

Constitutional, legal and regulatory imperatives for the renewed care and prevention of congenital disorders in South Africa

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Medical genetic services for the care and prevention of congenital disorders have declined significantly in recent years due to competing health priorities, with previously developed services becoming compromised. With an infant mortality rate of 28/1 000 live births, South Africa (SA) has passed the threshold of 40/1 000 when such services should be implemented. This article outlines the international background and SA legislative framework for medical genetic services and their implementation. International, regional and national conventions, legislation, and policy were studied for relevance to genetic services and their implementation was evaluated, including a comparison of sector capacity between 2001 and 2015. A comprehensive legislative and regulatory framework exists in SA for the provision of medical genetic services, but implementation has been fragmented and unsustainable. Congenital disorders and genetic services are not prominent in national strategies and excluded from interventions aimed at combating child mortality and non-communicable diseases. Capacity today is at a lower level than in 2001. The failure to recognise the burden of disease represented by congenital disorders is the underlying reason for the implementation and service shortfall. Child mortality rates have stagnated since 2011 and can be significantly further reduced by prioritising healthcare issues other than HIV/AIDS, including congenital disorders. It is now an imperative that SA responds to World Health Assembly Resolution 63.17 and prioritises congenital disorders as a healthcare issue, providing services to uphold the dignity and human rights of the most vulnerable members of society.

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Congenital disorders (CDs) are an underestimated critical health issue. Competing health priorities have resulted in the neglect of medical genetic services for the care and prevention of CDs. While a comprehensive national legislative and regulatory framework exists for these services, providing for the fundamental and socio-economic rights enshrined in the constitution, poor implementation has resulted in shortfalls in service for those affected by CDs, many of whom are living with disability.

The internationally agreed definition of CDs are abnormalities of structure or function present from birth, which may be evident at birth or manifest later in life.^[1] Although CDs are a global problem, over 90% occur in middle- and low-income countries (MLIC), where 95% of those affected consequently die.^[2] In South Africa (SA), one in every 15 live births is affected by a CD.^[3] Modelled data indicate a minimum of 6.8% of SA births, of which 80.5% are caused by genetic factors and 19.5% by

teratogens.^[2,3] CDs are often undiagnosed, or misdiagnosed due to a lack of awareness of attending clinicians to make appropriate diagnoses.^[2,4,5] Mortalities as a result of CDs are often incorrectly attributed, burying CDs as an issue.^[2,4,5] However, up to 70% of CDs can be prevented, cured or the resulting disability ameliorated through appropriate,

timely treatment.^[2,6] Lack of data resulting from poor diagnoses results in the CD burden being under-reported in many MLIC countries, where CDs are substantially under-reported.^[3,4,5,7] In SA, only 2 174 CD cases were documented in 2012 via the Birth Defect Collection Tool administered by the Department of Health,^[8] compared with the

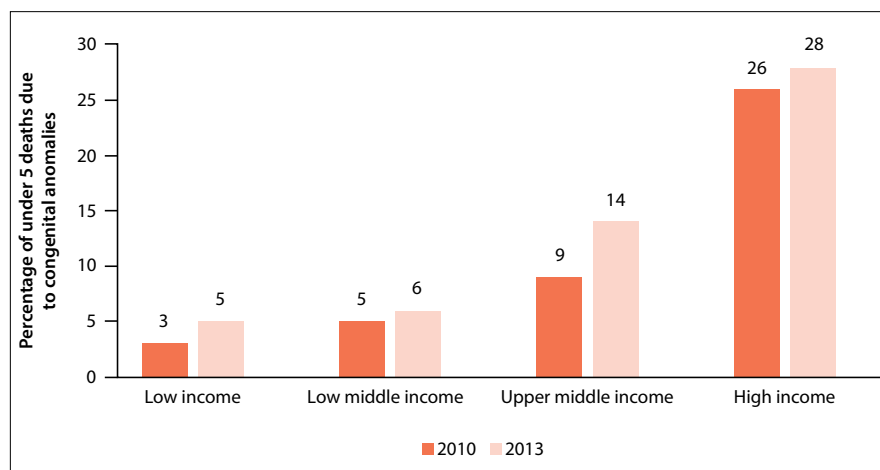


Fig. 1. Comparison of the percentage of under-five deaths resulting from congenital anomalies for World Bank Country Classifications.^[9]

83 118 (6.8%) expected, indicating under-reporting of 97%.^[3,9]

As SA develops, the proportion of childhood deaths from CDs is increasing, as mortality from communicable diseases decreases.^[3,9] This follows the epidemiological trend in industrialised countries where CDs emerged and remain as the leading cause of child death and disability.^[2,3] Fig. 1 compares the percentage of under-five deaths resulting from congenital anomalies (a subset of CDs) between the World Bank Country Classifications according to Gross National Income (GNI) per capita.^[9] The proportion of deaths from congenital anomalies in all GNI groups increased between 2010 and 2013, and in high-income countries they are the leading cause of deaths, accounting for 28% in children under five.

Significant reductions in the infant mortality rate (IMR) and the under-five mortality rate (U5MR) were seen between 2008 and 2011 from comprehensive HIV/AIDS interventions and the childhood Expanded Programme of Immunisation.^[10,11] However, both the IMR and the U5MR have stagnated since 2011 and the neonatal mortality rate since 2009.^[12] This indicates that health issues other than those being addressed, such as CDs, require prioritisation.^[3,9,10,12] In 2013, congenital abnormalities (obvious structural abnormalities) overtook infection as the third leading cause of death in early neonates, accounting for 11.24% of early neonatal deaths in infants weighing >1 000 g compared with 8.84% from infection.^[13] When the IMR falls below 20/1 000 live births in a country, CDs emerge as the leading cause of infant death.^[14]

Fig. 2 illustrates the IMR decreasing as countries develop and transition epidemiologically, and the increasing proportion of CD-related infant deaths.^[9] The need for medical genetic services is usually recognised by nations when their IMR falls below 40/1 000 live births, as infant and child mortality can be significantly further reduced through such measures.^[4,5,14,15] Up to 40% of serious congenital malformations which may be fatal if untreated, can be cured by surgical intervention, and in 30% of cases the degree of resulting disability can be reduced through relevant treatment.^[4,6] With an IMR of 28 live births in 2014^[12] SA is beyond the point when appropriate services should be implemented to further reduce child mortality and to better care for those who are disabled by CDs.^[3,4,9,14,15]

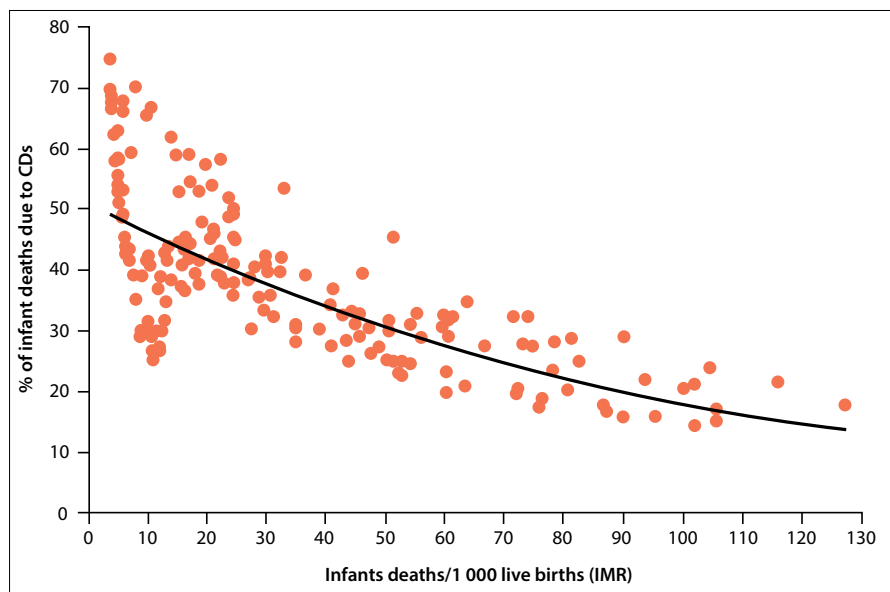


Fig. 2. Relationship between infant mortality and percentage of infants dying from CDs, based on global country figures.^[9]

Medical genetic services for the care and prevention of CDs ensure that people with CDs, or at reproductive risk of having children with CDs, can live and reproduce as normally as possible.^[5,16] They are key in reducing the contribution of CDs to the disease burden and should provide the 'best possible patient care' in the prevailing circumstances for those affected or at risk of CDs.^[9,15,16] Medical genetic services established at human genetics departments and medical schools have been the mainstay in SA since the 1970s.^[9] Access to these services has been limited to urban areas with some rural outreach.^[9]

Services began to improve in the late 1990s and early 2000s, when CDs first emerged as a health issue and the Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities were published.^[17] However, service implementation continued through the established framework of academic centres, rather than integrating services into primary healthcare and extending clinical genetic services beyond urban areas, as was recommended in the 2001 Policy Guidelines.^[9,17]

Constitutional, legal and regulatory framework in SA

Several international treaties and resolutions indicating global political commitment towards CDs are applicable to SA and summarised in Table 1. Notable is World Health Assembly (WHA) Resolution 63.17 of 2010.^[18] This recognises the importance

of CDs as a cause of stillbirths and neonatal deaths, and their contribution to the U5MR and attaining Millennium Development Goal (MDG) 4 to reduce child mortality by two-thirds.^[19] Achieving MDG4 required 'accelerated progress in reducing neonatal mortality including the prevention and management of birth defects.'^[9,18] Although progress was made towards an U5MR of 20/1 000 live births, MDG4 was not achieved.^[9]

SA is a signatory to international protocols and conventions that have resulted in national legislation (Table 1).^[9] Most relevant to medical genetic services is the United Nations Convention on the Rights of Persons with Disabilities (UNCRPD).^[20] Signed and ratified in 2007, the UNCRPD promotes, protects and ensures full and equal enjoyment of all human rights and fundamental freedoms by all persons with disabilities and promotes respect for their inherent dignity.^[20] The United Nations Convention on the Rights of the Child (UNCRC), ratified by SA in 1995, concerns the rights of the disabled child without discrimination, while ensuring dignity, promoting self-reliance and community participation.^[21]

The Constitution and National Legislation

A comprehensive, national legislative framework exists in SA for the provision of medical genetic services (Table 2).^[9] The constitution of SA underpins all other

legislation and provides for fundamental rights to equality, dignity, freedom and security of the person, education and life.^[22] Section 27 provides for the socio-economic right of everyone to access healthcare services, including reproductive healthcare, and social security: women, children and people with disabilities are entitled to these s27 rights with the qualification that:

'[t]he State must take reasonable legislative and other measures, within its available resources, to achieve the progressive realisation of each of these rights.'^[22]

Children's rights not subject to the concept of progressive realisation are listed in s28(1)(c), which states that every child has the right:^[9]
'to basic nutrition, shelter, basic health care services and social services.'^[22]

Table 1. International treaties, conventions, declarations, protocols of relevance to medical genetic services^[9]

Document	Article/rule/overview
World Programme of Action Concerning the Disabled (1982) ^[35]	Prevention, rehabilitation and equalisation of opportunities
Standard Rules on the Equalisation of Opportunities for Persons with Disabilities (1993) ^[36]	Rules: 1. Awareness raising; 2. Medical care; 3. Rehabilitation; 4. Support service; 5. Accessibility
United Nations Convention on the Rights of the Child (signed 1993 and ratified 1995) ^[21]	Articles: 2. No discrimination; 6. Right to life; 23. Disabled child; 24. Healthcare; 26. Social Security
International Covenant on Economic, Social and Cultural Rights (1966) (signed 1994 and ratified 2015) ^[37]	Article: 12. Physical and mental health
United Nations Millennium Declaration (signed 2000) ^[19]	Goal 4: Reducing under-five mortality by two-thirds by 2015
United Nations Convention on the Rights of Persons with Disabilities (signed and ratified 2007) ^[20]	Articles: 5. Equality/non-discrimination; 6. Women with disabilities; 7. Children with disabilities; 8. Awareness raising; 9. Accessibility; 10. Right to life; 19. Living independently; 20. Personal mobility; 23. Respect for home and family; 25. Health; 26. Habilitation and Rehabilitation
African Charter on the Rights and Welfare of the Child ('Children's Charter') (signed 1997 and ratified 2000) ^[38]	Articles: 5: Right to life; 13. Protection of physically/mentally disabled to ensure dignity; 14. Physical/mental health and healthcare
The New Partnership for Africa's Development (2001) ^[39]	Healthcare provision and delivery
African Youth Charter (signed and ratified 2009) ^[40]	Articles: 16. Health; 23. Girls and young women; 24. Mentally/physically challenged youth
World Health Assembly Resolution 63.17 (signed and ratified 2010) ^[18]	Urges member states to address CDs as a healthcare issue through specific actions

Table 2. Key national legislation of relevance to medical genetic services^[9]

Title	Overview and sections relevant to CDs
Constitution of the Republic of South Africa (108 of 1996) ^[22]	Chapter 2: Bill of Rights. Sections: 9. Equality; 10. Human dignity; 11. Life; 27(1)(a). Access to healthcare services, including reproductive healthcare; 27(1)(c). Access to social security including appropriate social assistance; 28(1)(c). Every child has the right to basic healthcare services
Health Professions Act (56 of 1974) ^[28]	Regulates the health professions through the Health Professions Council of South Africa
National Health Act (61 of 2003) ^[23]	Sections: 4(3)(a). Free healthcare to pregnant/breastfeeding women, children under six not members/beneficiaries of medical aid schemes (c) free termination of pregnancy; 21(2)(b)(vii). Genetic services; 21(2)(k) & 25(2)(w). Management, prevention and control of communicable and NCDs; 23 (1)(a)(ix) & 27(1)(a)(ix). Epidemiological surveillance/monitoring of national and provincial trends; 21, 23, 25 & 27. Implementation of national/provincial policy and compliance; 39(2)(a)&(d) and 70(2)(d) Health needs of vulnerable groups including children and people with disabilities; 48. Development and provision of human resources in national health system; 52. Regulations relating to human resources; 70. Identification of health research priorities
Choice on Termination of Pregnancy Act (92 of 1996) ^[24]	Sections: 2(b)(ii) and minors 5(5)(a)(ii) Termination of pregnancy (TOP) between 13 - 20 weeks inclusive if substantial risk that the fetus would suffer from a severe physical/mental abnormality Sections: 2(c)(ii) and minors 5(5)(b)(ii) TOP after the 20th week if the continued pregnancy would result in a severe malformation of the fetus
The National Health Laboratories Service Act (37 of 2000) ^[26]	Laboratory services for the public health sector Sections 4 & 5: Cost-effective and efficient health laboratory services including training
Mental Health Care Act (17 of 2002) ^[25]	A legal framework for mental health in SA with an emphasis on human rights
The Nursing Act (33 of 2005) ^[27]	Regulates the nursing profession through the South African Nursing Council
Children's Act (38 of 2005) ^[30]	Sections: 11. Children with disability or chronic illness; 156(1)(g). Care and protection
Social Assistance Act (13 of 2004) ^[29]	Sections: 7. Care dependency grants; 9. Disability grants

This includes children with CDs, and those living with disability caused by CDs.

National Health Act

The National Health Act (NHA) 61 of 2003 provides a framework for a single healthcare system for the country, rectifying the socio-economic imbalances and inequities of the health services of the past and provides for many of the rights outlined in the Bill of Rights.^[23]

The NHA is the only piece of national legislation in which genetic services feature prominently. A clear directive is included in Chapter 3, under Main functions of the National Department in s21(2)(b)(vii):^[9]

'the Director-General must, in accordance with national health policy, issue and promote adherence to, norms and standards on health matters including genetic services.'^[23]

Genetic services are listed among other vital services, including sterilisation and termination of pregnancy, and the provision of health services/healthcare services for convicted persons and persons awaiting trial, highlighting the considered importance of these services.

The NHA stipulates that the national policy is executed via the provincial departments of health in a top-down approach. National and provincial health councils ensure national policy is implemented provincially and that provincial health plans comply with national policy (NHA s21, 23, 25, 27).^[23] To date, no provincial policies or health plans for genetic services have been developed, with the exception of the Western Cape (Prof. Raj Ramesar and Dr Mike Urban, personal communication), and most provinces refer to the 2001 national policy.^[17]

Other sections of the NHA relevant to medical genetic services include:

- management, prevention and control of non-communicable diseases (NCDs) through healthcare services (s21(2)(k) and s25(2)(w))
- epidemiological surveillance and monitoring of disease trends (s23(1)(a)(ix) and s27(1)(a)(ix))
- identification of priority health problems and research priorities relevant to the burden of disease (s70)
- health needs of vulnerable groups, including children and people with disabilities (s39(2)(a)&(d), s70(2)(d))

Table 3. National policies, strategies, guidelines, charters and initiatives relevant to medical genetic services

Document	Year	Overview
Integrated National Disability Strategy White Paper ^[41]	1997	The right of people with disabilities to play a full, participatory role in society
Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities ^[17]	2001	Goals, objectives, strategies and delivery of clinical and laboratory services for the care and prevention of CDs, including human capacity recommendations
Guidelines on Ethics for Medical Research. Book 2. Reproductive Biology and Genetic Research ^[42]	2002	Section 3.3: Genetic screening and testing
Department of Health. Strategic Framework for the Modernisation of Tertiary Hospital Services ^[43]	2003	Human genetics throughout document in proposed models. Appendix 4: Revised human capacity recommendations
National Guidelines for the Care and Prevention of the Most Common Genetic Disorders, Birth Defects and Disabilities ^[31]	2005	Targeting primary health care providers, describing common CDs and strategies for their care/prevention
National Perinatal and Neonatal Morbidity and Mortality Committee ^[45]	2008	Audit perinatal and neonatal deaths and produce annual reports and final report in 2011
National Patients' Rights Charter ^[46]	2008	Common standard to realise, uphold, promote and protect the constitutional right of access to healthcare services Section 2: Access to healthcare; 2(3)(c) Provision for special needs (newborn, children, pregnant women, disabled); (d) Counselling; (e) Palliative care; (g) Health information
Negotiated Service Delivery Agreement. Outcome 2: A long and healthy life for all South Africans ^[47]	2010	Strategic outputs: 1. Increasing life expectancy, NCDs and burden of disease; 2. Decreasing maternal and child mortality; 4. Strengthening health system effectiveness
National Health Insurance in South Africa Policy Paper ^[48]	2011	To transform existing institutions/organisation in the healthcare system to make the system more equitable, offering universal coverage to a defined comprehensive package of services
South Africa's National Strategic Plan for a Campaign on Accelerated Reduction of Maternal and Child Mortality in Africa ^[49]	2012	To rapidly reduce maternal and child mortality
Strategic Plan for Maternal, Newborn, Child and Women's Health and Nutrition in South Africa 2012-2016 ^[33]	2012	Reducing maternal and child mortality. Long-term health conditions in children (p24)
Committee on Morbidity and Mortality in Children Under 5 Years ^[50]	2012	Reviews and monitors maternal, perinatal and childhood mortality and morbidity data in SA
National Department of Health: Strategic Plan 2014/15-2018/19 ^[32]	2014	Programme 3: Maternal and child health. NCDs
Guidelines for Maternity Care in South Africa ^[44]	2015	Chapters: 4. Antenatal care; 9. Pregnancy problems; 10. Intrauterine, neonatal deaths and stillbirths; 15. Screening for congenital anomalies

- human resources including adequate resources for education and training of healthcare personnel (Chapter 7).^[23]

Other national legislation

Other key national legislation provides for different aspects of genetic services and are summarised in Table 2.^[9] Notable are:

- The Choice on Termination of Pregnancy Act (1996), used as part of a preventive strategy when severe abnormalities are detected *in utero* (s2(b)(ii), 2(c)(ii), s5(5)(a)(ii), s5(5)(b)(ii)).^[24]
- The Mental Health Act (2002) provides a legal framework for mental health, emphasising the human rights of the mentally ill.^[25]
- The National Health Laboratories Service (NHLS) Act (2000) provides for laboratory services as an essential component of genetic services.^[26]
- The Nursing Act and the Health Professions Act provide for statutory bodies regulating these professions.^[27,28] In SA, genetic counsellors require registration under the Health Professional Council of South Africa.^[28]
- Social Assistance Act (2004) provides for care dependency and disability grants.^[29]
- Matters concerning the disabled child are specified in the Children's Act (2005).^[30]

National policy

Two key policy documents focus solely on CDs: National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities^[17] and National Guidelines for the Care and Prevention of the Most Common Genetic Disorders, Birth Defects and Disabilities.^[31] The National Policy Guidelines outline goals, objectives, strategies, personnel requirements and delivery of clinical and laboratory services appropriate for the care and prevention of CDs.

Despite these policies, CDs are not regarded as a healthcare issue in SA strategic plans (Table 3), despite the emphasis on reducing child mortality in response to MDG4. The Department of Health (DoH) Strategic Plan 2014/15-2018/19 recognises mental health disorders as a NCD, aiming to improve access to services through screening and treatment, but does not acknowledge the significant genetic component of many of these disorders.^[32]

In the DoH Strategic Plan for Maternal, Newborn, Child and Women's Health and Nutrition in South Africa 2012 - 2016, CDs are mentioned as a cause of neonatal death, contributing to 15 - 20%

of children affected by a long-term/chronic health conditions who are 'not receiving the care they require'.^[9,33] No responding interventions are outlined.^[9] NCDs are a key strategic focus in these policies, but there is no cognisance that CDs are an NCD, the first experienced in life, contributing to the 43% of NCD related deaths annually in SA.^[14,34]

A draft Disability Rights Policy, a first step to implementing the UNCRPD^[20] was published for public comment in early 2015. A review of several health and disability policy documents is underway, including the 2001 Policy Guidelines^[17] and accompanying clinical guidelines proposed.

Implementation

Despite the existence of a comprehensive legislative framework for the development of medical genetic services, implementation has fallen short and medical genetic services are in decline.

In Table 4, personnel capacity is compared between 2001 and 2015, demonstrating that these services are at a lower base today than in 2001.^[3,9]

Of over 1 000 healthcare providers, mainly labour ward nurses, trained through the Medical Genetics Education Programme (MGEP), less than 100 remain in services for care and prevention of CDs.^[9] The lack of continued support forced trainees to discontinue their genetic nursing role and move to non-related fields. This has resulted in reduced clinical capacity to identify and diagnose CDs and has influenced the surveillance of CDs.^[9]

SA is the only country (of eight emerging economies) where positive development in improving medical genetic service structures has ceased and indeed regressed.^[3,7]

Reasons for this decline since 2001 include competing health priorities that have redirected political commitment and funding.^[3,9] The lack of investment in medical genetic services has resulted in insufficiently trained personnel, inadequate capacity at all levels, and severely compromised laboratory services.^[3,9] These shortfalls make it impossible to uphold the constitutional rights of those affected by CDs, including children and those living with disability, through the practical provision of the services they require.

Conclusion

While the SA constitution is admired globally for its protection of human rights, there has been a failure to translate these constitutional imperatives into effective, accessible services for the care and prevention of CDs. Failure to recognise the burden of disease represented

Table 4. A comparison of medical genetic services capacity in 2001 and 2015^[9]

Category	Recommended*	2001 [‡]		2015	
	Number/ratio (Pop = 46 13 m) [†]	Number	Ratio (Pop = 44 82 m) [†]	Number	Ratio (Pop = 54 96 m) [§]
Medical geneticists	20/1 per 2 m	4	1 per 11.2 m	12 [¶]	1 per 4.6 m
Genetic counsellors	80/1 per 580 000	<20	1 per 2.2 m	9	1 per 6.1 m
Medical scientists/technologists	100/1 per 450 000	50	1 per 900 000	26**	1 per 2.1 m

* Department of Health. Strategic Framework for the Modernisation of Tertiary Hospital Services. Discussion Document. Pretoria, South Africa: Department of Health, 2003;86.

† Department of Health. Policy Guidelines for the management and prevention of genetic disorders, birth defects and disabilities. Pretoria, South Africa: Department of Health, 2001.

‡ Statistics South Africa. South African Statistics 2014. Pretoria, South Africa: Statistics South Africa, 2014.

§ Statistics South Africa. Mid-Year Population Estimates 2015. Pretoria, South Africa: Statistics South Africa, 2015.

¶ No medical geneticists are employed by the State in Gauteng. Personal communication: A. Krause, 11 February 2016.

|| Figure increased to 9 in April 2016 plus 6 in private practice. Personal communication: T. Wessels, 25 February 2016.

** NHLS academic medical scientists only. Personal communication: H. Soodyall, 27 July 2015.

by CDs is the underlying cause of this unethical shortfall in services.¹⁹ Despite the comprehensive legal framework remaining unchanged, the implementation of this legislation has declined. 'We have the constitution, the law, the guidelines and policy – but good laws are being lost in translation' (Prof. Marylyn Christianson, Special Olympics Disability Summit, Shanghai, 2007). In the words of the 2013 GenTee Report,¹⁷ 'the continuing lack of commitment to the country's constitutional, legal and regulatory framework [has resulted in] an implosion of genetic services in the public domain due to very limited public will, commitment and funding [resulting in] inequitable genetic services [impacting] the ability of the poorer population to utilize services according to their needs.'

It is an imperative that SA responds to WHA 63.17¹⁸ and prioritises CDs as a healthcare issue by providing the required medical genetic services to uphold the dignity and human rights of people with CDs, the most vulnerable of our society.

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