



# Human Genetics and Genomics in South Africa: **Ethical, Legal and Social Implications** Concise Report



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The Academy of Science of South Africa (ASSAf) was inaugurated in May 1996. It was formed in response to the need for an Academy of Science consonant with the dawn of democracy in South Africa: activist in its mission of using science and scholarship for the benefit of society, with a mandate encompassing all scholarly disciplines that use an open-minded and evidence-based approach to build knowledge. ASSAf thus adopted in its name the term 'science' in the singular as reflecting a common way of enquiring rather than an aggregation of different disciplines. Its Members are elected on the basis of a combination of two principal criteria, academic excellence and significant contributions to society.

The Parliament of South Africa passed the Academy of Science of South Africa Act (*No 67 of 2001*), which came into force on 15 May 2002. This made ASSAf the only academy of science in South Africa officially recognised by government and representing the country in the international community of science academies and elsewhere.

# Table of Contents

Foreword	1
Acknowledgements	3
Preamble	5
Background	6
Human Genetics and Genomics	6
Study Rationale	7
Methodology	8
Summary	8
Conclusions	14
Appendix A	17



# Foreword

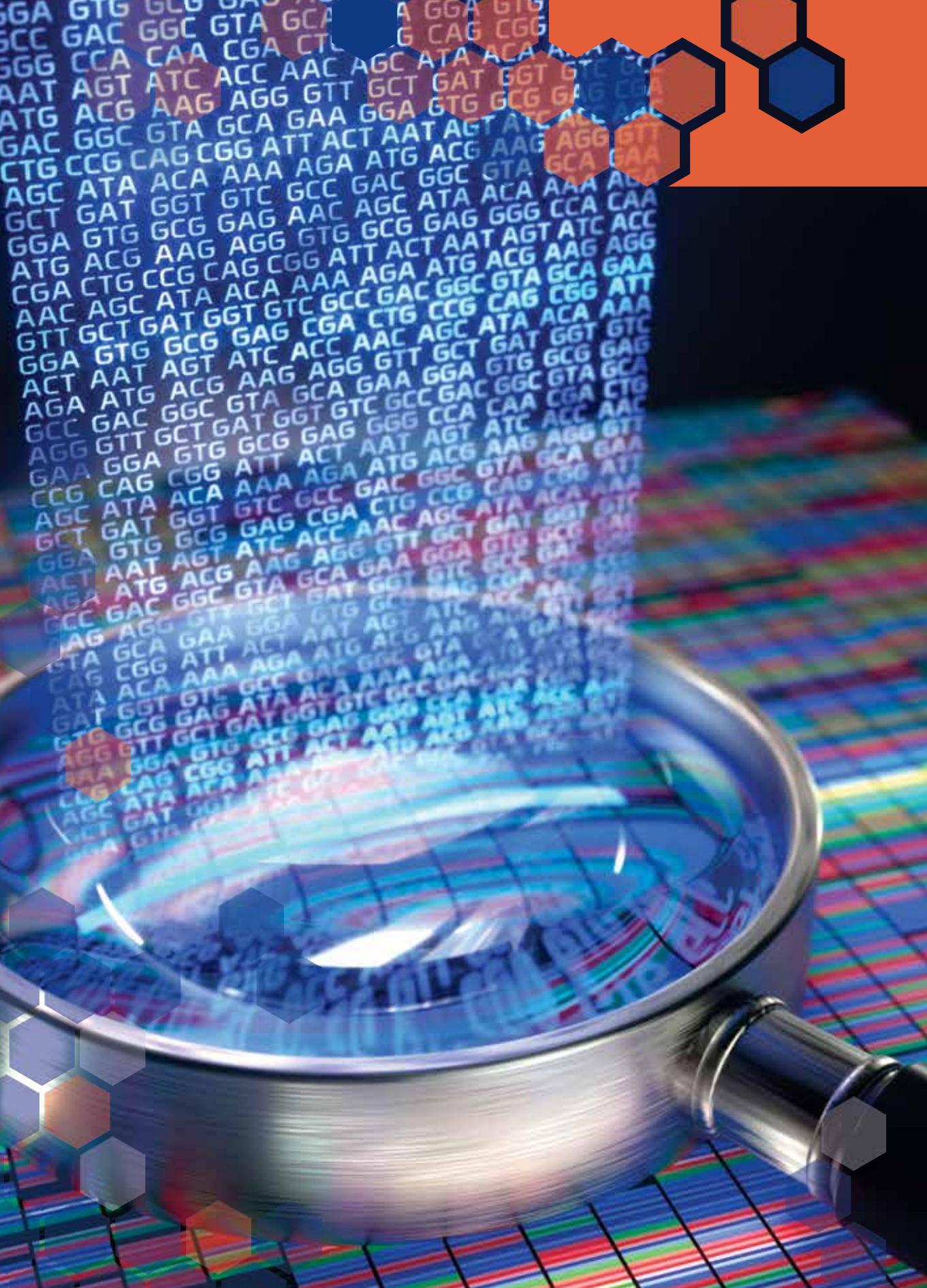
The Academy of Science of South Africa (ASSAf) is mandated to provide evidence-based science advice to government on matters of critical national importance. This consensus report is in fulfilment of this mandate. Great benefit is to be derived from the work done in the fields of genetics and genomics, which has the potential to impact positively on the health and quality of life of all members of society. Work in these fields is also likely to impact positively on the economy through job creation and formation of new businesses including small to medium enterprises. The report provides valuable insights into the ethical, legal and social implications (ELSI) of genetics and genome work in South Africa and sets out a number of recommendations to address the challenges. This document may also serve as a model for international stakeholders.

The ELSI of genetics and genomics impact on all. One of the recommendations is for the development of a code of conduct and best practice for professionals working in the field of genetics and genomics in South Africa to ensure that the work is conducted with integrity, honesty, collegiality, accountability and sharing. The findings and recommendations of this study are thus likely to be of interest to a wide-ranging audience, over and above policymakers and researchers.

The 13-member consensus study panel, under the leadership of Prof Michael Pepper, is to be commended on their diligence and on both the volume and quality of evidence that they have amassed to inform the recommendations they have made. This report is a product of their voluntary commitment and I thank them for their dedication to the task and look forward to the debates that will ensue following the release of the report.

I thank all those who were involved in the preparation and production of this report, particularly the Academy staff that supported the panel in their work. The ASSAf Council would like to extend its sincere appreciation to the panel for the service that they have rendered to the Academy. Funding from the Department of Science and Technology is also hereby acknowledged.

**Professor Jonathan Jansen**  
**President: Academy of Science of South Africa**



# Acknowledgements

This consensus study report is the result of the collaborative work of many people who are acknowledged as follows:

Panel members: Prof Michael Pepper, University of Pretoria (UP) (panel chairperson); Prof Collet Dandara, University of Cape Town (UCT); Prof Jantina de Vries, UCT; Prof Ames Dhai, University of the Witwatersrand (Wits); Prof Melodie Labuschaigne (formerly Slabbert), University of South Africa (Unisa); Dr Freddy Mnyongani, University of KwaZulu-Natal (UKZN); Prof Keymanthri Moodley, Stellenbosch University (SU); Dr Antonel Olckers, DNAbiotec®; Prof Anne Pope, UCT; Prof Raj Ramesar, UCT; Prof Michèle Ramsay, Wits; Prof Himla Soodyall, Wits; Prof Wayne Towers, North-West University (NWU). These members contributed their time and expertise to this study on a voluntary basis. They are sincerely thanked for their highly valuable input and commitment to ensuring that the best interests of the country were always of paramount importance in all of their deliberations.

A special thank you goes to the group who wrote the report comprising Prof Anne Pope, Dr Antonel Olckers, Prof Jantina de Vries and Prof Michael Pepper, and also to Prof Ames Dhai who oversaw the Relationship Building Group, Dr Freddy Mnyongani who oversaw the Respect for Persons Group and Prof Anne Pope who oversaw the Good Stewardship Group.

As part of the information gathering process, six consultative workshops were held with the following groups: The pharmacogenomics community, genetics service providers (two meetings held at Wits and UCT), the forensics community and the rare diseases community and advocacy groups. All participants and contributors are gratefully acknowledged. In addition, a presentation and discussion took place at the 2017 Joint Meeting of Research Ethics Chairs and the National Health Research Ethics Council in Pretoria.

The report was peer-reviewed by: Emeritus Professor Daniel Ncayiyana, UCT; Professor Ma'n H Zawati, McGill University; The National Intellectual Property Management Office; Mrs Glaudina Loots and Ms Modiegi Selematsela, Department of Science and Technology (DST). Their valuable suggestions are gratefully acknowledged.

Support and contributions from Academy staff, Dr Khutso Phalane-Legoale, Prof Roseanne Diab and Mr Ian Shendelana, are highly appreciated. Ms Patsy Scholtz is thanked for the editing and production of the study report.

The DST is sincerely thanked for providing financial support.

**Prof Michael Pepper**  
**Panel Chairperson**





# Preamble

We live in an era of rapid growth in the fields of genetics and genomics. Great benefit is to be derived from this work, which has the potential to impact positively on the health and quality of life of all members of society. Work in these fields is also likely to impact positively on the economy through job creation and the formation of new businesses including small to medium enterprises. For these benefits to materialise, however, skills development and clear guidance on how to implement these new and upcoming technologies are required. In addition, boundaries must be defined clearly, and adherence monitored to ensure that benefits are shared by all and that no harm is done.

Our objective is to produce an authoritative report that responds to these needs. While the fields are complex, our intent is to promote interest and encourage development of sustainable and ethical initiatives in the practice of genetics and genomics. Practice at this level includes clinical, research, entrepreneurial and forensic activities. Furthermore, genomic research is closely linked to biobanking and requires storage of samples and associated data sets, and therefore such activities are also included.

Focusing on South Africa and Africa in general, the broad philosophical approach for this genetics and genomics consensus study centres on Ubuntu. This theme guided deliberations and is central to the message conveyed herein. Within an African context, the self only makes sense in relation to the community. Incorporating a wide range of values, Ubuntu expresses the inextricable intertwining of the individual and his or her community. Key principles of human dignity and respect, as well as equity and distributive justice have also guided our deliberations. We were mindful of the need to ensure that the notions indicated in the title of this report, namely "Ethical, Legal and Social" implications, emerge as guiding principles, particularly regarding the more pragmatic areas such as informed consent, privacy, confidentiality, management of samples and data, intellectual property and commercialisation. The tendency to want strict regulation, due to past as well as recent adverse experiences, is understandable. However, we are conscious of the negative consequences that would flow from over-regulating the field, including stifling innovation and entrepreneurship.

The report is divided into three main sections, namely Building Relationships, Respect for Persons and Good Stewardship, each of which contains a set of recommendations. The background to the study, rationale for the study and the methodology used are discussed in the next section.

## Background

From the moment of conception, deoxyribonucleic acid (DNA) dictates the nature of the structural and functional components that, once fully integrated, define who and what we are. Also contained within DNA is information that may indicate our predisposition to certain diseases, our potential to benefit from certain therapeutic drugs, and our ability to integrate into and function within the environment in which we live. While this information can be used to maintain health and manage disease for an individual or groups of people, it can also be misused and result in consequences that are detrimental.

## Human Genetics and Genomics

Human genetics and genomics are exciting and rapidly evolving fields, both in terms of the new knowledge generated and the rate at which such information is generated. Heritable traits, determined at conception, are transmitted through DNA from parent to child. "Genomics" refers to the totality of the information contained in the DNA of an individual as inherited from his or her parents, whereas "genetics" defines the field that applies this information in order to understand how characteristics of living organisms are transmitted from one generation to the next through DNA (i.e. the study of heredity). DNA information is useful in heredity studies in general, and specifically in the clinical setting in the management of health and of individual identification as applied in forensics and kinship establishment. Human genetics includes the study of heritable factors in individuals, families and populations.

Since the DNA of an individual is inherited from both biological parents, who in turn, inherited it from their biological parents, it is important to note that DNA is shared among biological family members. Also noteworthy is the fact that although an individual shares DNA information with family members, each individual nevertheless possesses a unique overall genome (except in the case of identical twins whose genomes are identical). That DNA is shared by biological family members raises the issue of the impact of discovery of any DNA sequences that may affect their health on the members who have not sought testing. With respect to confidentiality, this becomes problematic if some family members disclose information on their DNA that others do not wish to have disclosed. Furthermore, the information in DNA is often stored in databases, which can lead to some disclosure. Since an individual's genome is unique, if their data are in an existing database, a comparison to that database would identify a genetic match with high probability. Thus, access to such databases requires regulatory oversight.

Due to these unique characteristics of genetic and genomic work, there are unique ethical, legal and social implications (ELSI) that relate to this type of work. These ELSI could affect rights (to the extent that they exist), political capital, regulation and sharing. Conclusions inferred from research or testing results could be stigmatising or viewed as favourable or unfavourable at a

socio-cultural level. A complication is that social preference in communities and nations is dynamic and changes over time, which necessarily requires regulatory mechanisms to be responsive to changing socio-cultural norms.

Finally, it is important to recognise that asymmetrical power relationships in science and medicine, together with historically unfair exploitation and data mining in Africa, especially where genetics and genomics are concerned, are important precursors to the ethical, social and legal dilemmas that exist in this field today.

## Study Rationale

Currently, legislation in South Africa that deals with genetics and genomics is very limited. The National Department of Health (DoH) oversees implementation of the National Health Act (NHA) (*No 61 of 2003*) and its Regulations. Chapter 3 of the NHA mandates the Director-General to make provision for genetic services: "The Director-General must issue and promote adherence to, norms and standards on health matters, including – genetic services" (s 21(b)(vii)). Chapter 8 of the NHA deals with blood and blood products, assisted reproductive technology, cell-based therapy, transplantation, tissue banks and forensic medicine/pathology. Chapter 8 includes a section on reproductive and therapeutic cloning (s 57(1)(a) and (6)(a) & (b)) which states that manipulation of human genetic material from gametes, zygotes and embryos for purposes of reproductive cloning, is prohibited. The Criminal Law (Forensic Procedures) Amendment Act (*No 37 of 2013*) and its subsequent Forensic DNA Regulations of 2015 (*Government Gazette 38561 of 13 March 2015, Government Notice R 207*) only address the collection, use, storage, and destruction of DNA samples in forensics. The Medicines Control Council guidelines (August 2012) refer to genetically modified material including recombinant DNA technology in the section on biological medicines. Apart from the regulatory measures mentioned above, no specific legislation on genetics and genomics exists in South Africa. Thus, critical and highly topical fields of practice and research such as gene editing and gene therapy remain unregulated.

This document addresses the ELSI of genetic and genomic work, as it relates to research, health service provision and forensic applications (medical and legal). Many of the implications span all these areas, but some are specific to certain applications and will be indicated as such. For example, in a research setting, limited feedback of genetic results is provided to the participant, in line with the research design described in the informed consent documentation. In a clinical setting, however, a clinician must evaluate, often with the assistance of a medical scientist, what is of clinical relevance and utility and thus determine what is to be communicated to the patient or client.

The objective of this consensus study is to inform the drafting of policy documents, regulations and guidelines under the auspices of the DoH, the Department of Science and Technology (DST) and other relevant departments.

## Methodology

This consensus study began with a proposal submitted to the ASSAf Council. Once the proposal had been approved, a team was appointed that defined the objectives of the study and met regularly over a two-year period to collect and deliberate on the materials collected from diverse sources. Several stakeholder workshops were held and included scientists, health care personnel, legislators, bio-entrepreneurs, research ethics committee representatives, as well as special interest and advocacy groups.

## Summary

The deliberations in this report are centred on the broad philosophical approach of Ubuntu, a philosophical notion that refers to the essence or quality of being human. The report describes the benefits to be derived from genetic and genomics work, the need for boundaries to be clearly defined and adherence monitored to ensure that benefits are shared by all and that no harm is done. Each of the three major sections (Building Relationships, Respect for Persons and Good Stewardship) is followed by recommendations (**Recommendations R1 to R19**) which are ethically and legally sound, culturally appropriate, feasible, enforceable and sustainable, given the resources within the country, and balanced against competing national priorities.

This study aims to address the ELSI of genetics and genomics work, as it relates to research, health service provision and forensic applications (medical and legal) in South Africa. The study was undertaken by a 13-member panel appointed by ASSAf.

The section on Building Relationships focuses on the engagement between human genetics and genomics practitioners and the general public. This relationship ranges from academic research projects, to genetic testing in the private sector, but also includes the relationship between the public, the law and the forensic science sector of the country. It highlights South African experiences with community engagement for genomics and the importance of education and the translation of science. It further stresses the critical role that persons with vested interests in genetics and genomics have to ensure that the public is well informed on research projects, their roles, rights and responsibilities. Four recommendations (**R1 – R4**) are made for building relationships and community engagement:

### R1. Stakeholder engagement

- a) Promote understanding that a community (engaged in a specific research study) and the public at large are complementary stakeholders and that the development of engagement strategies needs to be considered separately for the two groups.

- b) In genetic and genomic research, reciprocal researcher and community relationships should be promoted through community engagement activities such as the use of meetings with gatekeepers, establishment of community advisory boards (CABs) and the implementation of the principle of participatory action research.
- c) The success of stakeholder engagements should be objectively evaluated on an ongoing basis by researchers and communities, or the public.

## R2. Education and training

- a) Implement effective measures to improve the public's knowledge and understanding of genetics, genomics and associated new technologies in a culturally sensitive and appropriate manner.
- b) Mere adherence to process is not sufficient; substantive engagement is necessary between researchers on the one hand, and their funders, the regulators, their ethics committee and research communities on the other.
- c) Liaise with the Department of Basic Education (DBE) and the Department of Higher Education and Training (DHET) on how best to integrate information about new health-related technologies in school curricula.
- d) Promote appropriate genetics and genomics training for health care professionals.
- e) Make a substantive investment in training of genetic counsellors and clinical geneticists and other relevant professionals to increase the national capacity to deliver genetics and genomics services (See also **R14**).
- f) Educate the public with regard to forensic DNA testing.
- g) Promote the integration of forensic DNA testing into the curricula of law degrees.

## R3. Protecting the public

Direct to consumer genetic marketing and testing must be regulated.

## R4. Accountability and transparency

- a) Promote an appreciation and understanding of the importance of research for improving health care services for all while protecting public trust in the scientific fields of genetics and genomics.
- b) Establish a clear and strong legal and ethical framework that includes sanctions for misconduct in all genetics and genomics work, including commercial activities.

- c) Ensure accountability and transparency in the practice of forensic science in all sectors (academic, public and private).

The topic of Respect for Persons is addressed in accordance with the South African Constitution, which recognises and protects both autonomy and self-determination in the Bill of Rights: the right to dignity (s 10), to life (s 11), to bodily and psychological integrity (s 12), which includes security and control over one's body, and, for women, control over reproductive decisions. It further highlights that legally and ethically, people are entitled to make free informed choices about their health care and research participation. The communitarian philosophical outlook and how it deepens respect for persons in Africa were taken into account. In this section the panel recommends the following **(R5 – R8)**:

### R5. Ubuntu philosophy

- a) The Ubuntu principle must be promoted in genetics and genomics research, health care delivery and forensics practice.
- b) Recognition must be given to the fact that while the concepts of autonomy and Ubuntu may be in tension, these are complementary rather than mutually exclusive principles and that all fundamental rights should be understood within the matrix of the community. Relative solidarity is an important component of Ubuntu.

### R6. Consent models for genetics and genomics work

- a) Empirical research should be conducted to establish South African participant views on consent models.
- b) It must be recognised that blanket consent is incompatible with South African legislation (e.g. The Protection of Personal Information (POPI) Act (No 4 of 2013)).
- c) The National Health Research Ethics Council (NHREC) should be encouraged to prepare an informed consent template for genetics and genomics. The informed consent template should include the following considerations: whether results will be returned; benefit sharing arrangements; sample and data storage and re-use, including governance thereof; limits to the withdrawal of samples and data once shared; details regarding export of samples; privacy protection in countries to which data and samples are exported; and the specific circumstances that limit confidentiality related to DNA data.
- d) The DoH Guidelines on Ethics in Health Research (2015) that permit broad, tiered and specific consent models should be fully implemented. The panel recognises however that there is lack of consensus regarding the impact of the Act (No 4 of 2013) on broad consent, and that the situation may change once clarity is obtained from the Regulator.

## R7. Protection of information and resources

- a) Oversight provided by Research Ethics Committees (RECs) on future use of genetic material (samples and data) must ensure that proposals indicate whether storage is desired and if so, informed consent documents must include the relevant information to permit a voluntary informed choice by participants.
- b) Researchers should not report their research findings in ways that may be, or may be perceived to be, harmful or offensive.
- c) Engagement with the Information Regulator, Department of Justice, is important to discuss the development of regulations in the POPI Act (*No 4 of 2013*) and how this will impact on genetics and genomics research.
- d) A policy should be put in place to guide decisions about disclosure of incidental findings.
- e) The challenges related to the timeframe of 30 days to remove a DNA profile from the National Forensic DNA Database (NFDD) should be revisited.
- f) The establishment of a South African Human Genetics Advisory Board (SAHGAB) should trigger discussions with civil society with regard to the implications of forensic practices related to genetics and genomics, including the NFDD.

## R8. Communities, families and vulnerable and marginalised individuals

- a) When working with small, identifiable groups that may already be socially or politically marginalised, researchers must include in the community engagement process a discussion on the manner in which the research process and outcomes will be managed to mitigate potential harm to the community, e.g. unintended perceptions of stigma.
- b) Researchers investigating certain conditions, phenotypes or behaviours must also include in the community engagement process a discussion on the manner in which the research process and outcomes will be managed to mitigate potential harm to the community.

In the Good Stewardship section the need for responsibility is highlighted in terms of sustainable and careful use of genomic resources (reflected as both a value and a practice) by individuals, communities, organisations, companies and governmental institutions. This section is intended to emphasise the inherent characteristics of integrity, honesty, collegiality, accountability and sharing that make up the notion of stewardship. It further stresses the need for professionals in the field of genetics and genomics to

take up the role of stewards in the interest of the people of South Africa, without fear or favour and to do so objectively. The following recommendations (**R9 – R13**) are made with regard to good stewardship:

## **R9. Code of conduct**

A code of conduct and best practice for professionals working in the field of genetics and genomics in South Africa should be developed by government and other appropriate entities to promote good stewardship of resources including data and biological specimens.

## **R10. Policy and guidelines appropriate for the South African context**

The following should be developed by government and other appropriate entities:

- a) Guidelines for oversight of responsible clinical genetic/genomic testing, including for appropriate accreditation of laboratories offering genetic/genomic testing and for monitoring of staff qualifications and expertise.
- b) An appropriate national policy that outlines considerations, obligations, mechanisms and circumstances for feedback of individual results.
- c) Policies and guidelines to promote good stewardship of resources in clinical and research settings to promote innovation and translation of research into clinical practice.
- d) A national framework for South African biobanks that includes integrated data storage systems that have the potential to enhance health care and justice (i.e. in forensic and legal contexts), and to maximise their value to society.
- e) A national framework for sample and data access to promote equitable and responsible sharing of genetic and genomic resources to enhance knowledge generation and translational science, drawing on existing international and continental policies.

## **R11. South African Human Genetics Advisory Board**

A South African Human Genetics Advisory Board should be established. The board should have appropriate expertise to provide guidance to policymakers and regulatory structures (See also **R16**).

## **R12. Open debate with stakeholders and policymakers**

Debate, explore and adapt the 'sociologically informed model' for the principles of (a) custodianship/ownership of samples and (b) benefit sharing in South Africa. Include relevant stakeholders like the National Intellectual Property



Management Office (NIPMO) and the South African Law Reform Commission, since the topics affect a cascade of implications: ethical values of equity and distributive justice; good governance principles of benefit sharing; whether intellectual property can exist if genomic resources are to be regarded as a 'common good'.

### R13. Legal framework

- a) Laws and regulations relating to genetics and genomics must be aligned and contradictions must be carefully and comprehensively addressed.
- b) The South African Health Products Regulatory Authority (SAHPRA) should regulate genetic tests under the Medical Devices Act (*No 14 of 2015*).
- c) The Criminal Law (Forensic Procedures) Amendment Act (*No 37 of 2013*) and its Forensic DNA Regulations (2015) must be updated.
- d) The potential value of a mutually beneficial memorandum of understanding between the South African Council for Natural Scientific Professions (SACNASP) and the Health Professions Council of South Africa (HPCSA) must be explored for forensic practitioners using DNA testing (See also **R15** and **R18**).

A set of overarching recommendations (**R14 – R19**) is listed below:

### R14. Capacity development in genetics and genomics in South Africa

South Africa is currently in short supply of appropriately trained and skilled personnel at all levels of genetics and genomics work. To establish, build and maintain a service platform and large scale, sustainable genomics programmes for the benefit of a healthy nation, bearing in mind ethical, legal and social responsibility, will require technical, scientific, computational, bioinformatics and statistical analysis, as well as financial, legal and ethical expertise. More resources are therefore required to support genetic and genomic work, including training of genetics nurses, genetics counsellors, medical geneticists, medical scientists, bioinformaticists, biostatisticians and forensic scientists for the public and private sectors in South Africa (See also **R2 (e)**).

### R15. Legal framework with policies and guidelines for genetics and genomics in South Africa

Legislation and policies should be developed in an inclusive and cross-cutting framework, taking into account national, regional and international contexts, and should avoid stifling innovation (See also **R13** and **R18**).

## R16. South African Human Genetics Advisory Board

The South African Human Genetics Advisory Board (SAHGB) should be adequately resourced and independent, with the aim of providing oversight in genetics and genomics at the national level and working in concert with ethics and legal regulatory structures (See also **R11**).

## R17. Ethical oversight

Ethical implications that are deemed problematic by Research Ethics Committees, researchers, patients/participants or the public should be brought to the attention of the NHREC whose direct involvement in policy-drafting should be sought.

## R18. Legal oversight

Legal implications should be brought to the attention of the South African Law Reform Commission whose direct involvement in policy-drafting should be sought (See also **R13** and **R15**).

## R19. Framework for non-compliance

Sanctions for non-compliance with current and future legislation must be defined, be implementable and be effective.

## Conclusions

The face of health care delivery, biomedical research and forensics in South Africa is rapidly changing as a result of advances in the fields of genetics and genomics. Commensurate with this rapid evolution is the need to consider the ethical, legal and social implications of these technological advances. Implicit in this consideration is an understanding of African philosophy, and in particular the notion of Ubuntu.

This report has addressed what we consider to be key imperatives in genetics and genomics ELSI, and we have developed a set of recommendations which we believe could inform the drafting of one or more policy documents that will guide the drafting of legislation, regulations and guidelines/standards to regulate genetics/genomics and associated areas (e.g. biobanking) in South Africa. The framework must demonstrate that governance processes and procedures are infused with integrity, honesty, responsibility, accountability and efficiency. It must be informed by legislation and regulations that govern human biological materials and are compliant with international standards. The framework should be reasonable, feasible, pragmatic and non-stifling for the South African context.

The important conceptual issue of whether biological samples, including DNA, may be recognised as 'property' under South African law must be rigorously debated and clarified, preferably by the South African Law Reform Commission. This issue is different from the current pattern of permitting intellectual property rights in terms of current law. The convention that 'donors' provide their samples voluntarily without reward (the gift model) is rejected increasingly by those who do not accept that researchers and other downstream entities may acquire enhanced credentials and wealth as a result of working with the samples and their associated data. The implication is that benefit sharing is a logical part of the genomic project. The 'sociologically informed model' for benefit sharing includes the idea that some commercialisation of genetic and genomic research is acceptable.

Finally, the involvement of the 'public' and 'communities' should be integral to the policy-drafting process. Deficiencies in our understanding of precisely what constitutes communities and how they should be implicated are recognised. This matter should be addressed as a matter of urgency to ensure that participation is optimal.



# Appendix A: Composition of Study Panel

## A.1 Chairperson of the Study Panel

### Michael Pepper

Prof Michael Pepper (MBChB (Cape Town), PhD (Geneva), MD (Geneva), Privat Docent (Geneva)) is Director of the Institute for Cellular and Molecular Medicine, Director of the South African Medical Research Council (MRC) Extramural Unit for Stem Cell Research and Therapy and a research Professor in the Department of Immunology in the Faculty of Health Sciences at UP. He is also Professeur Associé in the Department of Genetic Medicine and Development in the Faculty of Medicine at the University of Geneva, Switzerland. He obtained his MBChB in 1982 from the Faculty of Medicine at UCT, and moved to Geneva in 1986, where he obtained his PhD in 1990 and MD in 1992. In 1997, he obtained his Habilitation and had the title Privat Docent conferred on him. He returned to South Africa in July 2004. Prof Pepper has worked extensively in the field of clinically-oriented (translational) molecular cell biology, and his interests include stem cells and the human genome. He is co-responsible for the Southern African Human Genome Programme which was launched in January 2011. Prof Pepper is part of a team which assists the National Department of Health with legislation concerning human tissues. He is President and Chairman of the Board of the South African Tissue Bank Association. He was until recently a member of the National Advisory Council on Innovation which advises the Minister of Science and Technology. Prof Pepper has >260 medical and scientific publications with an H-index of 70/81 (Scopus/Google Scholar), and has received a number of awards for his research. He has been extensively involved in teaching at undergraduate and postgraduate levels and is frequently solicited as a speaker at local and international meetings. He is on several editorial boards and interacts regularly with the media and writes for the lay press on medical and scientific matters.

## A.2 Members of the Study Panel

### Collet Dandara

Prof Collet Dandara is a full Professor of Human Genetics, specialising in pharmacogenomics. He is one of the leading pharmacogenetics experts in Africa. With support from various funding organisations (MRC, NRF, Southern Africa Consortium for Research Excellence (SACORE)), and colleagues in academia, he has led pharmacogenomics research at UCT. He is a TWAS Young Affiliate/ Alumni (TYAN) and represents the sub-Saharan Africa region on the TYAN Exco.

Prof Dandara is a member of the African Society for Human Genetics (AFSHG) and the Southern Africa Society for Human Genetics. He is an advocate of transformation of the research landscape which has seen him train many postgraduate students from previously disadvantaged backgrounds.

## Jantina de Vries

Prof Jantina de Vries is an Associate Professor in Bioethics at the Department of Medicine of UCT. She has broad experience in investigating ethical challenges in African genomics research and has published on a range of topics including the effect of genetic attribution on disease stigma, perspectives on broad consent, community engagement and fair and equitable policy development. She led the H3Africa Working Group on Ethics from 2012 until 2016, is the Co-PI on an H3Africa ELSI Coordinating Centre award from the National Institutes of Health (NIH) and is a member of the Global Alliance Regulatory and Ethics Working Group. Prof De Vries obtained her DPhil at the University of Oxford. Her PhD explored questions around ethnic stigmatisation as a risk in population genomic research in Africa. Her papers have appeared in *Nature Reviews Genetics*, *Science*, *PLoS Medicine*, the *European Journal of Human Genetics*, *BMC Medical Ethics*, the *Journal of Medical Ethics* and other leading journals.

## Ames Dhai

Prof Ames Dhai is the Director of the Steve Biko Centre for Bioethics which she established in 2007. It has local and international recognition as a leading centre. She serves on several policymaking bodies in the country, including being a Past-President of the South African Medical Association (SAMA), and the board of the recently established South African Health Products Regulatory Authority. She also serves as an expert advisor for the World Medical Association, the World Health Organisation (WHO), and is on the WHO's African Advisory Committee for Health Research. She participated in activities of the Institutes of Medicine (USA), and the National Academies of Sciences (USA). She is Editor-in-chief of the *South African Journal of Bioethics and Law* and Associate Editor of the *South African Medical Journal*. She can be credited with entrenching bioethics as an integral aspect of health sciences in South Africa. She is the recipient of several awards including SAMA Gender Acclaim Award (2012), the SAMA Certificate Award (2012) in honour of patriotism, courage and contribution made in the struggle for liberation of the medical profession and a joint recipient of the Vice-Chancellor's Academic Citizenship Award (2017). Using an academic platform, Professor Dhai has taken a lead in advocacy including testimony at the Life Esidimeni Tragedy.

## Melodie Labuschaigne (formerly Slabbert)

Prof Melodie Labuschaigne, former Deputy Executive Dean of the College of Law and Director of the School of Law at Unisa, is presently a full Professor in the

Department of Jurisprudence in the School of Law, Unisa. She is the recipient of the Unisa's Chancellor's Award for Excellence in Research and the Women in Leadership Research Award, and holds both a DLitt and LLD degree. Her research interests straddle medical law and ethics, legal aspects relating to the application of biotechnology, and the intersection between law and literature.

## Freddy Mnyongani

Dr Freddy Mnyongani is Senior Lecturer in Law at UKZN, Durban Campus. He has lectured at Unisa where he also served as the Chairperson of the Research Ethics Committee of the university and later of the College of Law. He is an attorney who holds the following degrees: BTh (St. Joseph's Theological Institute), LLB (Wits), LLM (Wits) and an LLD (Unisa). His research interests include legal philosophy, ethics in all its facets, public international law and international human rights law.

## Keymanthri Moodley

Prof Keymanthri Moodley is a Professor in the Department of Medicine and Director of the Centre for Medical Ethics and Law, Department of Medicine, Faculty of Health Sciences, SU. In 2017, she was appointed Adjunct Professor, Department of Social Medicine, University of North Carolina-Chapel Hill, USA. Prof Moodley is a family physician and a bioethicist. In 2013, she was rated by the NRF as an established researcher based on her numerous national and international publications, conference presentations, her role on national bodies like the MRC Board and NHREC and her involvement in international organisations – WHO, International AIDS Society (IAS) and National Institutes of Health (NIH) Data and Safety Monitoring Boards (DSMBs). She has worked as principal investigator on clinical trials since 1999, and served on the University Research Ethics Committee. The centre has been designated as a Collaborating Centre in Bioethics by the WHO, one of ten in the world and the first on the African continent. The main activities of the centre include bioethics teaching, empirical research in bioethics and clinical ethics consultation. Since 2011, Prof Moodley has co-hosted an NIH Fogarty programme to develop capacity in Health Research Ethics in Africa in collaboration with the Bioethics Centre, University of North Carolina-Chapel Hill, USA and has graduated 40 postgraduate scholars from ten African countries over the past four years. In 2013, she was awarded a second NIH grant to examine the ethical and social issues associated with HIV cure research. In 2015, the centre was awarded its third NIH grant to explore ethical, legal and social issues related to genomic biobanking. She is a Member of the ASSAf and completed an Executive MBA in 2015. Prof Moodley served as Chair of the MRC REC from November 2016 to February 2018. In 2017, she was awarded her 4<sup>th</sup> NIH grant to develop a doctoral programme in Clinical and Research Ethics. Prof Moodley has recently been appointed to the Scientific Advisory Committee of the European and Developing Countries Clinical Trials Partnership (EDCTP).

## Antonel Olckers

Dr Antonel Olckers co-founded DNAbiotec® in 2001 and has served as its CEO since then. Her expertise in science, innovation and business is integrated in DNAbiotec®. She obtained her PhD in molecular human genetics at UP with research performed at the Johns Hopkins Medical Institutions in Baltimore, MD, USA. After graduation she became the head of the Molecular Biology Group in the Department of Human Genetics (UP), and subsequently was Professor and head of department of the Centre for Genome Research (CGR) at NWU. During her tenure she served on two human research ethics committees, and thereafter on the biosafety committee of Wits. Her research focus was population genetics of African populations, and genetic risk factors for Type 2 Diabetes in different populations.

During her academic career she graduated more than 30 MSc and PhD students. She received the BioFundi Award for Capacity Building for her training and education of scientists in the academic and private sectors. To date, DNAbiotec® has trained scientists and legal professionals from 39 countries, 23 from Africa. Dr Olckers received the Legal Aid South Africa *Pro Bono* Award for the *pro bono* service her company has provided, and continues to provide, to Legal Aid SA with regard to DNA evidence. She has testified as a forensic DNA expert in the high courts and regional courts of South Africa for over 18 years. DNAbiotec® was contracted to develop the first formal forensic science qualification in South Africa, and registered it on the National Qualification Framework (NQF) of the South Africa Qualifications Authority (SAQA).

She serves on the advisory boards of several national strategic bodies, e.g. NIPMO, and has served on the National Biotechnology Advisory Committee (NBAC), which was a sub-committee for the National Advisory Council on Innovation (NACI), as well as two other project teams of NACI in the past. She was previously an extra-ordinary Professor in two departments at UP: Immunology and Forensic Medicine.

For more than a decade she has worked with others to form the first independent forensic science professional body in South Africa. These efforts culminated in the formation of the South African Academy of Forensics Sciences (SAAFS) and she was elected as its first Chairperson in early 2018. Dr Olckers received the BioFundi Lifetime Achievement award in 2018 for her work in biotechnology and innovation across different sectors.

## Anne Pope

Prof Anne Pope (LDipLib, SU; BA LLB, RU; PG Dip (International Research Ethics), UCT) is Emeritus Associate Professor in the Department of Private Law, Law Faculty, UCT. She remains a member of the UCT Faculty of Health Sciences Human Research Ethics Committee; is the Deputy Chair of the Human Sciences Research Council Research Ethics Committee; and is the current Chair of the National Health Research Ethics Council.



## Michèle Ramsay

Prof Michèle Ramsay is the Director of the Sydney Brenner Institute for Molecular Bioscience (SBIMB) and Professor in the Division of Human Genetics in the Faculty of Health Sciences at Wits. Her research interests include studying African population genetic diversity and environmental factors to better understand their role in health and disease. She also does research into the genetic basis of rare monogenic eye and skin disorders (including albinism and keratolytic winter erythema), African population genetics, pharmacogenomics and complex disease traits in African populations. She is co-responsible for the Southern African Human Genome Programme (SAHGP) with a view to exploring precision medicine in an African context. As an active member of the Human Heredity and Health in Africa (H3Africa) Consortium, she promotes ethical genomic research and capacity development in Africa. She is Principal Investigator of an NIH-funded Collaborative Centre under the H3Africa Consortium for Genomic and Environmental Risk Factors for Cardiometabolic Diseases in Africans. She teaches, supervises postgraduate students, hosts postdoctoral fellows and mentors young African scientists. She holds a South African Research Chair on Genomics and Bioinformatics of African Populations and is President of the African Society of Human Genetics.

## Raj Ramesar

Prof Raj Ramesar is Professor and head of the Division of Human Genetics at UCT. He also serves as Director of the MRC Human Genetics Research Unit, and Cancer Association of South Africa's (CANSA) Colorectal Cancer Research Consortium. Prof Ramesar is principal investigator on the Retinal Degenerative Disorders research project. Apart from being involved directly with several established research projects aimed at elucidating the genetic basis of diseases in South Africa, he is currently channelling his energy in setting up research into understanding the genetic basis of the more complex, yet common chronic disorders (e.g. hypertension) in our populations.

## Himla Soodyall

Prof Himla Soodyall is a Medical Scientist at the National Health Laboratory Service (NHLS) and Professor at Wits. Her research has used a molecular genetic approach to elucidate the evolutionary history and genetic affinities of sub-Saharan African populations. She was nominated as a Member of ASSAf in 2003, and currently serves as General Secretary on the ASSAf Council. She is the Chairperson of the Research Development Committee at the NHLS, and actively contributes to the public engagement of science. She received the Order of Mapungubwe, Bronze medal, from President Mbeki in 2005 for her contribution to science.

## Wayne Towers

Prof Wayne Towers is employed as the Academic Advisor in the Faculty of Health Sciences Ethics Office for Research, Training and Support at NWU, South Africa. He trained in the field of molecular human genetics and received his PhD in 2005 after which he completed a postdoctoral fellowship at the Centre for Genome Research, NWU in 2008. Although his basic training was in human molecular genetics, Prof Towers made a career change into the field of research ethics in 2015, after completing a postgraduate diploma in health research ethics at SU. His research focus is currently on genetic epidemiology of non-communicable disease, but he is building a future research track in the ELSIs of genetic and genomic research. Prof Towers is the chairperson of the Health Research Ethics Committee of the Faculty of Health Sciences at NWU and is also an advisory member on other ethics committees within the university.





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