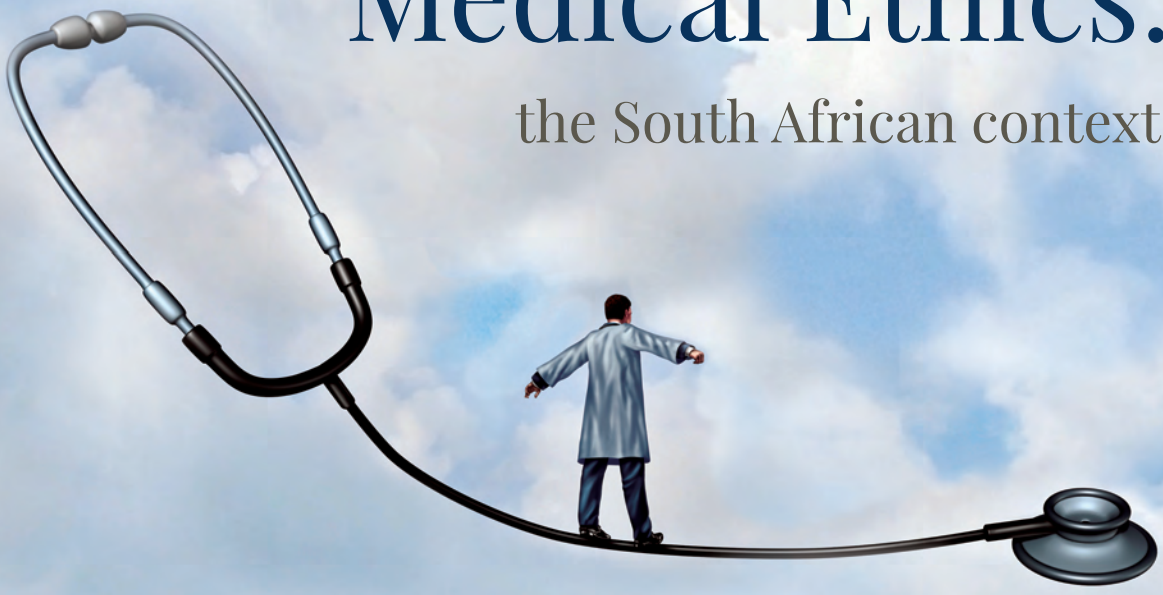
C. Jones, M. Kruger, J. van den Heever & A. van Niekerk (eds.). Challenges in Medical Ethics: the South African context. Stellenbosch: African Sun Media

Challenges in Medical Ethics:

the South African context



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12

How to Treat Private and Sensitive Medical and Genetic Data

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Keywords: confidentiality; consent; data sharing; electronic health records; genetic ancestry testing; genetic data; incidental findings; non-paternity; privacy

12.1 INTRODUCTION

Genomics research has significantly contributed to the improvement of diagnosis, treatment, and delivery of health care. Fundamental to the success of these interventions is public trust and confidence in researchers and medical professionals, underpinned by a range of ethical challenges particularly those related to consent, privacy, ownership of samples, and data sharing.

Seeley and Parker (2020) have explored issues related to feedback on findings and the nature of the relationship between genome research and medical practice, while Fisher and Layman (2018) have addressed the roles of "genetic and environment[al] factors on the development of mental health and behavioural risk, resilience and individual responsivity to prevention" and intervention programmes. Fisher and Laymen (2018) have argued that the progress of utilising biospecimens in prevention science parallels the rise in the integration of large datasets, commonly referred to as 'big data', across all scientific disciplines. The use of big data "is marked by increased access to and use of individual-level data across administrative data systems with the potential to link genomic data to health, public benefits, child welfare, criminal and juvenile justice, and educational records and other personal information" (Fisher & Layman, 2018) educational, and other information stored in large integrated datasets. These advances have created a new frontier of ethical challenges for scientists as they

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collect, store, or engage in secondary use of potentially identifiable information and biospecimens. To address challenges arising from technological advances and the expanding contexts in which potentially identifiable information and biospecimens are collected and stored, the Office of Human Research Protections has revised federal regulations for the protection of human subjects. The revised regulations create a new format, content, and transparency requirements for informed consent, including a new mechanism known as broad consent. Broad consent offers participants a range of choices regarding consent for the storage and future use of their personally identifiable data. These regulations have important implications for how prevention scientists and oversight boards acquire participant consent for the collection, storage, and future use of their data by other investigators for scientific purposes significantly different from the original study. This chapter describes regulatory changes and challenges affecting traditional informed consent for prevention research, followed by a description of the rationale and requirements for obtaining broad consent, and concludes with a discussion of future challenges involving ongoing transparency and protections for participants and their communities.

Along with the many benefits genomics and big data analytical tools have provided to society, many concerns such as re-identification of previously de-identified participant information have been raised. The collection, storage, and secondary use of biospecimens are compounded by further challenges. An area of intense debate has been the "model of consent that would be appropriate for genomic and biobanking studies in low and middle-income countries, and what type of governance mechanism is required for safeguarding the interest of research participants and their communities in such studies" (Seeley & Parker, 2020).

From an ethical point of view, respect for the principles of autonomy (informed consent, confidentiality, truth telling, and communication), beneficence (do good), non-maleficence (do no harm), and justice (rights justice, legal justice, and distribution justice) need to be considered together when addressing questions related to how we treat private and sensitive medical and genetic data.

This chapter discusses some of the ethical issues related to the privacy of sensitive medical and genetic data.

12.2 HOW SENSITIVE IS GENETIC DATA?

Essentially, the challenge with genetic data is the type of information it may contain such as ethnicity and health relevant information, together with their re-identification potential (Sariyar, Shur & Schlünder, 2017). Generally, genetic

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data can be globally unique, but as soon as data becomes linkable to a specific person, it becomes an issue for that individual and their relatives, making the problem of privacy even more complex. These authors have reviewed the sensitivity of genetic data based on criteria such as the type of genetic markers used, its stability and distinguishability as inherent characteristics.

Based on these criteria, they conclude that single nucleotide polymorphisms (SNPs) are mostly useful when combined with other gene-related information, which when used alone are less critical than other forms of genetic data – adding support for their use and value in public-domain databases such as *dbSNP* (Sariyar et al., 2017; Sherry et al., 2001). Short tandem repeat (STR) data, which are mostly used for genetic fingerprinting purposes, especially in forensic applications, are less relevant for inferring further re-identification unless used in conjunction with other information. Also, copy number variants (CNVs) can contain many genes and are therefore useful for many purposes and may play a bigger role than SNP and STR data in disclosure issues. DNA methylation as an epigenetic phenomenon is less stable than other kinds of genetic markers and does not pose much risk in issues related to sensitivity. However, whole genome sequence (WGS) data is regarded as "exceptional" and require special protection. In their (Sariyar et al., 2017) view, "once the full genome of an individual has become public, it can be used in a variety of settings for unforeseeable and discriminatory purposes". Their main argument against genetic exceptionalism is that "the characteristics of genetic data are like other medical or health-related information (e.g., it can be used to disclose an individual's identity, disease risks, or drug response)".

12.3 CONSENT

Chapter 2 of the National Health Act (2003) deals with rights and duties of users and health care personnel. Section 7 focuses on informed consent, which is enshrined in one of the four ethical principles referred to as "respect for autonomy". As Moodley (2023) outlines, "autonomy literally means self-rule". It refers to the right of every individual to make decisions for himself or herself. In health care, this entails allowing the patient to make the final decision about his/her treatment after the relevant information was clearly explained to them. It is also suggested that this principle should not be used for persons who are incapacitated, ignorant, infants, young children, suicidal, some drug-dependent patients, and patients with severe psychiatric illness that would incapacitate them (Moodley, 2023).

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Informed consent is essential for enrolment of participants for research, clinical trials, and diagnostic testing for patients. However, questions arise about ethical issues related to divulging information about deceased patients. One case that highlights some of the controversy around the sensitivity of this issue relates to the publication of the book, *Mandela's Last Years: The True Story of Nelson Mandela's Final Journey* published by Dr Ramlakan, the head of his medical team (Ramlakan, 2017). Rule 13(2)(c) of the Ethical Rules of Conduct by the Health Professionals Council of South Africa (HPCSA) state that confidential information about a deceased person should only be divulged "with the written consent of his or her next of kin or the executor of the estate" (HPCSA, 2016a). "Next of kin" is not clearly defined which leaves open the question of whether a spouse or only immediate genetic relatives qualify as next of kin. As in this case, problems arise when there is conflict among family members and the executors about giving written consent. It is recommended that "in such cases the specific order of priority for consent by relatives in the National Health Act (2003) be followed" (McQuoid-Mason, 2017).

McQuoid-Mason (2017) recommended that certain questions be considered prior to divulging medical information on deceased individuals, such as (1) when is it ethical to publish such information? (2) which family members or next of kin can consent to publication? (3) when is it legal to publish such information? (4) does it make a difference if the deceased is a public figure? and (5) whether family members have legal standing to prevent or claim damages for such publication on behalf of the deceased.

Having evaluated these questions using principles in medical ethics and the law, McQuoid-Mason (2017) noted that if a doctor has made disclosures about a deceased person's medical treatment in breach of the rule 13(2)(c), the deceased person's next of kin or the executor of the estate may file a complaint with the HPCSA, and disciplinary action can be taken against the doctor for unethical conduct. The problem arises if some family members give consent and others do not, and actively oppose the publication. Unfortunately, the rule does not define "next of kin", nor clearly indicates which next-of-kin takes precedence over others.

McQuoid-Mason argues that the National Health Act [section 7(1)(b)] specifies the order of persons who can give consent on behalf of incompetent live patients and suggests that this could be used as a guide when determining which next-of-kin relatives should have the right to give written consent for the publication of personal medical information of a deceased person. This is based on people who are authorised to give consent on behalf of a patient to access medical treatment.

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In terms of the Act, "the order of precedence is a spouse or partner, a parent, a grandparent, an adult child or a brother or sister of the person" (McQuoid-Mason, 2017). However, the South African Law Reform Commission (SALRC) has noted that this precedence "could be interpreted to imply that a brother should be consulted before a sister" (SALRC, 2018). The SALRC noted that "while there is justification for consulting a spouse or partner before consulting a child and consulting a child before consulting a sibling due to marriage, an intimate partnership or degree of family relationship, there is no justification for determining that a brother should be consulted before a sister" (SALRC, 2018). The SALRC has therefore determined that the wording in the National Health Act (2003) is discriminatory based on gender and hence in breach of section 9 of the Constitution.

Regarding when it is legal to publish information about a deceased person, the constitutional and common-law right to privacy concerning their health status must be upheld. According to McQuoid-Mason (2017), "the right is not unlimited and may be infringed where the person concerned consents, where there is a statutory duty to make disclosure (e.g., in the case of child abuse), where the disclosure is true and in the public interest or where the disclosure is privileged". The defence of truth and public interest are relevant when addressing disclosures concerning health status of public figures.

Public figures are "people who have by their personality, status or conduct exposed themselves and their families to such a degree of the publicity as to justify public disclosures of certain aspects of their private lives. Such persons include politicians, actors, entertainers, sportsmen and sportswomen, war heroes, and others who are regarded as having a limited right to private lives" since they have sought or consented to publicity (McQuoid-Mason, 2017). Should such personalities fail to carry out their professional duties or make false statements, then the media has a duty to inform the public.

President Mandela was a world-famous public figure and was always at the centre of much publicity. However, he was entitled to a private life and the public needed to respect that unless his private life contradicted what he stood for as a statesman. His and his family's wish for privacy after he retired from political life should have been respected.

Conduct that is unethical under the rules of the HPCSA may not necessarily be actionable under the law. There is no general protection for the personality rights of deceased persons. Consequently, if a doctor intentionally discloses private information about a deceased patient, there would be no action for damages in law because the person is deceased (McQuoid-Mason, 2017). Even the next-

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of-kin have no grounds to sue the doctor under these circumstances. "This is because the right to sue for breach of confidentiality or invasion of privacy vests in deceased persons during their lifetime, and not in their next of kin or executors after their death" (McQuoid-Mason, 2017).

This situation was resolved by the publishers withdrawing the book from circulation and never went to court. Despite the legal right to sue for breach of confidentiality or invasion vesting with deceased persona during their lifetime, the sharing of confidential health details of deceased persons raises profound ethical issues and arguably constitutes a moral harm.

12.4 CONFIDENTIALITY

The National Health Act (2003) makes it an offence to disclose patients' information without their consent, except in certain circumstances. Sections 14, 15, and 16 of the Act are pertinent with regard to confidentiality. Sections 15 and 16 describe how patient information may be disclosed by a health care worker "for any legitimate purpose within the ordinary course and scope of his or her duties where such access or disclosure is in the interests of the user" (National Health Act, 2003).

Respect for confidentiality is important to safeguard the well-being of patients and to ensure the confidence of society in the doctor-patient relationship. "Health information is not only based on objective observations, diagnoses, and test results, but also subjective impressions about the patient, their lifestyle, habits, and recreational activities. The improper disclosure of such highly sensitive information could harm patients' reputation or result in lost opportunities, financial commitments, and even personal humiliation (Beltran-Aroca et al., 2016).

The HPCSA places confidentiality as central to the doctor-patient relationship and a core aspect of the trust that holds the relationship together. The HPCSA's official guidance, *Confidentiality: Protecting and Providing Information* (2008), emphasises these key principles:

1. Patients have a right to expect that information about them will be held in confidence by health care practitioners. Confidentiality is central to trust between practitioners and patients. Without assurances about confidentiality, patients may be reluctant to give practitioners the information they need to provide good care.
2. Where health care practitioners are asked to provide information about patients, they should:

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a. Seek the consent of patients to disclosure of information wherever possible, whether or not the patients can be identified from the disclosure; Comprehensive information must be made available to patients with regard to the potential for a breach of confidentiality with ICD10 coding.
b. Anonymise data where unidentifiable data will serve the purpose;
c. Keep disclosures to the minimum necessary.

3. Health care practitioners must always be prepared to justify their decisions in accordance with these guidelines.

There are two general exceptions where it is necessary to question whether it is essential to maintain confidentiality: (1) when the safety of others or (2) public health is threatened (Beltran-Aroca et al., 2016). The HPCSA advocates that the risk of harm must be serious enough to outweigh the patient's right to confidentiality and recommends that consent of the patient should be sought first before disclosure unless this is not forthcoming.

Given the emphasis on confidentiality in the National Health Act (2003) and the HPCSA guidelines, the Constitutional Court ruling on the case of *NM, SM and LH v Charlene Smith, Patricia de Lille and New Africa Books* has been criticised by Mark Heywood representing the Aids Law Project in this case (Section27, 2007).[1]

This case was taken to the Constitutional Court by three women (NM, SM, and LH) against Charlene Smith, Patricia de Lille, and New Africa Books since the defendants had published their full names and HIV status without their consent in the biography of De Lille, written by Charlene Smith and published by New Africa Books. The women had participated in clinical trials and raised concerns about the illness and fatalities among the participants. They submitted that the consent forms for the trials were not properly explained to them. De Lille was contacted to help investigate the complaints. Professor SA Strauss was appointed to conduct an inquiry into the allegations of misconduct and subsequently issued a report on the trials (the Strauss Report). This report contained the applicants' names and HIV status.

"The women argued that the disclosure of their names and HIV status in the book was an invasion of their rights to privacy, dignity, psychological integrity and mental and intellectual well-being." They asked the Court to grant them relief on the following issues: "(1) an order directing the defendants to issue a private apology to each plaintiff, (2) an order directing the defendants to cause the

1 https://bit.ly/3VhTTr8

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offending passages to be excised or removed from all unsold copies of the book, and (3) an order directing the defendants to pay damages of R200,000 to each plaintiff" (Parker & Lucassen, 2018; Section27, 2007). In this judgment, Judge Schwartzman (Section27, 2007) referred to previous case law which confirmed that the "right to privacy entitles an individual to decide when and under what circumstances private facts may be made public". He further acknowledged that "because of the ignorance and prejudices of large sections of our population, an unauthorised disclosure [of HIV status] can result in social and economic ostracism" and "can even lead to mental and physical assault".

In summary, Mark Heywood (Aids Law Project) presented some explanation for Judge Schwarzman's decision as follows (Section27, 2007):

- De Lille and Smith have, by their long-standing involvement with people living with HIV/AIDS, demonstrated that they are unlikely to intentionally invade the privacy of a person living with HIV.
- The women's names and HIV status was contained in the report of an official inquiry, commissioned by a public body into a matter of public interest. Nothing in the report suggested that any part of it was confidential. Accordingly, the defendants were entitled "to assume" that in mentioning the plaintiffs' names and HIV status, they were acting in terms of a consent furnished by the plaintiffs to the inquiry.
- There was no legal duty on the defendants to establish the nature of the consent provided by the women for the use of their names in the report.
- Although he noted that publishing the names without informed consent had "transgressed the norms and standards of the profession" ethically, the dispute concerned legal issues and was "not a question of journalistic ethics or standards."
- The fact that the plaintiffs had not consented to the disclosure of their names and HIV status and had asserted this right through legal action after the publication of the book in March 2002, entitles the plaintiffs to damages and to an order for the removal of their names.
- In this respect, only the publishers are liable for the damages which occurred after the women asserted their rights by starting legal action in April 2002.

Judge Schwartzman nevertheless awarded limited damages and gave the following reasons (Section27, 2007):

- The women's fear that their family and community would come to learn of their HIV status through the book did not materialise. This finding is in fact incorrect since NM testified how her boyfriend had come to know of her

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status through his friends who read the book. Their relationship deteriorated and he subsequently burned down her house.

- The women's fears "of a likelihood that the disclosure of their status in the book will lead to this fact becoming well known is more imagined than real" because:
  - The plaintiffs have little formal education and are illiterate in English. "The English literacy level of the community in which they live was not explored in evidence. What is known is that some of the people with whom they associate read magazines and newspapers."
  - Since the publication of the book, apart from the first plaintiff's boyfriend, the plaintiffs were not confronted by anyone else who had read the book.
  - "Biographies, especially those read by politicians, are directed at, and read by a limited number of people. This limited readership is unlikely to include people with whom the plaintiffs come into regular contact."
- All three plaintiffs "in any event led stressful lives that are linked to the economically strained circumstances in which they grew up and in which they appear to continue to live, their lack of education and economic opportunity, and their HIV positive status."
- The publishers are only liable for damages caused by the book subsequent to its publication and the publisher having become aware of the fact that the plaintiffs had not consented to their names and HIV status being disclosed.

While the AIDS Law Project welcomed the recognition that their clients' rights to privacy were violated and the order that their names be deleted from the book, they

> were unhappy with other fundamental aspects of this judgment both in terms of its findings and reasoning. It remains our opinion that Patricia De Lille and Charlene Smith should have been held liable precisely because they are acutely aware of the impact of stigma and discrimination on people living with HIV. The disclosure of our clients' full names and HIV status in De Lille's book did not add anything and no harm would have been done had their initials or pseudonyms been used instead. It was, for example, not disputed that neither De Lille nor Smith approached the plaintiffs to ascertain whether or not their names could be used in the book even though they both acknowledged that the plaintiffs had the right to determine the ambit of the disclosure of their names and HIV status. (Section27, 2007)

The AIDS Law Project also expressed its disappointment with the low award of damages made in this case which seems to completely disregard large portions of the plaintiffs' testimony about the impact of the publication of the book on their lives.

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This case once again highlights some discrepancies in the way ethical issues and the law are interpreted.

12.5 HOW DOES THE DUTY OF UPHOLDING CONFIDENTIALITY APPLY WHEN SHARING GENETIC INFORMATION WITH FAMILY MEMBERS?

Genetic information from family members can be important in the identification, treatment, and counselling of patients at risk of inherited disorders. Despite this, some patients are not happy to share relevant information with other family members making it difficult for medical professionals to treat other family members who may benefit from the information, fearing breaching patients' confidentiality. Parker and Lucassen (2018) discussed

> four ways in which genetic information can fail to be communicated to families; (1) where a patient explicitly refuses to share genetic information with a family member for whom it might be useful; (2) where a patient, for one reason or another, simply never gets around to telling their relatives about it; (3) where a patient is overwhelmed by their own diagnosis and does not feel able to cope with sharing just yet; or (4) where a patient is unaware of the fact that there are relatives who might benefit or has lost contact with relatives.

They contend that "a duty of confidentiality should not prevent the use of familial genetic information in the care and treatment of family members. Although a familial factor may first be identified in a single individual, this does not necessarily make it sensitive to, or identifying of, that person" (Parker & Lucassen, 2018). Finally, notwithstanding the importance of confidentiality in medicine, in situations where there is a risk of death or serious harm, they suggest sharing of relevant information.

While the default position for health care professionals to treat patient information as confidential and disclosure without consent as the exception, not the rule, Dheensa, Fenwick and Lucassen (2016) argue that this approach to confidentiality and autonomy is based on an "inaccurate conceptualisation of patients as separate from others, free from social and familial constraints". By contract, they suggest that "relational approaches to autonomy stress that patients develop their autonomy through social embeddedness and engagements with others; that one person's choices affect other people's autonomy; and that despite wanting to protect their personal interests, people are interested in maintaining family and community relationships" (Dheensa et al., 2016).

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Along the same lines is the joint account model of confidentiality, where the genetic information is conceptualised as familial and "belonging" to all the relatives it might affect, not just the individual in whom it was first identified. These authors interviewed 33 patients at large genetic centres in the United Kingdom. The interviews included questions related to hereditary conditions regarding consent, confidentiality, and information sharing in genetic medicine. They found that generally the participants had altruistic tendencies and thought of genetic information as familial, which aligns with the relational joint account model. Overall, participants supported sharing, but noted two reasons where health care professions might find it difficult to share if a patient had refused consent. First, the result had been generated from one person's blood, which might confer special rights for them over it and second, personal information (the condition and its effect on an individual) and familial information (the familial mutation) were entangled. However, they could not articulate any special rights a tested person may have over the result that would justify non-disclosure. Based on these inputs, the authors recommend that health professions inform participants about sharing information before seeking informed consent for their engagement.

Sharing of clinical and genetic data has many benefits like promoting research efficiency, expediting translational efforts of research results, ensuring traceability and transparency of published data, and maximising the utility of existing databases and lessening the burden to participants. Successful collaborations and sharing of data include genomic research for cancer (e.g., International Cancer Genome Consortium [Zhang et al., 2019]) and rare diseases (e.g., Global Alliance for Genomic Health [GA4GH, 2013] and the International Rare Diseases Research Consortium [IRDiRC, 2011]) (Takashima et al., 2018). In genomic research for cancer and rare diseases, patient family members (e.g., parents, children, siblings, aunts, or uncles) can provide important information and their genomic information is submitted to a repository or database and shared broadly. Privacy protection, security, and respecting participants' consent, however, remain ethical concerns. Takashima et al. (2018) argue that when sharing clinical and genomic data, there is a need to consider protection of the patients' own data and their family members' data. To overcome the risk of re-identification of individuals from data when sharing clinical and genomic data, the authors reported that their survey showed that the public and patients considered that familial data sharing in research should be protected more strictly. They conclude that all stakeholders – researchers, sample collectors, laboratory scientists, data analysts, database operators, institutional review boards and public and research

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participants, fulfil their own responsibilities to protect patients' and their families' privacy and interests while advancing genomic science and medicine.

12.6 CONFIDENTIAL GENETIC TESTING AND ELECTRONIC HEALTH RECORDS

Although paper records and human interpretation of laboratory results have sufficed in the past, the synthesis and interpretations of large amounts of complex genomic information requires electronic tools capable of delivering to patients and health care professionals the right amount and type of information at point of care (Hazin et al., 2013). Electronic health records have more recently been introduced to facilitate these processes. Many electronic medical record systems allow patients to access their results online without the presence of professional help. Electronic medical record systems allow sharing of health information and test results with other health professionals, some of whom may or may not know the patient, leading to inadvertent disclosure.

Black, Barton and Perlmutter (2021) commented on their review of 10 well-known centres that conducted presymptomatic testing for Huntington's disease and found that there were still many unresolved issues between the "virtues of a shared medical record and the goal of patient control of sensitive medical information". The authors propose that medical record systems adapt to and respect the patient's desires for confidentiality and allow people undergoing presymptomatic testing to restrict access to this sensitive information.

Huntington's disease, an autosomal dominant neurodegenerative disorder, is typically late-onset, most commonly between the ages of 30-50, but individuals of any age can present with it if the disease is diagnosed in the family. Patients present with severe progressive decline in motor control and cognitive faculties and exhibit behavioural disturbances. Due to the nearly 100% penetrance of this adult-onset condition, the decision for an individual to undergo testing, especially prior to the onset of symptoms, is difficult, as the results may affect their life and family and also produce serious psychosocial reactions and lead to future financial planning, educational, and employment complications (Eno et al., 2020)patient requests for increased confidentiality may be in conflict with the legal obligations of medical providers to document patient care activities in the electronic health record (EHR. Although some Acts (e.g., The Genetic Information Non-discrimination Act of 2008 [Code of Federal Regulations, 2012]; Genetic Information Nondiscrimination Act of 2009 [GINA, 2009] and the Affordable Care Act [ACA, 2010] in the USA) protect individuals from health insurance discrimination, other insurance products such as disability, life, and long-term care are not protected by these laws (Eno et al., 2020). Pre-

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test counselling and confidential testing to protect the well-being of those who choose to be tested are highly recommended.

Arias and Karlawish (2014) reached similar conclusions with preclinical studies of Alzheimer's disease. They concluded that "current legal and regulatory mechanisms are not sufficient to protect against harms that could have very real consequences for subject of preclinical Alzheimer's disease trials" (Arias & Karlawish, 2014). They suggested that research institutions must address shortcomings in the design of the electronic medical recording system, revise laws to limit discrimination, and disclose details of risks associated with the loss of confidentiality.

12.7 INVOLUNTARY DISCLOSURE OF CONFIDENTIAL GENETIC INFORMATION TO RELATED THIRD PARTIES

When an individual has been diagnosed with a hereditary genetic condition, their immediate family members may wish to know the probability of themselves or their lineage developing the condition. It has been argued that an individual has a moral obligation to share their diagnosis of a hereditary condition with relevant genetically close family members so that these individuals can choose whether to be tested themselves (Singh, 2023). This moral duty to disclose is not necessarily confined to the immediate family. Some have argued that adults who have been diagnosed with a hereditable condition, or parents of a child diagnosed with a hereditable condition, have an ethical duty to inform relatives in the extended family about the condition (Andrews, 1987). This moral duty arises from kinship bonds (Wertz, Fletcher, Berg & WHO Human Genetics Programme, 2003). The moral duty to disclose to relevant parties becomes challenging when the patient diagnosed with the condition (or in the case of a minor child diagnosed, his or her parents) refuse to voluntarily disclose the diagnosis to relevant family members. Section 16 of the HPCSA's Guidelines, *Seeking Patient's Informed Consent: The Ethical Considerations* – which governs consent to genetic screening and testing – states that informed decision-making involves a health care practitioner explaining "any significant medical, social or financial implications of screening or testing for the particular condition or predisposition" (HPCSA, 2016b). 'Social implications' could be interpreted as including the need to consider the implications of the test for related third parties (Singh, 2023).

In a report, *Review of Ethical Issues in Medical Genetics*, the World Health Organization noted the stance of the US President's Commission – a multi-disciplinary body of legal experts, ethicists, and clinicians – on the issue of genetic

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confidentiality (Wertz et al., 2003). The Commission held that the genetics professional may, unless it is prohibited by law, override individual confidentiality provided the following conditions are met:

- All efforts to persuade the individual to disclose the information voluntarily have failed.
- There is a high probability of harm to the relatives (including future children) if the information is not disclosed, and there is evidence that the information could be used to prevent harm.
- The harm averted would be serious.
- Only genetic information directly relevant to the relatives' own medical status would be revealed. Information relevant to the individual must remain confidential.

The duty to warn blood relatives came up in the UK case of *ABC vs St Georges Healthcare NHS Trust and Others* (YIP DBE, 2020). In this case, a patient, XX, was diagnosed with Huntington's disease while serving a prison sentence. An offspring of an individual who has Huntington's disease has a 50% chance of developing the condition. XX refused to disclose his diagnosis to either of his daughters, despite learning that one of his daughters (ABC) was pregnant. The health care professionals treating XX were also aware that ABC was pregnant. However, they took the position that they should not override XX's confidentiality. The health care providers thus did not notify ABC that she was at risk of inheriting the gene for Huntington's disease timeously enough for her to terminate her pregnancy. ABC had her baby, and a few months later, a member of XX's medical team accidently disclosed XX's diagnosis to ABC. Several years later, ABC was diagnosed with the genetic mutation for Huntington's disease. She subsequently developed a psychiatric condition due to her diagnosis and became concerned about her daughter's health, as she too had a 50% chance of inheriting the gene for Huntington's disease. ABC brought a negligence claim against the three Trusts responsible for the relevant health care professionals who treated XX, arguing that the Trusts breached their duty of care to her by failing to notify her timeously that she was at risk of inheriting the gene for Huntington's disease, to enable her to terminate her pregnancy.

In deciding the case, the court had to establish: (i) whether a duty of care existed between XX's clinicians and ABC; and (ii) if so, whether the clinicians breached this duty of care (i.e., acted negligently by omitting to inform ABC), and whether this omission caused ABC to suffer loss/harm (i.e., whether the element of causation was satisfied). In determining whether a duty of care existed between XX's clinicians and ABC, the court held that while ABC was not a patient of the

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three defendants, there was a close proximal relationship between ABC and the second defendant (D2) as ABC was involved in the family therapy arranged by D2's clinicians and they had material information about ABC and her particular circumstances. The court held that it was foreseeable, and D2 actually foresaw that ABC was at risk of suffering harm if they withheld information about her genetic risk from her. As such, the court held that D2 owed a duty of care to ABC to "balance her interest in being informed of her genetic risk against her father's interest in preserving confidentiality in relation to his diagnosis and the public interest in maintaining medical confidentiality generally". The court underscored that this duty of care was not "a broad duty of care owed to all relatives" of the patient in relation to genetic information. Instead, this duty of care was likely to arise only in prescribed circumstances where there was a close proximal relationship between the medical professionals and the at-risk person.

Having established that a duty of care existed between D2 and ABC, the court noted that ABC's negligence claim against D2 would succeed if: (i) D2 had breached their duty of care to ABC; and (ii) ABC would have terminated her pregnancy had she been timeously informed of the risk during her pregnancy. The court held that while a duty of care existed, D2's decision not to disclose XX's condition to ABC did not amount to a breach of the duty of care as the decision was "supported by a responsible body of medical opinion and was a matter of judgment open to [D2] after balancing the competing interests". The court also held that ABC had not established that she would have tested and undergone a termination of her pregnancy had the risk been timeously disclosed to her during her pregnancy. Given these factors, the court dismissed ABC's negligence claim against D2.

While ABC was unsuccessful in her claim as she had not satisfied the court that she would have undergone testing for the Huntington's gene and terminated her pregnancy based on a positive result, this case is significant in England – and arguably other settings which have legal systems based on English common law, such as South Africa – as it established that clinicians have a duty of care to at-risk relatives of their patients who have been diagnosed with hereditary diseases.

12.8 INCIDENTAL FINDINGS: THE RIGHT TO KNOW AND THE RIGHT NOT TO KNOW

Every so often, during biomedical research, researchers obtain information on participants that are outside of the scope of the research study. These findings have been referred to in the literature as "incidental findings" and has engaged some debate among researchers as to whether these findings should be disclosed to participants. For example, researchers collecting imaging data for a particular

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purpose may find an abnormality that poses a health risk to the participant, or genetic screening can reveal variants that are associated with higher relative risks of certain conditions. It is also possible to detect non-paternity from some studies.

Schaefer and Savulescu (2018) hold the view that reporting of incidental findings "should be based primarily on the possibility of medial benefit, factoring in the findings' validity, clinical actionability, and significance to health or reproduction" (Schaefer & Savulescu, 2018). They direct attention to participants' autonomy, privacy, and promoting the interests of participants. To assist with dealing with such scenarios, it is suggested that researchers should anticipate the possibility of incidental findings in advance of the study and should have a plan as to how they would deal with them. This plan should be conveyed to participants during the consent process so that participants are informed of such possibilities.

While various researchers have debated what incidental findings to disclose, there are three criteria – validity, significance to health (and sometimes reproduction), and clinical actionability – which have been described as the best-mediated-interest standard as outlined by Schaefer and Savulescu (2018). According to these authors, validity refers to the accuracy and reliability of the findings. In other words, a mere suspicion of a problem would not merit reporting, but a finding informed by a number of clinical studies might. Significance to health and reproduction refers to whether the finding could have a substantial impact on someone's health or (via reproduction) that of his/her offspring. Clinical actionability refers to the potential for a clinical intervention to alleviate the health issue, e.g., treatment for breast cancer.

The best-mediated-interest standard has been viewed as problematic since it is overly narrow in its clinical focus. It is argued that participants may have an interest in receiving incidental findings that are not related to health or are health-related but not actionable (Schaefer & Savulescu, 2018). They qualify their view by stating that "misattributed paternity would be of crucial interest to a purported father paying child support. And some people will have a nonmedical interest in learning about a genetic predisposition to currently incurable conditions like Alzheimer's on grounds that the knowledge can let them better plan their lives".

Several factors have contributed towards the argument for non-disclosure of incidental findings. Some commentators have suggested that such information can sometimes by psychologically harmful – "it may induce anxiety, fear, survivor guilt, depression, or other negative mental states" (Schaefer & Savulescu, 2018). Even if there is a right to know, it may not be absolute, and the negative effects of some findings, especially those which are not actionable, may override that right.

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It is suggested that the decision about disclosure should involve weighing the positive reason(s) to provide information, respecting the right to know, against uncertain negative reason(s) against it (psychological harm). Also, disclosure should be limited to circumstances, when after appropriate explanations, participants would understand the implications of such information. It is recommended that researchers respect people's rights to know about themselves (Schaefer & Savulescu, 2018).

12.9 HOW TO DEAL WITH GENETIC TESTING WHEN IT REVEALS NON-PATERNITY?

An ethical issue that may cause medical providers extreme anguish is what to do when genetic testing reveals non-paternity as an incidental finding. The rate of this misattributed parentage is estimated to be between 1% to 10% of all births (Prero et al., 2019). When this outcome is discerned and disclosed, the result can be devastating to the entire family. The child, other children, and both parents may be severely and permanently harmed. This disclosure may also jeopardise mothers' safety, result in child support being withdrawn, result in family members' losing inheritance, and cause bitter custody disputes (Howe, 2021).

The value conflicts this poses for providers are exceptionally difficult. Providers' views on what they should do when this question arises have differed. Some have favoured, for example, never telling parents this outcome. Others spell out what they will do in advance. Still others tailor their responses to families' different needs (Howe, 2021).

Mothers may also find out that they are not biological parents. This may come about when children are born because of in vitro fertilization (IVF). Mistakes in IVF procedures may be made. With IVF, it may then also benefit parents if providers anticipate these incidental findings and discuss this with parents in advance. Providers informing these parents that a mistake could occur with IVF may, on the other hand, scare them. Thus, due to the most remote chance that this could occur, it may be better here to not inform them (Howe, 2021).

Howe (2021) discusses other concerns parents may have about the effects of genetic information on their relationship with their children in other contexts. An example occurred "when a transgender parent did not want his child to know that after he had given birth to his child, he changed his gender. He feared that his child might find this out after viewing his father's medical records. Here, even if this wouldn't occur, he still may have had this underlying concern".

Providers' options may be limited in what they can do to relieve parents' concerns in some instances. Then, they refer these parents to counsellors in the hope that

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this might help them reduce their fear. These parents too might decide, because of this counselling, to share information about themselves with their children that they wanted to keep secret, after all.

Parents' culture may also affect what providers can and should disclose. In some cultures, for example, a woman convicted of consented infidelity may face most severe adverse consequences. Providers should keep in mind, though, that even when parents share the same culture, as when they are next door neighbours, their views may wholly differ as much as they would if they were from different cultures. Providers must initially then explore both parents' views fully and separately so that they do not inadvertently favour one parent's views over the other, by not exploring each to the degree that they could and should. If two parents have irresolvable differences, mediators or, again, counsellors may help (Howe, 2021).

Perhaps the most common approach providers take is, when possible, medically, to inform, first and only, mothers of this risk of discovering non-paternity (Howe, 2021). This can occur if these mothers initially come in alone. Providers then can leave it up to these mothers to decide whether anyone will be tested and if so, who this will be. If the mother suspects that her child's father may not be the biological father, she may choose to not ask this father to be tested. This respects her autonomy and may be most beneficial to all. Providers may be willing to do this, however, only if there are no risks of adverse medical consequences to anyone. This approach also allows the provider to sustain a more positive relationship with the mother since it allows her to decide. The mother, too, then, may not know or even ever know whether her husband – or another former partner – is their child's biological father. This may be what she would want. The provider might, then, take initiative to explore this with her before she decides. Providers may not want to offer mothers this decision-making option on the ground that the mother chose to engage in what they see as an indiscretion. They may believe that she should bear this decision's consequences. This view is for several reasons ethically problematic. Chief among these problems is that the consequences may be profoundly destructive to the child and family. This harm may far outweigh all other considerations. The provider, also, has no ethical basis for taking on this judging role (Howe, 2021).

Another approach discussed by Howe (2021), quite the opposite of the one above, may occur after providers discover non-paternity incidentally. These providers may then tell these mothers that they must tell these fathers, as, for instance, within a week. Further, they may inform the mothers that if they have not told the father by then, these providers will.

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Providers may do this for several reasons. They may think that those in the medical profession should not keep secrets. They may think that fathers have an absolute right to know. They may also use these two above reasons to "justify their imposing personal view that the mother should be punished for her past indiscretion. They may do this without their knowing that they are using these reasons to rationalize their imposition of their morally unjustifiable personal bias" (Howe, 2021).

A third approach is for providers to inform both parents initially that any information they find incidentally they will not disclose unless it is medically relevant. This practice of total non-disclosure has been used to protect family members' relationships by keeping them intact in other contexts. Transplant centres, for example, may inform family members before a transplant is carried out that if for some reason the transplant is cancelled, the staff will not tell the family why. A reason for this policy is that parents and other donors may refuse to donate their kidney at the last minute. There may also be other reasons for cancelling the transplant than these parents' own interest, but the staff's policy not to inform the family will prevent the family from a possibly devastatingly effect. This transplant policy is controversial. It illustrates, however, another instance in which medical staff may agree to keep a family secret to themselves (Howe, 2021).

A fourth approach sometimes carried out is for providers not to forewarn either parent that genetic testing can produce this incidental finding. Some parents may not know this unless their provider tells them. They may not be aware that genetic testing conducted for an entirely different medical reason could also reveal non-paternity. If, though, the provider tells the parents this, such that they learn this for the first time, the alleged fathers – who almost always will be the biological fathers – may, in response, want to have a paternity test. They may seek this out even at another institution. Then, the same harmful outcomes for their family may come about (Howe, 2021).

Therapeutic privilege (also known as therapeutic exception) is an exception to the general rule of informed consent and applies in the uncommon instance where a health professional may be excused from revealing information to a patient if disclosing it could pose a serious psychological threat to the patient (Singh, 2023). The notion of therapeutic privilege allows health professionals to withhold 'psychologically sensitive information' in situations where the nature of the information to be conveyed to the patient (for example, XY genotype in a female patient) could cause 'grave psychological harm' to that patient or their family. The

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therapeutic privilege presumes full disclosure, but is delayed until the individual is psychologically and cognitively competent to process the information (Wertz et al., 2003).

Some experts advise obtaining a second opinion about the likelihood of psychological harm before opting to defer or delay disclosure (Wertz et al., 2003). However, in the absence of timely access to mental health professionals, and when assessment shows that emotional harm is possible, nondisclosure or delayed disclosure of the full scientific facts may be justified (Wertz et al., 2003). The notion of therapeutic privilege is recognised in South African law. Section 6(1) of the National Health Act (2003) states: "Every health care provider must inform a user (patient) of (a) the user's health status *except in circumstances where there is substantial evidence that the disclosure of the user's health status would be contrary to the best interests of the user*." The decision to invoke therapeutic privilege and determining what is in the best interests of the patient is left to the discretion of the health profession.

As a medical scientist doing population genetic research and offering genetic ancestry tests to the public and sharing individual results with subjects, one of the authors (Soodyall) has had to withhold Y-chromosome DNA results when returning reports. In one study, when examining the transmission of Y-chromosome DNA in conjunction with genealogical information in an isolated clan community, there were a few instances when the inferred Y-chromosome lineage linked with a particular founding father and his descendants did not match and could not be explained by new mutations but could be attributed to non-paternity. With input from the Chief in the district, some cases were attributed to a scenario in which a woman conceived out of wedlock and the child was then registered with her fathers' surname to protect the family, resulting in a different Y-chromosome lineage being introduced in the family not concordant with the family surname. Due to the sensitivity of these situations, Soodyall would leave out the Y-chromosome result in the report and only discuss the mitochondrial DNA results with the individual.

Another scenario presented when she had done an ancestry test from a profiled person and presented his results to him. Years later, another male with the same surname came for a genetic ancestry test, but they did not detect a match to this individual in their database. Soodyall was subsequently informed that these individuals' father was the profiled person that they had previously tested, and when the two results were compared, the 'son' had a very different profile to the father. Soodyall did not disclose this to the son and did not mention to him that she had compared the two results. However, in her explanation of the results

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to him, she informed him that sometimes when these tests are done, some changes are known to occur in a stepwise fashion with STR markers during spermatogenesis. Whether the son discussed the result with his father, and they compared results, was never disclosed to Soodyall and neither came back to her for further explanations. In both examples above, non-disclosure of the test results was in the best interest of the subjects.

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