Research proposal for minor dissertation for the degree of M.Sc (Med) Genetic Counselling

Forensic genetic research on sudden unexpected death in an infant (SUDI) at Salt River Mortuary: experiences and perceptions of parents

By

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DECLARATION

I, Susan Louw, hereby declare that the work on which this dissertation/thesis is based is my original work (except where acknowledgements indicate otherwise) and that neither the whole work nor any part of it has been, is being, or is to be submitted for another degree in this or any other university.

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ABSTRACT

The unexpected and sudden death of an infant (SUDI) is a traumatic event. SUDI is defined as all deaths occurring suddenly and unexpectedly in infants under the age of one year. Molecular autopsy is used to determine the potential genetic contribution to SUDI and may lead to screening and interventions of at-risk family members. However, the potential of this may only be realised if the family members are willing to engage in the follow-up process. Next-of-kin experiences of participating in molecular autopsy research are unknown, and not previously done in South Africa. This study explored the experiences and perceptions of bereaved next-of-kin participating in forensic genetic research on SUDI at Salt River Mortuary, Cape Town.

Methods

Eleven participants, including the mothers and other family members for six SUDI cases participated in the study. These participants were recruited from a larger forensic molecular autopsy study conducted at the University of Cape Town. In order to explore the experiences and perceptions of next-of-kin, a qualitative approach was used and semi-structured interviews were conducted. The interviews, transcribed verbatim, were analysed through thematic analysis. The perspective from the main researcher in the larger forensic molecular autopsy study was included to holistically explore the setting in which the genetics research took place.

Results

Four major themes were identified, namely (i) old wounds, (ii) my booboo, (iii) the sudden death and (iv) afterthought. Their main reasons for participating in the research were to find answers and to be of value in future cases of SUDI. Grief seemed to play a significant role in their understanding and engagement with regards to their research participation.

Conclusion

This study found that the grief and loss of at the time of obtaining consent may play a significant role in understanding and willingness for further engagement with molecular autopsy results. Understanding has previously been implicated in the willingness to engage with genetics results, however, it has not been explored in a mortuary setting. The understanding of genetics research is critical for further engagement that may have implications for the screening of other family members and future offspring. These findings may allow researchers to better engage with participants in genetics research on sensitive topics, including SUDI.

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LIST OF TERMS AND ABBREVIATIONS

CDC	Centre of disease control and prevention
RTI	Respiratory tract infection
SIDS	Sudden infant death syndrome
SUD	Sudden and unexpected death
SUDI	Sudden and unexpected death in an infant
UCT	University of Cape Town

GLOSSARY

Autopsy – Part of post-mortem investigations and consists of macroscopic post-mortem examination and additional investigations. Both forensic and clinical autopsies share the same methods.

Clinical autopsy – autopsy mostly performed in a clinical setting, such as through a hospital. The death was due to natural causes, but the autopsy is performed in order to better elucidate the underlying mechanism of the natural death. Consent is needed from the next-of-kin to perform the autopsy, as it is not mandated by law to investigate further.

Forensic autopsy – autopsy performed in a forensic setting, such as forensic mortuary, which means the death was thought to be unnatural and it is required by law to perform an autopsy.

Molecular autopsy – the molecular investigation of the cause of death through DNA analysis of a sample obtained during autopsy. There is a debate about the use of molecular autopsy as it is similar to post-mortem genetic investigations. The term has been coined in order to promote the use and understanding of the tool as part of post-mortem investigations on sudden death cases (Edwards, 2005). When used in a forensic setting, the term used is forensic molecular autopsy. In the current study, the participants consented to forensic molecular autopsy in a research setting and it is therefore referred to as either forensic molecular autopsy research or the larger forensic genetic study in short.

Post-mortem investigation - include assessment of the medical history of the infant, death scene information and an autopsy.

Sudden infant death syndrome - Where the cause of death remains unknown even after extensive investigations, including death scene investigation, clinical history review and full autopsy (Krous et al., 2004). It is defined by a method of exclusion of all other possible causes of the death.

Sudden and unexpected death - SUD refers to all sudden and unexpected deaths, regardless of the age of the deceased.

Sudden and unexpected death in an infant - SUDI is the umbrella term used for all deaths occurring suddenly and unexpectedly in infants under the age of one year, and includes all cases, whether explained or not (Krous et al., 2004).

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Chapter 1: Foreword

Research plays an integral part in the improvement of evidence-based practice. For the advancement of scientific understanding and intervention to occur, it is not only necessary to subject participants to research, but it is also important to understand how participants experience research participation and their subsequent expectations. This is especially true in settings where the participants may be in a vulnerable position when consenting to research, such as after the sudden loss of an infant.

This chapter serves as a short introduction to the topic of the current study and situates this study within the larger setting of genetics research. A definition of sudden unexpected death of an infant (SUDI) and sudden infant death syndrome (SIDS), and the distinction between these terms are provided here, as well as the rationale, aims and objectives of the current study. The chapter is concluded with an outline of this minor dissertation.

1.1 Background of the umbrella study

The concern of asking for consent for research participation of the next-of-kin was found to be of interest in an ongoing larger forensic molecular autopsy study being undertaken at The University of Cape Town (UCT) (HREC: 445/2015). This forensic molecular autopsy research, referred to as the larger forensic genetic study throughout this paper, formed part of a Doctoral study, which aimed to investigate the underlying genetic contribution towards sudden infant death in a local forensic setting. These infants were admitted to Salt River Mortuary, Forensic Pathology Services, Western Cape, for determination of the cause of death. Genetic analysis was performed on blood samples obtained during a forensic autopsy on these infants who had died suddenly and unexpectedly. Post-mortem forensic genetics refer to the process where presumed human remains or biological samples are investigated using molecular methods to aid in the identification of the deceased individual or to assist in determining a cause of death (Sajantila, Ström, Budowle, Karhunen, & Peltonen, 1991).

Parents at Salt River Mortuary were approached, by Sarah (pseudonym), the researcher of the larger forensic genetic study, to consent to the obtainment of a blood sample from their deceased infant for genetic analysis to investigate any possible genetic contributions to the sudden infant death. This type of post-mortem genetic testing is known as molecular autopsy (Edwards, 2005; Tester & Ackerman, 2012). Molecular autopsy is not, yet, implemented in South Africa. Therefore, this was the first study of molecular autopsy conducted in South Africa and aimed to determine the feasibility of implementing it as standard procedure in a forensic mortuary setting.

An autopsy is performed in both forensic and clinical settings. A forensic autopsy is required by law to investigate deaths which are thought to be *unnatural* according to the National Health Act (South African Government, 2003). This is in contrast to a clinical autopsy where next-of-kin need to provide consent for the autopsy to be performed on a *natural* death in order to elucidate the cause of death. Molecular autopsy refers to genetic analysis performed on samples obtained during the conventional autopsy and is a type of post-mortem genetic testing (Edwards, 2005). Molecular autopsy has not yet been internally validated in South Africa as part of standard practice. Conventional autopsy in South Africa only includes genetic analysis as part of the ancillary tests in a limited number of sudden death cases for identification purposes or determining the presence of a genetic disease. During the larger forensic genetic study, the molecular autopsy was performed as part of research, rather than as part of standard post-mortem investigations, and therefore consent was needed to obtain a biological sample in a forensic setting.

1.2 Sudden infant death

The unexpected and sudden death of an infant is a traumatic event. It can have a profound effect on the parents, as well as other family members and even extending to the surrounding community (Wong & Behr, 2014). SUDI is the umbrella term used for all deaths occurring suddenly and unexpectedly in infants under the age of one year, and includes all cases, whether explained or not (Krous et al., 2004). SIDS, however, is where the cause of death remains unknown even after extensive investigations, including death scene

investigation, clinical history review and full autopsy (Krous et al., 2004). The sudden and unexpected loss of an apparently healthy infant may invoke severe emotional distress which can be exacerbated if the cause of death remains unknown (Vaartjes et al., 2009).

Through decades of research, several risk factors contributing to SUDI have been identified and 'safe sleeping' awareness campaigns have led to a reduction in SUDI rates (Carlin & Moon, 2017). However, the burden of SUDI still remains high, particularly in low- and middle-income countries (Lawn, Cousens, & Zupan, 2005; Stanton, Lawn, Rahman, Wilczynska-Ketende, & Hill, 2006). In attempts to reduce SUDI even further, research has recently expanded to identify possible genetic variants that may predispose to or cause sudden infant death through the use of molecular autopsy (Opdal & Rognum, 2004). This has largely been possible due to massive parallel sequencing and the generation of large-scale genomic data, which has greatly advanced our understanding and the molecular diagnosis of complex conditions and rare diseases (Bloss et al., 2015).

Recognising the genetic contribution to SUDI and being able to determine pathogenic variants through molecular autopsy has potential value for the family, as it may lead to positive health interventions for at-risk family members (Bloss et al., 2015), which will be discussed in Chapter 2. However, such research also has several ethical concerns pertaining to the vulnerability of the participants, the emotional burden on the researchers, as well as the interpretation, validation and feedback of the genetic results to the family, as discussed elsewhere (Heathfield et al., 2017). Furthermore, as the molecular understanding of SUDI is improving through research, it is imperative to reflect on the experiences and perceptions of the participants of genetics research in order to understand which factors may play a role in the understanding of and future engagement in the case of positive results of the next-of-kin. The full potential of a molecular autopsy, including translation of genetic findings back to families, may only be realised if the family members engage in the full process; however, their grief, bereavement and understanding of the impact of these molecular findings may play a significant role in their willingness to engage in follow-up.

1.3 Rationale

Several studies have investigated the perceptions and experiences of family members that had lost an infant through SUDI (Krueger, 2006; Martin, 1998; Merlevede et al., 2004; M. Price, Carter, Shelton, & Bendell, 1985; Sullivan & Monagle, 2011; Vance et al., 1995; Vennemann, Rentsch, Bajanowski, & Zimmer, 2006), however, only a few studies have qualitatively explored the experiences of family members participating in research after the death of a child, including, but not limited to SUDI (Brabin & Berah, 1995; K. Dyregrov, 2004; Kentish-Barnes et al., 2015; Lee & Renzetti, 1990; Newman & Kaloupek, 2004). The overall findings from these studies indicated that research participants had a positive experience from their participation. The overarching motivation identified in these studies indicated that participation was mostly due to altruistic reasons, in that the research can benefit others in the future. Participation in various genetic research studies, has shown similar results, with finding a possible answer to the genetic cause for disease or death as another main motivator (Etchagary, Green, Parfrey, Street, & Pullman, 2013; Forrest, Delatycki, Skene, & Aitken, 2007; Harris et al., 2012; Henneman, Timmermans, & Van Der Wal, 2004; Lakes et al., 2013; Rankin, Wright, & Lind, 2002; Sexton & Metcalf, 2008). Studies on parents' experiences of autopsies performed after infant loss were conducted in developed countries (Rankin et al., 2002; Sullivan & Monagle, 2011; Vennemann et al., 2006) and are discussed further during the literature review. However, to the best of the author's knowledge, no qualitative study to date has explored the experiences or perceptions of *molecular autopsy*, including, but not limited to a forensic mortuary setting in South Africa.

The question arises as to how the parents felt about participating in the larger forensic genetic study that is investigating molecular autopsies at Salt River Mortuary and if they really understood the need for the research. As South Africa is a developing country with a diverse culture and population groups, the emotional response of socioeconomically disadvantaged population groups and cultural differences may play a significant role during autopsy-based research (Odendaal et al., 2011). Furthermore, the marked social differences and lack of access to higher education in South Africa may impact on the understanding of

basic genetic concepts of the research participants (Greenberg, Kromberg, Loggenberg, & Wessels, 2012).

Obtaining consent from parents who just lost a child needs to be strongly justified, as the possibility exists that there is a risk of harm in dealing with such a sensitive topic. The nature of the research and the way in which the participants interact with the researcher may impact on their grieving process, as well as the way the bereaved participants will make meaning of their loss. A further concern is how the participants relate the consent for genetic research to possible future implications for themselves, their families, and possible recurrence risks in sibs of the deceased infant. The research question of this study therefore was: what are the perceptions and experiences of the parents and/or next-of-kin of deceased infants who are participating in a SUDI molecular autopsy research study?

This study hopes to inform current scientific research on molecular autopsies by exploring the experiences of the participants with regards to genetics research in a forensic mortuary setting. These results are intended to help current researchers reflect on their own practices and to inform the ongoing design and execution of forensic genetic studies at UCT and beyond. Through the current study, I hope to demonstrate the need for a genetic counsellor to assist bereaved family members to understand and come to terms with any genetic implications that may be indicated by molecular autopsy, and that the results from this study will motivate for the need for specialised genetic counselling services in a medico-legal setting. I further expect that the knowledge gained through this study will help researchers become more aware and accommodating of vulnerable participants, such as grieving family members and lead to better insight into the motivations, concerns, expectations and perceptions of the participants of a genetics research programme. The overall value of this study will be to minimise the distress of participants and maximise the potential benefits of genetics research, including screening and possible prevention.

1.4 Aim

This research aimed to explore the parents' perceptions, experiences and understanding of consenting to molecular autopsy research on their infant after a sudden death for cases reported at Salt River Mortuary in Cape Town. The rationale for this is described in section 1.2 and relates to the paucity of literature, the importance of the researcher and setting in contributing to the understanding and full engagement in molecular autopsy research of bereaved next-of-kin, and the emotional vulnerability of the participants due to a sudden infant loss.

1.5 Objectives

With the above in mind, an interview guide was developed to explore the experiences, perceptions and knowledge of mothers and other family members participating in forensic molecular autopsy research.

The objectives of this study were to investigate the parents':

a) experience of being asked for consent in a forensic mortuary setting where they have to undergo extensive questioning with regards to the death of their child;
b) reasons/motivations for consenting to the molecular autopsy;
c) perceptions of the value of molecular autopsy on their infant;
d) understanding of the genetics involved in the molecular autopsy and SUDI.

1.6 Outline of this study

Chapter 2 consists of a literature review that entails a comprehensive overview of the existing literature with regard to the need for genetic research participation, research involving bereaved parents, ethics, SUDI/SIDS and molecular autopsy; Chapter 3 discusses the methodology and process of the research study. Chapter 4 and 5 consists of a comprehensive overview of the results obtained using the methods as described in Chapter 3. The greater research setting as a contextual factor of the participants' experiences,

perceptions and knowledge is explored in Chapter 4. This chapter is presented to highlight the researcher's voice in this study. The participants' experiences and their voices are presented in Chapter 5. Both Chapter 4 and 5 are further informed by a discussion on the existing literature. A conclusion is drawn based on the findings and discussions presented in Chapter 4 and 5. This conclusion, limitations and future possibilities of the research are discussed in Chapter 6. References follow the conclusion.

Chapter 2

Sometimes I will hug you far too tight

Check on you ten times a night

Sometimes I will kiss you a little too much

Cling to you, sing to you, tickle you and such

As you grow up I will kiss your nose

Touch your hair and feel your toes

I will cry when you walk, laugh when you talk

Worry as I sit and wait and scream when you come home late

I do not want you to have my fears

So I will often pull you close to hide my tears

Because you see I'm not just your mother

A long time ago, I lost another

(S. Saunders, 2012)

2.1. Introduction

The purpose of this chapter is: to present the literature pertaining to research participants' experiences in genetics *or* SUDI research, including their perceptions, knowledge and concerns; to explore the current gap in knowledge with regard to the experiences of research participants that experienced the unexpected death of an infant. The focus of the literature is on previous research participation in the field of genetics as well as in sensitive topics, ethical aspects, and the experiences of research participation. However, in order to contextualise this discussion, background information is provided on trends in epidemiology, risk factors and investigative procedures related to sudden infant death and the process of grief following an infant death. Sudden infant death is a well-studied phenomenon and literature dates back a few decades. As a result, a number of these older that are invaluable in informing the field, were referenced.

Research articles were obtained by searching several databases employing the following terms: *SIDS, SUDI, genetics research participation, bereavement research participation, genetic counselling, infant death experiences* and variations of these terms. The databases searched included PubMed, Google Scholar and PRIMO, a UCT search engine. The American Psychological Association (APA) sixth edition referencing style is used (American Psychological Association, 2010).

2.2 Background

2.2.1 Experiences of sensitive topics research participation

There has been some concern about the experience of being interviewed about the death of a child. The loss of a child has been described as a sensitive topic, as the bereaved parents may be in a period of grief. Wheeler (2001, p.52) describes parental bereavement as 'a search for meaning that has been shattered by crisis, along with the development of a new set of assumptions about themselves and the world.' Participants are in a vulnerable state and their situation demands special consideration from researchers (K. Dyregrov, 2004). Research participation may impact on the meaning-making and grief process of the bereaved next-of-kin, which may lead to potential ethical issues when using qualitative approaches in such sensitive areas (Krueger, 2006). Such ethical issues include concerns about whether the participants experience emotional distress, and if this influences the informed consent process, which is essential to participation. However, it should not just be assumed that subjecting bereaved parents to an interview would unavoidably cause them harm. It has been demonstrated that bereaved parents may benefit from sharing their stories with a considerate and emphatic, informed researcher (A. Dyregrov & Dyregrov, 2004).

A number of studies have aimed to explore the experiences of family members participating in bereavement research (Brabin & Berah, 1995; Cook & Bosley, 1995; Decker, Naugle, Carter-Visscher, Bell, & Seifert, 2011; K. Dyregrov, 2004; Neimeyer, 2000; Newman & Kaloupek, 2004). Across these studies, minimal participants reported high levels of distress.

Participants also reported some sort of benefit or gain through participation. This was found across a variety of research topics and methodologies (Newman & Kaloupek, 2004). Some benefits of participating in research as described on sensitive topics, included: gaining self-awareness; clarity of thought with regard to their traumatic experience; possibly helping someone else; and promotion of public awareness with the potential to lead to positive social change (Decker et al., 2011). Positive effects were also reported in the case of bereaved parents who were asked to share their experiences of participating in extensive interviews after the stillbirth of their baby (Brabin & Berah, 1995). This was also the case in a qualitative study with bereaved parents who previously participated in research in order to determine the experiences of the parents in an Intensive Care Unit (Kentish-Barnes et al., 2015). This positive effect was mainly ascribed to the feelings of being able to assist others in the future and contributing to healthcare professionals' understanding of grief.

K. Dyregrov (2004) also cites a growing body of research that showed research participation in bereavement studies may be enlightening, enriching, remedial or empowering for vulnerable populations. Through these research findings, awareness has been created relating to maladaptive behaviours to bereavement, as well as the development of possible intervention strategies to decrease the likelihood of developing psychological stress after bereavement.

Participating in a research study on SUDI can also be classified under bereavement research, as the research would typically take place just after the loss of an infant. Sudden infant death has occurred for many centuries and as such, numerous studies have been undertaken to understand the effect of sudden infant death on several affected individuals, including the mothers, parents, family members, professional and health services (Garstang, Griffiths, & Sidebotham, 2014; Krueger, 2006; Lepore, Silver, Wortman, & Wayment, 1996; Merlevede et al., 2004; M. Price et al., 1985; Vance et al., 1995). However, only a few studies have explored these individuals' experiences of participating in research following the sudden death of an infant.

A comprehensive and informative review study was done by Garstang et al. (2014) on bereaved parents' needs after the loss of a child. This study used thematic analysis and a

narrative process across 52 papers from Europe, North America and Australasia and detailed the experiences of 4 000 bereaved parents. They concluded that parents need to be allowed time to say goodbye to their child and it is essential that parents are able to understand why their child died. Furthermore, parents value any emotional support during the time and after the time of their child's death (Garstang et al., 2014). The following figure was provided by Garstang et al. (2014) as recommendations for healthcare professionals in dealing with child death, including SUDI.

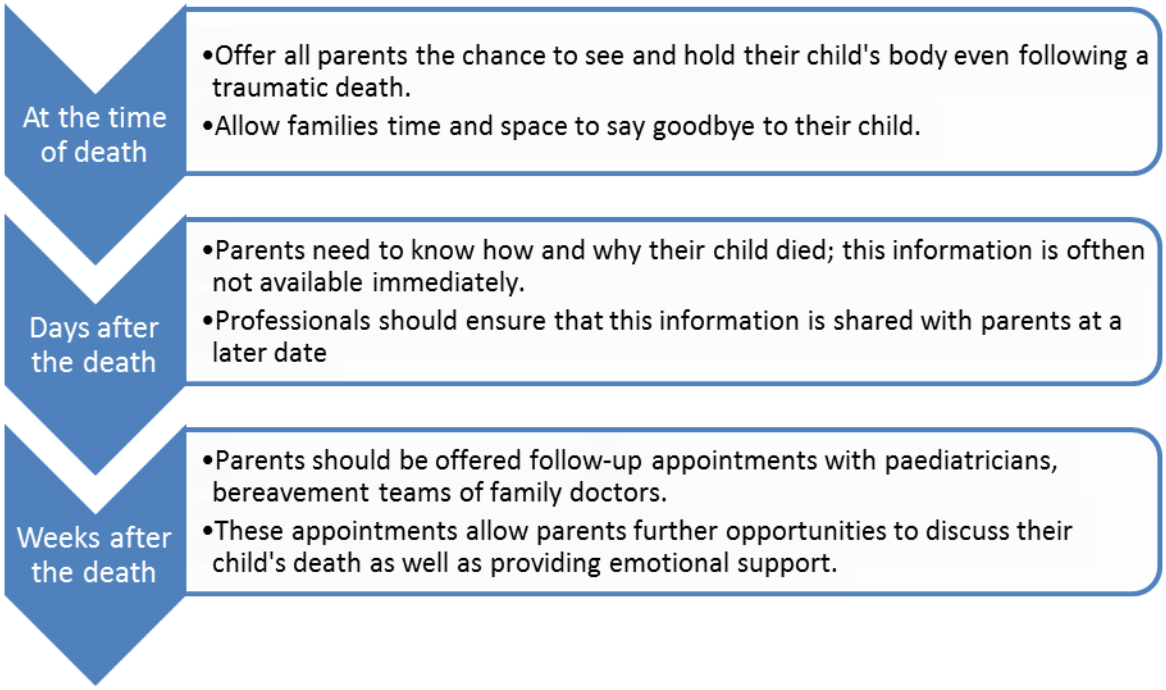


Figure 1 Summary of recommendations of health care professionals as provided by Garstang et al.(2014).

2.2.2 Epidemiology of sudden unexpected death in an infant

The incidence of SUDI and SIDS differs across countries, ethnic groups and sex (Moon, Horne, & Hauck, 2007). It is important to note that the terms SIDS and SUDI are unfortunately often used interchangeably in older literature. As the definition of SIDS and SUDI were updated in 2004, the terms are used as defined. However, internal procedures with regards to the classification of SIDS and SUDI may be interpreted differently (Corey, Hanzlik, Howard, Nelson, & Krous, 2007). This review uses the term sudden infant death for articles published before 2004, in order to circumvent the complexities in the methodology

of determining SIDS by definition of exclusion and to create consistency. SUDI includes SIDS cases as stated by the current use of the definition.

Due to the interchangeable use of SUDI and SIDS in the literature, there were difficulties in determining the exact incidence of SUDI. In England and Wales the average rate of SUDI/SIDS was 0.45 per 1 000 live births from the period 2004 to 2014 (Office for National Statistics, 2016). In America, the estimated incidence is approximately 0.49 per 1 000 live births, with SIDS reported as the main cause of death in infants under one year of age (Carlin & Moon, 2017). The rate of SUDI is noticeably higher, as it encompasses all SIDS cases as well as those infants who have a cause of death determined at autopsy (Carlin & Moon, 2017).

In South Africa, a retrospective study performed at medico-legal mortuaries in Pretoria and Tygerberg in Cape Town reported 813 (6.7%) infant deaths (younger than 1 year) out of a total 12 181 cases for the period of 2000 to 2004 (Du Toit-Prinsloo, Dempers, Wadee, & Saayman, 2011). Of these, 99/344 (28.8%) and 413/469 (88.1%) were admitted as SUDI to Pretoria and Tygerberg mortuaries, respectively. For the five-year period between 2005 and 2009, medico-legal mortuaries in Bloemfontein, Durban, Johannesburg, Pretoria and Tygerberg processed 80 399 cases with 3 295 (4.1%) of these being infants. Out of the infant group, 2 704 (82.1%) were identified as SUDI (Du Toit-Prinsloo et al., 2013). A current ongoing investigation by the University of Cape Town (UCT) determined an average of about 260 SUDI cases per year, with an overall decrease in the SUDI rates (Mathews et al., 2016).

2.2.3 Global investigation into SUDI

Numerous investigations have been undertaken to try and determine the cause of the sudden demise of an apparently healthy infant. Many different theories have been proposed, which include social, physiological, environmental and genetic risk factors. Recently an influential global collaborative effort was made to establish further research priorities to better understand and reduce the incidence of SUDI (Hauck et al., 2017).

One of the most well-known and accepted theories on SIDS, is the triple-risk model as proposed by Filiano and Kinney (1994). This model states that the risk for SIDS is most likely when three overlapping factors occur simultaneously, including: (i) a vulnerable infant; (ii) a critical period of homeostatic control during development; and (iii) an extrinsic stressor or precipitating event (Filiano & Kinney, 1994). A critical review of this hypothesis, together with other existing triple risk models for SUDI was published in the Journal of Paediatric and Child Health (Spinelli, Collins-Praino, Van Den Heuvel, & Byard, 2017). Overall, the theories stipulated that SUDI is a multifactorial disorder with interplay between intrinsic and extrinsic factors that may either result in or predispose the infant to a sudden death. When multiple risk factors are present, it can act in a cumulative way to further increase the risk for SUDI. Determining the underlying factors of SUDI can create the possibility to actively intervene to reduce the incidence of sudden deaths in infants (Campuzano et al., 2014; Hauck et al., 2017).

2.2.4 Risk factors

An in-depth review of risk and protective factors that have been commonly associated with SUDI, are provided by Carlin and Moon (2017) and a further review on factors that are less often addressed are provided by Hauck and Tanabe (2017). Table 1 provides an overview of the well-known environmental risk factors (Hunt & Hauck, 2006). In addition, protective factors include breast feeding, vaccinations and the use of pacifiers. Even though there are extensive research studies and informed risk reduction campaigns, SIDS still remains one of the leading causes of infant mortality in developed countries (Carlin & Moon, 2017; Hauck et al., 2017; Moon et al., 2007).

Table 1 Environmental risk factors for sudden infant death; adapted from Hunt and Hauck (2006).

Maternal and antenatal risk factors	Infant risk factors
Smoking	Colder season, no central heating in cold climates
Intrauterine hypoxia	Age (peak 2 - 4 months)
Illegal drug use (especialy opiates)	Male sex
Inadequate prenatal care	Prematurity
Fetal growth retardation	Soft sleeping surface, soft bedding
Young maternal age	No pacifier ('dummy') used at bed time
Low level of education	Exposure to tobacco smoke
Single marital status	Prone or side sleeping position
Increased parity	Recent febrile illness
Short interval between pregnancies	Thermal stress/overheating
Alcohol use (especially periconceptionally and in first trimester)	Race/ethnic background (e.g., Black, Native Indian, other indigenous group)
Low socioeconomic status (SES)	Face covered by bedding
	Sharing bed with parents or siblings
	Sleeping in own room rather than parents' room

A retrospective study of SUDI cases in Canada from 1987 to 1996 (n=625) determined the following main causes: 80% remained SIDS, 7.1% infections, 2.7% cardiovascular abnormalities, 2.6% child negligence or abuse, and 2.1% metabolic or genetic disorders. In South Africa in 2009, an estimated 37 974 infants died and 97.3% were declared as death due to natural causes. The leading cause of death was intestinal infections (17.4%), closely followed by respiratory and cardiovascular conditions (15.2%) and influenza and pneumonia (15.2%) (Statistics South Africa, 2009). The incidence of SUDI versus SIDS could not be clearly established. Retrospective studies in Western countries have revealed that without a forensic autopsy, as much as 41% of cases are classified as SIDS (Turs, Crost, Gerbouin-Re'rolle, & Cook, 2010).

2.3 Post-mortem investigations

2.3.1 The traditional autopsy

The medico-legal investigation of SUDI has been implemented in most countries (Du Toit-Prinsloo et al., 2013). In South Africa, any case of sudden, unexpected or unexplained death has to be admitted to a medico-legal mortuary for further investigation in order to determine a cause of death (South African Government, 2003).The Salt River Mortuary is responsible for performing all autopsies of all deaths thought to be unnatural in the western part of the Western Cape metropole and works in association with UCT as a learning and research facility (Western Cape Government, n.d.)

Post-mortem investigations include assessment of the medical history of the infant, death scene information and an autopsy (Ludwig, 2002). The nature of the autopsy can be divided into a macroscopic post-mortem examination and additional investigations. The macroscopic autopsy can further be classified into different categories ranging from no autopsy, as cause of death is most likely due to an underlying disease, external examination only, known as 'viewings', partial evisceration and dissection where the process is stopped once the cause of death is ascertained, and full macroscopic autopsy with complete evisceration of the body and dissection of the organs. The additional investigations includes histology, toxicology, bacteriology, virology, biochemistry, metabolic studies, radiology and genetic studies in some cases (Du Toit-Prinsloo et al., 2011).

At Salt River Mortuary, international guidelines are followed for the investigation of SUDI (Centers for Disease Control and Prevention (CDC), 1996). As part of the protocol for the autopsy, the next-of-kin are asked to provide information on a multitude of factors, including known risk factors (refer to Appendix A). Most SUDI cases at Salt River Mortuary are found to have demised from respiratory causes (Groenewald et al., 2016; Mathews et al., 2016). Due to most infant deaths being classified as *natural* deaths, resources are not allocated for further investigation.

There remains a burden of infant death across all mortuaries in South Africa, and some SUDI cases remain as having an undetermined cause of death even after extensive post-mortem investigation (Du Toit-Prinsloo et al., 2011). As conventional autopsy is unable to provide insight into the underlying genetic mechanisms which may contribute to SUDI, new methods have been developed to investigate possible genetic contributions. These include molecular autopsy, which has shown potential in resolving cases which were previously unresolved.

2.3.2 Molecular autopsy

Molecular autopsy has been defined as a tool that can be used in post-mortem investigations in sudden death cases to explore a number of genetic variants that may cause or contribute to a sudden death. This created the possibility of resolving the uncertainty left by the lack of a cause of death or SIDS (Ackerman et al., 2011).

Developments in genomics research and the genetic analysis of SIDS have greatly advanced the knowledge of the underlying genetics risk for these conditions (Campuzano et al., 2014; Männikkö et al., 2018; Tester et al., 2018). Studies using a molecular autopsy were initially limited to cardiac channelopathies and epilepsy genes in all sudden deaths under the age of 45 years. These studies uncovered a genetic diagnosis in approximately 25% of previously unresolved cases of sudden deaths. Later molecular autopsy research include inborn errors of metabolism, the central nervous system, infections and nicotine response in sudden and unexpected death (SUD) cases (Van Norstrand & Ackerman, 2010). For a full review on the genetic contribution to SUDI, see Bloss et al. (2015), Van Norstrand and Ackerman (2010) or Odal and Rognum (2004).

2.3.3 The potential value of the molecular autopsy

As described in the section 2.2.6, the aim of the molecular autopsy is to use genetic testing to help resolve the cause of death (Ackerman et al., 2011). With the cause of death undetermined, it may leave the next-of-kin with incomplete and ambiguous information

with regards to their family history (Bloss et al., 2015). With a determined cause of death, the emotional burden of the parents may be lessened as it may assist with meaning-making during the grieving process, leading to acceptance and peace of mind. It may also lessen feelings of guilt of their own responsibility towards their child's death. The potential exists for early testing and treatment for blood relatives if a pathogenic variant is found (Forrest et al., 2007; Sexton & Metcalf, 2008).

2.4 Research participation

2.4.1 Experiences of autopsy research participation

Studies have been conducted to determine parents' perceptions of having an autopsy performed on infants (Rankin et al., 2002; Sullivan & Monagle, 2011; Vennemann et al., 2006). In a study where autopsies were performed on SIDS cases in Germany from 1998 to 2001, a follow-up survey study was done four to seven years after the parents (n=141) lost their infant. This survey indicated that 83% of the bereaved parents found that having an autopsy performed helped them come to terms with and cope better with the death (Vennemann et al., 2006).

In South Africa, the Safe Passage Study performed by Odendaal et al. (2011) found the reasons for participation in autopsy research is similar to other bereavement research findings. The reasons included the desire to know the specific cause of death and to help others in similar circumstances in the future. This study included 12 000 woman and their infants from a multiracial population group in Cape Town and American Indians from the Northern Plains described that autopsy-based research in socioeconomically disadvantage groups may raise unique ethical concerns (Odendaal et al., 2011). However, little is known with regard to molecular autopsy.

2.4.2 Experiences of genetics research participation

Participation in genetics-related research encompasses a very broad spectrum of research. Nature.com defined genetics research as the scientific discipline that deals with the study of the role of genes in traits, including, but not limited to, the development of disease. It has an essential role in understanding genetically based variation in populations and identifying potential targets for therapeutic intervention ("Genetics Research," 2018). Genetics research has led to an increased understanding of the pathogenesis of a large variety of genetic conditions and created the opportunity for improving health (Begleiter, 2002; Etchagary et al., 2013). However, there are still concerns about how genetics information will be used to realise its full potential in improving health and health care practices. Exploring how the public perceives and understands genetic information and how it influences their health decisions, is essential for the implementation of genetic services (Etchagary et al., 2013) .

A key aspect of realising the potential of genetics is linked to public acceptance (Etchagary et al., 2013). For genomic medicine to be successful, the public will have to share their family history, medical and lifestyle history and genomic information with health-care providers, as well as be willing to engage in self-monitoring and management of health-related behaviours. Examples of successful implementation of genomic medicine in clinical practice include malignant tumour genotyping in cancers and the use of pharmocogenomics in medication selection for patients whose genotype influence the way they metabolise specific drugs (Etchagary et al., 2013). Even though it is generally accepted that genomic research has the potential for the advancement of treatment and improving healthcare, some risks and public concerns have been recognised. General concerns relate to appropriate consent models, the feedback of genomic research results, ownership of genetic information and data sharing policies (Lemke, Wolf, Herbert-Beirne, & Smith, 2010).

There is a growing body of literature focusing on public attitudes towards genetic research (Etchagary et al., 2013). Through the community sessions it became clear that the participants felt that they lacked knowledge with regards to genetics and related research. The community members proceeded to ask numerous questions in these sessions. They

were found to be optimistic about the potential of genetics research to lead to improvement in health care. The implication for their families and the return of individual results were brought up as a concern (Etchagary et al., 2013).

A further online survey on public opinions in the USA explored different influences on the public's motivation to participate in genetics studies (n=4 659). The aim was to improve the understanding of motivating factors that would increase the recruitment and retaining of participants in large genetic cohort studies. The results from the study indicated that 84% of respondents supported a large genetic cohort study and 60% indicated their willingness to participate. The largest motivator for participation was receiving feedback on the genetic results, with 75% of the respondents indicating that they would be less likely to participate if the research results were not returned. Increased compensation was the second leading factor to increase motivation to participate (Kaufman, Murphy, Scott, & Hudson, 2008).

2.5 The ethics of conducting molecular autopsies on the young

As molecular autopsies are now becoming more integrated in mortuary settings, recommendations have been provided on ethical issues that arose with conducting molecular autopsies in the young. These recommendations were developed in order to resolve ethical and policy issues and to promote the responsible conduct of molecular autopsies (Mcguire et al., 2016). These recommendations are clearly outlined by Mcguire et al. (2016) and cover consent, confidentiality, genetic analysis and disclosure of the results. These recommendations were informed by a multidisciplinary consortium in the United States of America, with experts in the fields of pathology, medical genetics, genome science, genetic counselling, bioethics and law. Recommended guidelines are presented in Box 1 of their publication (Mcguire et al., 2016) and are also provided in Box 4.1 below for ease of reference.

BOX 4.1 Summary of recommendations

Before conducting molecular autopsies in cases of sudden death in the young, a multidisciplinary team of medical examiners or coroners, clinical geneticists, genetic counsellors and cardiologist should be established. The team should consult other specialised medical professionals, bioethicists and lawyers when appropriate. All of the recommendations require communication and collaboration among the team.

Consent

Recommendation 1.1 Reasonable efforts should be made to notify the decedent's family before conducting genomic analysis as an aspect of death investigation or when a positive result is obtained.

Recommendation 1.2 When notifying families about the planned analysis, members of the team (as appropriate) should (1) solicit consent to obtain samples from biological relatives, if available, for confirmatory testing, (2) offer family and biological relatives an opportunity to opt out of receiving genetic tests results, and (3) discuss opportunities for research participation.

Confidentiality

Recommendation 2.1 Although genetic test results may become part of the cause of death statement and autopsy report, the public release of post-mortem genetic test results should generally be treated as an 'unwarranted invasion of personal privacy' and exempted from disclosure under state and federal Freedom of Information Acts.

Analysis

Recommendation 3.1 Causality of any given gene variant should be determined through a process that includes (1) review of the literature, (2) careful manual curation of multiple databases, (3) validation of the finding by an independent method (usually Sanger sequencing), (4) if carried out in a research laboratory, confirmation of the result in a certified diagnostic laboratory, (5) multidisciplinary collaboration, and (6) other useful methods.

Recommendation 3.2 Validation testing of available first degree biological relatives should be carried out whenever possible.

Disclosure

Recommendation 4.1 Positive results that are confirmed and determined to be causative or likely causative should be disclosed to the next-of-kin and first degree biological relatives who have submitted samples for validation testing. The potential risk to other biological relatives should be clearly communicated.

Recommendation 4.2 When communicating negative findings, families should be counselled on the limitations of our current knowledge and technology to detect all genetic risk factors for SDY.

Recommendation 4.3 The disclosure discussion with the family and biological relatives should include a genetic counsellor or clinical geneticist.

Recommendation 4.4 All first-degree relatives of the decedent with a presumed pathogenic variant should be referred to a genetic specialist familiar with issues in diagnostic DNA sequencing and the interpretation of the lab reports, and a clinical specialist who has familiarity with the specific disorder and expertise in evaluation, follow-up, and management of these rare disorders.

Recommendation 4.5 The ME/C should remain involved in the case even after disclosure because follow-up genetic testing of biological relatives could affect the determination of the cause and manner of death of the decedent.

When conducting research on molecular autopsy, these guidelines can assist with addressing ethical concerns in research and can ensure that data collection can proceed without any harm to the research participants (Decker et al., 2011). These recommendations informed the consent model in the research implementation of molecular autopsies in South Africa (Heathfield et al., 2017). The guidelines may be applicable in a research setting and can be implemented in the ethical framework of the research study. In order to obtain a DNA sample for the molecular autopsy research informed consent is needed from the parent(s) or next-of-kin of the deceased infant.

Obtaining consent from parents after the loss of their child then comes with a complex set of additional ethical considerations which were explored by Heathfield et al. (2017). Some of the ethical issues raised in the molecular forensic autopsy research setting included the feedback of results, incidental findings and the time of obtaining informed consent. Potential incidental harms of molecular autopsy findings could include revealing alternative parentage, or raising unnecessary concern and anxiety about genetic mutations in future pregnancies or previous children (Mcguire et al., 2016).

Even though results from both genetics research and research on sensitive topics have indicated no harm to the research participants, there is a lack of investigations into the experiences of participating in molecular autopsy research. As the family may be implicated through the use of the molecular autopsy, it is necessary to explore what participants understand with regards to the genetic testing of their deceased infant.

Chapter 3: Materials and Methods

The aim of the current study was to capture and explore the perceptions, experiences and knowledge of parents participating in forensic molecular autopsy research after the loss of their baby. This is important, as the participants' story with regard to their involvement in genetics research in a forensic mortuary setting in South Africa has not been told before. In order to elucidate how the aims of the research study were achieved the methodology of the study is presented in this chapter. I conclude the chapter with a short self-reflection and how I view my position within this research study.

3.1 Study design

In order to ascertain the experiences and perceptions of the parents who consented to molecular autopsy research after the sudden death of their infant, an in-depth qualitative approach was deemed the most appropriate. This approach aims to understand the experiences of the participants' from their own point of view (Leedy & Ormrod, 2005). The qualitative approach was chosen as it allowed me to enter the world of the participants and hear their own point of view of their experiences participating in the larger forensic genetic study on SUDI. Qualitative research is a holistic approach to research and aims to explore and recognise the meaning that an individual assign to their personal experiences. The role of the researcher in qualitative research is to clarify the complexity of individual experiences that may be poorly understood (Hesse-Biber & Leavy, 2010). It further requires the researcher to engage with the personal experiences of the research participants (Morse, 2012). This in turn may lead to better understanding of the social phenomenon (Silverman, 2015). The current research drew on the principles of Phenomenology, which is concerned with understanding phenomena through the lived experiences and descriptions of a social group, without the prior assumptions and experiences of the researcher influencing the data (Denscombe, 2008). It was essential to engage in self-reflexivity throughout the process in order for me to be aware of any prior or emerging personal beliefs that may influence the data. An important aspect of qualitative research is that social phenomena

may be viewed from a myriad of perspectives, however, the qualitative researcher has a duty to use an honest approach without any preconceived ideas and prejudice (Beeson, 1997).

Additional criteria of qualitative research is based on contextualisation and authenticity as described by Damico and Simmons-Mackie (2003). This pertains to the need to study certain social phenomena in its natural setting as the specific context have the ability to influence the behaviours of the research participants. This approach allowed for the exploration of the larger context in which the participants' experiences were created. Therefore, factors that may have had an influence on the capturing of the data of this study were recorded and analysed in greater depth (Damico & Simmons-Mackie, 2003). The experience of asking for research participation in a mortuary setting was therefore also investigated from the viewpoint of the researcher of the larger study, Sarah.

3.2 Study population

3.2.1 Participants

Participants were recruited from a larger forensic genetic study that started recruitment on 1 March, 2016. The study is titled 'Sudden unexpected death of infants: Genetic contributions in a South African cohort' and has updated their ethical approval (HREC: 445/2015) to include the current study. Full informed consent was obtained for this genetics study performed at Salt River Mortuary on infants who passed away unexpectedly. At the time of study, 145 families had been recruited for the larger study, of which 47 had also consented to being contacted again for follow up research.

Ethics approval and informed consent was also obtained for the current study, this is discussed further in section 3.2.4. The parents had been asked for a DNA sample from their deceased infant to be obtained at autopsy in order to investigate the genetic variants possibly contributing to the sudden death of their infant. All bereaved mothers who had consented to follow up study (n=47) and other family members also present for the initial

consent process, were approached to participate in this project. Of those, six mothers consented to be interviewed for the current study. Although it is a small sample, it is acceptable in qualitative research (Braun & Clarke, 2006).

Through the unexpected difficulties encountered in the recruitment of the participants, it became necessary to explore the larger context in which the participants functioned. As such, the main researcher (Sarah) of the forensic molecular autopsy study was also asked to participate in order to provide her view and perceptions of asking for research participation from parents who had just lost their infant. Sarah is completing her PhD in Forensic Genetics and had asked the parents for informed consent for the larger research study. She therefore has valuable knowledge to contribute to this study as a participant.

3.2.2 Research setting

Participants' infants underwent an autopsy at Salt River Mortuary. Salt River Mortuary is responsible for the medico-legal investigation of all deaths thought to be unnatural in the Western metropole of the City of Cape Town, as discussed in Chapter 2 section 2.3.1. This mortuary conducts approximately 3 500 autopsies per year (Heathfield et al., 2017). This was also the place where families were recruited by Sarah to participate in the larger forensic molecular autopsy study.

The sociodemographic information of all deceased infants in the current study is captured as part of the investigations performed by Salt River Mortuary (Appendix A). This information was obtained from the next-of-kin during their time at the mortuary, before they had to identify and see their baby for the last time. This may take a variable amount of time. Sarah obtained permission from Forensic Pathology Services, Western Cape Government to access the information of all sudden infant death cases reviewed at Salt River Mortuary and is presented in Table 2.

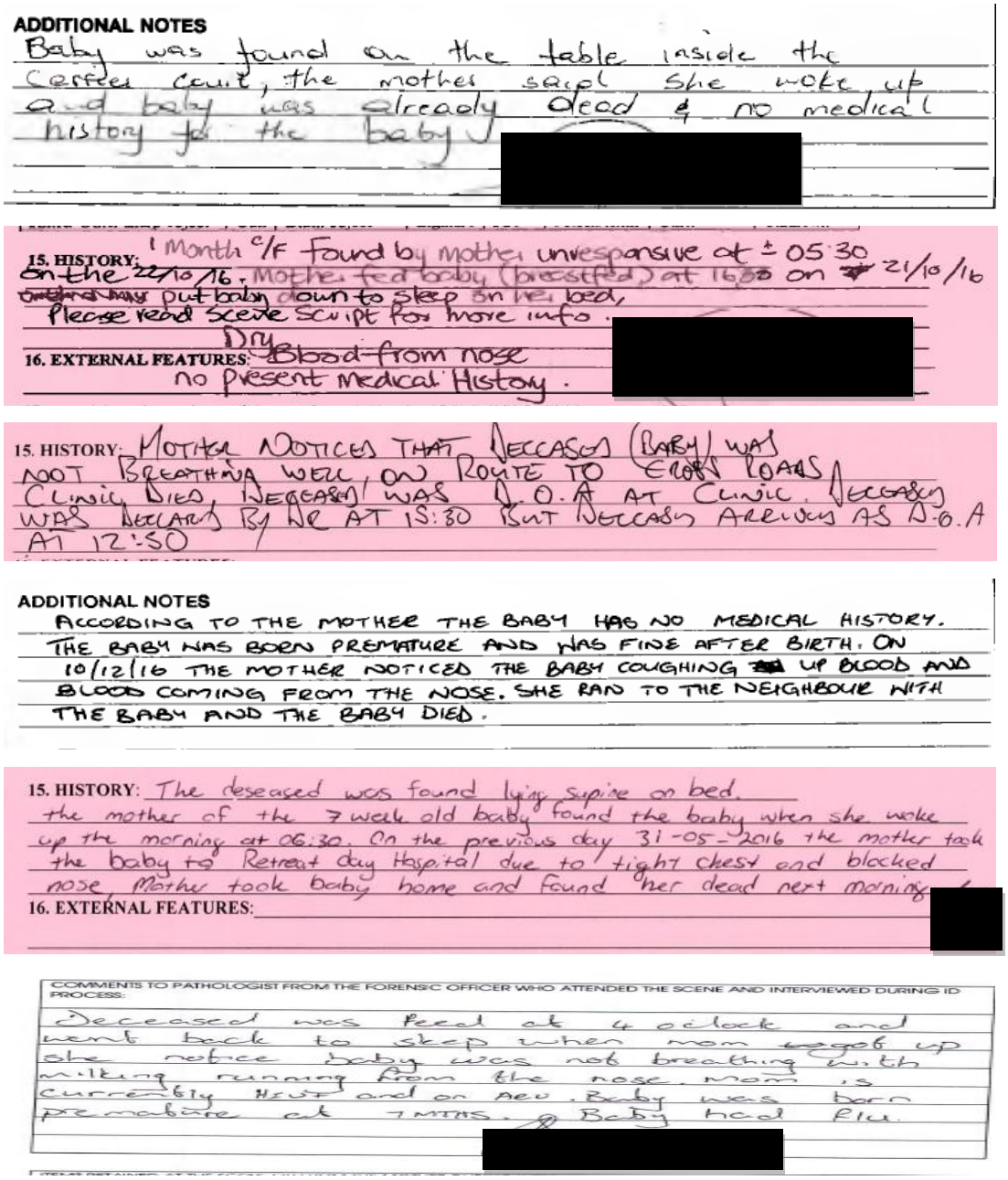
Table 2 Demographic information from Salt River mortuary as per CDC requirements, including race, time of death, sex, age, cause of death, possible SUDI risk factors, area of death and extra information.

Nr.	Infant	*Race	Time of death	**Sex	Age	Cause of death	Possible risk factors	Area of death	Extra information
1	Baby of Mary	C	2017, morning	F	2 months	RTI***	Breastfeeding, born at home, slept on side, foam mattress, slept with mother and 3 other sibs	Mitchel's Plain	No disease symptoms
2	Baby of Sarah	C	2016, morning	F	1 month	Natural	Premature, underweight, co-sleeping, slept on side, foam mattress, recently went to hospital	Atlantis	No formal housing, backyard dwellers, shack well ventilated, one big bed for family of 4 and baby,
3	Baby of Louise	B	2016, afternoon	F	3 months	RTI	Coughing, shortness of breath, Co-sleeping with mum and twin, side-sleeping, inner spring mattress, bottle-fed	Nyanga	
4	Baby of Dianne	C	2016, morning	F	2 months	RTI	premature, baby coughing up blood, co-sleeping, prone sleeping (stomach), 2 pillows, bottle-fed	Manenberg	
5	Baby of Nosi	C	2016, morning	F	7 weeks	RTI	prone-sleeping (stomach), inner spring, breast feeding,	Grassy Park	Took to hospital - tight chest
6	Baby of Vuyelwa	B	2016, morning	M	2 months	RTI	Premature		Forms not completed

***Race: C – Coloured, B – Black, W; **Sex: F – Female, M – Male; ***RTI – respiratory tract infection**

From Table 2, it is clear that these infants were in a critical period of development, where infants less than 4 months old that have been well described as a risk factor (Hunt & Hauck, 2006). The cause of death was found to be due to respiratory tract infection (RTI) in five of the cases. This would fall under natural causes of death, as no external factors were suspected that may have caused the death.

As well as this demographic information, the SUDI questionnaire also requires a description of the last moments before the infant death. Figure 2 captures the last notes on the babies' lives. The signatures are blocked out for confidentiality reasons.


ADDITIONAL NOTES
Baby was found on the table inside the coffee cart, the mother said she woke up and baby was already dead & no medical history for the baby [redacted]

15. HISTORY: 1 Month C/F Found by mother unresponsive at ± 05:30 on the 22/10/16. Mother fed baby (breastfed) at 1600 on 21/10/16 and then put baby down to sleep on her bed, Please read Scene Script for more info. [redacted]
16. EXTERNAL FEATURES: Dry Blood from nose
no present medical History.

15. HISTORY: MOTHER NOTICES THAT DECEASED (BABY) WAS NOT BREATHING WELL ON ROUTE TO EROS LOANS CLINIC, DIED. DECEASED WAS D.O.A AT CLINIC. DECEASED WAS DECLARED BY NR AT 15:30 BUT DECEASED ARRIVED AS D.O.A AT 12:50

ADDITIONAL NOTES
ACCORDING TO THE MOTHER THE BABY HAS NO MEDICAL HISTORY. THE BABY WAS BORN PREMATURE AND WAS FINE AFTER BIRTH. ON 10/12/16 THE MOTHER NOTICED THE BABY COUGHING [redacted] UP BLOOD AND BLOOD COMING FROM THE NOSE. SHE RAN TO THE NEIGHBOUR WITH THE BABY AND THE BABY DIED.

15. HISTORY: The deseased was found lying supine on bed. the mother of the 7 week old baby found the baby when she woke up the morning at 06:30. On the previous day 31-05-2016 the mother took the baby to Retreat day Hospital due to tight chest and blocked nose, Mother took baby home and found her dead next morning.
16. EXTERNAL FEATURES: [redacted]

COMMENTS TO PATHOLOGIST FROM THE FORENSIC OFFICER WHO ATTENDED THE SCENE AND INTERVIEWED DURING ID PROCESS:
Deceased was feed at 4 o'clock and went back to sleep when mom woke up she notice baby was not breathing with milking running from the nose. Mom is currently HIV and on ARV. Baby was born premature at 7 mths. Baby had flu. [redacted]

Figure 2 Last notes captured on the death of the infants as part of the investigations as Salt River Mortuary.

The majority of participants were asked for consent before the extensive questionnaires as per CDC guidelines and viewing the body. However, due to logistical reasons, the minority of participants were asked for consent after these procedures.

In order to immerse myself in the world of the participants, I visited Salt River Mortuary (see Figure 3 below). An ethnography of my experience/observations is provided in Appendix B. All the interviews for the current study were conducted at UCT. Therefore, the research setting for the current study is in the Western Cape Metropole, South Africa.

Figure 3 The Salt River Mortuary in Cape Town, Western Cape, South Africa.

3.2.3 Participant recruitment

Purposive sampling was used in this study. Purposive sampling involves the selection of participants that are to be expected to have experience in the particular phenomenon being investigated and therefore likely to provide in-depth information (McMillan & Schumacher, 2001). The potential participants had experienced the sudden loss of their baby and consented to participation in the larger forensic genetic study and as such, were suitable for selection of the current study participation. The sampling method was used to gain deeper understanding into the experience of participants, rather than to generalise to a larger population (Denscombe, 2008).

Due to the sensitive nature of the topic, participants were first contacted telephonically by the genetics researcher, Sarah, who had asked for their consent for molecular autopsy. This

was done to continue the trusting relationship that had been built with Sarah, as opposed to a stranger contacting them. Potential participants were asked if they were still willing to participate in further research following their consent after participating in the larger study. They were told that the interview pertains to their experiences and expectations of their involvement in the larger molecular autopsy study. If they were willing to participate in my study, a time and location was arranged for an interview. Due to financial and demographic difficulties, telephone interviews were added to the protocol to accommodate family members who wanted to participate but were not able to travel to the interview location. Four in-person interviews with the mothers of the deceased infants took place at UCT and two interviews were conducted telephonically. Of these, five mothers were joined by one family member during the interview. The following flow diagram is presented as a visual representation of the recruitment process undertaken in the current study.

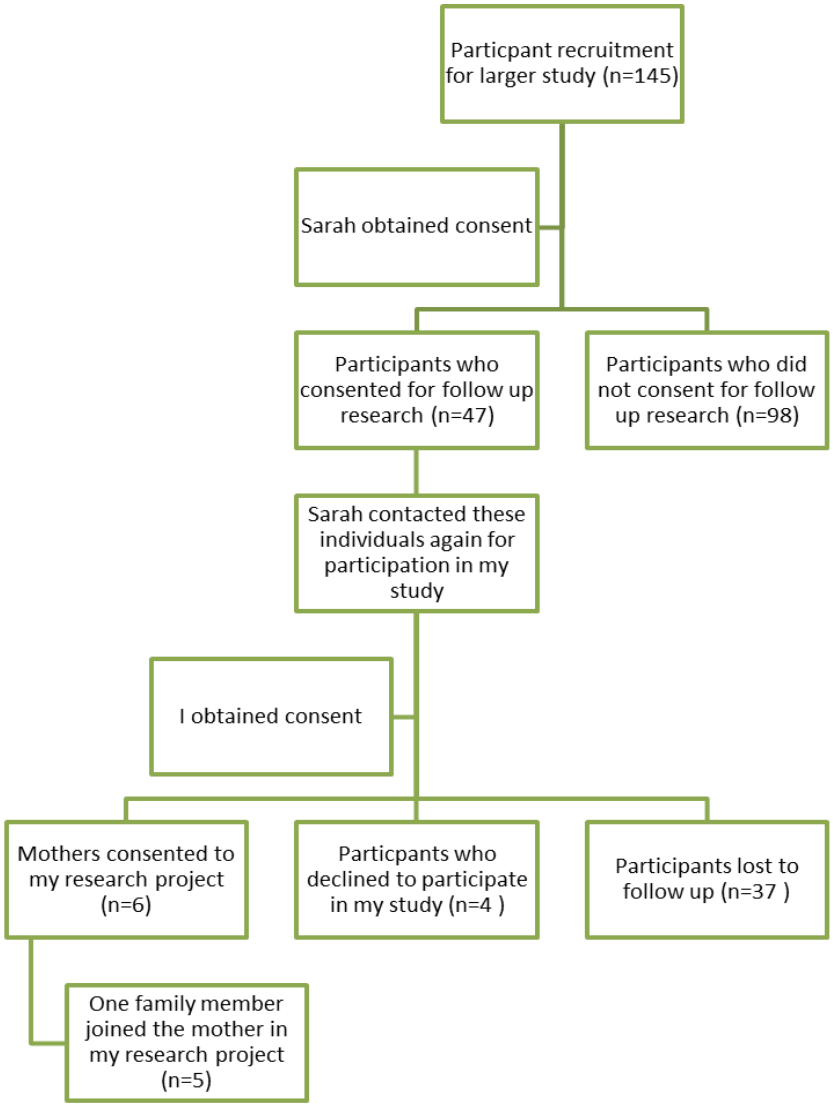


Figure 4 Flow diagram depicting the process of participant recruitment in the current study

Recruitment of participant took place from August 2017 to May 2018. Once all potential participants were contacted and no further participants were available for participation, recruitment ended. The recruitment of participants was challenging for a number of unanticipated reasons: out of the 145 participants in the larger study, 47 had consented to follow up research; however, it was extremely hard to contact them and eventually, only ten were reached by telephone; of which six consented to participate in my study. For the remainder (n=37) several different reasons for non-contact were as follows: number did not exist; wrong number; went straight through to voicemail, and if possible to leave a voicemail, no follow-up contact was achieved; no number or contact details were provided; there was no answer. Figure 5 visually depicts the number and percentage of the contact history and achieving the final number of participants. From the potential cohort of participants that consented to the larger study (n=145), 4% (n=6) participated in the current study.

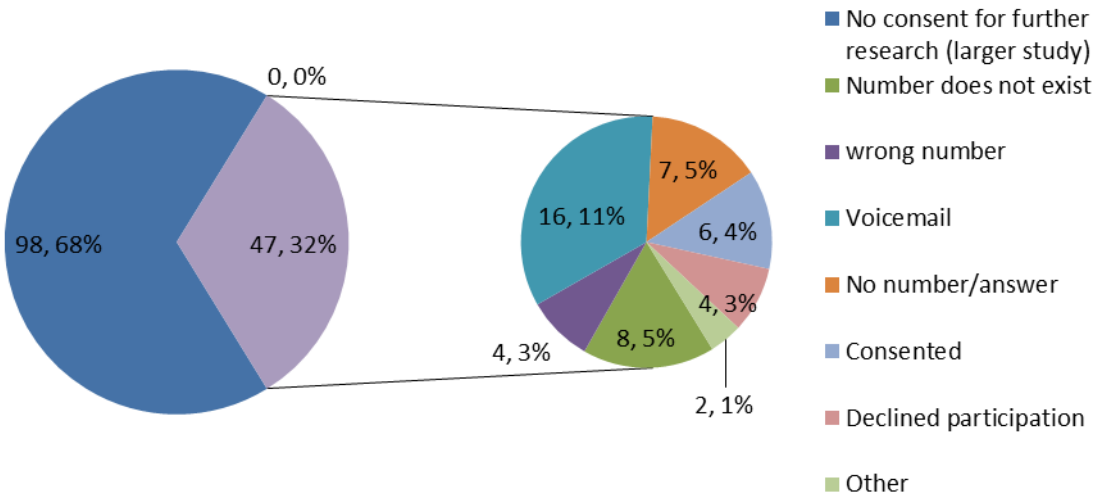


Figure 5 Visual representation of participant recruitment

It is interesting to note that the mortuary requires an affidavit stating that the next-of-kin provide a working phone number. As can be seen by the results found during participant recruitment, this was not the case. Of the 47 potential participants contacted, 12 had either a wrong number or the number did not exist. There may be various reasons for this. People may not be able to afford phones, airtime or data; phones might be stolen or lost; phones

might have changed hands; people may not be available to answer their phones; prospective participants who receive multiple voicemails were not interested in participation in further research.

The difficulty in recruitment brought an interesting dimension to the research and was necessary to explore further. The small sample size and the context behind it prompted the inclusion of the interview with Sarah, the main investigator in the larger forensic molecular autopsy research. This interview shed some light on difficulties found in research in a mortuary setting and were explored in Chapter 5.

3.3 Data collection

An open-ended question guide was used for the purpose of this qualitative research project (Appendix C), which allowed for the exploration of perceptions and experiences of participating in molecular autopsy research as described in the participants' own words (Williams, 2007). The lack of pre-set categories for the questions allowed the participants to express their thoughts and feelings more freely and in greater depth. This is critical as I, as the researcher, should be open and free of any preconceptions, but as I bring my own experiences into the session, this may be difficult to achieve. Critical appraisal and self-reflexivity was essential to this process in order to gain awareness on possible biases that I may have brought to the research. The open-ended question guide allowed me to be more flexible and provided room to adapt to the responses of the participants (M. Saunders, Lewis, & Thornhill, 2007).

In order to ensure the validity of the interview guide, information was obtained from the existing literature on the topic and previous interview schedules were evaluated in the field of qualitative genetic research. The interview guide was reviewed by the supervisors and practise interviews were conducted which allowed questions to be refined and altered as needed.

Three practise interviews were performed in order to provide a safe environment so that I could become familiar with interviewing techniques and the practical application of the recording equipment. Feedback was provided by the supervisors on how to further improve the quality of the interview and the interview guide. I had previous experience in dealing with vulnerable populations in a clinical setting, as part of the course work of my degree. My interview skills were further developed through my theoretical course work in the Genetic Counselling course. I have also attended workshops on grief and ethical principles in dealing with vulnerable populations.

The face-to-face and telephonic interviews were conducted at UCT by myself, at a time that was suitable for both the participants and me. The interviews were conducted in a quiet, private room without any distractions in order to facilitate the sharing of sensitive information. It was anticipated that the Interviews would reveal emotional information, perceptions and experiences of the participants and sensitive topics potentially including privileged information (Denscombe, 2008).

Participants were given a choice with regard to their preference to which language the interview was conducted. Since the researcher is fluent in Afrikaans and English, those interviews were conducted solely by the researcher. For Xhosa-speaking participants, in conjunction with the researcher, a translator with a background in genetics was used. The information sheets with informed consent forms (Appendix C), as well as the interview schedule (Appendix B) were explained to the participants in a language of their choice.

The interviews ranged from 20 to 39 minutes and were audio recorded which allowed for a record of the specific words and language used by the participants. Audio recordings allowed the researcher the opportunity to engage with the participants and to minimise distractions (Patton, 2002). Field notes were used during the interview process to assist in remembering points to explore further and in recording participant behaviours, activities and other features of the setting that may have been relevant during data analysis. Note-taking was kept short and concise so as not to interrupt the flow of the interview. Field notes assisted in producing meaning and understanding of the social situations that were

being studied (Mack *et al*., 2005). After the interview, immediate reflection on the session took place. All the audio recorded interviews were transcribed verbatim by me.

Further contextual data was captured on the deceased infants' from medico-legal case files through the Salt River Mortuary archives. This allowed for a further opportunity to situate the data within its original context. These were extensively examined to allow the researcher to become familiar with the details of what the parents experienced during the time leading up to the baby's death and provide a holistic and unbiased view of the research setting as explained in section 3.2. The medico-legal case files contain the questionnaires that were completed by next-of-kin at The Salt River Mortuary are based on CDC guidelines (Center for Disease Control and Prevention, 2018) (Appendix A), as well as clinical history and autopsy findings. The interview with Sarah was recorder and transcribed verbatim and was included in order to explore the setting of the larger forensic genetic study. Therefore, no themes were identified, as it was included to provide specific answers.

3.4 Data analysis

Data, including the demographic forms received from the larger study, the field notes and the audio recordings, were evaluated and used to inform and enrich this study. The audio recordings were deidentified and participants were provided with a pseudonym to ensure confidentiality. These verbatim transcripts were then subsequently used to critically evaluate and identify possible themes using the framework approach.

The framework approach has shown good utilisation in healthcare research settings (Smith & Firth, 2011). This approach facilitates meticulous and clear management of data through the use of a systematic process which involves the organisation of the data into themes and related sub-themes (Spencer, Richie, & O'Connor, 2003). Thematic analysis within this framework provided a rich and complex account of the experiences and perceptions of the parents who lost an infant. Thematic analysis involves sifting through the data in order to recognise patterns and similarities. These are the categorised and grouped together to link into overarching themes (Braun & Clarke, 2006). Thematic analysis has been described as a

useful method for investigating the perspectives of different research participants in order to identify similarities and differences, and also to discover unexpected insights. It is a well-structured approach to handle datasets, and assists in providing a clear and organised evaluation (Braun & Clarke, 2006).

In the current study, I evaluated the data and identified similar ideas in the observations and words of the participants. The data was then reread to facilitate the grouping of ideas into categories, which later allowed me to identify and describe themes. This was used to infer meaning to the experiences and perceptions of the participants, which are further illustrated by using quotations from the participants. In order to enhance trustworthiness in the data analysis, the interpretation of the dated was validated by two independent research supervisors.

3.5 Reliability and validity

As qualitative research becomes more accepted and valued in the research field, it is important that it is conducted in a way that it provides meaningful and useful results. In order for the research to be seen as trustworthy and valid, it is necessary to demonstrate that the analysis of the data was conducted in a methodical and precise manner (Nowell, Norris, White, & Moules, 2017). Validity refers to the credibility of the interpretation of the data. In order to achieve this in the current study, all anecdotal data were supported within the context of previous research. The data for the current study was carefully and objectively analysed in order to make valid interpretations. Two independent supervisors examined two of the interviews individually, which were then discussed with me to ensure correct data interpretation. This was done in order to eliminate research bias and improve validity of the findings (Silverman, 2015). A further session with two supervisors and peer at a similar stage of analysis was held where we discussed our emerging themes and received feedback in order to improve validity and reliability.

Reliability refers to the replicability of the research findings and level of consistency of data interpretation (Silverman, 2015). Thematic analysis has been found to be a reliable research

method through transparent use (Stockmeier et al., 1998). The interview questions and probes were reviewed by two supervisors, one who is an expert on forensic molecular autopsy research and the other who has a background in qualitative research. This was done to ensure the relevancy and comprehensiveness of the questions in order to reach the aim of the study. It was also done to ensure that the questions would be easily understood by the participants. The use of field notes, verbatim transcripts and the inclusion of the interview guide in this research study, were all used to ensure that the findings are reliable (Silverman, 2015).

3.6 Ethical considerations

3.6.1 Ethical approval

The current study was granted ethical approval by the Human Research Ethics Committee of the University of Cape Town (HREC: 627/2017). According to the declaration of Helsinki (as amended in 2013) the essential principles for medical research involving human subjects is respect for the individual, the individual's right to autonomy and their right to make informed decisions with regards to participating in the research (World Medical Association, 2013). These principles applied both at the start of the study and throughout the course of the research. The researcher kept in mind that the subject's well-being must always take priority over the interests of science and society and ethical considerations must always take priority over laws and various regulations (World Medical Association, 2001).

3.6.2 Consent

Informed consent is of the utmost importance to ensure autonomy during research. Informed consent can be obtained by providing information that is understandable and without deception, and subsequently providing the prospective participants the opportunity to autonomously accept or decline the invitation of research participation (Silverman, 2015). Participants had given their consent in the larger study to be contacted again for research

purposes as discussed previously. Additional consent was obtained for participation in this study as discussed in Chapter 1 section 1.1.

All participants were required to sign consent form. A copy of the consent form and information sheet is available in Appendix D. All participants were provided with an information sheet. In the case of telephonic interviews, the consent form was read to the participant and telephonic consent was noted on the form. The participant had a choice to have the information sheet read to them or to provide a contact address for either an electronic or hard copy. All information with regards to the study and the process of asking for informed consent were explained in a language that was understandable to the prospective participant. Consent was completely voluntary and participants were free to withdraw from the study at any stage. The hallmarks of achieving informed consent is that information pertaining to the research should be presented in a manner that is understandable (which may include possible translation and further clarification of the consent form) to the participant, and not forcibly persuasive (Mack, Woodsong, MacQueen, Guest, & Namey, 2005). Contact details of the researcher as well as the supervisors were provided if the participants had any concerns or questions after the interview.

3.6.3 Privacy and Confidentiality

Confidentiality and privacy were assured through several measures. Interviews were audio recorded and transcribed by myself. The data obtained was assigned a code and the participants' names or identifying information did not appear in the data. All audio and written data generated during the research process were locked away in a cupboard or stored on a password-protected computer within the Human Genetics division at UCT. Only supervisors and I had access to this information. All recordings will be destroyed once the research is completed.

All data collected remained confidential and were used for research purposes only with all personal information remaining anonymous during data publication. The participants were assured that all their information is private and confidential and were treated as such throughout the study. With regard to the participation of Sarah, she was given a

pseudonym, but anonymity cannot be assured in a small setting where her colleagues may identify her. However, every effort was made to protect her privacy. Sarah also consented to participation and understands the risks

3.7 Risks and benefits to participants

No physical risks were foreseen for participating in this study. Due to the sensitive nature of the unexpected death of a child, and in revisiting some aspects following the period after this death, the participants may have experienced some emotional difficulties with the interview. During the interview, participants displayed some emotion and they were informed that they could stop the interview at any time. One interview was particularly difficult for the mother and any further involvement and genetic counselling follow-up was declined. I was aware and sensitive to the emotions that the participants experienced during the interview and all participants were treated with empathy and sensitivity. If the participant felt uncomfortable at any point during the interview they had the choice to refrain from answering certain questions and could withdraw from the study. If the participants would have required further support, they would have been referred appropriately e.g. for grief counselling or support groups.

Expected benefits to participants were that they get to share their story, which may have allowed them to make sense of their world more effectively (Bailey & Tilley, 2002). Story telling has been seen to be therapeutic to patients and allow for marginalised groups to be heard (Koch, 1998). However, the potential benefits are only anticipated and have not been assessed through any feedback from the participants. The information received through the interview process may also help improve our understanding of asking for consent and the possible emotional burden experienced with regard to sensitive topics. This is important for health care professionals, including genetic counsellors, to objectively assess the efficiency of various psychosocial and medical measures as well as the outcomes of specific medical and therapeutic intervention (R. Price et al., 2014).

3.8 Self-reflexivity and position in research

I am a 35-year old Afrikaans, South-African female and I am currently in my second year of my Master's degree in Genetic Counselling. I was approached by the supervisor of my previous Master's project in Behavioural Genetics to help explore the possible involvement of a genetic counsellor in a molecular autopsy research setting.

I had had a conversation with my aunt and uncle who lost their 18 month old baby due to a heritable metabolic disease in the 1990's. Unfortunately for them, genetics was still a relatively young and unknown field in South Africa at that stage, as they really could have benefitted from this service. They only received a probable answer from a neurologist towards the end of their baby's life. They refused an autopsy as they did not want to put their child through any more harm. It is now more than 20 years later and they are still carrying their grief with them.

This has touched me deeply I was immediately intrigued by the invitation of my supervisor on how a genetic counsellor can contribute in a setting where parents have just lost their child. As I became more immersed in the research and was confronted with the deeply moving stories of the participants about the loss of their baby, it has given me more insight in the special role a genetic counsellor can play in the lives of parents, not only those who lose their loved ones, but also who have family member diagnosed with genetic conditions and often very serious syndromes.

Chapter 4: Results and Discussion – the participants' story

The following chapter is presented as an exploration of the perceptions, experiences and knowledge through thematic analysis of all the genetics research participants who lost an infant during 2016 and 2017, as described in Chapter 1 section 1.1. In addition, a discussion is provided in order to fit the findings of the current study within a wider body of knowledge described in previous research. The chapter concludes with a summary of the pertinent findings.

4.1 Sample demographic

Six mothers who lost an infant within the past two years and their five family members participated in this study. All 11 participants received a pseudonym in order to maintain confidentiality. The participants' were all present at the mortuary at the time of the infant's death and during the consent process of the larger forensic genetic study. The information is presented in Table 3.

Table 3 Participants and demographic information

Family	Participant pseudonym	Relation to deceased infant	Interview method	Ethnicity	Time since bereavement (years)	Number of other children and losses
1	Patricia Malcolm	Mother Father	In person	Coloured	1	2
2	Felicia Stephan	Mother Grandfather	In person	Coloured	2	2 1 miscarriage
3	Dimpho Benni	Mother Uncle	In person	Black	2	2, 1 deceased child (twin of SUDI)
4	Bernice Rayvan	Mother Father	In person	Coloured	2	3
5	Nicci Ettiene	Mother Father	Telephonic	Coloured	2	2
6	Vuyelwa	Mother	Telephonic	Black	2	1 1 stillborn

4.2 Themes

After thorough, inclusive and comprehensive analysis, themes emerged that were mostly related to the experience of the loss of the baby, rather than the actual research participation. Throughout the themes there was a distinct contrast between the participants' poor recall and memories of the *research* versus the vivid memories of their *baby* and procedures relating to the infant death. The salient point throughout the themes and discussion is the sudden death of the infant, rather than the participation in the genetics research. This was an unexpected finding that emerged from the data and was explored in depth in order to create a holistic understanding of the participants' experiences. Four major themes emerged from the data and form the foundation of the analysis and are presented in Table 4.

Table 4 Major themes and sub-themes that emerged through thematic analysis

Major themes	Sub-themes
Old wounds	Emotions Meaning making
'My booboo'	Yesterday Today Tomorrow
The sudden death	'Because, why' 'It's God's case' Expectations
Afterthought	

Direct quotes of the participants are provided to share their thoughts and feelings and to provide greater insight to the reader. All of the quotes are presented in English, however, a few phrases or words are also presented in the home language of the participant to capture the essence of the participants' thoughts.

4.2.1 THEME 1 - Old wounds

Throughout the interviews of the participants expressed strong emotions and there was an overarching need to find meaning in the loss of the babies. Some of the emotions were expressed verbally, whereas others were through their behaviour and the way a voice would break when talking about the baby.

Subtheme 1.1: The emotions

> ***Yes, it was just very sad for me to see that I brought a child into life and then she just died.*** - Nicci

> ***I would cry, but now I don't even cry now, I am just alright.*** - Vuyelwa

> ***I told my mom it won't be forever, but she will always stay in my heart, but some peace will have to come.*** - Felicia

All the mothers became a little emotional during the interviews. The overall feelings expressed by all the participants were that of grief and sadness for the loss of their baby. Whilst Felicia was saying how "*peace will have to come*", she became very emotional.

Sometimes Vuyelwa would just go quiet over the phone whereas Nicci passed the phone to her husband after talking about the time leading up to the death of her daughter. A particularly sad moment for me was when Bernice showed a picture of her deceased baby and her voice broke as she talked about her and when I mentioned "*she was so beautiful*".

> Vuyelwa: ***"…it was almost like yesterday."***
> Interviewer: ***"So it doesn't go away?"***
> Vuyelwa: ***"No."***

Another commonly expressed emotion in the mothers was that of confusion, in that they did not understand how this could happen. Patricia described how her child started to laugh out loud the day before her death, *'prontuit',* and that she had no problems. Even for Nicci who described how she took the baby to the hospital, did not expect her baby to pass away and was planning to take her to hospital again the next day..

Dimpho seemed to be the most agitated and also refused to engage fully during the interview, which can be seen by the limited number of quotes from her during the subsequent theme discussions.

> ***That was a traumatic experience and I am not feeling well, even now.*** - Dimpho

The following was expressed by her brother who accompanied her to the interview, after she spoke to him quietly:

> ***…coming back here is unnecessary, because it had happened and it's just reviving, and like what was already starting to heal, by coming back and by doing anything to do with that traumatic experience, because it would not change the fact that the children have passed on.*** - Benni

> ***But, as time goes on, we, I mean, I experienced that my sister didn't accept. She didn't accept and now she was traumatised that, especially after the second one, when she passed away. I mean, she passed away also.*** - Benni

Benni also mentioned that the family went through a very traumatic time. He felt that that may have impacted on what they remembered about the research. There has been a

concern that research participation after the passing of a loved one may reactivate the pain of the loss and contribute to additional emotional stress (Kentish-Barnes et al., 2015). This might have been what happened in Dimpho's case. She might have thought that she was ready to talk about the experience, but the interview could have triggered her loss. However, this is not unique to this study. Even though studies mostly indicate that participants find research participation beneficial, there are some that experience it in a negative way. Individual differences or experiences may compound participant distress (Decker et al., 2011). Dimpho experienced the subsequent loss of the other twin, shortly after the loss of the first infant who was included in the larger forensic genetic study. Dimpho's mother also passed away during that time. All of those traumatic losses may have increased Dimpho's distress and she felt that the research would not change anything for her. Dimpho's case is discussed further in Chapter 5 section 5.3.

... the experience that we've lost our baby, it hurts me very much. - Rayvan

In the above quote, Rayvan is expressing his feelings of pain due to the loss of his baby. The emotions expressed by the participants represented their feelings of grief. Traditional and contemporary theories on grief have acknowledged the immense emotional pain and suffering that all grieving individuals experience (see Krueger (2006) for a full review of these theories).

Through the participants' accounts or behaviour, the following emotions were therefore present: confusion, distress, sentimental memories of the baby, disbelief, crying, sadness, a sense of guilt and agitation. These feelings were not always admitted out loud, and I found it to be only moderate in its intensity. Merlevede et al. (2004) described similar emotions in bereaved next-of-kin of patients that demised suddenly and unexpectedly in a hospital setting due to natural death, accidental death or suicides. Most of the deceased demised outside of the hospital after failed resuscitation attempts. The relatives of the deceased patients also described sleeping problems, decreased concentration and the feeling of a spiritual presence. The participants who experienced the loss due to natural causes displayed less psychological problems than the participants who experienced a death due to trauma or suicide (Merlevede et al., 2004).

For bereaved parents, and mostly mothers, the expression of feelings of anxiety, fear, guilt, regret, remorse, anger, resentment, jealousy, envy, rage, loneliness, sorrow and depression are common during the grieving process (Simonds & Rothman, 1992). Grief is a universal feeling experienced in the wake of a great loss.

> ***Uhm, for me, to be honest, for me, it was a shock for me, because my child was okay en just the next day cold.*** - Felicia

The cascade of events following the sudden death of an infant is different from that than any other death. Parents are in extreme shock. Family members and the community may show confusion and also suspicion, as discussed further in subtheme 2.3. On the other hand, bereaved parents may experience guilt as they make the connections between the risk factors of sudden infant death and their own circumstances leading up to the death of their child (Mitchell, Tuohy, & Brunt, 1997). Vuyelwa displayed so much self-blame during our interview.

> ***I was traumatised…I was sad and I was angry at myself also, I don't know why. Because the baby was not sick so he was fine. It just happened… I don't know, I just blame myself, I don't know why.*** - Vuyelwa

Further events after a sudden infant death include the involvement of ambulance services and police, and also the mortuary process, may increase feelings of being overwhelmed (Martin, 1998). These were described by Vuyelwa when she mentioned all the people and ambulances around her and how traumatised she felt. All of these factors pertaining to a sudden infant death may play a critical role in the meaning-making of the participants.

Each participant was different in the way they expressed their grief, and not all participants expressed this negatively. Both the experience and expression of numerous emotions is fundamental to grief (A. Dyregrov & Dyregrov, 1999).Through these grief reactions and emotions, the way in which some of the participants were still searching for meaning, was evident.

Subtheme 1.2: Meaning making

Meaning making in the current participants were expressed as both a *question* of how or why did the baby have to die, or as an *answer* in that the participants' life changed in response to the death of their baby. The first group, consisting of Patricia, Vuyelwa, Dimpho and Felicia were still in the process of finding a meaning, whereas Bernice and Nicci have already found some meaning.

> ***I just personally want to know what, what took place that morning? What? What is the cause, why? Because it's just, you don't have answers. You can't say it can be this, it can be that. I wanna know personally what. Is it something different that happened to my child, or is it the same that happens to other people's children. What?*** - Patricia

> ***So, we lay together on the bed. That really grabbed my attention, because how the…I don't really understand. I don't really know how it can be that, because, what do they really mean about the child together with you, I mean, there are children that it happens to, even when they sleep in a cot? So…*** - Patricia

Patricia struggled to understand how sleeping in the same bed as her baby may have caused the sudden death. She was questioning why it happened to her, but not to others, and how this sleeping together could have caused her this great loss.

> Patricia: ***"There was something that I found on the web. Where they talk about sleeping together in one bed, I don't know what they call it."***
> Interviewer: ***"Yes, co-sleeping, Did you …"***
> Patricia: ***"No, she slept with us on the bed…next to us, not in the middle. I slept in the middle and she slept next to me."***

Even after Patricia 'googled' cot deaths, she still expressed her struggle to understand how this could have contributed, or how this could have happened and this also forms part of her meaning-making process.

> ***…we wonder about how the baby dies. I was a little bit worried about the way, but I thought, I don't have anything to worry about something that I don't know about.*** - Bernice

In contrast to this, Bernice did not understand it, but she also decided that she does not have to worry about something that she does not understand. Bernice kept reiterating that they tried very hard to bring up the baby and that they also stopped their drug use for some time.

> ***It is almost as if God came to speak to us…*** - Bernice

She mentioned that she tried really hard with that baby and that her mother agreed with that statement. In a way I perceived her to still carry some guilt, but that she kept trying to convince me, and maybe herself, that she did not do anything wrong.

> ***We just want to do the best that we must do, but the Lord must decide…because our future changed. I just wish that I did something good (by participating in the research)*** - Bernice

When Bernice described her reasons for agreeing that a blood sample be obtained from her baby at autopsy, she mentioned that she told her mother that she was doing it for the future of other babies. She then stated that that made her feel good about herself. This is part of her meaning making process.

> ***Like that one doctor told us, is, how can I say? Like she said, it is 5 minutes, how can I say? When the earth rotates, then a person doesn't get oxygen. And that is the reason that most babies also go. And then there is no oxygen in the air, and then they can't breathe, and so. And when those 5 minutes are over, and then are those who survived, then that is how it is, that is what they told us at Westfleur hospital when I asked for my mild tablets to stop my milk. I asked her then. I don't understand.*** - Felicia

Some parents will continue to attempt to find meaning, sometimes when there is no meaning to be found. This may result in frustration and prolonged pain (Krueger, 2006). Other parents with a SIDS experience have described that they have struggled to make meaning and for some it never happens or it may take significantly longer than what they or close others, expected (A. Dyregrov & Dyregrov, 1999).

Martin (1998) believes parents follow a path after SIDS where they start with a search for reason, as often expressed through the question, 'why did this happen?'. This is followed by 'this doesn't make any sense, the body was perfectly healthy', as an expression as a search for logic. The process concludes with a search for meaning, 'what does this mean for my life in the future?'. This reconstruction of meaning after the trauma of a sudden infant death is a part of the grieving process that is not well understood (Krueger, 2006). The way in which each of the participants make meaning of their loss, are unique to them and can range from everyday adjustments to existential questions (Krueger, 2006).

4.2.2 THEME 2: *'My booboo'*

An interesting theme that that came up was the participants' reference to their baby, ('booboo'), time and time again.

Where is our booboo? – as asked by Felicia's children about the baby.

The current research study did not set out to explore the days leading up to the demise of the baby or the time after and therefore the initial interview guide (Appendix C) did not include questions on this time. However, most of the mothers or family members referred back to the time before the baby died. The participants also remembered how difficult it was after their experience of the sudden death of their baby and what they went through in subsequent days. The mothers could describe these times in great detail and could remember the day and other experiences they had at the mortuary after the death of the baby very well, in contrast to their genetics research participation.

Subtheme 2.1: Yesterday

There was great variety in how the participants described the day leading up to the death of their baby. Dimpho and Benni (family 3) were the only participants who did not refer back to this time, as Benni said that it was just opening old wounds for Dimpho. Felicia described her baby's personality and how the baby was, whereas Stephan (Felicia's father), did not say anything about the child, only that they saw the baby the day before she died.

> ***She was, how can I say, she was a very quiet little girl, she was very curious, and if, I would have talked to her, then it almost looked if she looked for where I was. And you may say she was a happy person. That was how she was.*** - Felicia

> ***…the 22nd, oh, so the 21st I was there at their place and the 22nd we received the news in the morning that the baby passed away. And that's the reason why we immediately went over to the house…*** - Stephan

Nicci recounted her baby's life from her birth. Her baby was sick the day before, as he 'caught the flu' when friends came to visit. So she took her to the hospital. The doctors asked her to wait, but as she already had a long day and could not cope any longer on her own at the hospital, she decided to go home.

> ***And then, the next day when I wanted to go, she was already dead.*** - Nicci

Participants became rather emotional during the recounting of this time and remember it with sadness. Vuyelwa expressed some shock over the unexpectedness of the event. She also displayed some grief by saying how short her baby's life was and the limited time that she had with her.

> ***Yes, I wasn't expecting it. The baby was fine and the baby just sleep and never wake up. So I didn't expect it. Yes and it was so short.*** - Vuyelwa

Bernice described how her baby was underweight and was just getting better. She described with difficulty when she found her child.

> ***It was very difficult. I ran to my neigbour, because I had a fright when I saw her, then I ran to my neighbour. When I ran to my neighbour, I told my husband, I just had to run, I could see the baby was already gone. It was warm, still alive…*** - Bernice

The way in which the mothers or other relatives described the day before the death, could be possibly attributed to the attachment of significance to specific events that they remembered and thought about retrospectively (Garstang et al., 2014). This was found to be the case in a study on the experiences of a stillbirth (Nuzum, Meaney, & Donoghue,

2018). These parents often referred back to the baby and the baby as part of the family was found to be very important to them.

Subtheme 2.2: Today

Most of the participants remembered the day as not too long, but very traumatic and difficult. Vuelwa described the day as if she was living in a dream:

> ***…it was like a long day for me, it was like a dream. All the people that was around, like, they surrounded me, the ambulances, all the things, man. It was just a busy day for me.*** – Vuyelwa

Vuyelwa mentioned how the hardest part on that sad day, was "*having to see my baby for the last time*". It has been found that taking leave and saying goodbye for the last time is often an intense emotional event (Merlevede et al., 2004).

> ***The first thing that they did, I was taken to identify the body, but they didn't show me my child. They showed me a number of bodies, and asked whether that was mine, until they got to the one that was mine.*** - Dimpho

For Dimpho, this was an extremely emotional day, as previously described, especially when she had to go through the experience of having to identify her baby from a number of deceased babies. It seems that viewing the body is beneficial in the grieving process as relatives are afforded the opportunity to fully realise that the deceased has passed on (Garstang et al., 2014; Walters & Tupin, 1991), which may not be the case for Dimpho. There are four viewing rooms at the Salt River Mortuary and unfortunately, the possibility exists that she was shown to the wrong viewing room. It is not standard practise to identify a SUDI case from a number of bodies. This was in contrast to how Vuyelwa viewed her experience her, which was more cathartic.

The other participants also remembered a busy day, but most of them did not feel that it was very long. Stephan recalled that he went to the mortuary to help his daughter and her husband with directions, as they did not know how to get there.

> ***We, I will say the first person took about 20 minutes. And with Sarah we took about 20 minutes, or half an hour, more or less. What we then whatever we did, but we also had to fill in other papers before then, en still had to sign in and had to go through security as well. But, uhm, that didn't take so long.*** - Stephan

Bernice also felt that it was not a particularly long day and even though Nicci recounted the day leading up to the death of her baby in vivid detail, she did not want to talk about her time spent at the mortuary. She only mentioned how she felt the questions with regard to how the death occurred as intruding.

> ***Yes, they asked over and over again how it happened. I only told the truth, what we did and how it happened. But I was on my nerves, because I don't know much about a child that goes.*** - **Nicci**

Three participants specifically mentioned the researcher that asked for consent to the larger forensic genetic study on the day.

> ***How can I put it now? She, almost like she wants to, she wants to give the people closure, man, she feels more than connected, more than just, because she, she gets the cases like that often. So your case is not gonna be any different to the person she meets tomorrow. So she gets more. She makes you feel more, more, how can I say, she doesn't make you feel like she is a stranger.*** - Patricia

> ***So when 'Sarah' came in that day, it was almost like she could feel my hurt, she could feel my pain. And that's the reason why I said yes… it was almost like if felt as if she had a piece of me.*** - Bernice

Both Patricia and Bernice could remember the name of the researcher, Sarah, and how she spoke to them at the mortuary, whereas Benni could remember the face of the researcher, but not the name. The other participants did not specifically mention Sarah. Numerous studies have shown that parents felt better supported if professionals, such as Sarah, showed some emotion, especially after a child death, as cited in Garstang et al. (2014). Lemmer (1991) describes how mothers who experienced a neonatal death perceived staff or professionals who lacked emotions as uncaring and unfeeling. Other studies have also described that bereaved parents remember the professionals that they interact with after the death of their child, and that these memories may either cause comfort or distress (Meert, Briller, Schim, Thurston, & Kabel, 2009; Nordby & Nohr, 2009).

Subtheme 2.3: Tomorrow

After the time spent at the mortuary and seeing their baby for the last time, the participants had to proceed with their lives as part of a family and wider communities. Their experiences after the death of the infant included references to the stigma they experienced within their communities.

The participants' narratives showed that a sudden infant death may involve misunderstanding with regard to SUDI at a greater societal level.

> ***Nosy, man, a lot of gossipmongers. Yes, man, they make their own stories, because it was so sudden, now everyone is throwing in their two cents. Everyone says no, it's this, it's that, it's like this. And because you can't give answers, it looks as if you are hiding something… Then I tell them it is none of their business.*** - Patricia

This type of death may cause suspicion and misunderstanding in both family members and neighbours. As Vuyelwa described:

> ***They like, like it is almost like they blame me. Or they say I killed my own baby and such stuff like that. It was my fault. Even if they didn't know and they were not there, but they just judge. They always did that, but I just carry on.*** - Vuyelwa

She then went on to say the following:

> ***Yeah, I feel sad sometimes but it is better to open up and speak about it than to keep it for yourself. And then ... so it is better to speak about it.*** - Vuyelwa

Vuyelwa found that speaking about the death after the loss of her baby assisted her in processing the loss. In a study of SIDS-mothers, the bereaved mothers were found to have a strong need to talk about the experience of SIDS in the weeks following the death. It was also found that the opportunity to discuss the death, as afforded by positive social relationships, would lead to better psychological adjustment (Lepore et al., 1996). Unfortunately, a lack of supportive networks to discuss trauma-related thoughts and feelings could have the opposite effect (Lepore, 1992).

It has previously been described that the trauma survivors could be subjected to negative responses from their social networks when they want to talk about it (Lepore et al., 1996). For Bernice, Patricia and Vuyelwa, they experienced outright negative reactions in their communities.

> ***Some of them said yes, that one said a lot of things, very ugly things as well. I know I tried really hard. I looked after her well. I tried my best with that baby…maybe, some of them don't know us. Some of them even said the child had a blue face, this and that, just talked their own stuff.*** - Bernice

This stigma could be further compounded, as the sudden death of an infant is not well understood. The community responses in the participants' cases may show an underlying misconception about the risk factors for sudden infant deaths. Mothers with sudden infant deaths who experienced negative social responses that constrained them to talk about it, were more likely to be distressed by unsettling thoughts about the death than those who had more supportive social interactions (Lepore et al., 1996). It has been argued that if someone does not discuss traumatic events, they may be less likely to find meaning in it (Tait & Silver, 1989). However, this is not true for everyone and some do not need to discuss it to recover from the experience.

4.2.3 THEME 3: The sudden death

The sudden death of the infant was the main discussion point, although the participants hardly, if ever, referred to it as SUDI. Only Patricia and Vuyelwa mentioned that it is a known occurrence and Stephan, the grandfather of a deceased infant, mentioned that he also lost a baby when he was younger. Participants could give their reasons for participating in the genetics research, which they mostly associated with sudden infant death research, rather than genetics-related. Two fascinating findings that emerged from the data were the participants' understanding of the death notification form that provided a cause of death from the autopsy findings on all the sudden infant deaths, and the answers and expectations from the genetics research in lieu of that. The cause of death was therefore important to discuss, as it linked with the participants understanding of the 'answer' for

their baby's death. In addition, as they mainly participated 'to find an answer' it influenced which 'answers' they wanted from the genetics research.

Subtheme 3.1: Because, why

> ***I just wanted an answer. I wanted to know how my baby died. I wanted to know what's the cause of it.*** - Vuyelwa

The reasons for participating in the larger study, was verbalised clearly by five out of the six mothers, even though they only had a very basic understanding of what they had consented to. Five families understood that they participated in research in order to find out more about the cause of death, whether genetics-related or not, and participants consented for two main reasons: to find an answer and to help future families.

As clearly illustrated by the above comment by Vuyelwa, all the participants just wanted to find an answer for why their babies died, irrespective if it was through the traditional autopsy at Salt River Mortuary or the molecular autopsy research performed by the larger study.

> ***Man, because why, people will ask me what the cause was, and then I can't tell them. Then I tell them I also don't know. Now some people make up their own stories. But, that is why I told her it is fine, she can that, because I also want to know for myself and I don't know.*** - Patricia

This most salient reason expressed by the participants related to their limited understanding of finding an answer as described in the previous section. As all the participants thought the genetics research may provide an answer to the cause of death of the infant, although the 'answer' would be different in the case of the genetic findings, than that of the traditional autopsy. Their reasons for participation reflected the hope that this cause, any cause, can be found in their cases. As seen by Patricia's statement, if there is something that can provide a reason for the infant's sudden passing, then they felt that they would participate. Felicia echoed this sentiment after explaining how shocked she was at her baby's death.

> ***She then said, she is going to query, do after tests, so I then said I would really like to know why it happened, and the circumstances, what made this come to pass and so. That is then why I participated.*** - Felicia

In this sentiment, Felicia described her reason for participation as she wanted to know more, not just about the cause of death, but also how external circumstances may contribute. She also expressed her shock of her child being 'orraait' (alright) and then just cold the next day. So when the researcher said that she will investigate through follow-up testing, she jumped at this opportunity. For Etienne and Nicci, the reasons were different, as they did not seem to realise that they were participating in *research* at that time they consented.

> ***If we didn't, for me, if I didn't do it, I wouldn't have been able to carry on. Just for me to find out, so they are looking for it. Why is she dead and what caused it?*** - Nicci

The above statement referred to the general post-mortem evaluation, including traditional autopsy. For them the reasons why they were going through this process was only to find out the cause of death. This is in line with the reason for post-mortem investigations for all sudden and unexpected deaths as described by Garstang et al. (2014). Their review study found that parents want to know and understand the cause of death, specifically in cases of sudden infant death. The current findings are also consistent with the survey of 892 parents with a sudden infant death that determining the cause of death was of the utmost importance to the parents (Royal College of Pathologists, 2004).

The only participants who could not remember that they participated in a research study, were Etienne and Nicci. As they were not clear with regard to their research participation, Etienne could only postulate a reason for any possible research participation. He said that he would do it to help any other person that has to go through the same thing that he had to go through. He said he is even willing now to speak to anyone and act as a support for them. This would link to the altruistic reasons, also provided by some of the other participants. This altruistic reason was also described by Bernice and Vuyelwa. Bernice said that she is willing to give some of her baby's blood, as her child is not there anymore, but it may possibly help a next baby in the future.

> ***So if the tests can help for children in the future to, maybe the doctors can, like, pick up something earlier, what maybe caused it and maybe can prevent it, that's the reason why I agreed that she can…*** - Patricia

> ***That is also why I agreed. Maybe they can save another baby. You never know.*** - Vuyelwa

Some provided alternative reasons. Stephan described the importance of research and how he feels that it is something that has to be done.

> ***Because who is going to know the truth, if there is no research being done? Know what I mean? And we get different types of research. But look, many people always discover something according to research.*** - Stephan

He further expressed that research is important to find the truth and that there are a lot of different types of research. He also felt that research may lead to discovery. His daughter, Felicia, echoed his thoughts and also mentioned that if you know that you made a mistake, then you can change things. The other participants to mention the importance of research to further knowledge were Etienne and Vuyelwa.

> ***It is very Important, because it can help a lot, because the things that they ask you, will be giving information.*** - Vuyelwa

These sentiments have been reflected in other studies, both in genetics research and post-mortem studies (Brabin & Berah, 1995; Decker et al., 2011; Kentish-Barnes et al., 2015; Rankin et al., 2002). A cross sectional survey of 166 participants by Rankin, Wright and Lind (2002) explored the reasons of parents for agreeing to a post-mortem examination following the death of their baby. A total of 244 responses were recorded for the 166 participants and participants could provide one or more of reasons. This study found the majority of respondents wanted more information about what happened (108 responses). This is in line with the current study that found that all the participants wanted to know more about the cause of the demise of their baby. As with the current study, the help to improve medical knowledge and research was an important finding and was indicated in 59 responses. Other reasons provided by this survey included that it was recommended by the

person asking for the consent (40 responses), the respondents felt a need for closure after their loss (25 responses), they wanted to know the risk to future pregnancies (7 responses) and they needed to know if they had done anything to cause the loss of their baby (2 responses). The respondents in this study included mothers who lost their babies due to miscarriages, late terminations, stillbirths, neonatal and infant deaths. In contrast to the mothers in the current study, these respondents did not agree to help other babies, but they felt it necessary to find out more about their own future risks (Rankin et al., 2002). There is a paucity of literature specifically pertaining to the experiences of next-of-kin who had a deceased individual undergo molecular autopsy. All the participants in the current study failed to make the connection with regards to the risk to future pregnancies. This is discussed further in the last major theme.

Subtheme 3.2: *' It's God's case'*

The 'death due to natural causes' as an answer for the babies' death was a discussion point during all the interviews. The participants assigned different meanings to this concept. Therefore this subtheme was presented in relation to the sudden death.

> ***What I know about natural death is the statement natural death, if it is not through an accident or a shooting or a knife wound or what. It is almost, I will say it like this: it is God's case.*** – Stephan

> ***A natural cause is only, it is the Lord's management. There's nothing that people or anyone could do, it just is.*** - Bernice

Both Stephan and Bernice described a natural cause as 'God's case' or 'the Lord's management' and therefore ascribe it to a higher power and that there is nothing one can do about it. Five out of six mothers have received a death certificate stating that the death of their baby was due to natural causes.

> ***No, as they told us on the death certificate, it was natural causes.*** – Nicci

> ***Look, I think this is the answer (natural cause). According to me, that's the answer, but the fact remains, we do not really know what the cause is, understand what I mean? Perhaps, out of everything, every, every death has a problem, how can I***

> ***say, has a mistake. I think it might have been a vein that burst, know, in her head, or something like that, do you understand what I mean? Now natural death is just something inside you, what went wrong, understand what I mean? That is how I see it. See what I mean? But I know that is the answer according to the natural death. Look, many people say of heart attacks, they get strokes, understand what I mean, all these things are natural death, yes.*** - Stephan

As part of the forensic death investigations in South Africa, next-of-kin receive a death notification form stating the cause of death. As Vuyelwa described, she struggled to understand the meaning of natural causes as a cause of death as provided on the death notification.

> *Interviewer: **"…have they given you a reason why your baby passed away?"***
> *Vuyelwa: **"No, they just say it is a natural cause of death. They didn't explain it like that. I don't understand a thing about that. I feel that I am still wondering what it is."***

Vuyelwa's sentiment has been replicated in previous studies where parents sometimes find it challenging to comprehend the autopsy results, even after it was explained to them by professionals in the field (Dent, Condon, Blair, & Fleming, 1996). Forensic Pathology Services, South Africa, convey the cause of death via the death notification form. Personal follow up from the forensic pathologist is seldom due to the immense caseload experienced in South African mortuaries. Therefore, the participants in this study also received the cause of death results via the death notification form and no contact from the medical professional. As with Vuyelwa, bereaved parents with a lack of understanding of the results, felt that their questions remained unanswered. In contrast to this, numerous studies have also indicated that *any* type of information may help parents to come to terms with the death of a child (as cited in Garstang et al. (2014)).

In response to how he felt about receiving the death notification, Etienne said the following:

> ***For me it is a little bit, it made her feel better, but for me, the way it went for me, I felt a little bit down…For the cause of death, I am satisfied with it.*** - Etienne

Even though he accepted the answer, he was left feeling a bit deflated. For Nicci, his wife, the answer from the death notification was enough. It has been found that autopsy results

may play a powerful role in assisting bereaved parents to reach some closure. In parents where mandatory autopsies were performed after a SUDI experience, 66% believed that the autopsy results helped to come to terms with their grief. This was also true for parents four to seven years after they suffered the loss of their child (Vennemann et al., 2006). In contrast to this, a few studies have found a small group of parents who regretted consenting to their child's autopsy. In the current study autopsy was mandatory as part of legal investigations into the sudden death.

Subtheme 3.3: Expectations

Participants described that their most salient reason for participation was to find an answer for the death of their infant. However, the participants were split in their opinions if they wanted to hear back from the researcher with 'an answer'. This difference in opinion was linked to their understanding the cause of death and having accepted it, as discussed in subtheme 3.1.

> ***I thought about it, but I thought I will probably hear from her in a year or so. Or I didn't, I didn't expect her to contact me again.*** - Patricia

This statement by Patricia is very interesting, as it states that she thought about the research again, and she thought that she would hear from the researcher again, but then contradicts herself by saying that she did not expect that the researcher would contact her again.

> ***Either way, it won't be a thing. Maybe in the future somebody else will find the answer, but she tried. I won't feel upset, because I mean, such things take time, even if she phoned me over the next five years.*** - Patricia

> ***According to me, how can I say, according to me I won't really worry if she doesn't have an answer, because, what has happened, has happened. Like I told you, I don't want to reopen old wounds. That is how it is for me, but I am not the mother or the father. I am only the child's grandfather.*** - Stephan

His daughter felt very much the same. She said that you cannot change anything, *'mens kan nie yster breek met kale hand nie. Dis al'*, in that no amount of research or answers would actually bring her baby back from the dead.

> ***She told us she'd let us know, just for, because why, she also wants us to know why the child really, it is natural causes, but they should do further research about is. That she told me and I can still remember that.*** - Bernice

A contrast emerged in the data where the same participants wanted to hear back from the researcher, but also did not want to be reminded of that traumatic day at the mortuary. This was clearly illustrated by Dimpho who wanted answers, but who also went through the trauma of losing the twin of the deceased infant, and the trauma prevented her from wanting to engage further.

The systematic review by Garstang et al. (2014) describes the need of the parents to know how and why their child died. This echoed the sentiment found in the current study.

> ***I still want to hear… I want to know anything about the baby.*** - Vuyelwa

Even though participants admitted to hardly thinking about the research or revisiting it after that day at the mortuary, they still carried the expectation that the research will find answers. This main expectation related to their most salient reason for participating in the genetics research, which was to receive an answer. As the perception of the research was mainly that it would lead to finding a cause for the sudden death, the expectations of the research flowed from that.

Information following any type of death, including infant deaths, may contribute to the ability of parents to make sense of the death and assist with the grief process as described in the subtheme about God's case. There was disconnect in the minds of next-of-kin about the 'answer' that they might receive from the genetics study and, for some of them, the cause as ascertained by the traditional autopsy findings were enough.

A molecular autopsy identifies potential causal genetic variants that either directly contributes or act as a predisposition for SUDI and these genetic variants may also be present in family members (Bloss et al., 2015). None of the participants remembered the connection that there could be familial implications as well as the chance for recurrence if a genetic result is found. When the subject was raised in the interviews with regards to the familial implications and to the understanding of the underlying genetic contribution to sudden death, the participants became really concerned and asked me, as the researcher, what it all meant.

A study performed by Lakes et al. (2013) on the expectations of mothers on the return of genetic results of their child, found that the mothers would indicate that the decision to receive the results would depend on the context and specific circumstances. These mothers struggled to explain this and just stated that in some cases they would like to know, whereas in other cases they would not want the results (Lakes et al., 2013). This could be true for the current participants, especially Vuyelwa and Dimpho, where they wanted answers, but it depends on their individual contexts.

4.2.4 THEME 4: Afterthought

The main aim of the current research study was to determine the participants' feelings, thoughts and knowledge of genetics research participation. However, through the exploration of the transcripts, it became clear that the research participation was almost an afterthought to the participants, which is an underlying concept throughout all of the themes. Participants, notably the mothers, could recall in a very detailed manner the events and experiences leading up to the time of death of their infants. In contrast to this, their recall of participating in genetics research was sparse and lacked in depth descriptions. They could recall the reasons why they participated, but then confessed to not really thinking about the research after that, with an exception here and there. The tragedy surrounding the loss of their baby and their bereavement took center stage, whereas research participation was just another part of the process that the participants were exposed to during their time at the mortuary.

Throughout this theme, there is a stark contrast between how little the participants remembered about the genetics research, but how they could remember something if it pertained specifically to their baby. It also became apparent that some participants wanted to forget that sad time, and also their research participation.

> ***Yes, she told me, she talked about how there was stuff and whatever. Like that.*** - Bernice

Participants really struggled to explain what they understood about genetics research and related concepts. All the participants described some lack of clarity and understanding about what the main purpose of the genetics study was. As can be seen by the above statement of Bernice, she cannot exactly say what was explained to her or her understanding of it, just that it was 'stuff and whatever, like that'.

> ***I can't remember much. I just know she asked if she may have a sample for research, because there are many such cases, and they, how can I say, they want to find answers, man, for the people that, because why, it is actually mind-boggling, because you don't really know what it is.*** - Patricia

From the above, it is clear that she remembers that she was being spoken to and that the researcher wanted a blood sample to find out about the cause of death of her baby. The following statements by Etienne and Bernice illustrate a similar finding.

> ***I can just remember when (the researcher) came in there, she asked me a lot of questions. But all the questions that she asked, all, can she get some of the baby's blood, to find out what was really, the, the cause of the death.*** - Felicia

> ***Like they told me, this can maybe help in the future, but the other baby then, how can I say, that goes from the natural cause. That's why they wanted a little bit of my baby's blood before we went. And that is now finding the answers.*** - Bernice

> ***Mmm, if I remember clearly, she told us that the research of the cause of the suddenly death and so on. Ja, that she was trying to explain something about that.*** - Benni

Most participants said that the genetics research was performed in order to determine a possible cause of death, as illustrated by the above accounts. Bernice also mentioned how

the researcher explained that the research may contribute to further understanding in order to help other babies.

Benni explained how they did not quite remember what the research was about, especially his sister (the mother whose infant passed away). But also that he remembered that it was about investigating the cause of death. Benni futher elaborated by saying that

> ***…to how exactly they would do determine the cause of death, I am not quite clear about that.*** - Benni

He also confessed that he did not remember the name of the researcher and he agreed to come because he thought that maybe if he saw the face, he would remember, which he did. So he remembers the face, but not that much else.

Data analysis further highlighted how little the participants knew and understood with regards to the underlying genetics, including concepts such as DNA and the need for a blood sample and how a possible cause could be determined.

> ***What do I know about DNA? A little. Can't exactly say what I know.*** - Patricia

> ***Yes, she talked about DNA, she then told me she has to (take) the blood, the blood has to be done through the DNA, understand what I mean?*** - Stephan

Stephan could remember that the researcher asked for blood to look at DNA and he also had a basic understanding of DNA. Stephan was not the only one looking for an explanation from me. Patricia could not explain what she knew and also wanted to know more about the tests. This was similar to what Vuyelwa wanted to know after she said that she did not think about the research again and just wanted to put it out of her head.

> ***Ja, but what, what does she test, what is it? Which type of tests with the DNA, because there is a lot of tests that you can do?*** - Patricia

> ***If I may ask what was the reason for taking the blood of my baby...I can't remember.*** - Vuyelwa

Bernice said that she cannot tell how they would find answers with regards to the cause of death and in taking the blood, but that maybe through that they can find something to keep other babies safe.

> ***She told me for the future, for when it maybe will happen again, a baby die, then they know what the cause is.*** - Bernice

In contrast to the participants that could recall some part of the research procedure, Nicci and Etienne could not remember anything about the research, except that there was blood taken, possibly as part of the autopsy process, according to their recall. It might be possible that the answer they received that the sudden infant death was due to natural causes was the answer for them. That they did not have to think about the research at all, as they received an answer that made sense to them and could assist in their meaning-making.

> ***We can't remember everything anymore. What did they ask? That what they said, that it is a little bit difficult to 'do this', but they have to do it. It is to find the cause of what she died from.*** - Etienne

This was their recall about the consenting process:

> ***Uh-uh, they didn't give us any papers. No, we didn't sign anything.*** - Etienne

With regards to the consent as part of ethical recommendations, the importance of obtaining informed consent before genetic testing is highlighted by Mcguire et al. (2016). The concept of informed consent is based on respect for autonomy and to keep the consequences of testing in mind (MacLeod, Demo, Honeywell, & Rutberg, 2013). Even though additional consent was asked to perform the molecular autopsy, it is possible that that whole period, with the loss of the baby, the procedure at the mortuary and the research participation, fused into one procedure for the participants. Vuyelwa also said that she did not take note of the whole procedure. Vuyelwa explained that she could not remember anything that the researcher explained to her with regards to genetic testing or how the cause would be found. She said that it was two years ago and that at that time, she was traumatised and there is a possibility that the information just did not register for her.

No, she didn't tell me anything about that. Maybe she did tell me, but I didn't take note at the time of that. – Vuyelwa

That could have been the same for Etienne and Nicci. Therefore, from the data analysis there seemed to be a clear lack of recall and understanding of what the genetics research entailed.

Although public awareness of genetics is growing, these participants may not have had exposure to genetic concepts (Greenberg et al., 2012). Several studies have investigated the level of understanding of both the general public and genetic research participants (Etchagary et al., 2013; Henneman et al., 2004; Kaufman et al., 2008; Lemke et al., 2010). Participants considered themselves to have limited knowledge of genetics in a survey study in the Netherlands (Henneman et al., 2004). This was also the case in community session held in Canada. A large number of participants mentioned that they had not previously thought much about genetics (Etchagary et al., 2013). This was also true for Nicci and Vuyelwa in the current study. As Vuyelwa said:

I think that DNA is all about, wait, it is all about the child and the mother, everything. I don't know really really really what it is. - Vuyelwa

Knowledge of genetic testing could be influenced by education and literacy level of participants, and this is of particular concern in South Africa where there are striking social differences and access to higher education. This could lead to lower comprehension of genetic concepts (Greenberg et al., 2012). The literacy level of participants for the current study was not reported. This could have played a role in their understanding of genetics.

In this study, the longer time since the death, the less the partcipants could recall about the genetics research. Both Vuyelwa and Dimpho stated how traumatised they were during that time at the mortuary. Several studies, as cited in Garstang, Griffiths and Sidebotham (2014), have noted that, following the death of a child, the parents were too distressed to understand detailed and complicated information.

It was such a long time ago. - Nicci

The time of consenting and their emotional state may influence both their understanding and recall of the specific details of the research. This could further be compounded by the time lapse since participating in the genetics research, which was between 1 and 2 years ago for the participants.

Chapter 5: Results and discussion – a researcher's story

The initial prospective sample that was accessible to the researcher comprised a total of 47 participants. Some difficulties were expected in the recruitment for the current study. However, only being able to recruit six participants was not expected. This low response rate of about 13%, is in itself an interesting finding and worth exploring further. Difficulties were also experienced by Sarah regarding the non-engagement of families when genetic results were actually found in the SUDI cases.

This chapter is therefore included as an exploration of the loss to follow-up and potential reasons for the barriers to further engagement in this particular study population as informed by contextual factors and the experiences of the researcher, Sarah, who obtained consent for the larger molecular autopsy study. This chapter creates a context in which the experiences and perceptions of molecular autopsy research, as discussed in the previous chapter, are explored holistically. This chapter is presented as a narrative from Sarah's viewpoint, as well as the experiences of Dimpho and Vuyelwa, and is further informed by existing literature.

5.1 Loss to follow-up

As the current study was conducted within 1 to 2 years after consent to the larger study, it is possible that earlier follow-up may have had a different result.

> ***So the loss to follow up, is mainly because the contact details that they provide to the mortuary, even under oath, when we utilise those contact numbers they go straight to voicemail and … only four have actually said no, no, they don't want to engage further, the rest have all said yes.*** - Sarah

Reasons for the contact history labelled as 'Others' mentioned in Fig. 5.2, included some examples as follows:

- Sarah spoke with the foster mother of a SUDI case. She was willing to pass the message on to the biological mother, but unfortunately the mother was living on the street and not in a very good condition. She would not have been able to come to an appropriate setting for an interview, and as she was living in an unsafe area, I felt it prudent not to go there.
- The family was from Zimbabwe and the father of the deceased infant had moved back to Zimbabwe. It was decided not to pursue contact with the mother, as she felt that she could not participate without her husband.

I was also interested in exploring the possible reasons why prospective participants in the larger study, declined molecular research participation. Knowing and understanding these reasons may lead to possible improvements in addressing these and therefore further research participation. As these participants were not available to ask, Sarah postulated potential reasons.

> ***… just thinking about what they had said during the session which led up to them saying no, was mostly that they, that they accepted what had happened, and… a lot of them were religious reasons. It was God's will that my baby has passed away. And there is nothing more that can be done. And the second reason was the fact that… the forensic pathologist taking a sample from their infant, from the infant for the study, would mean an extra needle going into their baby. Which was perceived as an invasive procedure; they didn't want to cause their infants any more hurt or pain and so said no…*** - Sarah

Rankin, Wright and Lind's cross-sectional survey (2002) provided certain reasons why bereaved next-of-kin agreed to a post-mortem examination on their infants (n=148). Out of this group, 120 parents agreed to the autopsy and provided similar reasons for consenting to autopsy as in the current study. These reasons included the need for more information and to improve research and subsequent medical knowledge. The researchers further evaluated the reasons of the 28 parents who declined the post-mortem examination and 27 of those 28 agreed to provide reasons. These reasons are presented in Table 5. More than one reason could be given for non-participation.

Table 5 Reasons for respondents not consenting to a post-mortem examination (Rankin et al., 2002)

Reason	Number of responses (%)
I felt my baby had already 'suffered enough'	22 (44)
I did not feel it would help me	13 (26)
I was concerned about the effects of the examination on my baby's appearance	5 (10)
I didn't want my baby cut	3 (6)
I was concerned it may delay funeral arrangements	2 (4)
For religious reasons	0 (0)
Other	5 (10)
Total responses	50

Not all of these responses would be comparable between the consent of a molecular autopsy and a conventional autopsy. As can be seen between the description of Sarah's account and the findings of Rankin et al., (2002), there is some overlap, but not with the religious reasons that Sarah postulated. Other studies have also found similar reasons in a mortuary setting as cited in Odendaal et al. (2011).

> ***… so in a few cases, the family had already gone to view their infant and filled in the questionnaire, and I felt those individuals were more emotional...*** - Sarah

The setting and time of asking for consent may therefore play an important role in agreeing or not agreeing to molecular autopsy research and also in the way bereaved parents would experience research participation if they consented.

So, one thing that we must consider is that the rates of follow-up with parents in South Africa are very low. - Sarah

This is not a unique finding to this study. A study exploring the effectiveness of long QT molecular autopsy in sudden infant deaths also found that the families showed little interest in participating in follow-up (Glengarry et al. 2014). For the failed family screening, 32 cases were due to families who did not wish to engage further, did not respond to messages or did not attend the appointments. These family members were from mainly adverse social circumstances with a high incidence of co-sleeping, which is comparable to the participants in this study. Sarah expressed her wishes for the molecular autopsy research to add value for the affected families.

> ***… our testing in South Africa is still in its infancy. And there are many cases reported in the literature where these results have been delivered to families, and they were able to identify blood relatives at risk and link them with appropriate medical help. So I think there is still value… I think it's important to engage families early on and only perform a molecular autopsy if they are engaged and willing to give samples themselves which would facilitate co-segregation studies*** - Sarah

Glengarry et al. (2014) and Mcguire et al. (2016) recommend that family members of the deceased who had molecular autopsy must agree to further contact and be willing to engage in full screening and further genetic evaluation. This would allow for possible interventions to be realised. This might not have been feasible in the larger molecular forensic study, but the sentiments were echoed by Sarah.

5.2 A sensitive topic

> ***I think what made it difficult was not knowing how families were going to react and how I would be able to handle a wide range of reactions. I was nervous of people being offended by me asking for samples, I was worried that they wouldn't fully understand what I was asking of them in this period of grief.*** - Sarah

As discussed in Chapter 2 section 2.2.1, the death of a child is a particularly sensitive topic and therefore a sensitive area of research. Determining the aspects of participation that may potentially create distress or benefit for the participants are important to inform future studies on how it may be designed to diminish the distress and enhance the potential benefit (Decker et al., 2011). The way the researcher interacts with the bereaved parents at this vulnerable stage, should not increase their distress.

> ***And then the two, upon saying yes, they don't want to know the genetic results.*** - Sarah

As can be seen by the above statement, two bereaved mothers said yes to participate in the current study. However, they were not willing to engage further once results were available. Lakes et al. (2013) also described this confounding result where mothers would want the answers from genetic studies on their child in certain contexts, but not in others. This was

made even more complicated by the fact that the mothers could not clarify in which circumstances they would prefer the results. The mothers also stated that the researchers should communicate the urgency of the results if there was a certain level of risk, but they were unable to determine this level of risk (Lakes et al., 2013). This could be comparable to the current study where Vuyelwa was willing to engage with the risk up to a certain point. However, this point was not clarified through the current research.

5.3 The genetics behind it

> ***In interacting with the family, the common thing seems to be that they want to know what happened. But I think each family also has different views, but that for me is the feeling that I get that is common between these families, that they want to know what happened to their baby. I don't think they all fully realise the implication (of the molecular autopsy) that it has on them and future children or siblings. Some more than others.*** - Sarah

The larger forensic genetic study discovered pathogenic variants in two infants that could have contributed to their sudden death. Sarah found possible underlying genetic variants in Dimpho and Vuyelwa's babies.

Dimpho's baby

I have found a homozygous pathogenic mutation in a gene called GALT. - Sarah

In the case of the Dimpho's baby, a homozygous mutation was found in the *GALT* gene. This mutation causes galactosemia, which is an autosomal recessive metabolic disorder that affects galactose metabolism in the body which means that infants cannot metabolise lactose (milk) Complications of galactosemia include feeding problems, failure to thrive, hepatocellular damage, bleeding and *E.coli* sepsis in untreated infants. Another type, known as clinical variant galactoseamia, is characterised by the same clinical features as the general type, but may be missed during newborn screening, as the hypergalactoseamia is not as evident and breath testing is normal (Berry, 2000). This type is the most common type found in the South African Black population (Henderson, Leisegang, Brown, & Eley, 2002).

With the adherence to a lactose-restricted diet during the first ten days of the infant's life, severe acute neonatal complications and possible death can be prevented.

Dimpho's baby was a girl and was three months old when she passed away. She was from the Black South African population, possibly suffered from galactoseamia, and several other risk factors, including co-sleeping and having milk, was present (see Table 4). She also had an upper respiratory tract infection. All of these factors could have increased her risk for sudden infant death (Campuzano et al., 2014). The sad part of this story was that she was part of a set of twins and the other twin subsequently passed away as well. This was one of the reasons why Dimpho was so traumatised.

> ***That was a very interesting case because the, the infant for which we found the homozygous mutation and therefore had to have had galactosaemia, was also an identical twin. And for me this motivated to make contact with the family even more then if the infant wasn't an identical twin.*** - Sarah

As this is a recessive disorder, a couple has a recurrence risk of 25% to have another affected child but the risk is lower when there is a different partner (Berry, 2000). This information would benefit Dimpho as she could plan a future pregnancy but Dimpho refused to attend this, as seen by the end of the conversation between her and her brother:

> Dimpho: "***I am fine now.***"
> Benni: "***Shall we then just leave this?***"
> Dimpho: "***Mmm***"

Vuyelwa's baby

> ***And what it means for them as a family? I think for the cases which are natural and then could provide a genetic explanation, in those two families, they might be less willing to engage further. That it might almost unsettle their acceptance and moving on from their infants' death. Because there is now new evidence and they have to rethink everything at that time, which is not a nice feeling. So they might have a more negative perception towards it.*** - Sarah

Vuyelwa's baby was another interesting, complicated case. A homozygous variant was found in the *SCN10A* gene which is implicated in Brugada Syndrome and long QT syndrome.

Brugada syndrome is a cardiac channelopathy that is characterised by cardiac conduction abnormalities with a high risk of ventricular arrhythmias. Treatment includes the use of an implantable cardioverter defibrillator. This syndrome typically presents during adulthood, however, age of diagnosis may range from infancy to late adulthood. It is mostly inherited in an autosomal dominant manner, except in the case of *KCNE5*-related Brugada syndrome, which is X-linked. Most individuals will have an affected parent and there is a 50% recurrence risk to future children to be affected (Brugada, Campuzano, Sarquella-Brugada, G Brugada, Brugada, & Hong, 2005). The influential review by Carlin and Moon (2017) describes cardiac arrhythmias as a potential contributor to sudden infant death.

There was very little information available from the forms collected from the mortuary on Vuelwa's little boy. We know that he was a Black South African who was born prematurely. He demised when he was 3 months old and had an upper respiratory tract infection. At the time of this study, there was a functional study on a similar amino acid residue, but not on the variant of *SCN10A* found in the larger forensic genetic study. We were also in contact with the parents, as they are both carriers of this mutation and potentially at risk for Brugada syndrome and offered them clinical and genetics screening. They had another baby who is currently 2 months old and the mother also had a stillbirth prior to the birth of the infant that passed away. It is difficult to provide recurrence risks in this case, as the mechanism of this mutation in Brugada syndrome is not well-known. The parents seemed unwilling to engage in the screening and further genetic testing at the moment, even though Vuyelwa's following response initially indicated interest in further findings:

> Vuyelwa: "***No, I still want to hear."***
> Interviewer: "***What is your reason for that?***"
> Vuyelwa: "***I want to know anything about the baby."***

Vuyelwa also mentioned that she is better now and that she is very happy with her new baby. Maybe she is just not ready to deal with any new information or possible risks to her new baby.

Chapter 6: Afterword

This study was performed in order to complement genetic research studies and focused on the experiences of research participants in a forensic molecular autopsy study. Therefore, this section is presented in order to understand the grief reactions of bereaved parents and how that may have played a role in creating the contrast between their memories of the research participation and that of the sudden infant death, which they are still living with everyday. This could have impacted on the participants' engagement with forensic molecular autopsy findings.

6.1 Conclusion

The study examined participants' consenting to *molecular autopsy research* in a *forensic mortuary* setting. The burden and impact of a sudden infant death is an immense tragedy for parents and family members. The passing of their baby has an ongoing influence on their lives. Only a few studies have qualitatively explored the experiences of participating in research after bereavement in general (Brabin & Berah, 1995; K. Dyregrov, 2004; Kentish-Barnes et al., 2015; Lee & Renzetti, 1990; Newman & Kaloupek, 2004) and the overall finding from these studies indicated that participants found participation to be beneficial to them.

As is apparent throughout this study, grief formed part of the participants' journey. A sudden death is not just devastating to the parents of the baby, it may even result in the shattering of themselves and the ideas they held about their world (Krueger, 2006). Mourning and grief are natural and deeply personal reactions to the experience of a loss. As this study was cross-sectional, it was difficult to say how the participants' grief has changed over time, however, grief from their babies loss even a year ago was still present during the current study. These emotions and feelings colour the way that the participants experienced and perceived the genetics research that they consented to and is essential to explore. Unfortunately, I lost many nuances of emotions during the telephone interviews and it was harder for me to infer feelings of grief. The phone interviews also prevented me from

assessing facial expressions and body language. However, the in-person interviews had a palpable feeling of loss in the air.

It is important to note that mourning through meaning construction is a unique experience and broad generalisations are not really possible across a bereaved population (Krueger, 2006). Some of the participants may experience that they find meaning in the death sooner than others and some may never find meaning. These emotions and grief reactions did not emerge in isolation during the thematic analysis and were also apparent throughout the themes as created by the unexpected loss of an apparently healthy infant. It is therefore postulated that the setting and time of research participation is overshadowed by grief and the great loss that the family suffered. Several studies have noted that, following the death of a child, the parents were too distressed to understand detailed and complicated information (Garstang et al., 2014). The time lapse since the time of obtaining consent may also have impacted on their recall.

This has ethical implications for the conducting of research in this setting. Research is an essential step towards the implementation of better practises, and as such, the researcher of the larger forensic genetic study, together with supervisors and The Ethics Committee of UCT, chose the time of obtaining consent. Even though this was not the most ideal time, it created the opportunity for maximum participation of the population who may receive the potential benefit from the research. The value of obtaining informed consent before the process of genetic testing in both a research and clinical setting has widely been accepted as a matter of adhering to ethical principles. Obtaining consent is based on a respect for autonomy and to keep the consequences of testing in mind (Mcguire et al., 2016). In the case of molecular forensic autopsy in a research setting, the results may be of great relevance to the surviving biological relatives (Forrest et al., 2007). There are both benefits and potential harm involved in the findings from molecular autopsies, as cited in Mcguire et al (2016). The disclosure of the intention to conduct a molecular autopsy may negate some of these harms, and is an acknowledgement of the bereaved family's interests and possible concerns, as was done in the larger forensic genetic study. This is particularly important in our setting where the parents often do not want to revisit the trauma of their baby's death and we saw a lack of follow-up on the part of the parents, even though none of the

participants in the larger forensic genetic research reported any harm due to their research participation.

Therefore, this study found that the correct approach and timing of obtaining consent is a paramount time to prepare parents for possible genetic results and the resultant implications of that. Family members should also be offered the choice if they would like to receive those results.

6.2 Limitations and strengths

There were several limitations to the current study. The sample size was small which was due to availability of participants. The sample was not selected randomly, but rather identified from a pre-existing sample that previously provided consent to participate in research and gave permission to be recontacted. Responders and non-responders may have different perceptions in the particular setting, thus there may be a selection bias. The experience from their point of view could have enriched the data of this study. In order to address this fact, the researcher asking for consent at the mortuary setting, were asked for insight into the reasons that the participants provided in some cases for declining to participate in the study.

The data of the current is informed from the subjective experiences of the participants and the researcher of the larger forensic genetic study, and no previously validated, objective measures were used in this study. In addition, as the researcher of the current study was a genetic counselling student, she may have lacked in her counselling and interview skills to explore all sensitive areas fully.

Despite the limitations of the current study, the findings are supported by previous research and some new information has come to light. By allowing the interviews to be conducted in the language of choice for the participants, authentic responses were recorded and a better understanding was developed between the researcher and participants. By allowing the participants to recount their story from beginning to end, it may facilitate their process of

reconstructing meaning in their loss. This reconstruction of their child's life has been described as essential to processes of identity reconstruction in a period of grief (Riches & Dawson, 1996). There is a paucity of research on the perceptions and experiences of participants in forensic molecular autopsy research. To the best of the author's knowledge, this is the first study to describe this in a diverse South African population. These findings therefore make a valuable contribution towards the existing body of literature and have the potential to inform a larger audience, such as university ethics review boards.

6.3 Recommendations

The current findings elucidate the importance of both sensitive and unambiguous consenting procedures in a mortuary setting during a time of grief and bereavement for family members. The emotional reaction towards autopsy-based research may raise unique issues when dealing with socioeconomically disadvantaged population groups (Odendaal et al., 2011) and these issues were explored in the Safe Passage Study as described in Chapter 2 section 2.4.1. Specific conclusions from their and other research studies were that research involving autopsies is acceptable if the families are approached in a sensitive and respectful manner. It is preferred that they be approached by an informed and experienced counsellor that forms part of the medical community (Elliott et al., 2008; Millar et al., 2007).

Furthermore, researchers, healthcare providers and geneticists may better understand the needs of the participants in order to promote engagement in the case of positive results obtained through molecular autopsy. These findings may indicate the need for a genetic counsellor in such a setting and can assist in how genetic counselling services may be implemented in such a setting. Genetic counsellors have training in medical genetics, as well as psychosocial counselling. This training provides them with the skills to obtain and assess medical and family history, to facilitate and explain genetic testing and test results, to organise referrals and also to provide psychosocial support to families and individuals (McCarthy Veach, Le Roy, & Bartels, 2003).

A further recommendation is for more extensive public health initiatives to increase the awareness of risk factors pertaining to SIDS/SUDI. Bereaved parents of previous bereavement studies have stressed that the opportunity to share their thoughts and feelings about a loss may help them to heal to some extent of the disruption that they experienced due to the traumatic event (A. Dyregrov & Dyregrov, 2004; Neimeyer, 2000). Stigma may prevent this and it is essential that these campaigns target high-risk South African populations (Odendaal et al., 2011).

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APPENDIX A

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
Centers for Disease Control and Prevention
Division of Reproductive Health
Maternal and Infant Health Branch
Atlanta, Georgia 30333

CDC

Sudden Unexplained Infant Death Investigation

SUIDI Reporting Form

INVESTIGATION DATA

Infant's Last Name	Infant's First Name	Middle Name	Case Number

Sex: ☐ Male ☐ Female Date of Birth: _____ Age: _____ SS#: _____

Race: ☐ White ☐ Black/African Am. ☐ Asian/Pacific Isl. ☐ Am. Indian/Alaskan Native ☐ Hispanic/Latino ☐ Other _____

Infant's Primary Residence:

Address: _____ City: _____ County: _____ State: _____ Zip: _____

Incident Address: _____ City: _____ County: _____ State: _____ Zip: _____

Contact Information for Witness:

Relationship to deceased: ☐ Birth Mother ☐ Birth Father ☐ Grandmother ☐ Grandfather

☐ Adoptive or Foster Parent ☐ Physician ☐ Health Records ☐ Other Describe: _____

Last: _____ First: _____ M.: _____ SS#: _____

Address: _____ City: _____ State: _____ Zip: _____

Work Address: _____ City: _____ State: _____ Zip: _____

Home Phone: _____ Work Phone: _____ Date of Birth: _____

WITNESS INTERVIEW

1 **Are you the usual caregiver?** ☐ No ☐ Yes

2 **Tell me what happened:**

3 **Did you notice anything unusual or different about the infant in the last 24 hrs?**

☐ No ☐ Yes Specify: _____

4 **Did the infant experience any falls or injury within the last 72 hrs?**

☐ No ☐ Yes Specify: _____

5 **When was the infant LAST PLACED?**

Date: _____ Military Time: _____ Location (room): _____

6 **When was the infant LAST KNOWN ALIVE(LKA)?**

Date: _____ Military Time: _____ Location (room): _____

7 **When was the infant FOUND?**

Date: _____ Military Time: _____ Location (room): _____

8 **Explain how you knew the infant was still alive.**

9 **Where was the infant - (P)laced, (L)ast known alive, (F)ound** (write **P**, **L**, or **F** in front of appropriate response)**?**

☐ Bassinet	☐ Bedside co-sleeper	☐ Car seat	☐ Chair
☐ Cradle	☐ Crib	☐ Floor	☐ In a person's arms
☐ Mattress/box spring	☐ Mattress on floor	☐ Playpen	☐ Portable crib
☐ Sofa/couch	☐ Stroller/carriage	☐ Swing	☐ Waterbed
☐ Other - describe:			

10 **In what position was the infant LAST PLACED?** ☐ Sitting ☐ On back ☐ On side ☐ On stomach ☐ Unknown
Was this the infant's usual position? ☐ Yes ☐ No What was the usual position? _____

11 **In what position was the infant LKA?** ☐ Sitting ☐ On back ☐ On side ☐ On stomach ☐ Unknown
Was this the infant's usual position? ☐ Yes ☐ No What was the usual position? _____

12 **In what position was the infant FOUND?** ☐ Sitting ☐ On back ☐ On side ☐ On stomach ☐ Unknown
Was this the infant's usual position? ☐ Yes ☐ No What was the usual position? _____

13 **Face position when LAST PLACED?** ☐ Face down on surface ☐ Face up ☐ Face right ☐ Face left

14 **Neck position when LAST PLACED?** ☐ Hyperextended (head back) ☐ Flexed (chin to chest) ☐ Neutral ☐ Turned

15 **Face position when LKA?** ☐ Face down on surface ☐ Face up ☐ Face right ☐ Face left

16 **Neck position when LKA?** ☐ Hyperextended (head back) ☐ Flexed (chin to chest) ☐ Neutral ☐ Turned

17 **Face position when FOUND?** ☐ Face down on surface ☐ Face up ☐ Face right ☐ Face left

18 **Neck position when FOUND?** ☐ Hyperextended (head back) ☐ Flexed (chin to chest) ☐ Neutral ☐ Turned

19 **What was the infant wearing?** *(ex. t-shirt, disposable diaper)* _____

20 **Was the infant tightly wrapped or swaddled?** ☐ No ☐ Yes - describe: _____

21 **Please indicate the types and numbers of layers of bedding both over and under infant (not including wrapping blanket):**

Bedding UNDER Infant	None	Number	Bedding OVER Infant	None	Number
Receiving blankets			Receiving blankets		
Infant/child blankets			Infant/child blankets		
Infant/child comforters (thick)			Infant/child comforters (thick)		
Adult comforters/duvets			Adult comforters/duvets		
Adult blankets			Adult blankets		
Sheets			Sheets		
Sheepskin			Pillows		
Pillows			Other, specify:		
Rubber or plastic sheet					
Other, specify:					

22 **Which of the following devices were operating in the infant's room?**
☐ None ☐ Apnea monitor ☐ Humidifier ☐ Vaporizer ☐ Air purifier ☐ Other - _____

23 **What was the temperature in the infant's room?** ☐ Hot ☐ Cold ☐ Normal ☐ Other - _____

24 **Which of the following items were near the infant's face, nose, or mouth?**
☐ Bumper pads ☐ Infant pillows ☐ Positional supports ☐ Stuffed animals ☐ Toys ☐ Other - _____

25 **Which of the following items were within the infant's reach?**
☐ Blankets ☐ Toys ☐ Pillows ☐ Pacifier ☐ Nothing ☐ Other - _____

26 **Was anyone sleeping with the infant?** ☐ No ☐ Yes

Name of individual sleeping with infant	Age	Height	Weight	Location in relation to infant	Impairment (intoxication, tired)

27 **Was there evidence of wedging?** ☐ No ☐ Yes - Describe: _____

28 **When the infant was found, was s/he:** ☐ Breathing ☐ Not Breathing
If not breathing, did you witness the infant stop breathing? ☐ No ☐ Yes

WITNESS INTERVIEW (cont.)

29 **What had led you to check on the infant?**

30 **Describe the infant's appearance when found.**

Appearance	Unknown	No	Yes	Describe and specify location
a) Discoloration around face/nose/mouth				
b) Secretions (foam, froth)				
c) Skin discoloration (livor mortis)				
d) Pressure marks (pale areas, blanching)				
e) Rash or petechiae (small, red blood spots on skin, membranes, or eyes)				
f) Marks on body (scratches or bruises)				
g) Other				

31 **What did the infant feel like when found?** *(Check all that apply.)*

☐ Sweaty ☐ Warm to touch ☐ Cool to touch ☐ Limp, flexible ☐ Rigid, stiff ☐ Unknown

☐ Other - specify:

32 **Did anyone else other than EMS try to resuscitate the infant?** ☐ No ☐ Yes

Who? Date: Military time: :

33 **Please describe what was done as part of resuscitation:**

34 **Has the parent/caregiver ever had a child die suddenly and unexpectedly?** ☐ No ☐ Yes

Explain:

INFANT MEDICAL HISTORY

1 **Source of medical information:** ☐ Doctor ☐ Other healthcare provider ☐ Medical record ☐ Family

☐ Mother/primary caregiver ☐ Other:

2 **In the 72 hours prior to death, did the infant have:**

Condition	Unknown	No	Yes	Condition	Unknown	No	Yes
a) Fever				h) Apnea (stopped breathing)			
b) Diarrhea				i) Decrease in appetite			
c) Excessive sweating				j) Cyanosis (turned blue/gray)			
d) Stool changes				k) Vomiting			
e) Lethargy or sleeping more than usual				l) Seizures or convulsions			
f) Difficulty breathing				m) Choking			
g) Fussiness or excessive crying				n) Other, specify:			

3 **In the 72 hours prior to death, was the infant injured or did s/he have any other condition(s) not mentioned?**

☐ No ☐ Yes - describe:

4 **In the 72 hours prior to the infants death, was the infant given any vaccinations or medications?** ☐ No ☐ Yes

(Please include any home remedies, herbal medications, prescription medicines, over-the-counter medications.)

Name of vaccination or medication	Dose last given	Date given Month	Day	Year	Approx. time (Military Time)	Reasons given/comments:
1.						
2.						
3.						
4.						

5 **At any time in the infant's life, did s/he have a history of?**

Medical history	Unknown	No	Yes	Describe
a) Allergies *(food, medication, or other)*				
b) Abnormal growth or weight gain/loss				
c) Apnea *(stopped breathing)*				
d) Cyanosis *(turned blue/gray)*				
e) Seizures or convulsions				
f) Cardiac (*heart*) abnormalities				

6 **Did the infant have any birth defects(s)?** ☐ No ☐ Yes

Describe:

7 **Describe the two most recent times that the infant was seen by a physician or healthcare provider:**
(Include emergency department visits, clinic visits, hospital admissions, observational stays, and telephone calls)

	First most recent visit	Second most recent visit
a) Date		
b) Reason for visit		
c) Action taken		
d) Physician's name		
e) Hospital/clinic		
f) Address		
g) City		
h) State, ZIP		
i) Phone number		

8 **Birth hospital name:** Discharge date:

Street address:

City: State: Zip:

9 **What was the infant's length at birth?** inches or centimeters

10 **What was the infant's weight at birth?** pounds ounces or grams

11 **Compared to the delivery date, was the infant born on time, early, or late?**

☐ On time ☐ Early - how many weeks? ☐ Late - how many weeks?

12 **Was the infant a singleton, twin, triplet, or higher gestation?**

☐ Singleton ☐ Twin ☐ Triplet ☐ Quadrupelet or higher gestation

13 **Were there any complications during delivery or at birth?** *(emergency c-section, child needed oxygen)* ☐ Yes ☐ No

Describe:

14 **Are there any alerts to the pathologist?** *(previous infant deaths in family, newborn screen results)* ☐ Yes ☐ No

Specify:

INFANT DIETARY HISTORY

1 **On what day and at what approximate time was the infant last fed?**

Date: Military Time: :

2 **What is the name of the person who last fed the infant?**

3 **What is his/her relationship to the infant?**

4 **What foods and liquids was the infant fed in the last 24 hours** (include last fed)**?**

Food	Unknown	No	Yes	Quantity (ounces)	Specify: (type and brand)
a) Breastmilk (one/both sides, length of time)					
b) Formula (brand, water source - ex. Similac, tap water)					
c) Cow's milk					
d) Water (brand, bottled, tap, well)					
e) Other liquids (teas, juices)					
f) Solids					
g) Other					

5 **Was a new food introduced in the 24 hours prior to his/her death?** No Yes
If yes, describe *(ex. content, amount, change in formula, introduction of solids)*

6 **Was the infant last placed to sleep with a bottle?** Yes No - if no, skip to question **9** below

7 **Was the bottle propped?** (i.e., object used to hold bottle while infant feeds) No Yes

If yes, what object was used to prop the bottle?

8 **What was the quantity of liquid (in ounces) in the bottle?**

9 **Did the death occur during?** Breastfeeding Bottle-feeding Eating solid foods Not during feeding

10 **Are there any factors, circumstances, or environmental concerns that may have impacted the infant that have not yet been identified?** *(ex. exposed to cigarette smoke or fumes at someone else's home, infant unusually heavy, placed with positional supports or wedges)*

No Yes

If yes, - describe:

PREGNANCY HISTORY

1 **Information about the infant's birth mother:**

First name: Last name:
Middle name: Maiden name:
Birth date: SS#:

Street address: City: State: Zip:

How long has the birth mother been at this address? Years: Months:

Previous Address:

2 **At how many weeks or months did the birth mother begin prenatal care?** No prenatal care Unknown

Weeks: Months:

3 **Where did the birth mother receive prenatal care?** *(Please specify physician or other healthcare provider name and addresses.)*

Physician/ Provider: Hospital/clinic: Phone:

Street address: City: State: Zip:

PREGNANCY HISTORY (cont.)

4 **During her pregnancy with the infant, did the mother have any complications?** ☐ No ☐ Yes
(ex. high blood pressure, bleeding, gestational diabetes)

Specify:

5 **Was the birth mother injured during her pregnancy with the infant?** *(ex. auto accident, falls)* ☐ No ☐ Yes

Specify:

6 **During her pregnancy, did she use any of the following?**

	Unknown	No	Yes	Daily		Unknown	No	Yes	Daily
a) Over the counter medications					d) Cigarettes				
b) Prescription medications					e) Alcohol				
c) Herbal remedies					f) Other				

7 **Currently, does any caregiver use any of the following?**

	Unknown	No	Yes	Daily		Unknown	No	Yes	Daily
a) Over the counter medications					d) Cigarettes				
b) Prescription medications					e) Alcohol				
c) Herbal remedies					f) Other				

INCIDENT SCENE INVESTIGATION

1 **Where did the incident or death occur?**

2 **Was this the primary residence?** ☐ No ☐ Yes

3 **Is the site of the incident or death scene a daycare or other childcare setting?** ☐ Yes ☐ No - If no, skip to question **8**

4 **How many children (under age 18) were under the care of the provider at the time of the incident or death?**

5 **How many adults (age 18 and over) were supervising the child(ren)?**

6 **What is the license number and licensing agency for the daycare?**

License number: Agency:

7 **How long has the daycare been open for business?**

8 **How many people live at the site of the incident or death scene?**

Number of adults (18 years or older): Number of children (under 18 years old):

9 **Which of the following heating or cooling sources were being used?** *(Check all that apply)*

☐ Central air ☐ Gas furnace or boiler ☐ Wood burning fireplace ☐ Open window(s)
☐ A/C window unit ☐ Electric furnace or boiler ☐ Coal burning furnace ☐ Wood burning stove
☐ Ceiling fan ☐ Electric space heater ☐ Kerosene space heater ☐ Floor/table fan
☐ Electric baseboard heat ☐ Electric (radiant) ceiling heat ☐ Window fan ☐ Unknown

☐ Other - specify:

10 **Indicate the temperature of the room where the infant was found unresponsive:**

☐ Thermostat setting ☐ Thermostat reading ☐ Actual room temp. ☐ Outside temp.

11 **What was the source of drinking water at the site of the incident or death scene?** *(Check all that apply.)*

☐ Public/municipal water ☐ Bottled water ☐ Well ☐ Unknown ☐ Other - Specify:

12 **The site of the incident or death scene has:** *(check all that apply)*

☐ Insects ☐ Mold growth ☐ Smoky smell *(like cigarettes)*
☐ Pets ☐ Dampness ☐ Presence of alcohol containers
☐ Peeling paint ☐ Visible standing water ☐ Presence of drug paraphenalia
☐ Rodents or vermin ☐ Odors or fumes - Describe:

☐ Other - specify:

13 **Describe the general appearance of incident scene:** *(ex. cleanliness, hazards, overcrowding, etc.)*

Specify:

INVESTIGATION SUMMARY

1 **Are there any factors, circumstances, or environmental concerns about the incident scene investigation that may have impacted the infant that have not yet been identified?**

2 **Arrival times**

	Military time
Law enforcement at scene:	:
DSI at scene:	:
Infant at hospital:	:

Investigator's Notes

1 **Indicate the task(s) performed**

- ☐ Additional scene(s)? (forms attached)
- ☐ Materials collected/evidence logged
- ☐ Notify next of kin or verify notification
- ☐ Doll reenactment/scene re-creation
- ☐ Referral for counseling
- ☐ 911 tape
- ☐ Photos or video taken and noted
- ☐ EMS run sheet/report

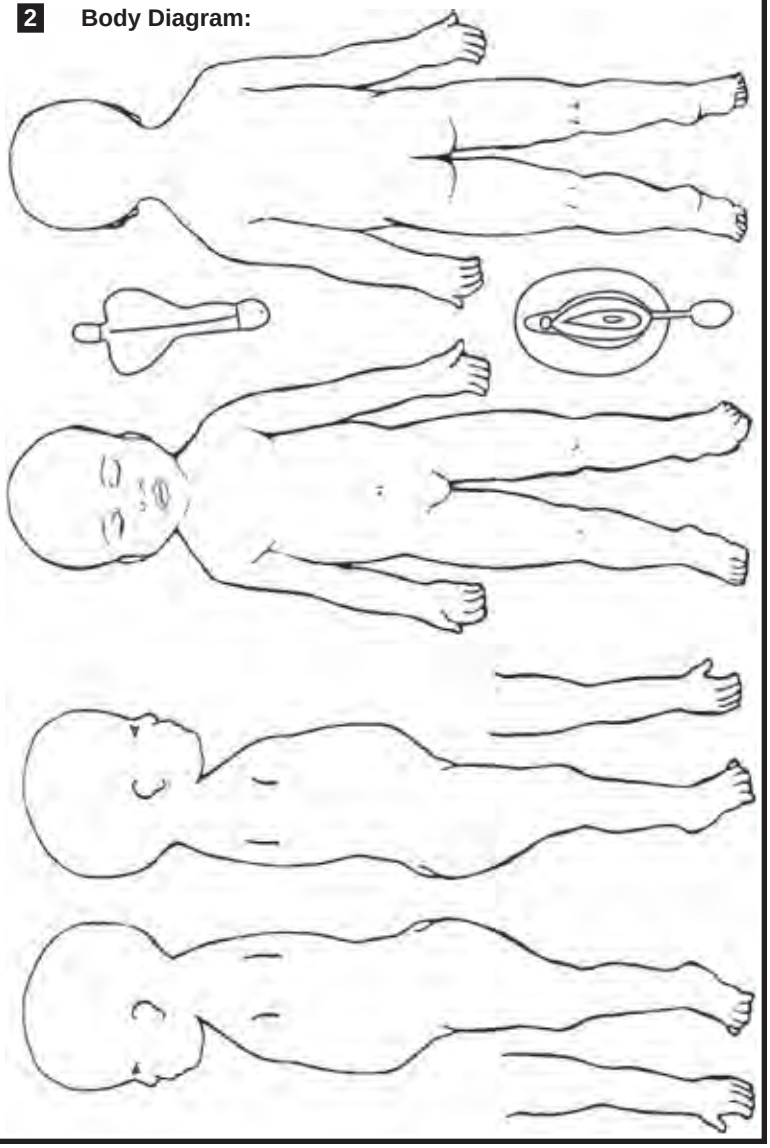
2 **If more than one person was interviewed, does the information differ?** ☐ No ☐ Yes

If yes, detail any differences, inconsistencies of relevant information: *(ex. placed on sofa, last known alive on chair.)*

INVESTIGATION DIAGRAMS

1 **Scene Diagram:**

2 **Body Diagram:**



SUMMARY FOR PATHOLOGIST

Case Information

1 **Investigator information** Name: Agency: Phone:

	Date	Military time
Investigated:		:
Pronounced dead:		:

2 **Infant's information:** Last: First: M: Case #:

Sex: ☐ Male ☐ Female Date of Birth: Age:

Race: ☐ White ☐ Black/African Am. ☐ Asian/Pacific Islander

☐ Am. Indian/Alaskan Native ☐ Hispanic/Latino ☐ Other:

Sleeping Environment

1 **Indicate whether preliminary investigation suggests any of the following:**

Yes	No	
☐	☐	Asphyxia *(ex. overlying, wedging, choking, nose/mouth obstruction, re-breathing, neck compression, immersion in water)*
☐	☐	Sharing of sleep surface with adults, children, or pets
☐	☐	Change in sleep condition *(ex. unaccustomed stomach sleep position, location, or sleep surface)*
☐	☐	Hyperthermia/Hypothermia *(ex. excessive wrapping, blankets, clothing, or hot or cold environments)*
☐	☐	Environmental hazards *(ex. carbon monoxide, noxious gases, chemicals, drugs, devices)*
☐	☐	Unsafe sleep condition *(ex. couch/sofa, waterbed, stuffed toys, pillows, soft bedding)*

Infant History

☐	☐	Diet (*e.g., solids introduced, etc.*)
☐	☐	Recent hospitalization
☐	☐	Previous medical diagnosis
☐	☐	History of acute life-threatening events *(ex. apnea, seizures, difficulty breathing)*
☐	☐	History of medical care without diagnosis
☐	☐	Recent fall or other injury
☐	☐	History of religious, cultural, or ethnic remedies
☐	☐	Cause of death due to natural causes other than SIDS *(ex. birth defects, complications of preterm birth)*

Family Info

☐	☐	Prior sibling deaths
☐	☐	Previous encounters with police or social service agencies
☐	☐	Request for tissue or organ donation
☐	☐	Objection to autopsy

Exam

☐	☐	Pre-terminal resuscitative treatment
☐	☐	Death due to trauma (injury), poisoning, or intoxication

Investigator Insight

☐	☐	Suspicious circumstances
☐	☐	Other alerts for pathologist's attention

Any "Yes" answers above should be explained in detail (description of circumstances):

Pathologist

2 **Pathologist information** Name:

Agency: Phone: Fax:

APPENDIX B

As we entered the large, steel gates, I could feel apprehension wash over me. We tell security that I am part of the research team led by Sarah and we don't need to sign in. This is the place where people come to identify their deceased loved ones for legal purposes. Not just the mothers who just lost their infant such as in the current study, but also others who come to say goodbye to their mother, father, brother, sister, grandparent, spouse or child.

It might be my imagination, but you can feel the anxiety and the sadness in the air. This is in contrast to the warm sunny day. At the big security gate that we just entered, there is a family of four waiting to sign in and I wonder why they are here. We walk into what looks like a side door, but is the main entrance. I didn't notice the large signs declaring it as 'Forensic Pathology Service'. It is quiet in the reception area. It is a Wednesday afternoon. There is not a lot of natural light and the walls are painted a pale yellow. There are wooden benches for people to sit on. The officers behind the reception desk know Sarah. They are friendly and are talking about everyday matters.

To the left of the reception desk, there is a narrow hallway. We walk down it and speak in hushed tones. I didn't even realise that we passed the little hallway to the four viewing rooms. We turn left into a room. There is a brown wooden desk. On the table I can see a computer, box of tissues and ink pad. There is a plant in the corner. Sarah would sit behind the desk and explain her study to the bereaved parents. It is not a memorable room, not to me. There are no pictures on the walls to create even a semblance of warmth. Sarah explains that there are often a number of people with her in this room. Sometimes they are quiet and sometimes they are crying. Religious leaders sometimes also join the family here.

Next to this room, there is another small waiting room. This is the main waiting room. The walls are also yellow. There are a couple of coffee tables with pamphlets. I didn't look at the

pamphlets. There are paper posters on the wall from the Western Cape Government. The only bright point is the display of children's drawings.

Sarah needs to pick up a blood sample for her study. We go to the back. It is busy and confusing, with a lot of smaller rooms. These are the offices of the Forensic Pathology Officers and teaching rooms. I never even went to the viewing rooms. I don't like the feeling of the place, but as well as the bereaved and the dead, there are also hard-working people there that want to provide answers and closure to many families.

As we leave, an undertaker's van pulls in.

APPENDIX C

OPEN-ENDED INTERVIEW GUIDE

1.) What can you remember about the process at the morgue? (Time spent there, forms, staff)

2.) How did you feel when you were asked to participate in research and they needed DNA from your baby?

3.) What can you remember about what you were told about the research? (What is it for, what will it do, what do they need from you)

4.) Why do you think that you said yes?

5.) Have you thought about the research since then? What are you expecting back from the research? How would you feel if you hear from them again?

6.) What are your concerns about taking part in research?

7.) What have you gained from participating in the research?

APPENDIX D

REQUEST FOR M.Sc RESEARCH PARTICIPATION

Genetic Counselling

Division of Human Genetics

UCT Medical School, Observatory 7925

Tel: (021) 404 6235

Dear participant

You are hereby invited to participate in a study for the completion of a Master's Degree in Genetic Counselling with the title:

Forensic genetic research on sudden unexpected death in an infant (SUDI) at Salt River Mortuary: experiences and perceptions of parents

<u>About the study</u>

The main reason that I am doing this study is to explore parents' perceptions, experiences and understanding of agreeing to provide a DNA sample of their baby who passed away suddenly. I am interested in your reasons for having DNA testing on your child; what do you remember about what the researcher told you; and how you feel about having participated in a research study.

This study will not have any impact on the previous study that you participated in. The results from this study will help us to see if we should do research in a different way, or explain it in a different way, to make it as easy as possible for parents in a similar situation to participate. It will also help us to see what people expect from such research and if we can meet some of those expectations. It would also help us to decide if there is a need for someone with special training to explain DNA studies and ask for participation.

<u>Participation</u>

To take part in this study, you will be interviewed at a place convenient for you. I will only interview you and you will not have to do any other examinations or tests. You are free to withdraw from the study at any time, and if you do decide to stop, there will be **NO** negative consequences. It is completely voluntary and will not cost you any money. Similarly, you will also not receive any money or any other compensation if you decide to do the interview.

You will be asked to provide a few personal details, but it will be treated with the strictest confidence and in line with the highest ethical standards. All recordings will be stored in a locked cupboard. Electronic versions will be password protected. Only the researcher and her supervisors will have access to the information. All recording will be destroyed once the study is completed and identities will not be made known. Also, the information will only be used for research at the University of Cape Town, and will **NOT** be available to any other person or company. The results of this study may be published in a scientific journal, but no personal information will be printed. It will not be possible to link the information from the interviews to you. Your identity will remain secret and only the researcher will know your personal information. All in all, participating in this study should not take more than 1 hour of your time.

Contact information

This study will be done by a Master's student at the University of Cape Town, under the supervision of Ms. L. Heathfield (Lecturer: Forensic Genetics) and Dr. T. Wessels (Senior lecturer: Genetic Counselling).

Please feel free to contact Susan Louw (021 404 6235), Ms. Heathfield (021 406 6569) or Dr. Wessels (021 406 6373) with any questions you may have regarding this study. You may contact the HREC Human Research Ethics Committee (HREC) at the Faculty of Health Sciences at the University of Cape Town, telephone number (021) 406 6492 if you have questions about your rights as a research participant.

Thank you for your cooperation.

Susan Louw

(Office nr: 021 404 6235)

REQUEST FOR M.Sc RESEARCH

PARTICIPATION

Genetic Counselling

Division of Human Genetics

UCT Medical School

Observatory 7925

Tel: (021) 404 6235

Please fill in all the information requested:

Surname:_____ First name(s):_____

Sex: M ☐ F ☐ Date of Birth: Year:_____ Month: _____Day: _____

Number of children: _____

Contact Address: _____Town: _____

Tel:_____

For Research Use Only:

*Participant number:*_____

*Date Received: Year:*_____ *Month:* _____ *Day:* _____

CONSENT FOR INTERVIEW AND RECORDING

1. I, _____ , have been asked to take part in a research study to find out about my perceptions, experiences and understanding of agreeing to provide a DNA sample of my baby who passed away suddenly. The study has been approved by the Human research Ethics Committee at the Faculty of Health Sciences at the University of Cape Town (HREC:).

2. I understand that taking part in this study involves being interviewed. This interview will be about one hour and it will be voice recorded.

3. I understand that taking part is my choice and all the information from this study will remain confidential. Information will be used for research purposes only.

4. I understand that the interview questions may cause some emotional pain and I may choose not to answer a question of I do not wish to. I also have the right to stop the interview at any time.

5. The results of the study may give a better understanding of research methods and the ethical issues of research of sensitive topics.

6. I agree / do not agree [DELETE WHERE APPLICABLE] to the recording of this interview. All recordings will be stored in a locked cupboard. Electronic versions will be password protected. Only the researcher and her supervisors will have access to the information. All recording will be destroyed once the study is completed and identities will not be made known.

7. I have been told that there will be no payment for taking part in this study.

8. I understand that I may withdraw from this study my at any time without this affecting my future medical care.

9. **ALL OF THE ABOVE HAS BEEN EXPLAINED TO ME IN A LANGUAGE THAT I UNDERSTAND AND MY QUESTIONS ANSWERED BY:**

   _____. DATE: _____

   PATIENT SIGNATURE: _____
   WITNESS SIGNATURE:_____

NOTE - PLEASE INSERT A FAMILY PEDIGREE DRAWING ON THE REVERSE OF THIS FORM

UNIVERSITY OF CAPE TOWN
Faculty of Health Sciences
Human Research Ethics Committee

Room E53-46 Old Main Building
Groote Schuur Hospital
Observatory 7925
Telephone [021] 406 6492
Email: sumayah.ariefdien@uct.ac.za
Website: www.health.uct.ac.za/fhs/research/humanethics/forms

04 October 2017

HREC REF:627/2017

Ms L Heathfield
Division of Forensic Pathology
Falmouth Building-FHS

Dear Ms Heathfield

PROJECT TITLE: FORENSIC GENETIC RESEARCH ON SUDDEN UNEXPECTED DEATH IN AN INFANT (SUDI) AT SALT RIVER MORTUARY: EXPERIENCES AND PERCEPTIONS OF PARENTS- LINKERD TO 445/2015 (MSc candidate-Ms Susan Louw)

Thank you for your response letter dated 25 August 2017, addressing the issues raised by the Human Research Ethics Committee (HREC).

It is a pleasure to inform you that the HREC has **formally approved** the above-mentioned study.

Approval is granted for one year until the 30 October 2018.

Please submit a progress form, using the standardised Annual Report Form if the study continues beyond the approval period. Please submit a Standard Closure form if the study is completed within the approval period.
(Forms can be found on our website: www.health.uct.ac.za/fhs/research/humanethics/forms)

We acknowledge that the student: S Louw will also be involved in this study.

Please quote the HREC REF in all your correspondence.

Please note that the ongoing ethical conduct of the study remains the responsibility of the principal investigator.

Please note that for all studies approved by the HREC, the principal investigator **must** obtain appropriate institutional approval, where necessary, before the research may occur.

Yours sincerely

PROFESSOR M BLOCKMAN
CHAIRPERSON, FHS HUMAN RESEARCH ETHICS COMMITTEE

Federal Wide Assurance Number: FWA00001637.
Institutional Review Board (IRB) number: IRB00001938

This serves to confirm that the University of Cape Town Human Research Ethics Committee complies to the Ethics Standards for Clinical Research with a new drug in patients, based on the Medical Research Council (MRC-SA), Food and Drug Administration (FDA-USA), International Convention on Harmonisation Good Clinical Practice (ICH GCP), South African Good Clinical Practice Guidelines (DoH 2006), based on the Association of the British Pharmaceutical Industry Guidelines (ABPI),and Declaration of Helsinki (2013) guidelines.
The Human Research Ethics Committee granting this approval is in compliance with the ICH Harmonised Tripartite Guidelines E6: Note for Guidance on Good Clinical Practice (CPMP/ICH/135/95) and FDA Code Federal Regulation Part 50, 56 and 312.