. Participants reported targeted treatments as the main means to incorporate genomics in practice however some nurses believe that they have no input into targeted treatments. Two participants stated “*That’s not nursing; that’s doctor*” (P3), “*We can’t dictate anything. We’ve chosen a*

career path that doesn't allow that" (P3) and "Because they're [the doctors] the ones who prescribe the care..." (P9). Similar findings were seen for specialist nurses, for example "I think probably them [breast care nurse], because they have a lot more to do with the patients..." (P5) and "Prostate [nurses], yeah. The [clinical] trial nurses, yeah. Not to my own [practice] – what I do." (P6). Other participants believed genomics was the responsibility of senior nurses for example "I think, to the lower level nurse, not that important ... versus if you were a nurse practitioner or a CNC - someone whose ... at a higher level..." (P3). These statements show that many nurses see genomics as the responsibility of others.

Participants' perceptions of genomics and its role in oncology nursing practice and the wider nursing profession are varied. The general tone is one of dubious optimism. One participant stated "I think you've got an uphill battle on your hands to make this [genomics] seem like a priority to nurses" (P3). This sentiment is mirrored in terms of this individual research study itself, with two participants stating "I'd be interested to see how you sell this" (P3) and "I don't think that there's really anyone – apart from you, but I mean there's not a great deal of a push here to learn about it, and I think it's something that probably should be..." (P4).

Theme 3: Applying knowledge to practice

Participants' reports on the frequency with which they use genomics in nursing practice is varied. Most participants reported using genomics daily or weekly. Two participants reported rarely using it. For some participants, genomics is not a part of daily practice, as they stated "I don't think genomics came into play at all" (P3) and "It's not particularly nursing practice" (P8).

Targeted treatments or 'precision medicine' was by far the most commonly cited application for genomics in healthcare. Participants appreciated the use of genomics in directing treatment, as indicated by statements such as "cancers ...are often treated based on their genetics or genomics" (P7) and "The more information you have, the more tailored you can provide your treatment and your care" (P2). Participants overwhelmingly stated that improved treatments (targeted treatments) is the main way genomics improves patient care. Many participants reported using genomics in their own nursing practice via targeted treatments, for example "...we started him [patient] on a certain treatment...once his genetic status was known, we dropped one part of the treatment which wasn't indicated for that certain type of mutation..."(P2). Other participants reported using genomics in their nursing practice by taking a family history, for example "...I suppose taking family histories of patients who have prostate cancer or something in their family" (P1) and "...you usually take a history of patients and stuff..." (P5). Some participants articulated the familial risk associated with certain cancers in her/his statement "...[woman] had breast cancer. She's got two daughters...she was worried about her children" (P6). Participants also spoke about the psychosocial aspect of patient care, and the use of genomic information to allay patient fears and provide reassurance and counselling, for example "...some people who do have

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that genetic mutation... it can be quite daunting knowing that this could happen to someone else in their family....being compassionate... it could be a bit of psychology as well” (P4).

Some participants reported patients asked them genomic-related questions about their care, frequently based on information available in the public domain and/or media, for example *“Oh yes. As soon as something comes on the telly, I can say to my husband, I’m going to get people all the time, yeah, all day long tomorrow or whatever. That happens all the time” (P7)* and *“Yeah, they do, especially if people...[see] these wonder drugs and this is happening or they see new trials, and they come and ask us about that...” (P8)*. Participants believed they were rarely able to answer patient questions related to genomics satisfactorily. One patient reported referring the patient to the doctor *“No...and a lot of the time it was like – well, just ask the doctor – because I didn’t know” (P4)*. Another participant reported providing simplistic answers such as *“Well I can answer from a child point of view” (P7)*.

Some participants believed that genomics would not change the care delivered, as indicated in the statements *“I don’t know if it impacts what I do for them, you wouldn’t treat them any differently ...” (P5)* and *“It doesn’t make any difference to how I treat the patient, which is doing the doing that has to be done. ...I don’t think it changes my day-to-day work in that regard...” (P9)*.

Discussion

This study is the first to report in-depth findings regarding registered nurses applying genomics in oncology nursing practice in Australia, therefore comparisons will be limited. Study demographics are comparable to Australian nursing workforce statistics with participants predominantly female and in higher age ranges (Australian Institute of Health and Welfare, 2016a). International studies addressing genomic literacy and/or competency had similar demographics in terms of gender and age, as did a recent Australian study (Wright, Zhao, Birks, & Mills, 2018).

Participants’ familiarity with the term ‘genetics’ indicates that it has made its way into nursing discourse. Conversely, no participant was able to adequately describe ‘genomics’, indicating that its “broader and more ambitious reach” (Guttmacher & Collins, 2002, p. 1512) is unrecognized. Participants’ knowledge is consistent with other genomic literacy studies. Studies that assessed actual (Calzone et al., 2013; Calzone, Jenkins, Culp, Caskey, & Badzek, 2014; McCabe, Ward, & Ricciardi, 2016; Read & Ward, 2016; Seven et al., 2015) and perceived (self-reported) genomic knowledge (Calzone et al., 2013; Calzone et al., 2014; Calzone et al., 2012) largely reported nurses’ knowledge as limited. In spite of this lack of familiarity with the term, most participants in this study were interested in learning more about genomics.

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Oncology is a rapidly changing specialty. The development and incorporation of genetic/genomic information and technologies into oncology patient care requires individual registered nurses and the existing nursing workforce to continuously update their knowledge (Eggert, 2017). Most nurses receive minimal genomics education (Bancroft, 2013) with genomics rarely addressed beyond the basic biological concepts and/or terms. Most applications of genetics to oncology patient care and other specialties has occurred since 2003 following the complete sequencing of the human genome, therefore more recent discoveries such as epigenetics, molecular signaling, and targeting present in modern oncology will be new to many working in the specialty (Beamer et al., 2013). Improving genetics education for nurses and all health professionals is essential if they are to fully integrate genetic/genomic advances into everyday patient care (Bancroft, 2013).

The high perceived relevance and/or importance of genomics to nursing is similar to that reported in several other studies. Calzone et al. (2012), Calzone et al. (2013) and Calzone et al. (2014) all reported that the majority of nurses believed genomics was important to nursing practice, a finding similar to that recorded by Godino, Turchetti, and Skirton (2013) where most nurses believed genetics was relevant to their daily work. Negative participant comments are perhaps most revealing. That genetics does not even 'cross the mind' of some participants is concerning. That participants are only interested in a patient's immediate physical problems shows that there is a lack of understanding of the role genomics plays in terms of patient physical and emotional health.

Health professionals including nurses tend to consider genetics as a medical specialty only, and not as a part of their daily practice (Flória-Santos et al., 2013). The study findings indicate that many nurses believe that they have limited or no input into decisions about a patient's care and therefore see genomics as relevant only to those that do, that being doctors and specialist nurses. It is important that oncology nurses recognize the boundaries of their knowledge and refer to another colleague with expertise in a particular area (Eggert, 2017) however many registered nurses believe all genomics falls outside their practice. Advances in medicine mean it is likely that genetic information will be integrated into routine health assessments (Bancroft, 2013). This routine integration will "move genetics away from being a specialty and to become a standard part of the care pathway" (Bancroft, 2013, p. 560).

Integrating genomics into the nursing profession requires nurses to recognise its applications in clinical nursing (Hickey et al., 2018). The study findings show that participants recognised the various applications of genomics in oncology nursing practice, especially targeted treatments. Targeted treatments was by far the most commonly cited application for genomics in healthcare. The role of genomics in cancer must be understood in the broader context of the 'personalised medicine' era now frequently referred to as 'precision medicine' (Otlowski, 2016). Precision medicine is a new term and concept in health care that "designs treatment and diagnostics based on individual needs and genetics

and genomics of patients” (Eggert, 2017, p. 14). Precision medicine is moving healthcare from a “one-size-fits-all” approach (Hickey et al., 2018) to more ‘tailored’ care.

Participants’ reports on the frequency with which they used genomics in nursing practice is varied. Assessing genomic utilisation in practice is challenging given its varied applications. International studies have shown that nurses believe family history to be important so it is not surprising that this is the most common means by which participants incorporated genomics into their practice. Coleman et al. (2014) reported that over half of respondents believed family history was important to nursing, and Pestka, Meiers, Shah, Junglen, and Delgado (2012) and Pestka, Meiers, Shah, Junglen, and Delgado (2013) each reported nurses as having a positive perception of the use of family pedigrees in practice. Calzone et al. (2012) used family history assessment as a specific measure of genomic utilization. They found that in the three months prior to the study, only 3% (n = 4/148) of nurses reported always taking a family history, while 77% (n = 114/148) had rarely or never assessed a family history. This is consistent with later findings by Calzone et al. (2013) where 6% (n = 22/359) of nurses always reported taking a family history, while 60% (n = 216/359) had rarely or never assessed a family history in the three months prior to the study.

Genomic information and technology has the potential to improve healthcare outcomes, quality, and safety, and reduced costs (Alexander, 2017; McCormick & Calzone, 2016). Nursing is moving forward into “new and unfamiliar territories of genomics” (Emon, 2014, p. 3) and we need to be current in our practice to ensure quality and safe patient care. The nursing profession needs to make a concerted effort to improve the use of genomics in oncology nursing practice to provide the best outcomes for patients and their families.

Limitations and recommendations

This study had some limitations. There were a small number of interviews conducted. Recruiting participants to a research study can be challenging, especially in an unfamiliar subject area such as genomics, and this may have reduced participation in the interviews. It is recommended that future studies be conducted as part of an ‘education initiative’ to raise awareness about the topic and improve engagement with the study.

The authors elected to interview nurses working in oncology-based settings. Genomics has a higher presence in oncology therefore it is likely that nurses working in oncology would be more familiar with the terms genetics and genomics, incorporate genomics into their practice, and have opinions about relevance and/or utility of genomics in nursing practice, than their colleagues working in other nursing specialties. The findings of oncology nurses may therefore not adequately reflect those of the wider nursing profession. It is recommended that future studies be conducted in various clinical settings. The

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findings can then be compared across individual clinical specialties and subsequently combined to create a full picture for the entire nursing profession.

Implications for nursing

Genomics is central to the care of most patients with cancer (Flória-Santos et al., 2013) and yet it does not appear to be central to the practice of current oncology nurses. The increasing significance of genomics across the oncology care continuum requires nurses to improve their genomic knowledge and apply this knowledge to patient care to achieve better patient outcomes.

Conclusion

Study findings indicate that oncology nurses are applying genomics in practice though in a reduced capacity. Nurses' limited application of genomics in practice may be the product of a lack of understanding as to what constitutes 'actionable genomics'. Genomics is becoming an everyday component of cancer care (Beamer et al., 2013) and oncology nurses must accept and incorporate genomics into their practice if they are to provide competent, evidence-based nursing care.

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6.5 Summary

This chapter has addressed genomics in oncology, including an overview of new technologies that have advanced practice, as well as the various applications of genomics in oncology nursing practice. This chapter has also presented an article submitted for publication that describes the findings of semi-structured interviews undertaken with oncology nurses. This paper discussed how genomics is applied in oncology nursing practice in a regional hospital in Queensland, Australia. These findings will be collated with the survey findings presented in the previous chapter as a combined 'case' in Chapter 7 of this thesis.

Chapter 7: Discussion

7.1 Introduction

The aim of this study was to determine how Australian nurses engage with genomics in nursing practice. A single-case, holistic, critical realist case study was used to examine the ‘case’ of genomics in nursing practice within the context of the Australian healthcare setting. The preceding two chapters outlined the findings from the cross-sectional survey and semi-structured interviews. The findings of the individual studies outlined in these chapters will be combined to produce a description of the ‘case’ of Australian nurses’ engagement with genomics in practice. This chapter presents a discussion of the case in the context of the literature. Three categories—*point of learning*, *point of reference* and *point of care*—will be used as a framework to describe the case. Building on this discussion, recommendations for improving nurses’ engagement with genomics, along with suggestions for future research, will be presented in the following chapter.

7.2 The Case

A case study ‘investigates a contemporary phenomenon (the “case”) in depth and within its real-world context’ (Yin, 2014, p. 16). More simply stated by Anthony and Jack (2009), a case study allows for comprehensive research into complex issues. Therefore, this case study is the comprehensive investigation of genomics in nursing practice within the Australian healthcare setting, conducted with the aim of determining how Australian nurses engage with genomics in practice. A single-case holistic study design was used in this research. Data were collected via a cross-sectional survey of Australian registered nurses and midwives, and semi-structured interviews with a selected subset of Australian registered nurses working in oncology-based units at a regional hospital in Queensland. Key case findings were generated using thematic analysis, and these are presented in Figure 7.1. As stated in Chapter 1, the term ‘genomics’ is largely used throughout this thesis to refer to both the study of single genes (genetics) and the study of an individual’s entire genetic makeup (genome). Exceptions to this rule occur when it is necessary to differentiate between the terms, such as where the more familiar term ‘genetics’ is most appropriate, or in reference to the literature where a particular term is considered more consistent with the author’s intentions.

Cross-sectional survey	Semi-structured interviews	Elements of the case description
<ul style="list-style-type: none"> • Nurses and midwives believe genomics is relevant to nursing and/or midwifery practice • Nurses' and midwives' genomic knowledge is poor or average • Nurses and midwives are most knowledgeable about mutations and genomic healthcare applications • Nurses and midwives have the least knowledge in the area of human genome basics 	<ul style="list-style-type: none"> • Genomics is rarely incorporated into nursing education beyond simple biology • Nurses believe genomics is relevant to nursing practice • Genomics is seen as the responsibility of the doctor, nurse specialist or senior nurses • Nurses' genomic knowledge is poor or average; however, most believe it is sufficient for practice • Nurses are applying genomics in practice infrequently and in a reduced capacity 	<ul style="list-style-type: none"> • Genomics education in nursing is inadequate and, as a result, nurses' genomic knowledge is limited • Nurses believe genomics is relevant to nursing practice; however, they are unclear about their roles and responsibilities • Nurses are not adequately incorporating genomics into nursing practice and are subsequently not achieving genomic competency

Figure 7.1: Key Findings for the Case of Australian Nurses' Engagement with Genomics

While the survey described in Chapter 5 gathered data from both the nursing and midwifery professions, the focus narrowed to nurses specifically in the interviews and related findings discussed in Chapter 6. Consistent with the aims of this research, elements of the case description presented in Figure 7.1, as discussed in this chapter, relate specifically to nursing.

Figure 7.2 summarises the elements of the case description presented in Figure 7.1 into three categories: *point of learning*, *point of reference* and *point of care*. *Point of learning* refers to education and its associated nuances, such as what is learnt and how nurses are learning (the means or mode of learning). *Point of reference* refers to the roles and responsibilities or professional expectations outlined in nursing policy and procedures. *Point of care* refers to clinical practice and how nurses are incorporating genomics into nursing care. These three categories (Figure 7.2) will be used as a framework to describe the case of Australian nurses' engagement with genomics.

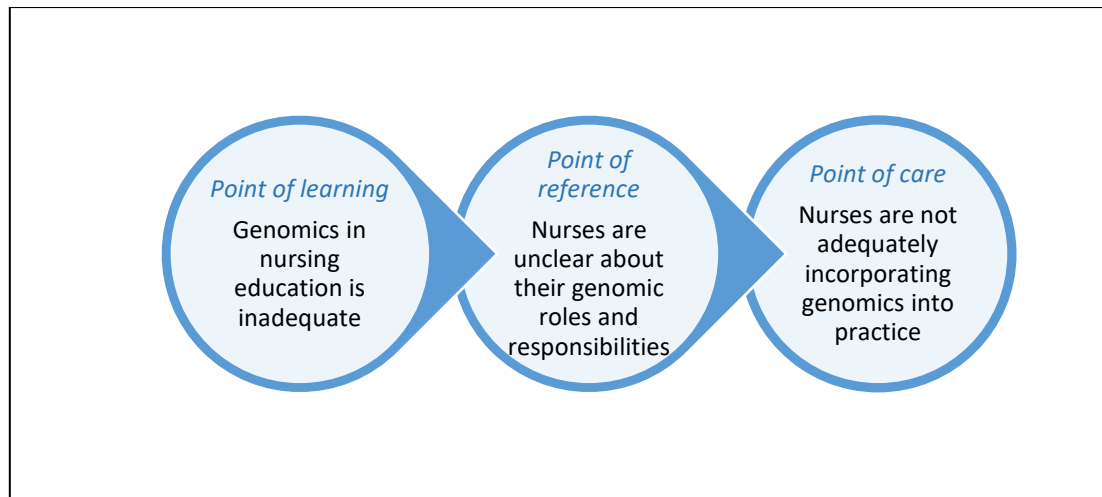


Figure 7.2: Relationship between Genomic Education, Roles/Responsibilities and Competency

As suggested by Figure 7.2, there is a logical linear relationship between these three elements, in that inadequate genomics education has led to nurses being unclear about their genomic roles and responsibilities, and nurses subsequently not demonstrating genomic competency through incorporation of genomics into their practice.

7.2.1 Point of Learning

7.2.1.1 Key Case Finding: *Genomics in Nursing Education in Australia is Inadequate*

The integration of genomic information and skills into nursing practice is contingent on the inclusion of genomics in nursing education programs. Including genomics at this *point of learning* is necessary to equip nurses with the knowledge and skills they need to incorporate genomics into their practice. The case is made throughout this thesis that genomics in nursing education in Australia is inadequate. Genomics is not being adequately addressed in Australian nursing curricula, with nursing students rarely learning about genomics beyond the basic biological concepts and/or terms, and reference to healthcare applications is largely absent.

The call to integrate genomics into nursing education is not new. Brantl and Esslinger (1962) addressed the genetics implications for the nursing curriculum in 1962, arguing that:

Contemporary nurse educators would not consider teaching students the nursing care of patients with neuropathology without first having taught basic neurophysiological concepts. Why, then, should we not consider it of equal importance to prepare students in basic genetics when teaching care of patients with inheritable disabilities and disease processes? (p. 91)

Their point has particular resonance in the post-HGP era. The HGP was an international research initiative with the aim of sequencing the entire human genome (National Human Genome Research Institute, 2016). It produced a ‘human blueprint for health’ showing that many common diseases have a genetic basis (National Human Genome Research Institute, 2016). It is now known that chronic diseases (such as arthritis, asthma, back problems, cancer, chronic obstructive pulmonary disease, cardiovascular disease, diabetes and mental health conditions) are caused by multiple factors, including a person’s genetic makeup, as well as lifestyle and environment (Australian Institute of Health and Welfare, 2016). This is a salient point, since around one in two Australians have a chronic disease, and around one in five have multiple chronic diseases (Australian Institute of Health and Welfare, 2015), thereby positioning chronic disease as the leading cause of illness, disability and death in Australia. This growing understanding of the genetic/genomic contribution to common diseases is increasingly facilitating genetic and genomic medicine entry into all areas of healthcare (Battista, Blancquaert, Laberge, van Schendel, & Leduc, 2012), meaning that all nurses will at some stage care for a person with a condition that is inherently genetic or has a genetic component.

Since the publication of Brantl and Esslinger’s (1962) seminal work, various individuals, consensus panels and organisations have promoted genomics education in nursing. The Consensus Panel on Genetic/Genomic Nursing Competencies (2009) argued that ‘each nursing curriculum preparing registered nurses for practice (at any and all levels) should include genetic and genomic learning experiences’ (p. 38). The National Coalition for Health Professional Education in Genetics (2007) has called for all healthcare professions to integrate genetics content into ongoing education. Publications addressing the integration of genetics and genomics into nursing curricula (Daack-Hirsch et al., 2011; Garcia et al., 2011)—especially those addressing faculty readiness (Jenkins & Calzone, 2012; Read & Ward, 2016; Williams et al., 2011)—have appeared frequently in the literature. These publications continue the call for the integration of genetics/genomics into nursing education.

These frequent calls for the integration of genomics into nursing curricula are largely going unanswered, with genomics still not fully integrated into nursing education (Kirk, Calzone, et al., 2011). Curriculum and textbooks do not include adequate genetic content, and the genetic content that does exist is generally grouped with information about maternal and child health, and lacks information about ethics in relation to genetic care (Aiello, 2017). This restricted

genomic content does not reflect the scope of genomics in healthcare. Genomics is fundamental to nursing care and should be ‘woven into the fabric’ of the nursing curriculum (Skirton, 2017, p. 401), rather than isolated among select nursing specialties, where complexities such as the ethics, legal and social implications are largely ignored.

Incorporating genetics into the undergraduate curriculum has its challenges. These challenges include contracting curricular time (curriculum crowding), limited access to education opportunities, inadequate resources in the educational institution (Calzone et al., 2018a; Williams et al., 2011), lack of genomic competency assessments (Calzone et al., 2018a) and low genetic literacy among faculty staff (Calzone et al., 2018a; Secretary’s Advisory Committee on Genetics Health and Society, 2011). Other challenges stated by the Secretary’s Advisory Committee on Genetics Health and Society (2011, p. 9) include minimal representation of genetics on certifying examinations; limited numbers of training experiences that incorporate genetics; a lack of evidence-based practice guidelines; and the false perception that genetics entails only rare genetic disorders, as opposed to more common disorders. Although several challenges exist, curriculum crowding and faculty confidence are the reasons most frequently cited for the continued exclusion of genetic content from nursing curricula.

Genomics competes with other evolving content areas for limited space in the nursing curriculum (Prows, Glass, Nicol, Skirton, & Williams, 2005, p. 198) and is frequently excluded from nursing curricula in favour of more traditional nursing topics. Similarly, genomics is considered by many as a specialty area and subsequently viewed as an unnecessary component of standard curriculum content. This theme was reflected in the current study’s qualitative findings, where nurses perceived genomics to be the responsibility of the doctor, nurse specialist or senior nurse. There is no disputing that genomics nursing *is* a specialty practice. However, while genomics nursing is a specialty, this does not mean that generalist nursing is void of genomics knowledge, skills and technologies. Cardiovascular/cardiothoracic nursing is a specialty, yet all patients have a heart; therefore, all nursing curricula prepare nurses to have the required basic knowledge and skillsets to care for all patients. The same logic applies to genomic nursing. Genomic nursing is a specialty, yet all patients have a genomic profile; thus, all nurses require basic genomics knowledge and skillsets to care for all patients.

Nurse educators’ lack of confidence in their knowledge of genomics presents a more fundamental problem: ‘Faculty cannot teach what they do not know’ (Ward, 2011, p. 39). Consequently, nursing programs frequently include limited genetic/genomic content. Studies

have shown that many nursing faculty are uncomfortable with genomic content (Read & Ward, 2016) and that current nursing faculty received little formal genetic education themselves, resulting in the potential for them to feel unprepared to teach genetic content (Sharoff, 2015).

In the *Genetics/Genomics in Nursing and Midwifery: Task and Finish Group Report* to the Nursing and Midwifery Professional Advisory Board, Kirk, Campalani, et al. (2011) presented a vision that ‘prepares a pathway to take the professions forward to embrace current and future developments in genomic healthcare’ (p. 9). The vision is that every nurse and midwife:

- (i) recognises and acts on the importance of genetics/genomics in the care they provide to people and families
 - (ii) is competent to a minimum standard in genetics/genomics through education provision
 - (iii) recognises that genetics/genomics is important and relevant because of the implications
- (Kirk, Campalani, et al, 2011, p.9).

Advances have been made. There is movement away from mere justification of genomics education to considering how best to deliver education and training (Tonkin, Calzone, Jenkins, Lea, & Prows, 2011, p. 331). The nursing professions in the US and UK are making steps towards the integration of genomics into their nursing curricula. Daack-Hirsch et al. (2011) and J. Jenkins, Prows, Dimond, Monsen, and Williams (2001) provided recommendations to achieve this goal in these countries. However, the same cannot be said of Australia, where there has been no clear contribution towards the advancement of genomics in nursing education. As discussed in Chapter 1, a desktop analysis was conducted by the current author in 2016. The author accessed the public websites of 34 universities, colleges and institutions that offered a nursing course in Australia. The words ‘genetics’ or ‘genomics’ did not appear in any subject titles. These terms were present in the aims and/or synopses of 10 subjects, and in the learning outcomes or specific content of 16 subjects. Genetics or genomics appeared in the content of one or more subjects at 15 of these universities. In cases where genetics was visible in a subject (whether in the aim, synopsis, learning outcomes or specific content), it was generally related to physiology and pathophysiology.

Multiple resources are available to assist with the integration of genomics into nursing education. The Global Genomics Nursing Alliance—frequently referred to as G2NA—is a global genomic resource initiative (Calzone et al., 2018b). The G2NA’s vision is ‘to serve as the unified international voice for advancing and integrating genomics into nursing practice’ by supporting nurses to realise their full potential to improve healthcare for all (Global

Genomics Nursing Alliance, 2018). The genomic resources that already exist are not readily accessible or discoverable to the international nursing community and are subsequently underused (Calzone et al., 2018b). Genetics has begun to feature in a few medical/surgical and anatomy/physiology-related nursing texts. For example, the latest edition of *Medical-Surgical Nursing: Critical Thinking for Person-Centred Care* (LeMone, 2014) has increased its genetic content and includes a valuable short chapter titled ‘Genetic Implications of Adult Health Nursing’, along with a series of ad-hoc textboxes titled ‘Genetic Considerations’ scattered throughout the text. Similarly, the latest version of *Smeltzer and Bare’s Textbook of Medical-Surgical Nursing* (Farrell, 2017) includes an excellent genetics chapter. However, despite these examples, there remains inadequate genetics/genomics information in general nursing textbooks. Texts devoted exclusively to the genetic aspects of nursing have been appearing for at least a decade, such as *Lashley’s Essentials of Clinical Genetics in Nursing Practice* (Lashley, Kasper, & Schneidereith, 2016), *Genetics and Genomics for Nursing* (Kenner & Lewis, 2013) and *Genetics and Genomics in Nursing and Healthcare* (Beery & Workman, 2012). However, such specialised texts are generally not prescribed in undergraduate nursing programs.

There is a call for all healthcare professionals to be appropriately prepared to integrate genetic and genomic knowledge into their practice (Sharoff, 2017). The Australian Government recently published the *National Health Genomics Policy Framework 2018–2021* (Australian Health Ministers’ Advisory Council, 2017a), which presents a shared commitment to leveraging the benefits of genomics in the health system for all Australians. Their vision is to help Australians live longer and better by integrating genomics into the health system through taking coordinated action across agreed strategic priority areas. It has five priority areas, the second of which is to build a skilled workforce that is literate in genomics, advising that ‘Upskilling the workforce through increasing capacity and capability in genomics and bioinformatics is necessary to effectively and efficiently support improved health outcomes for the individual and population’ (Australian Health Ministers’ Advisory Council, 2017b, p. 7). The framework shares a similar aim to that proposed by the authors of the *Genetic/Genomic Nursing Competencies*, which is to prepare the nursing workforce to deliver competent genetic- and genomic-focused nursing care (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 7). Such preparation is contingent on adequate preparation at the *point of learning*.

As a profession, we need to prepare for ‘the reality of tomorrow and not only for the needs of today’ (University of Glamorgan and University Hospital of Wales, 2003, p. 6). Nurses currently do not receive sufficient genetic and genomic education (Aiello, 2017)—an assertion supported by the case findings. Practice and curriculum change requires the commitment of nursing leaders and academic faculty to develop a long-term plan to incorporate genetic and genomic information to improve the public’s health (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009).

7.2.2 Point of Reference

7.2.2.1 Key Case Finding: Nurses are Unclear about Their Genomic Roles and Responsibilities

There is a responsibility for all nurses to be knowledgeable about genetics and genomics, and to incorporate genomics into their nursing practice (Camak, 2016). This responsibility extends to all registered nurses, regardless of academic preparation, practice setting, role or specialty (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). To meet this new responsibility, nurses require a clear ‘point of reference’. In general terms, a point of reference is defined as ‘something which you use to help you understand a situation or communicate with someone’ (Collins English Dictionary, 2018, para.1). In the nursing context, the point of reference for nurses to understand their ‘situation’ (expectations of registered nurse practice) is the professional practice documents issued by governing bodies. These documents take many forms, yet are significant in establishing and regulating the scope of nursing practice (Birks, Smithson, Lindsay, & Davis, 2018). One such document developed by the NMBA (2016) is the *Registered Nurse Standards for Practice*. These standards are universal to all registered nurses in Australia and do not align with specific nursing specialties. Standards for practice are described as the ‘expectations of registered nurse practice’ (NMBA, 2016). Standards for practice: (i) inform the educational standards for registered nurses; (ii) inform the regulation of nurses and determination of the nurse’s capability for practice; and (iii) guide consumers, employers and other stakeholders about what to reasonably expect from a registered nurse, regardless of the area of nursing practice or years of nursing experience. Fundamentally, these standards address what registered nurses do in their practice.

The findings of this case study indicate that Australian nurses are unclear about their genomic roles and responsibilities or expectations for practice. In terms of genomics nurses are unclear

about what registered nurses can do in their practice. As stated, the NMBA (2016) *Standards for Practice* are universal to all registered nurses in Australia, and are thus intentionally generic. However, it is likely that the generic nature of the NMBA *Standards for Practice* contributes to nurses' inability to appreciate the relevance of genomics to nursing practice. Some examples of this relevance are quite obvious. For example, the NMBA (2016) *Standards for Practice* state that registered nurse practice is person centred and evidence based. Genetics/genomic information is personal (pertaining to an individual) and contributes to the current evidence base for practice. Therefore, nurses must incorporate genomic knowledge and skills into their nursing care if they are to meet the 'expectations of registered nurse practice' outlined in the NMBA *Standards for Practice*. More specific illustrations of the relevance of the generic standards for practice can be explicated through a comparison with those documents that establish the standard for genomic competence. For example, there is a clear alignment between the NMBA's (2016) *Standards for Practice* and the *Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators* (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). This alignment is illustrated in Table 7.1, which displays the individual NMBA *Standards for Practice* and exemplar competencies taken from the *Essentials of Genetic and Genomic Nursing* document.

Table 7.1: NMBA Registered Nurse Standards for Practice and Alignment with Exemplar Essentials of Genetic and Genomic Nursing Competencies

NMBA (2016) Registered Nurse Standards for Practice	Consensus Panel on Genetic/Genomic Nursing Competencies (2009) Essentials of Genetic and Genomic Nursing
1. Thinks critically and analyses nursing practice	Accesses, analyses and uses the best available evidence, which includes research findings, for safe, quality practice Develops practice through reflection on experiences, knowledge, actions, feelings and beliefs to identify how these shape practice
2. Engages in therapeutic and professional relationships	Collaborates with healthcare providers in providing genetic and genomic healthcare
3. Maintains the capability for practice	Demonstrates an understanding of the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment and monitoring of treatment effectiveness
4. Comprehensively conducts assessments	Conducts comprehensive health and physical assessments that incorporate knowledge about genetic, environmental and genomic influences and risk factors Collects personal, health and developmental histories that consider genetic, environmental and genomic influences and risks
5. Develops a plan for nursing practice	Develops a plan of care that incorporates genetic and genomic assessment information
6. Provides safe, appropriate and responsive quality nursing practice	Provides comprehensive, safe, quality practice to achieve agreed goals and outcomes that are responsive to the nursing needs of people Identifies clients who may benefit from specific genetic and genomic information and/or services, based on assessment data
7. Evaluates outcomes to inform nursing practice	Evaluates influence and effectiveness of genetic and genomic technology, information, interventions and treatments for clients' outcomes

The alignment demonstrated in Table 7.1 indicates how genomics fits with the professional roles and responsibilities or 'expectations of registered nurse practice' required by the NMBA. This alignment between the NMBA *Standards for Practice* and the *Essentials of Genetic and Genomic Nursing* can be further refined. To articulate each competency, the *Essentials of Genetic and Genomic Nursing* document provides specific knowledge and clinical performance indicators to guide the nurse, an example of which is provided in Table 7.2.

Table 7.2: NMBA *Standards for Practice* and an Exemplar Competency with Associated Knowledge and Clinical Performance Indicators

NMBA <i>Standards for Practice</i>	<i>Essentials of Genetic and Genomic Nursing</i>	Specific Area of Knowledge	Clinical Performance Indicators
Standard 4: Comprehensively conducts assessments	Competency: Collects personal, health and developmental histories that consider genetic, environmental and genomic influences and risks	Fundamentals of genetic- and genomic-focused health assessment Basics of risk factors: • Indicators of disease susceptibility or a genetic condition: • Family history: ⇒ Red flags of genetic/genomic conditions, such as: – disease found primarily in males – early age of onset for chronic adult onset disease – multiple cases of rare disease ⇒ Confounders: – race and ethnicity • Physical findings • Health history: ⇒ environmental and lifestyle factors ⇒ social and emotional status	Demonstrates ability to collect personal, medical and family history that includes genetic/genomic as well as environmental risks

Source: Extract from Consensus Panel on Genetic/Genomic Nursing Competencies (2009).

Ensuring that nurses are aware of the NMBA’s *Standards for Practice* as a *point of reference* for genomics in nursing practice is not the only professional challenge. As aforementioned, a misconception exists that genomics is the domain of specialist nurses. Some nurse specialists—such as the breast care nurse specialist or prostate care nurse specialist—will have an extended genomic knowledge and skill base associated with their position, as will some senior nurses through their professional experience. This expanded role for selected nurses does not void the generalist nurses’ responsibility to meet the NMBA’s *Standards for Practice* by providing person-centred and evidence-based care. Table 7.2 further illustrates the alignment between the NMBA’s *Standards for Practice* and the equivalent genomic competency, and more importantly articulates the knowledge and skills required to achieve the expectations of practice associated with an individual standard. The arrival of the genome era creates uncertainty and role ambiguity for nurses; however, awareness of the responsibility to possess fundamental genomic knowledge and skills, as an element of the overarching *Standards for Practice*, can help reduce this ambiguity.

The case findings clearly indicate that the genomics *point of reference* for nurses in Australia is abstract, and nurses are unclear about their professional obligations. Thus, strategies are required that enable registered nurses to overcome this uncertainty, so they can practise to the standard expected by the profession.

7.2.3 Point of Care

7.2.3.1 Key Case Finding: Nurses Are Not Adequately Incorporating Genomics into Practice

Registered nurses are required to incorporate genetic and genomic information into their practice if they are to adequately care for individuals, families, communities and populations throughout the life span (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009). To achieve genomic competency, nurses must use genomics information and skills at the ‘point of care’. Point of care, frequently referred to as ‘clinical point of care’, is described as clinicians delivering healthcare products and services to patients. The term ‘point of care’ is frequently associated with clinical documentation and information technology needs at the bedside (Faithfull-Byrne et al., 2017). In this study, the term is used in the context of providing nursing services to patients, their families and health professionals. The findings of this case study indicate that nurses are not adequately using genomics at the point of care. Inadequate preparation at the *point of learning* and lack of clarity at the *point of reference* results in registered nurses not possessing the necessary competence at the *point of care*.

In the context of this thesis, competence refers to the ability of nurses to apply genomic knowledge in practice. Developing competency requires that health professionals master their discipline knowledge base, as well as understand why, when and how that knowledge should be used to improve health outcomes for their patients. Competence serves as the dominant framework for the education of health professionals (National Coalition for Health Professional Education in Genetics, 2007). Inclusion of genomics in education programs and development of a set of core competencies in genetics, as discussed earlier in this chapter, will assist health professionals to integrate genetics knowledge, skills and attitudes into routine healthcare, thereby providing effective and comprehensive services to individuals and families (National Coalition for Health Professional Education in Genetics, 2007). Such strategies are pivotal to developing genomic competency.

Genomic literacy is a similar, yet distinct, term to genomic competency. Genomic literacy is described as having the ‘knowledge of genetics and genomics as these topics relate to, and

affect, professional nursing practice' (Giarelli & Reiff, 2012, p. 529). According to Ward (2011), literacy is more closely aligned with knowledge, while competency infers the ability to apply that knowledge. This distinction can be seen in Ward's (2011) alternate definition for genomic literacy as 'the foundational knowledge ... necessary for nurses to achieve genomic competency' (p. iv). In other words, genomic literacy is necessary, yet not sufficient for genomic competence (Ward, 2011). The GNCI[®] has been described by its author as a 'valid ruler by which to measure genomic literacy' (Ward, 2011, p. v). The findings of the GNCI[®] survey presented in Chapter 5 returned a mean score of 13.3 (score range 3–29), equating to a 42.9% correct response rate. This score indicates that the genomic literacy of Australian nurses is low. Therefore, Australian registered nurses do not have sufficient knowledge of genetics and genomics as these topics relate to, and affect, professional nursing practice, nor the foundational knowledge necessary to achieve genomic competency.

The findings from the GNCI[®] survey in terms of best and least knowledge were largely reflected in the limited genomic competency reported by the participants in the interviews. The GNCI[®] survey scores indicated that nurses and midwives were most knowledgeable in the 'mutations' category (49.3% correct response rate). Given that nurses rarely learn about genomics beyond the basic biological concepts and terms, this positive performance in a biological area was expected. However, the finding contrasted with the information provided by nurses during the interviews. In the interviews, the respondents most frequently cited targeted treatments as the main application of genomics in practice. This reference to targeted treatments aligned with nurses' and midwives' performance in the GNCI[®] survey, where they performed well in the 'genomic healthcare applications' category (47.9% correct response rate). The 'genomic healthcare applications' category included the concept of 'pharmacogenomics' and its respective questions, which is an example of targeted treatments.

The GNCI[®] survey scores indicated that nurses and midwives have the least knowledge in basic genetics, with respondents performing least well in the theory-based 'human genome basics' category. Performance in the 'family health history' category was moderate. This category had two questions receiving a 23% and 75% correct response, equating to a category average of 43% (correct response rate). This moderate performance was contrary to the interview findings, where family history was identified as the most common means for nurses to apply genomics in practice. This use of family history in practice was more consistent with the positive respondent performance in the 'genomic healthcare applications' category in the GNCI[®]

survey, and perhaps suggested a closer alignment of this concept with that category. The leaning of the interviewees towards healthcare applications indicated nurses' preference for practical genomic knowledge. However, the case findings indicated that Australian nurses are unsure how genomics can be applied in practice other than through 'targeted treatments', and are not actively using genomics with any regularity beyond determining a family history.

The findings of this study suggest that genomic competency is maximised when genomic knowledge has a direct healthcare application. Given that nurses are not demonstrating genomic competency, it can be assumed that potential genomic healthcare applications are unclear to nurses. Thus, it follows that, if the healthcare applications of genomics were made clear to nurses, genomic competency would likely improve. Several genomic competency documents have been developed to guide nurses in applying genomics in practice, as presented in Table 7.3. These competency documents outline the necessary knowledge, skills and competencies required by nurses, and serve to guide nurses in the application of their professional skills and responsibilities. Specific competency documents are not intended to replace or recreate existing standards for practice, but are intended to incorporate the genetic and genomic perspective into all nursing education and practice (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 7).

Table 7.3: Nursing Competency Documents

Document	Publisher	Available
<i>Core Competencies in Genetics for Health Professionals</i>	National Coalition for Health Professional Education in Genetics (2007)	Last accessed 11.2.16 no longer available at the time of writing.
<i>Essentials of Genetic and Genomic Nursing: Competencies, Curricula Guidelines and Outcome Indicators</i>	Consensus Panel on Genetic/Genomic Nursing Competencies (2009)	http://www.genome.gov/pages/careers/healthprofessionaleducation/geneticscompetency.pdf . Accessed 25/3/19.
<i>Essential Genetic and Genomic Competencies for Nurses With Graduate Degrees</i>	Consensus Panel on Genetic/Genomic Nursing and the American Nurses Association; Greco, Tinley and Seibert (2011)	http://www.genome.gov/Pages/Health/HealthCareProvidersInfo/GradGenComp.pdf . Accessed 25/3/19.
<i>Genetics/Genomics in Nursing and Midwifery</i>	Genetics in Nursing and Midwifery Task and Finish Group; Kirk, Campalani, et al., (2011)	https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/215250/dh_131947.pdf . Accessed 25/3/19.
<i>Fit for Practice in the Genetics Era: A Competence Based Education Framework for Nurses, Midwives and Health Visitors</i>	Genomics Policy Unit, University of Glamorgan, and the Medical Genetics Service for Wales, University Hospital of Wales (2003)	http://genomics.research.southwales.ac.uk/projects/fitforpractice2003/ . Accessed 25/3/19.

Family history and targeted treatments are routinely addressed in the varied competency documents available to the nursing profession. Collecting a family history and drawing a pedigree ('family tree') frequently appears in competency documents. The *Essentials of Genetic and Genomic Nursing* document has many competency statements addressing family history. For example, the registered nurse 'demonstrates ability to elicit a minimum of three-generation family health history information' (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 11) and 'constructs a pedigree from collected family history information using standardized symbols and terminology' (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 12). The *Essentials of Genetic and Genomic Nursing* document also has competency statements addressing treatment selection, such as 'Uses genetic- and genomic-based interventions and information to improve clients' outcomes' (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 13) and 'Performs interventions/treatments appropriate to clients' genetic and genomic healthcare needs' (Consensus Panel on Genetic/Genomic Nursing Competencies, 2009, p. 13). Participant performance in the pharmacogenomic questions and concepts in the GNCI[®] survey, together with the frequent references to targeted treatments and family history in the interviews,

indicated that nurses appreciate genomic-based information and interventions to improve patient care, and are subsequently on their way to achieving the associated competencies.

Although targeted treatments and family history were raised by many participants, some genomic applications were not raised by any participant in this research in any capacity. For example, in the ‘practice domain’ of the *Essentials of Genetic and Genomic Nursing* document, it is recommended that a nurse ‘Facilitates referrals for specialized genetic and genomic services for clients as needed’ (p.12). Although many participants spoke of specialist nurses, such as the breast care nurse or prostate care nurse, during the interviews, no participants referred a patient to these services or reported speaking to a colleague (nurse or medical practitioner) about the suitability for a referral. Perhaps even more alarming, no participant mentioned genetic health services (such as Genetic Health Queensland) during their interview. In fact, no participant mentioned a genetic counsellor at all, whether from Genetic Health Queensland or any other genetic service. The researcher argues that this inability of nurses to recognise and enact the various applications for genomics in practice represents a failure to demonstrate genomic competency.

Many factors may discourage or even prevent nurses from achieving genomic competency. The first factor is complexity. Genomics is a complex subject, with leading researchers believing genomics to be the ‘epitome of a complex competency’ (Calzone et al., 2018a). Many nurses equate genetics with complex scientific concepts (Skirton, 2017) that are best placed in a medical laboratory. While translating genetic concepts into nursing practice may be challenging for nurses (Calzone, Jenkins, Culp, Caskey, & Badzek, 2014; Rogers et al., 2017), this need not be the case. Attention can be directed to the way in which everyday nursing care can be improved by an awareness of genetic/genomic concepts, such as the underlying genetic basis for many common conditions (Skirton, 2017). The second factor is lack of observability. Many of the health outcomes derived from genomics are not readily observable (Garrison, Mestre-Ferrandiz, & Zamora, 2016). For example, identifying an individual with a genetic predisposition to a disease such as cancer provides an opportunity for risk reduction or early detection, while using a pharmacogenomic test can inform treatment options, reduce adverse drug events and improve efficacy (Ciardiello et al., 2014). These are important skills; however, they are not particularly tangible in the sense that nurses can articulate them as having performed a ‘task’. There has been a shift away from task-oriented nursing to person-centred and value-based nursing; however, remnants of this task-oriented nature persist. Nurses want

to *do* genomics, and this *doing* requires an observable task or action, which is frequently absent in the more subtle genomic applications in practice. The third factor is scope of practice. As has been argued earlier in this chapter, genetic and genomic competencies are integral to the practice of all registered nurses, regardless of academic preparation, practice setting, role or specialty. The findings of this study confirm that many nurses believe all genomics falls outside their practice. These nurses believe that they have limited or no input into decisions about a patient's medical treatment, and view genomics as relevant only to those who do—that is, the doctors, specialists and senior nurses. While this may reflect the reality of contemporary healthcare, routine integration will move genetics away from being a specialty to becoming a standard part of the care pathway.

Genomics has a clearer presence within the public domain. There are frequent references to genetics/genomics with respect to healthcare in the media and other texts/publications that are freely available to the public. There are new terms, concepts and issues appearing in the media every day, such as stem cells, cloning, biobanks and 'saviour siblings', to name a few. Each of these concepts is becoming increasingly visible in healthcare, and the public may turn to healthcare professionals, such as nurses, for further information. As the public are becoming more aware of the relationship between genetics and health and disease, nurses in all areas of practice are being asked to address basic genetic- and genomic-related questions and service needs (Sharoff, 2017): 'Nurses will need to navigate this new information and comprehend the changes that genomics are bringing to the healthcare field' (Rogers et al., 2017, p. 56). While nurses need not be experts on all concepts and issues, they require some familiarity so they can alleviate fears, correct falsehoods and refer the patient onto reputable people and sources of information as required. Genetic testing is also more accessible to the public, with companies such as '23 and me™' offering personalised genome screening. Personalised medicine or 'personalised genomic healthcare' allows risk assessment of disease, health screening and promotion, and tailoring of disease treatments to the individual's genetic profile (Garcia et al., 2011). Direct-to-consumer testing is available, and is allowing the public to learn about their genetic makeup (Cashion, 2009). The successes of these companies indicate that the public is aware of genetic tests and their application to healthcare. However, public expectations that genomics will be incorporated into care are not being met. Increased public awareness will lead healthcare professionals to address and accommodate patients' desire for knowledge of how genomics affects their healthcare (Rogers et al., 2017).

Part of providing comprehensive care is acknowledging the limitations and potential negative implications associated with genomics in healthcare. As Amor (2017) stated, ‘all genetic knowledge is not necessarily helpful’. Genetic testing is a prime example of potential negative implications. As with any medical intervention, genomic testing carries risks as well as benefits. Genetic testing on people who are well has special implications. Newson and Amor (2016) asks the question ‘If lives could be saved by being “forewarned” by a genomic test, should we perform genomic testing of all babies at birth?’ (p.12) and encourages us to review the scientific and ethical issues involved in the use of genomic information as a ‘lifetime health resource’ (p.12) . These authors cautioned that, if a lifetime health resource is to come to fruition, we need to think more about cost-effectiveness, custodianship of the data and engagement with families over time. Common to debates about genetics are issues surrounding the privacy and disclosure of genetic information, and the storage or future use of test samples and data. In Australia at least, good legal and regulatory controls are in place (Newson & Amor, 2016).

Embracing genomic healthcare requires a prepared workforce that can ‘inform, educate, and empower people, address existing and novel ethical issues, and anticipate any potential negative impact on vulnerable populations’ (Badzek, Henaghan, Turner, & Monsen, 2013; Calzone, Jenkins, et al., 2018; Seven, Eroglu, Akyüz, & Ingvoldstad, 2017). There is a clear need to expand the workforce of professionals trained to understand, deliver and incorporate genetics into the care of patients (Rehm, 2017). As stated by Calzone, Jenkins, Culp, and Badzek (2018), ‘the speed in which genomic information and discovery are transitioning to the clinical setting is only going to continue to accelerate’ (p.244). Thus, we need to prepare our nursing workforce to employ genomics at the *point of care* so that they are able to inform, educate, and empower people with respect to their genomic healthcare.

Overall, nurses are not adequately using genomics at the *point of care*, as a reflection of their limited genomic literacy and competency. These genomic literacy and competency deficits contribute to lost opportunities to take advantage of the benefits of genomic healthcare, such as improved health outcomes, reduced healthcare costs and increased patient quality and safety (Calzone, Kirk, et al., 2018). To maximise the potential of genomics, we must support the delivery of information at the *point of care* (Rehm, 2017). By not using genomics at the *point of care*, the nursing profession will not deliver on the promises that genomic healthcare has to offer.

7.3 A Critical Realist Perspective

The central tenets of critical realism were presented in Chapter 3 being the primacy of ontology, the existence of a stratified reality, the search for generative mechanisms and the interplay between structure and agency. As the philosophical framework underlying the study, it is important to discuss the case in the context of critical realist philosophy.

7.3.1 Primacy of Ontology

Critical realism is primarily concerned with ontology. This ontological focus asserts that much of reality exists and operates independently of our awareness or knowledge of it (Archer et al., 2016). Thus, our human perceptions of the world (epistemology) are not synonymous with the world's objective state (ontology). Similarly, nurses' perceptions of the world (nursing care that excludes genomics) are not the same as the objective state (nursing care that includes genomics). This phenomenon presents a 'more than meets the eye' scenario, meaning that nurses do not always see all that comprehensive care in the genomic era involves. For example, a patient being admitted to a day unit for a colonoscopy informs the nurse that he is feeling pressured by family members to undergo genetic testing for bowel cancer that 'runs in his family'. A nurse without adequate genomic knowledge may complete the admission without providing further information and support to the patient about genetic testing, unaware of the implications for the patient and his family's health. The reality of genomics in this scenario—genetic testing and the implications for the individual and his or her family—exists independently of the nurse's awareness or knowledge of it.

Alternatively, the nurse can provide comprehensive nursing care according to the NMBA's (2016) 'Standard 2.4: Provides support and directs people to resources to optimise health-related decisions', 'Standard 3.2: Provides the information and education required to enhance people's control over health' and 'Standard 2.6: Uses ... consultation and referrals in professional relationships to achieve improved health outcomes'. As a means of achieving these standards, the nurse providing comprehensive care will understand that genetic testing has implications for both the patient and at-risk family members (Connors & Schorn, 2018), and will direct the patient to appropriate resources and services. In this example, the nurse's continuation of the admission without addressing the patient's concerns about genetic testing is evidence of the limited engagement of Australian nurses with genomics. The ontological concept—that our human perceptions of the world are not synonymous with the objective state

(Bhaskar, 1998a)—is at the forefront of the exploration of nurses and their engagement with genomics in nursing practice in Australia. Understanding the reasons for this limited engagement means accepting that there is ‘more than meets the eye’ and that there are underlying factors that are either responsible for or contribute to the limited engagement with genomics.

7.3.2 Stratified Reality and the Generative Mechanisms in Nursing Practice

Bhaskar (1978, p. 56) interpreted reality as existing at three different ontological domains—the *empirical*, *actual* and *real*. Table 3.2 presented in Chapter 3 is replicated below as Table 7.4, displaying the factors (experiences, events and mechanisms) present in each domain. Nursing practice can be considered to exist in terms of the stratified reality advocated by Bhaskar’s critical realist philosophy. Individual nursing practices loosely align with the *empirical*, *actual* and *real* levels in a stratified reality in terms of what nurses experience (*empirical*) or are able to experience (*actual*), and what is happening ‘behind the scenes’ (*real*) that is influencing nurses’ engagement with genomics. The stratified reality can be used to understand and, to some extent, explain nurses’ lack of engagement with genomics.

Table 7.4: Real, Actual and Empirical Ontological Domains

Domain	Description	Experiences	Events	Mechanisms
Empirical	Fallible human perceptions and experiences, including science	✓		
Actual	Events and actions that are more likely to be observed	✓	✓	
Real	Underlying powers, tendencies and structures that cause events in the actual domain	✓	✓	✓

The *empirical* domain comprises only human perceptions and experiences (Clark et al., 2008). This refers to what is happening that is visible. Genomics is perceived and experienced by the nurse in the *empirical* domain. Nurses experience genomics by engaging with patients, as well as their nursing, medical and allied health colleagues. A nurse may collect a family history, administer a targeted treatment and monitor for specific adverse drug reactions without being fully aware that they are providing genomic care. This constitutes genomics that is seen (experienced) by the nurse.

The *actual* domain comprises events that happen whether we experience them or not (Danermark, 2002). This refers to what is happening that may or may not be visible. Opportunities

for the nurse to engage with genomics are present in the actual domain, regardless of whether the nurse chooses to take the opportunity or not. Genomic information is relevant in a family history, while targeted treatments and adverse drug reactions are based on the patient's genomic profile, and collaborations with colleagues are available regardless of whether the nurse uses (experiences) them or not. Genomics underpins nursing practice even if it remains unseen (experienced) by the nurse.

The *real* domain contains both the structures (objects) and the mechanisms that generate phenomena (McEvoy & Richards, 2003), and exists independently of the thought, awareness and even existence of human beings (Schiller, 2016). This refers to what is happening that is invisible. Thus, the real domain is concerned with the mechanisms that underpin genomics in nursing practice. Generative mechanisms are the structures, powers and relations that explain how things work beneath a surface (observable) appearance (Bhaskar, 1975/2008). These generative mechanisms are active in the real domain or 'reality' of the nursing profession. Bhaskar (1975/2008) argued that the objective of science is to produce knowledge about those generative mechanisms and structures that combine to produce phenomena (Schiller, 2016) or events (Souza, 2014). Understanding the generative mechanisms that produce events is pivotal in critical realism (Souza, 2014), and understanding these mechanisms in the nursing context may help improve 'events', or, in this case, the *acts* of nurses engaging with genomics, in terms of frequency and quality. These generative mechanisms support the rejection of the rival theory.

7.3.3 Agency and Structure

Social phenomena can be analysed using structural and agential approaches. *Agential* approaches emphasise the way that human agents respond to their surroundings based on the meanings they give to things or events, and *structural* approaches emphasise the social worlds and organisations within which individuals are embedded (Blumer, 1969). The relative importance of individual ('agency') factors (such as beliefs, attitudes and personal meanings) and contextual ('structural') factors (such as social norms, culture, geography and environment) is frequently contested (Clark et al., 2008). Applied to the nursing context, the question presented by these opposing factors becomes how much freedom do nurses (actors) possess, and to what degree does society (nuances of the nursing profession) constrain their behaviour (Houston, 2014)? In simpler terms, how much freedom do nurses (with their own beliefs, attitudes and personal meanings) possess, and to what degree does the nursing

profession (with its social norms, culture, geography and environment) constrain nurses' practice?

Critical realists emphasise the interdependence of structure and agency. They acknowledge that social structures provide the resources necessary for individuals to act, and place limits on individual behaviour. The social structures of nursing form the context for nurses to provide nursing care, while placing limits on the way nurses act and the nursing care provided. At the same time, it is acknowledged that human agents (actors) are also able to transform social structures by responding to their circumstances (Connelly, 2000). According to Bhaskar, 'actors shape their social worlds but, in turn, are constrained by social structures embedded in the fabric of social life' (Houston, 2014, p. 222). This infers that nurses shape their nursing context, yet are in turn constrained by it.

7.4 Critical Realism and the Case of Genomics in Nursing

Critical realism emphasises the interdependence of structure and agency in that it leads us to consider the way education (*point of learning*), policy and procedure (*point of reference*) and nursing practice (*point of care*) form the context for nursing practice. The way that nurses respond to this context determines the extent to which they are able to transform education, policy and practice.

The underlying premise of a critical realist view of genomics in nursing practice is that there is 'more than meets the eye' at the *point of learning*, *point of reference* and *point of care*. The use of critical realism in this research facilitates an explanation of the relationship between these categories that create the reality of genomics in nursing practice. These generative mechanisms begin with limited education at the *point of learning*, leading to unclear professional standards of practice at the *point of reference* and ultimately contributing to inadequate engagement with genomics at the *point of care*. Figure 7.3 depicts the relationship between the key tenets of critical realism and the categories that were developed in this research. Assuming a perspective in which ontology is primary, this figure indicates the relevance of generative mechanisms and agency and structure at all points.

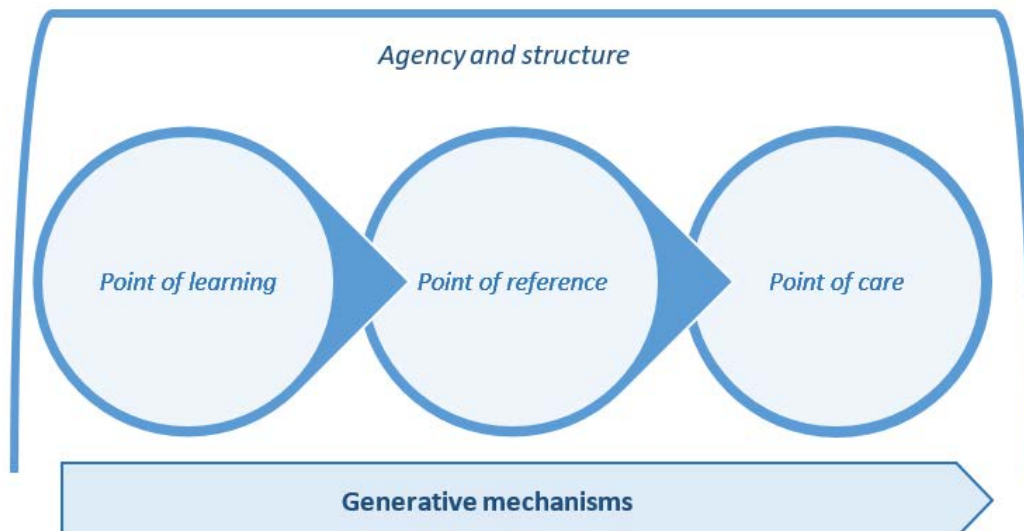


Figure 7.3: Critical Realism as Applied to the Case

7.5 Summary

The case of Australian nurses' engagement with genomics presents a less-than-ideal picture. The study findings indicated that Australian nurses are not adequately engaging with genomics at the *point of learning*, *point of reference* or *point of care*. If nurses are not knowledgeable about genomics and are unclear about their professional roles and responsibilities, they cannot be expected to adequately integrate genomics into their practice. Australian nurses' limited engagement with genomics has consequences for the nurse, the patient and the wider nursing profession. This limited engagement must be addressed if we are to meet our professional obligations to those in our care. Transforming nursing policy, practice, education and research is a global endeavour (Calzone et al., 2018a; World Health Organization, 2016) and Australia is well placed to contribute. The following chapter presents a final summary of the 'case', as well as recommendations for improving engagement with genomics at the *point of learning*, *point of reference* and *point of care*.

Chapter 8: Conclusion

8.1 Introduction

Advances in genetics and genomic science mean there is now a responsibility for all nurses to be knowledgeable about genomics in relation to healthcare (Camak, 2016). As described in this thesis, this research undertook a critical realist case study using a single holistic design to create a picture of genomics in nursing practice in Australia. The study findings presented in the previous chapters can be used to improve the integration of genomics into nursing practice in Australia. This final chapter summarises this research and evaluates the quality of the final case study. It discusses the implications of the study findings and study limitations, and presents recommendations for policy, education, practice and research.

8.2 Study Summary

At the beginning of this study, the researcher set out to determine how Australian nurses engage with genomics in nursing practice. Although existing research indicated that genomics has a limited presence in nursing practice in Australia, a comprehensive investigation had not been undertaken. It became the researcher's intent to create a picture of Australian nurses' engagement with genomics and to summarise the genomic knowledge and skills of nurses, including how these are used in the delivery of nursing care. The key elements of the research study are summarised in Table 8.1.

Table 8.1: Key Elements of the Research Study

Research question	How are nurses engaging with genomics in nursing practice in Australia?
Research aim	Determine how nurses engage with genomics in nursing practice in Australia
Objectives	Document instances of engagement with genomics (genomic knowledge, skills and technologies) in nursing practice in Australia Seek to gain a deeper understanding of: 1. the genomic knowledge and skills employed by nurses in nursing practice 2. nurses' perceptions of the role of genomics in nursing practice 3. nurses' experience of using genomics in daily nursing practice in terms of patient care and communication within the healthcare team 4. the barriers and enablers to nurses applying genomics in nursing practice
Research design	A single holistic case study underpinned by a critical realist framework
Case summary	Australian nurses are not adequately engaging with genomics at the <i>point of learning</i> , <i>point of reference</i> or <i>point of care</i>

The individual survey and interview findings were addressed in Chapters 5 and 6, respectively. These findings were synthesised into the case description and discussed in the context of the literature in Chapter 7.

8.3 Quality and Rigour

Case study research has been unfairly accused of lacking rigour (Stewart, 2014), likely because the analytical phases used to construct the final case are largely qualitative in nature. The quality of qualitative research cannot be judged comparatively with quantitative research (Houghton et al., 2013), as there are different criteria used to assess rigour in each approach. The most common criteria for evaluating qualitative research are those proposed by Lincoln and Guba (1985): credibility, dependability, confirmability and transferability. Yin (2014) proposed alternative criteria for assessing the quality of social research, including case study research: construct validity, internal validity, external validity and reliability. Lincoln and Guba's and Yin's terminology are qualitatively and quantitatively nuanced, respectively.

This single-case holistic study contained both a quantitative and qualitative phase; therefore, a hybrid approach for evaluating this research was appropriate. The quantitative terms used by Yin align with those of Lincoln and Guba—construct validity with credibility, reliability with dependability, internal validity with confirmability, and transferability with external validity. Yin (2014) and Lincoln and Guba (1985) each recommended different strategies (or tactics) to address these criteria. A summary of the strategies is presented in Table 8.2.

Table 8.2: Comparison of the Quality Strategies Proposed by Yin and Lincoln and Guba

	Yin	Lincoln and Guba
Construct validity	Multiple sources of evidence Chain of evidence Key informants review draft case study report	Prolonged engagement and persistent observation Triangulation Peer debriefing Member checking
Reliability	Case study protocol Case study database	Audit trail Reflexivity
Internal validity	Pattern matching Explanation building Address rival explanations Logic models	Audit trail Reflexivity
External validity	Theory in single-case studies Replication logic in multiple-case studies	Thick descriptions

Source: adapted from Houghton et al. (2013, pp. 12–17) and Yin (2014).

A selection of the strategies listed in Table 8.2 was employed by the researcher to assess rigour in this research study. These strategies are outlined in Table 8.3.

Table 8.3: Quality Strategies Employed in This Case Study

Construct validity	Employ multiple sources of evidence, establish a chain of evidence, have draft case study report reviewed by key informants, undertake peer debriefing
Reliability	Case study protocol, audit trail, reflexivity
Internal validity	Pattern matching, audit trail, reflexivity
External validity	Thick description

Yin (2014, p. 46) described his use of the terms as follows:

- *construct validity*—identifying correct operational measures for the concepts being studied
- *reliability*—demonstrating that the operations of a study can be repeated with the same results
- *internal validity*—seeking to establish a causal relationship (for explanatory or causal studies only, and not for descriptive or exploratory studies)
- *external validity*—defining the domain to which a study’s findings can be generalised.

The strategies listed in Table 8.3 are largely reflected in Yin's (2014, p. 118) four principles of data collection: (i) use multiple sources of evidence, (ii) create a case study database, (iii) maintain a chain of evidence (iv) exercise care when using data from electronic sources. The case study strategies to ensure the rigour of this research study will be discussed in the following section. Given that this study was based on Yin's design framework, this discussion will be structured using his terminology.

8.3.1 Construct Validity (Credibility)

First, the researcher used two data sources in this case study (cross-sectional survey and semi-structured interviews). Multiple data sources are important in developing converging lines of enquiry (Yin, 2014, p. 120) and allow the researcher to capture the complexities of phenomena, thereby enhancing the completeness of the case description (Houghton et al., 2013). Second, the researcher maintained a chain of evidence in conducting this case study. A chain of evidence allows the reader (or researcher) to 'trace the steps' of evidence from the research question to the case study conclusion (Yin, 2014, p. 127). Third, the researcher participated in regular peer debriefing with doctoral advisors and colleagues. Peer debriefing requires an external colleague or expert to support the credibility of findings (Casey & Houghton, 2010). The expectation is that an independent analyst will agree with the data labels and the logical paths taken to determine them (Graneheim & Lundman, 2004; Houghton et al., 2013).

8.3.2 Reliability (Dependability)

First, the researcher developed a case study protocol as a requirement for successful completion of candidature. The protocol was approved by the James Cook University higher degree research candidature committee, and was used to guide the researcher in conducting data collection (Yin, 2014, p. 84). The protocol addressed the key areas of case study research, as well as more generic research elements according to university requirements. The four case study protocol elements according to Yin (2014, p. 85)—case overview, data collection procedures, data collection questions, and guide for case study report—were addressed. Second, the researcher developed a case study database as a repository for all case study data (Yin, 2014, p. 123) using computer programs that included SPSS and NVivo, as well as generic word-processing files (Microsoft Word and Excel files). Third, the researcher maintained an audit trail during the study. An audit trail is an outline of the decisions made throughout the research process. It provides a rationale for the researcher's methodological and interpretative

judgements (Houghton et al., 2013). This was essential in the qualitative interviews, where NVivo was used to record decisions made during data collection and analysis (Yin, 2014). Readers may not share a researcher's interpretation of the data; however, they should be able to discern the means by which it has been reached (Koch, 2006). Finally, the researcher maintained a reflexive diary to record thoughts about decisions made throughout the research. Reflexive diaries consider the researcher's history and personal interests as contributors to the research (Toffoli & Rudge, 2006) by journaling the rationale underpinning research decisions (Rolfe, 2006).

8.3.3 Internal Validity (Confirmability)

Internal validity is primarily a concern for explanatory studies where the researcher is seeking to establish a causal relationship (Yin, 2014, p. 4); however, internal validity can still be addressed in exploratory studies by using 'pattern matching'. Pattern matching compares an empirically based pattern with a predicted pattern (Yin, 2014, p. 143). In this study, the researcher elected to use thematic analysis, as opposed to Yin's pattern matching technique. Thematic analysis involves discovering, interpreting and reporting patterns within the data (Spencer, Ritchie, Ormston, O'Connor, & Barnard, 2014, p. 271) and, in this way, is similar to Yin's approach. Thematic analysis or 'theming the data'—as Saldana (2016, p. 200) termed the process—is suitable for almost all qualitative studies, and provides a suitable (and more straightforward) alternative to pattern matching.

8.3.4 External Validity (Transferability)

A case study strives for findings or 'lessons learnt' that extend beyond the specific case (Yin, 2014). In this study, the researcher used 'thick' descriptions in describing the case to enable readers to make informed decisions about the transferability of the findings to their specific contexts (Lincoln & Guba, 1985). Thick descriptions include accounts of the context, details of the research methods and examples of raw data so that readers can consider their interpretations (Stake, 1995), as well as a 'rich and vigorous' presentation of the findings (Graneheim & Lundman, 2004). This research study provided 'thick' descriptions of the context and research methods, as well as quotations as examples of raw data, from which readers can make their own decisions about the fit with their particular context.

8.3.5 Summary of Evaluation

Yin (2014, p.200) described five characteristics for an exemplary case study. The processes by which these characteristics can be achieved have been described in the preceding section. Table 8.4 presents these characteristics and a summary of how they have been demonstrated in this thesis.

Table 8.4: Yin’s Characteristics of an Exemplary Case Study

Characteristic	Description	Evidence
Significant	Has the researcher focused the case on unusual topics of general public interest, or underlying issues that are nationally important—either in theoretical terms or policy or practical terms?	Chapters 4 and 7
Complete	Has the research clearly defined the case’s boundaries, collected extensive evidence and conducted the study absent of artefactual conditions?	Chapters 4 to 8
Consider alternative explanations	Has the researcher considered rival propositions and sought to collect evidence from differing perspectives in the case?	Chapters 4 and 7
Display sufficient evidence	Has the researcher reported the case in such a way that a reader can reach an independent judgement regarding its merits?	Chapters 5 and 6
Composed in an engaging manner	Has the researcher presented the case in a way that communicates the results widely?	Chapters 7 and 8

Source: Yin (2014).

8.4 Implications and Recommendations

The findings of this study have implications for nursing education, policy, practice and future research. Recommendations can be made based on the study findings in relation to these four areas, and are discussed in the following sections.

8.4.1 Education—Genomics at the *Point of Learning*

Key Study Finding: Genomics in nursing education is inadequate	
Implications for practice	Australian nurses do not possess the genomic knowledge and skillset necessary to adequately incorporate genomics into their clinical practice
Recommendations	Genomics must be embedded throughout the nursing curricula with healthcare applications made clear to the learner
Achieved by	<ul style="list-style-type: none"> • Addressing genomics as a biological ‘system’ (akin to the cardiovascular system or respiratory system) • Introducing a dedicated ‘Genomics in Healthcare’ or ‘Genomics in Nursing’ subject/study unit • Preparing nursing faculty to teach genomics

The evidence is clear from this research that genomics is not being adequately addressed in Australian nursing curricula, with nurses rarely learning about genomics beyond the basic biological concepts and/or terms. Thus, nurses do not possess the genomic knowledge and skillset that would allow them to adequately incorporate genomics into their clinical practice. This lack of genomics knowledge and skills represents a barrier to nurses providing comprehensive healthcare. To address this insufficiency, genomics needs to be embedded throughout nursing curricula, and the healthcare applications need to be made clear to the learner.

This recommendation can be achieved by addressing genomics as a biological ‘system’ within nursing curricula. Biological systems (such as the cardiovascular system) and their associated nursing knowledge and skills are standard across nursing curricula. It can be assumed that all nursing curricula address the cardiovascular system, where nurses learn about the heart and how to measure blood pressure, or the neurological system, where nurses learn about level of consciousness (alertness) and how to recognise a deteriorating patient. The *genomic system* would allow nurses to learn about genetics and how genetic and genomic information can be used in the delivery of nursing care. For example, nurses would learn about inheritance patterns and how to recognise ‘red flags’ when conducting a family history, understand pharmacogenomics and how individuals may respond to a particular medication, and appreciate ethical and legal factors affecting an individual’s decision-making process about genetic testing. These genomic healthcare applications would likely contribute to nurses using genomics in practice more frequently and with more confidence. Similarly, subjects dedicated to genomics could be considered for all nursing curricula. A specialist ‘Genomics in Healthcare’ or ‘Genomics in Nursing’ undergraduate or postgraduate subject/study unit—or, as a minimum, a single lecture, tutorial or workshop—should be included as standard in nursing curricula. Genomics is frequently presented as basic biology. Providing a subject/study unit dedicated to genomic healthcare applications may transform *theoretical genetics* into what the researcher terms *active genomics*.

Active genomics refers to genomic knowledge, skills and technologies that can be used by the nurse (or healthcare professional) in the delivery of patient care. Introducing a subject dedicated to genomic healthcare applications and *active genomics*—genomics that nurses can *use*—is sure to increase the frequency and confidence with which genomics is applied. Kronk, Colbert, and Lengetti (2018) reported on an undergraduate genetics course designed based on

the *Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics*. They identified themes about genetic and genomic transformational learning, as well as an overall appreciation of the content and applicability of the material. The findings indicated that students were able to readily detail how and why this content would contribute to their practice, discuss the specific skillset they achieved, and improve their understanding with regard to the scope of genetics and genomics in nursing practice (Kronk et al., 2018). Providing a subject/study unit, such as that reported by Kronk et al. (2018), would improve the clinical utility of genomics for Australian nurses.

Ultimately, addressing the inadequacy of genomics in nursing education can only be achieved by preparing nursing faculty to teach genomics. All nursing faculty are responsible for incorporating genomic content into their courses (J. K. Williams et al., 2011, p. 233). Integrating genomic content into nursing curricula requires faculty staff to recognise the relevance of genomics to nursing practice, and demonstrate adequate knowledge, confidence and comfort with the underlying concepts (Read & Ward, 2016). As stated by J. Jenkins, Bednash, and Malone (2011, p. 1), nurse faculty staff are the key to preparing future professional nurses to assist individuals, families and communities to traverse the complex personalised healthcare environment.

8.4.2 Policy—Genomics at the *Point of Reference*

Key Study Finding:	
Nurses are unclear about their roles and responsibilities in using genomics in practice	
Implications for practice	Nurses are unclear how genomics relates to their professional roles and responsibilities
Recommendation	Nursing policy must articulate the alignment between the NMBA’s <i>Standards for Practice</i> and genomic competencies
Achieved by	Providing nurses with clinical examples of nursing policy aligning with the NMBA’s <i>Standards for Practice</i> and genomic competencies

There is a clear alignment between the expectations of registered nurses as outlined in the NMBA’s (2016) *Standards for Practice* and generic genomic nursing competencies (an exemplar using the *Essentials of Genetic and Genomic Nursing* was provided in the previous chapter). However, as has been argued in the previous chapter, this alignment between the documents is not always clear. The implication for practice is that nurses are similarly unclear about how genomics relates to their professional roles and responsibilities. Nursing policy needs to more clearly articulate the professional responsibility of nurses with respect to

genomic competencies. This articulation can occur prior to registration, post-registration at the organisational (employer) level, or through larger governing bodies (such as AHPRA) making statements to address recognition of this alignment. Nurses need to be aware of this alignment if they are to acknowledge and understand their professional role and responsibilities. The NMBA’s (2016) *Standards for Practice* state that:

RNs [registered nurses] need to continue to develop professionally and maintain their capability for professional practice. RNs determine, coordinate and provide safe, quality nursing. This practice includes comprehensive assessment, development of a plan, implementation and evaluation of outcomes. (p. 1)

Thus, it is clear that the NMBA’s *Standards for Practice* accommodate genomics, explicating the way that genomics is integral to, rather than distinct from, the expectations of nurse practice set out by this document. The safe and quality nursing advocated by the NMBA includes genomic knowledge, skills and technologies, and registered nurses must acknowledge and adopt genomics if they are to maintain their capability for professional practice.

8.4.3 Practice—Genomics at the *Point of Care*

Key Study Finding:	
Nurses are not adequately incorporating genomics into practice	
Implications for practice	Nurses are not providing comprehensive, safe and quality healthcare, and in doing so may not meet public expectations
Recommendation	Nurses must incorporate genomics knowledge and skills into practice
This can be achieved by	<ul style="list-style-type: none"> • Increasing the awareness of nurses with respect to their obligation to possess and apply genomic knowledge, skills and technology • Leadership that promotes a cultural shift with respect to the significance of genomics in nursing practice

The inadequacy of genomics use at the point of care has been highlighted throughout this thesis. Consequently, nurses are not providing comprehensive healthcare, meaning that quality of care is lower, and patients may experience adverse consequences because of nurses’ lack of knowledge and understanding of basic genomics (Read & Ward, 2018). Thus, nurses must incorporate genomics knowledge, skills and technology into their practice to fulfil their professional obligation to their patients.

Increasing the use of genomics in nursing practice will go a long way towards meeting public expectations. Growing public awareness of genomics means that nurses in all practice areas will increasingly be asked to address basic genetic- and genomic-related questions, concerns

and service requirements. If nurses are not adequately incorporating genomics into practice, there is a clear risk that the public (our patients) will not be fully informed, or, perhaps more troublingly, be misinformed about the genomic issues related to their healthcare. Nurses must be genomically competent so they can alleviate fears, correct falsehoods and refer the patient onto reputable people and sources of information as required. Although the nurse need not be an expert on all concepts and issues, familiarity with generic genomic competencies will allow the nurse to meet the public's expectations.

Incorporating genomics into nursing practice represents a change in current practice—a change that can be accelerated with good leadership and advocacy. The 'diffusion of innovations' theory describes the process of social change (E. M. Rogers, 2003) and can be used to expand nursing practice. The diffusion of ideas is frequently a slow process; however, adoption can be accelerated using opinion leaders or *champions*. Opinion leaders 'act as gatekeepers for interventions, help change social norms, and accelerate behavior change' (Valente & Pumpuang, 2007, p. 881). These nursing champions are pivotal to changing professional and social norms and accelerating behaviour change with respect to genomics in nursing practice. The findings of a study by J. Jenkins et al. (2015) indicated that nursing champions can facilitate change in genomic nursing capacity. Supporting nursing leaders to become aware of, plan for and begin to incorporate innovation in practice can expand nursing capacity (J. Jenkins et al., 2015), thereby leading to an enhanced presence of genomics at the *point of learning*, *point of reference* and ultimately *point of care*.

8.5 Study Limitations

8.5.1 Data Sources

There were only two data sources used in this study—a cross-sectional survey and semi-structured interviews. Yin (2014, pp. 118–130) proposed four principles of data collection, the first being multiple sources of evidence. Triangulating from multiple sources of evidence (data) enables the researcher to capture the complexities of phenomena, thus enhancing the completeness of the case description (Houghton et al., 2017), as well as enhancing rigour (Houghton et al., 2013). Yin (2014) did not stipulate how many sources constitute 'multiple' sources. It is assumed that a third source of data is not necessary for case study research; however, it would have been preferable. The researcher considered observation to be a third data source for the case study. Observation is one of Yin's (2014) six recommended data

sources and is frequently used in case study research because it offers insight into behaviours and practices as they occur in their natural settings. The contemporary phenomenon under exploration in this case study was genomics—specifically, nurses using genomic knowledge, skills and technologies in practice. Consistent with the literature on this topic, it was expected that incidences of nurses using genomics would be limited and difficult to observe, given their subtlety. Therefore, observations were deemed impractical for this study.

8.5.2 Participant Numbers

There was a relatively small number of participants involved in this case study, with only 253 respondents in the survey. Three hundred and ninety-eight responses were recorded; however, more than 100 respondents did not complete the genomic literacy scale. Completed surveys (those with 30 or more questions completed) were included in the final analysis, with the expectation maximisation function used to replace the missing values and allow a total value to be calculated. For a national cross-sectional survey, 253 respondents was disappointing. It is worth noting that Read and Ward (2016), in a previous iteration of the survey (also administered online) were not able to report a response rate. Their completion rate was 73%, slightly higher than the 64% completion rate noted in this survey. As stated in the researcher's 'Genomic Literacy of Registered Nurses and Midwives in Australia' manuscript presented in Chapter 5, the reduced participation could be due to respondents' unfamiliarity with the topic and terminology. It is recommended that future studies using the GNCI[®] or similar instruments define key terms as a means to increase familiarity and improve engagement with the study. It is also acknowledged that most respondents were in Queensland, meaning the data may reflect the Queensland healthcare system as opposed to those of other states and territories. The varied state representation may be due to varying levels of engagement by branches and members of the relevant recruiting organisations. However, Australia has national accreditation standards that guide content of pre-registration nursing programs (ANMAC, 2012), and therefore the results can still be considered reflective of the wider nursing population.

The interviews involved nine participants. As stated in the researcher's 'Genomics in Oncology Nursing Practice' manuscript presented in Chapter 6, it is likely that lack of familiarity with the subject matter again contributed to reduced participation. It is recommended that future studies be conducted as part of an education initiative to raise awareness about the topic and improve engagement with the study. It is also possible that the perspectives of oncology nurses

may not adequately reflect those of the wider nursing profession; however, some of the findings of this study may have applicability in other contexts.

8.5.3 Mixed Participants

The participants for the survey and interviews were not selected from the same participant pool. The survey was open to registered nurses and midwives in Australia, while the interviews were open to registered nurses working in oncology-based units at a regional hospital in Queensland. The use of an alternative participant pool for the interviews was a deliberate decision made in response to the national survey findings. Genomics has a higher presence in oncology; therefore, it is likely that nurses working in oncology would be more familiar with the terms 'genetics' and 'genomics', incorporate genomics more regularly into their practice, and have more opinions about the relevance and utility of genomics in nursing practice than would their colleagues working in other nursing specialties. Thus, targeting oncology nurses maximised the chances of collecting usable data. Accessing oncology nurses at a single hospital was a practical decision to reduce the challenges associated with collecting data at multiple sites. The oncology nurses participating in the interviews represent a small subset of the larger and wider sample pool of health professionals used in the survey. Thus, it is possible that the findings derived from the interviews with oncology nurses may not adequately reflect those of the wider nursing community, as represented in the survey.

8.6 Recommendations for Future Research

Further research is required to ensure that all nurses have sufficient knowledge of genetics and genomics as these topics relate to and affect professional nursing practice, and are able to apply this knowledge to achieve genomic competency. The findings in this study indicate that genomics is not adequately addressed at the *point of learning*, *point of reference* and *point of care*. Nurses must learn about genomics and accept their professional responsibilities for practice if they are to adequately use genomics in their nursing care.

Future research focusing on the integration of genomics into nursing curricula at undergraduate and post-graduate levels is needed. This research should investigate barriers and enablers to the inclusion of genomics in nursing curricula. Research is also needed to examine the presence and value of genomics in accreditation and practice standards. Future investigations should also focus on practice adoption techniques, such as Rogers's diffusion of innovations theory

(Rogers, 2003). This theory has been used to explore the adoption of genomics with international nurses; however, there is no indication that it has been used in the Australian context.

Nursing leaders, policy makers and governing bodies, as well clinicians in everyday nursing practice, need to acknowledge the presence of genomics in the profession. Further research could address the limitations of this research with respect to sources of data, numbers of participants, and the professional and experiential profiles of the nurses who took part in this research. Alternative data sources, such as observations, may be included in future research; however, the reasoning that precluded them from use in this study may still apply and should be considered. The challenges faced in this research regarding interest and subsequent recruitment will likely persist for some time, although increased attention in academia and healthcare sectors may reduce these challenges.

8.7 Summary

This chapter has summarised the research study and concluded the thesis. This study sought to determine how Australian nurses engage with genomics in practice. A single-case exploratory case study with a critical realist framework was conducted. The ‘case’ indicated that Australian nurses have limited engagement with genomics, and genomics is inadequate at the *point of learning, point of reference* and *point of care*. These findings can now be used to inform an increased genomics presence in nursing education, policy and practice. Genomics has the potential to transform healthcare delivery by increasing quality and safety, decreasing costs, and improving health outcomes (Alexander, 2017; Calzone, Kirk, et al., 2018; McCormick & Calzone, 2016), yet this potential to transform healthcare delivery will only be realised if nurses begin to fully engage with genomics.

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Appendices

Appendix 1: Advertising flyer for GNCI® survey



**Are you a RN and/or RM?
What do you know about genetics?**

Do Australian registered nurses and midwives have the genetic literacy they need to practice in these modern times?

We want to know what you know about genetics.

Complete the survey via the link
https://jcuchscs.qualtrics.com/SE/?SID=SV_8uWPuDfKyiwcgWV
or scan the QR code below using your smartphone
**and go into the draw to win a
\$200 Pre-Paid Visa.**



The survey will be open until Friday 28th October.

Contact information:
Helen Wright
Nursing, Midwifery and Nutrition
College of Healthcare Sciences
James Cook University
Phone: (07) 4781 5352
Email: helen.wright@jcu.edu.au
HREC ID: H6587

Cairns
Singapore
Townsville

Appendix 2: JCU HREC documentation

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Appendix 3: Letter providing permission to use the GNCI® instrument

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Appendix 4: Advertising flyer for genomics in oncology interviews

JAMES COOK UNIVERSITY AUSTRALIA

Are you a registered nurse working in an oncology unit at the Townsville Hospital?

If the answer is yes, we would like to invite you to participate in a research study about genetics in oncology nursing practice

What's the research about?
James Cook University researchers are conducting a study about how genetics is applied in oncology nursing practice

What would I have to do?
You would be asked to participate in a 1 hour interview at a time and place that is convenient to you.

If you are interested in being involved in this study and/or would like more information please contact:

Helen Wright
Phone: (07) 4781 5352
Email: helen.wright@jcu.edu.au

This research study is being conducted by Helen Wright PhD Candidate as Principal Investigator and supervised by Professor Melanie Birks, Professor Jane Mills and Dr Lin Zhao, James Cook University (JCU) and approved by Queensland Health and JCU Human Research Ethics Committee (HREC) [HREC Reference number: HREC/17/QTHS/241].

Cairns
Singapore
Townsville

Appendix 5: THHS HREC/SSA documentation

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Appendix 6: THHS project protocol

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Appendix 7: Genomics in oncology nursing practice interview guide

Genomics in oncology nursing practice Semi-structured interview guide

Interview details	
Date	
Time	
Location	
Interview start time	
Interview stop time	
Interview duration	

Part 1: Nurses' perceptions of the role of genomics in oncology nursing practice			
Information	Key question	Probes/prompts/follow-up questions	✓/ comments
Terminology	<p>1. In the information sheet, you were provided with a definition for the terms <i>genetics</i> and <i>genomics</i>. Were you familiar with these term/s prior to this research study? (Prior to reading the information sheet or the flyers).</p> <p>2. What is your understanding of the terms <i>genetics</i> and <i>genomics</i>? Can you describe them to me in your own words?</p>	<p>Genetics: The study of individual genes and their impact on relatively rare single gene disorders</p> <p>Genomics: The study of all the genes in the human genome together, including their interactions with each other, the environment, and the influence of other psychosocial and cultural factors</p>	
Role of genomics in nursing	<p><i>The literature suggests that genomics is relevant to all areas of nursing practice.</i></p> <p>3. How <i>relevant</i> do you believe genomic information is to oncology nursing practice?</p> <p>4. Do you believe genomic information is more relevant to other healthcare professions, for example medicine or psychology?</p>	<p>Probe: Can you tell me why you believe it is relevant (or not relevant)?</p> <p>Probe: Can you tell me why you believe it is more relevant to this/these healthcare professions?</p>	

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	<p><i>In terms of importance, some nurses have said that genomics is very important to nursing practice, where as other nurses have said it is not important at all.</i></p> <p>5. How <i>important</i> do you believe genomic information is to your nursing practice?</p> <p>6. How <i>important</i> do you believe genomic information is to oncology nursing practice?</p>	<p>Probe: Can you tell me why you believe it is important (or not important) to your practice?</p> <p>Probe: Can you tell me why you believe it is important (or not important) to oncology nursing practice?</p>	
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Part 2: The application of genomic information and/or skills by nurses in oncology nursing practice			
<p>Personal use of genomics in practice</p>	<p><i>The literature suggests that to care for a person, their families, and communities, registered nurses will need to incorporate genetic and genomic information into their practice.</i></p> <p>7. How do you think genomic information can be used in oncology nursing practice?</p> <p>8. How do you incorporate genomic information into your practice?</p>	<p>Prompt: For example, do you think genomic information can be used to direct cancer treatment? Or perhaps prevention?</p> <p>Probe: Can you give me an example of how genomic information can be used in practice?</p> <p>Probe: Can you give me an example of how you incorporate genomic information into your practice?</p>	

	<p>9. How frequently would you say you incorporate genomic information into your practice?</p> <p>10. How do you think genomic information impacts patient care and/or health outcomes?</p>	<p>Prompt: For example, daily, once a month, rarely? Perhaps only in relation to a specific type of cancer?</p> <p>Prompt: For example, do you think genomics can be used to improve patient care by enabling earlier diagnosis? Perhaps you think genomics has no impact on patient care?</p>	
Observations of genomics in practice	<p>11. Can you tell me about a time when you saw or heard another registered nurse use genomics in their practice?</p> <p>12. Can you tell me about a time when you saw or heard a fellow healthcare professional use genomics in their practice?</p>	<p>Prompt: For example, perhaps you saw another registered nurse taking a comprehensive family history or drawing a family tree.</p> <p>Prompt: For example, perhaps you heard a doctor talking to a patient about their family members being 'at increased risk' of cancer? Or perhaps referring a patient to the genetic counsellor.</p>	
Genomic literacy	<p><i>Some nurses have described their knowledge of genomics as poor where as others have described their knowledge as excellent.</i></p> <p>13. How would you describe your genomic knowledge and/or skills?</p> <p>14. Do you believe your genomic knowledge and/or skills are adequate for practice?</p> <p>15. What opportunities for further education in genomics would you like to see offered?</p>	<p>Prompt: For example, limited, average, good, very good or excellent?</p> <p>Prompt: In other words, do you think you know enough about genomics to do your job?</p> <p>Prompt: For example, would you like to attend an in-service or an online course? Perhaps you would like to read a book or journal article about genomics in nursing?</p>	

Part 3: Nurses' experience of using genomics in 'day to day' nursing practice				
Public and genomics	<p><i>Some nurses have reported that patients and their family members have asked questions about genetics. For example 'Are my children at risk of developing the same cancer?' 'Does it matter that I was diagnosed with cancer at a young age?'</i></p> <p>16. Has a patient or a patient's family member ever asked you a question about genomics?</p> <p>17. Did you feel you had sufficient genomic knowledge to answer these question/s?</p>	<p>Probe: Can you give me an example of the question/s you have been asked?</p>		
Genomic material	<p>18. If a patient, their family member or a colleague asked you a question about genomics, where would you go to find more information?</p> <p>19. Can you tell me about a time that you sought out genomic information?</p>	<p>Prompt: For example, would you ask a colleague? Or perhaps search the hospital resources or a public website?</p> <p>Prompt: For example, perhaps a patient was found to have a genetic predisposition to breast cancer and you contacted the Breast Cancer nurse to get more information.</p>		
Support	<p><i>Some nurses have commented that they receive support from their nursing colleagues and/or other members of the healthcare team to use genomics in their practice, where as other nurses do not feel supported.</i></p> <p>20. What support do you receive to use genomics in practice?</p>	<p>Prompt: For example, your NUM may approved leave for you to attend a genomics seminar. Perhaps a colleague</p>		

	21. What support would you like to receive?	has shared a journal article about targeted cancer treatments. Prompt: For example, would you like to attend an 'in-service' about genomics, or a paid study day to attend a course or conference?		
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Participant details (participants may decline to answer)					
I will read out some age ranges and if you can tell me in which category you belong.	Age	18-24yrs	25-34yrs	35-44yrs	45-54yrs
Do you identify as male, female, other or prefer not to say?	Gender	55-64yrs	65-74yrs	75+yrs	
What unit/ward do you work in?	Unit	Male	Female	Other	Prefer not to say
In what year were you registered as a registered nurse?	Year registered				
How many years have you worked in an oncology setting?	Time worked in oncology setting				
Do you have any post-graduate qualifications?	Post-graduate qualifications	Grad Dip	Grad Cert	Masters	PhD Prof Doctorate
Do you have any post-registration education and/or training in oncology nursing?	Post-registration education and/or training in oncology nursing	No	Yes (if so, describe)		