

The contribution of congenital disorders to child mortality in South Africa

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Reduction in child mortality has been a priority issue in South Africa leading up to the Millennium Development Goals. However, the contribution of congenital disorders (CDs) to child mortality is yet to be recognised and acted upon.

Rapid reductions in child mortality have resulted largely from comprehensive HIV and AIDS programmes and interventions such as the childhood Expanded Programme of Immunisation. However, the Rapid Mortality Surveillance System reports that since 2011, reductions in child mortality rates “stopped abruptly”. This indicates that health issues other than those currently being addressed may require long-term prioritisation. In 2013, congenital anomalies (excluding many CDs) overtook infection as the third leading cause of early neonatal deaths, which account for one-third of all under-five deaths.

As South Africa transitions epidemiologically, the proportion of deaths caused by CDs is increasing, as mortality from communicable diseases drops, revealing the previously hidden disease burden of CDs. In South Africa, many CDs go undiagnosed or are misdiagnosed, resulting in the incorrect cause of death being reported. These inaccurate data result in an underestimation of the true disease burden of CDs in the country.

As up to 70% of CDs can be prevented or ameliorated, it is essential that they be prioritised and that relevant, accessible services for prevention and care be implemented. A good legislative and regulatory framework exists in South Africa for the provision of services, but implementation has been poor and fragmented. Current services are available at a lower base than in 2001.

This chapter argues for recognition of the role of CDs in child mortality and morbidity and the potential advantages of medical and genetic services for the prevention and care of CDs.

In South Africa many congenital disorders go undiagnosed or are misdiagnosed resulting in the incorrect cause of death being reported and in an underestimation of the true burden of congenital disorders in the country.

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Introduction

Since the Millennium Development Goals (MDGs) were set in 2000, there has been a global drive to reduce child mortality.¹ In order to achieve the MDG 4 target of cutting under-five mortality by two-thirds by the end of 2015, countries rapidly incorporated measures relevant to their specific healthcare challenges. As the MDG deadline loomed, South Africa focused on responding to the HIV and AIDS and concomitant tuberculosis (TB) epidemics, re-engineering primary health care and developing a strategic plan for Maternal, Newborn, Child and Women's Health (MNCWH) and Nutrition. Several ministerial committees were established to address underlying issues, including the National Perinatal Mortality and Morbidity Committee (NaPeMMCo) and the Committee on Mortality and Morbidity in Children (CoMMiC). The topic of newborn and child mortality and survival has also been examined in detail in previous editions of the *South African Health Review*.^{2,3}

None of these policies or initiatives has comprehensively recognised the contribution of congenital disorders (CDs) to neonatal, infant and child mortality and morbidity. This is despite the fact that in industrialised countries around the world, CDs are the leading cause of death in infants and children, contributing up to 28% of under-five deaths in high-income countries.⁴ Like many other middle- and low-income countries (MLICs), South Africa is following this epidemiological trend and the proportion of deaths and disability resulting from CDs is rising, especially as communicable diseases are better controlled.⁵

This chapter provides an overview of the health issue of CDs and their unappreciated role in child mortality and morbidity in South Africa. Epidemiological transition is described in relation to CDs and an outline is given of the growing role of CDs in the burden of disease in South Africa and why it has remained hidden. The chapter also identifies the benefits of recognising the contribution of CDs in the burden of disease, including potential reductions in mortality and morbidity through medical genetic services for the prevention and care of CDs. The current status of these services is reviewed and compared with what is required to address this growing health need. Where relevant, secondary data have been sourced from peer-reviewed literature and globally recognised data sources.

Millennium Development Goal 4

The adoption of the United Nations Millennium Development Declaration in 2000¹ and the time-based targets set for the following 15 years has resulted in varying degrees of achievement among participating nations. MDG 4 focused on improving child survival, and significant progress towards this two-thirds reduction in under-five mortality was achieved globally. By 2015, the global under-five mortality rate (U5MR) had been reduced by 53%, halving the number of children dying annually from 12.7 million in 1990 to 5.9 million in 2015.⁶ Many regions achieved or came close to the targeted two-thirds (66%) reduction in U5MR. In sub-Saharan Africa, the U5MR decreased by 54%.⁶ One in every 12 children still dies before his or her fifth birthday in sub-Saharan Africa compared with one in 147 in high-income countries.⁶

South Africa's progress towards MDG 4 has been varied. In 2005, South Africa was one of only four countries where the U5MR

was higher than the 1990 MDG baseline due to the negative epidemiological impact of HIV and AIDS and concomitant TB.⁷ Significant reductions were then seen in child mortality until 2011 due to a number of factors. These included scaled-up prevention of mother-to-child transmission of HIV, expanded roll-out of antiretroviral therapy, and the addition of the rotavirus and pneumococcal conjugate vaccines to the childhood Expanded Programme of Immunisation (EPI).⁷⁻⁹ Despite the rate of reduction tripling since 2000, the U5MR decreased by one-third only, to 39 per 1 000 live births,⁸ falling short of the South African MDG 4 target of 20 per 1 000 live births.

However, according to the Rapid Mortality Surveillance Report there has been no further decrease in infant and child mortality in South Africa since 2011.⁸ This stagnation indicates the need to address other health issues contributing to infant and child mortality, whilst continuing with ongoing efforts.^{5,7,8} Many issues related to social determinants of health and childhood illnesses are already being addressed, including malnutrition and infectious diseases associated with poverty such as measles, diarrhoea and malaria. A 2013 study identified 15 key interventions that were scaled up in South Africa for maximum impact on maternal and child mortality during the final two years of the MDGs.¹⁰ Preliminary measures to target non-communicable diseases (NCDs), now a component of South Africa's quadruple burden of disease, are also under way.¹¹ However, CDs, which are the first NCD experienced by infants and children,^{5,12} are yet to be recognised for their contribution to stillbirths, and neonatal, infant and child mortality in South Africa. This is despite the global call to action by the World Health Assembly (WHA) in 2010 through Resolution 63.17,¹³ which recognised the importance of CDs as a cause of stillbirths and neonatal mortality. To attain MDG 4, WHA 63.17 called for "accelerated progress in reducing neonatal mortality including the prevention and management of CDs".¹³

CDs are a common, costly and critical health issue. According to the 2006 March of Dimes report,¹⁴ serious CDs result in the death of 3.3 million children under the age of five globally every year. Although CDs are found in all populations throughout the world, over 90% occur in MLICs where 95% of CD-related deaths occur.¹⁴ Reasons for this unequal distribution of CDs include less-developed health services and a variety of poverty-related reasons that increase the risk of CDs occurring, such as a higher percentage of older mothers and consanguineous marriages, and the survival advantage against malaria for carriers of some single gene disorders.¹⁴ Despite this higher incidence of CDs in MLICs, the contribution of CDs to the burden of disease is yet to be recognised by many of these countries.

Definitions and terminology

CDs are defined as "any potential pathological condition arising before birth, including disorders caused by environmental, genetic and unknown factors, whether they are evident at birth or become manifest later in life".¹⁵ CDs that are caused before conception are genetic and the result of chromosomal abnormalities or single gene defects, or are multifactorial in origin. Post-conception CDs are the result of teratogens (alcohol, prescribed and recreational drugs, maternal infections and illnesses, exposure to environmental toxins

and radiation) or abnormalities of the fetal environment that deform or disrupt the developing fetus (e.g. constraint and amniotic band disorder). Teratogens – fetal environmental factors that cause CDs – may be chemical substances, physical agents or infections, and are more common in MLICs where the potential for exposure is higher and there are fewer preventative measures in place.¹⁴

The global lack of consensus on terminology related to CDs has plagued the medical genetics sector and resulted in lower priority being given to this healthcare issue. In 2006, after decades of uncertainty, agreement was reached by international experts on the use of basic terminology at a joint World Health Organization (WHO) and March of Dimes meeting.¹⁵ Participants recommended that the terms ‘CDs’ and ‘birth defects’^a be used synonymously. Use of the term ‘congenital anomaly’ was not advised, since it excludes around 40% of functional CDs including non-syndromic congenital disabilities, common single gene disorders and inborn errors of metabolism.^{b,12,15}

The limited uptake of this agreed terminology and the copious use of non-equivalent terms has led to confusion, fragmentation of effort and an inability to compare data sets.^b This has diluted the visibility of the significant contribution of CDs to neonatal, infant and child mortality, and prevented CD data from being evaluated comprehensively. CDs are often undiagnosed or misdiagnosed and the cause of death is wrongly attributed, contributing to under-reporting.¹⁶ The lack of accurate data on CDs has led to an underestimation of the true contribution of CDs to the disease burden.^{14,16,17}

Epidemiological transition

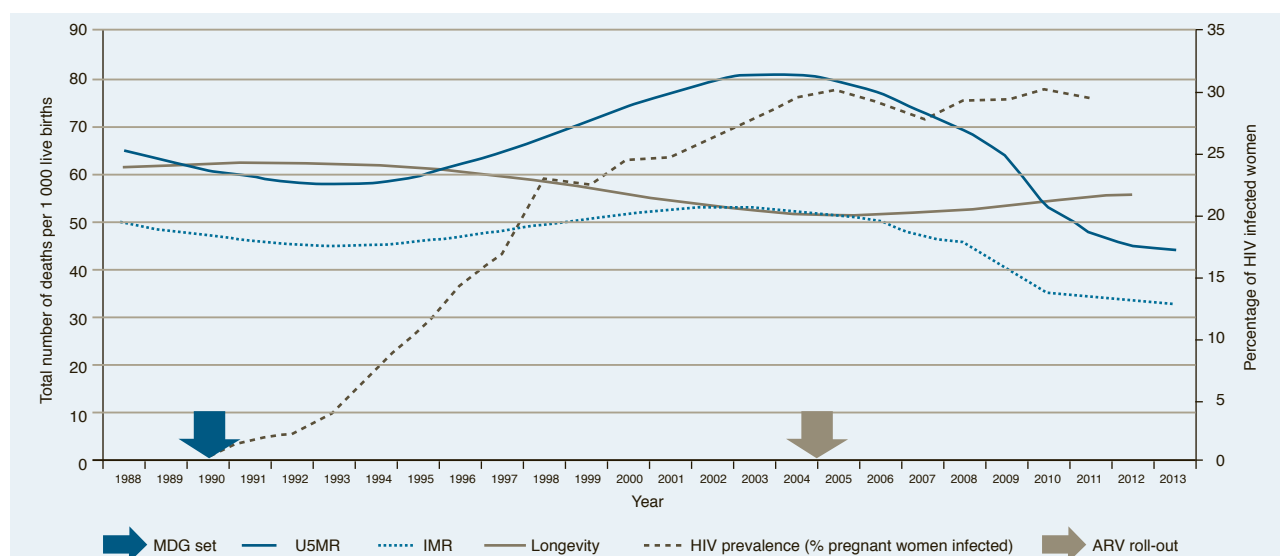
Epidemiological transition occurs when there is a change in population health statistics and pattern of disease in a region or country, resulting from changes in socio-economic, educational, infrastructural and healthcare development.¹⁴ The epidemiological

transition of countries has been described extensively using Omran’s model,¹⁸ which defines three stages of disease. As mortality rates decrease and longevity increases, countries move from Stage 1 ‘the age of pestilence and famine’ to Stage 2, ‘the age of receding pandemics’ and into the third stage, ‘the age of degenerative and man-made diseases’ characterised by low mortality rates and high life expectancy at birth (over 50 years). It is in this third stage that communicable diseases are well controlled or eradicated, and NCDs and degenerative diseases emerge.¹⁸

Like many MLICs, South Africa is not following Omran’s classic model¹⁸ of epidemiological transition that was completed by industrialised, high-income nations decades ago.¹⁹ From 1960 to the early 1990s, infant and child mortality in South Africa declined steadily and longevity increased.⁵ Figure 1 plots the U5MR,²⁰ infant mortality rate (IMR)²⁰ and life expectancy at birth (longevity) data,²¹ and the HIV epidemic (indicated by the percentage of HIV-positive pregnant women)²² for South Africa for the last 25 years. Life expectancy at birth peaked at 62.33 years in 1992, and in 1993 both the U5MR and the IMR were at an all-time low of 58.2 per 1 000 live births and 45.1 per 1 000 live births respectively.^{20,21} All indications were that South Africa was approaching the early phases of transition from Stage 2 (the ‘age of receding pandemics’) to Stage 3, the ‘age of degenerative and man-made diseases’ and was set to follow the classical model of epidemiological transition.^{5,18}

However, in the mid-1990s, epidemiological transition was interrupted and reversed by the HIV and AIDS and concomitant TB epidemics (Figure 1). As a result of these epidemics, South Africa has seen a counter-transition.⁷ HIV prevalence rates, indicated by the infection rate in pregnant women, climbed over the following decade from 7.6% in 1994 to 30.2% in 2005.²² The U5MR rose dramatically in response, peaking at 80.8 per 1 000 live births in 2003 and the IMR at 53.2 per 1 000 live births in 2002.²⁰ In 2005, longevity dropped to its lowest level since the 1960s at

Figure 1: Epidemiological transition in South Africa over the past 25 years, as demonstrated by data for childhood mortality,²⁰ longevity,²¹ and the HIV epidemic²²



Source: Malherbe, 2015.⁵

a Birth defects are defined as abnormalities of structure or function, including disorders of the metabolism, which are present from birth.¹⁵

b Personal communication: B. Modell, 19 January 2016.

51.56 years.²¹ The combination of a newly emerged communicable disease (HIV and AIDS) and the re-emergence of an old infection (TB) with an increasing burden of NCDs has resulted in an additional stage being added to Omran's original concept, called the 'age of emergent and re-emergent infections'.^{19,23,24}

Following the roll-out of comprehensive HIV and AIDS interventions in 2004, the HIV and AIDS prevalence rate plateaued at around 30% in the early part of the current decade (see Figure 1).²² The rapid reductions achieved in infant and child mortality between 2005 and 2011 have resulted in both the IMR and U5MR being lower today than prior to the HIV and AIDS epidemic, at 28 per 1 000 and 39 per 1 000 live births respectively.⁸ South Africa is now back in positive epidemiological transition. However, both the IMR and U5MR have stagnated since 2011, and the neonatal mortality rate (NMR) has stagnated since 2009 despite the continued implementation of HIV and AIDS interventions.⁸

A significant contributor to ongoing high mortality in children are deaths from unnatural causes, causing just over a third (18.5%) of deaths in children aged 1–4 years in 2014.²⁵ Natural deaths in the same age-group were attributed to intestinal infectious diseases (17.2%), influenza and pneumonia (9.1%) and malnutrition (8.6%), with TB and HIV ranked fourth and fifth.²⁵ Some neonatal deaths, which contribute the bulk of under-five deaths, are preventable through addressing modifiable factors that are intertwined with social determinants of health. CoMMiC reports that modifiable factors are 30% home-based, including seeking medical attention earlier, failure to recognise severity of illness, and inadequate nutrition.²⁶ The majority of health system-modifiable factors (80%) relate to health personnel.²⁶

The total contribution of CDs to stillbirths^c in South Africa is unknown. Data from the Perinatal Problem Identification Programme (PIPI) 2012/13 attributed 2.5% of stillbirths in non-tertiary settings and 7.7% of stillbirths in tertiary settings to CDs.^{27,28} These data are likely to be an underestimate, especially in primary health care settings due to restricted diagnostics and unavailability of screening. The Lancet Ending Preventable Stillbirths Series Study Group estimates a global median of 7.4% stillbirths attributed to CDs based on reliable data from 18 countries.²⁸

However, the role and contribution of CDs to the ongoing high level of mortality is not visible in the available data.

Congenital disorders and epidemiological transition

During the process of epidemiological transition, CD deaths remained invisible, essentially 'buried' among deaths due to communicable diseases, and only emerging as these diseases were adequately controlled.¹⁶ As industrialised countries moved through the second stage of epidemiological transition, there was a slight decrease in CD birth prevalence and deaths due to fetal environmental factors, essentially teratogens.¹⁴ This was because of improved care and prevention strategies for these disorders. However, since 85–90% of CDs have a genetic or partially genetic aetiology, their birth prevalence and resulting mortality remained high.¹⁴ Deaths from

these CDs became proportionately greater in overall neonatal, infant and child mortality as deaths from communicable diseases reduced. As industrialised countries completed the second stage of epidemiological transition, CDs emerged and have remained a leading cause of child death in these nations today.^{29,30}

CDs attained public health significance in these industrialised nations in the early 1960s when they moved into the third stage of epidemiological transition.^{14,18} This was demonstrated in a comparative study undertaken by McKeown³⁰ of death rates in England and Wales for 1901 and 1971, shown in Table 1. An overall reduction of 68% was seen in the death rate for all diseases over the 70-year study, including a 90% reduction in infectious diseases. Death rates for NCDs decreased by 45% overall, but the number of CD deaths remained the same.³⁰ This was due to the mainly genetic cause of CDs, which cannot be changed, resulting in CDs contributing a greater proportion of deaths as overall mortality decreased.

Table 1: Standardised death rates (per million of population) for England and Wales in 1901 and 1971

	1901	1971	% Reduction
Infectious Diseases			
Airborne Diseases			
Respiratory infection	2 747	603	78
Pulmonary TB	1 268	13	99
Whooping cough	312	1	100
Measles	278	0	100
Scarlet fever and Diphtheria	407	0	100
Smallpox	10	0	100
Upper respiratory tract infections	100	2	98
Sub-total	5 122	619	88
Food and Water-borne Diseases			
Cholera, Diarrhoea and Dysentery	1 232	33	97
Non-respiratory TB	544	2	100
Typhus, Typhoid	155	0	100
Sub-total	1 931	35	98
Other Infections			
Sub-total	1 415	60	96
90% Overall Reduction for Infectious Diseases			
Non-communicable diseases			
Birth defects	126	127	0
Perinatal problems	1 249	192	85
Heart disease	1 186	1 688	- 42
Rheumatic heart disease	487	88	92
Cancer	844	1 169	- 39
Other diseases	4 598	1 406	69
Sub-total	8 490	4 670	45
Overall reduction for non-communicable diseases 45%			
Total	16 958	5 384	68
68% Overall Reduction in Death Rate of All Diseases			

Source: McKeown, 1976.³⁰

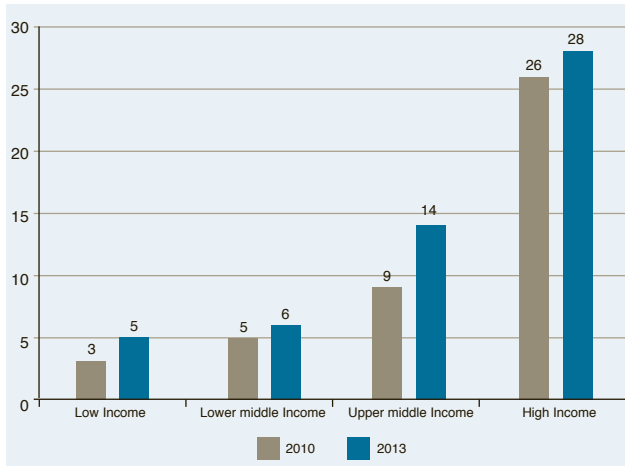
A further example of the increasing proportion of child deaths from CDs is shown in Figure 2, which shows the percentage of deaths due to congenital anomalies using the World Bank Country Classifications^d according to Gross National Income (GNI) per capita.⁴ As GNI increases, the percentage of child deaths from CDs

c In South Africa, a stillbirth is defined as a fetus of at least 26 weeks' gestation and born with no signs of life after complete birth. This definition varies between countries, making data compilation and comparison complex.

d South Africa is classified as an upper middle-income country.

(congenital anomalies only) increases, contributing a greater portion in the higher GNI classification. An increase can be seen in all groups between 2010 and 2013. Today, CDs account for 28% of child deaths in high-income countries and they are the leading cause of death in infants and children younger than five years.

Figure 2: Percentage of under-five deaths resulting from congenital anomalies using World Bank Country Classifications

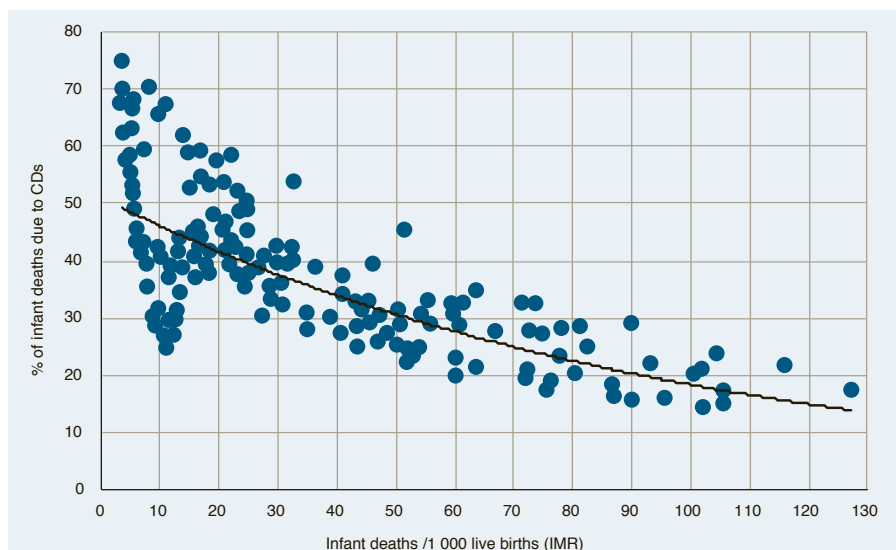


Source: WHO, 2015.⁴

Figure 3 plots global IMRs for countries against the percentage of infant deaths resulting from CDs, demonstrating that as infant mortality drops, the contribution (proportion) of CDs relative to infant mortality increases. This is particularly noticeable as the IMR drops to between 40 and 50 per 1 000 live births.

It is clear from the literature and mounting evidence that the contribution of CDs to child mortality increases as countries develop, and that MLICs, including South Africa, are following this epidemiological trend.^{5,14} The proportion of deaths resulting from CDs in South Africa will rise as overall infant and child mortality decreases.

Figure 3: Relationship between infant mortality and percentage of infants dying from CDs based on global country figures



Source: Modell, 2015.^e

Congenital disorders in South Africa

Prior to the HIV and AIDS epidemic in the early 1990s, CDs began to emerge as a healthcare issue in South Africa due to falling child mortality and increasing longevity. A national task force of experts was established in collaboration with the WHO to investigate the need for, and implementation of, services for the care and prevention of CDs. Following wide consultation, the National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities were published in 2001.³¹

The 2001 National Policy Guidelines³¹ outlined goals, objectives, strategies and delivery of clinical and laboratory services appropriate for the care and prevention of CDs in South Africa. Priority disorders were designated, which included Down syndrome, neural tube defects, fetal alcohol syndrome (FAS), albinism, cleft-lip and palate, and club feet. The financial cost to society and to the State resulting from burden of disease was estimated at several billion Rand annually at the time. Personnel requirements to implement these services were specified in the 2001 Guidelines, based on UK criteria, and were later revised using more relevant criteria for South Africa in the Strategic Framework for the Modernisation of Tertiary Hospital Services.³²

In 2004, the National Guidelines for the Care and Prevention of the Most Common Genetic Disorders, Birth Defects and Disabilities³³ were published, targeting Primary Health Care Providers (PHCPs), and describing common CDs and strategies for their care and prevention.

After this surge of policy generation, the HIV and AIDS and TB epidemics obscured the issue of CDs once more.⁵ As a result, the growing commitment and momentum towards CDs as a healthcare issue was redirected to these competing healthcare priorities along with the associated resources.

e Personal communication: B. Modell, 20 August 2015.

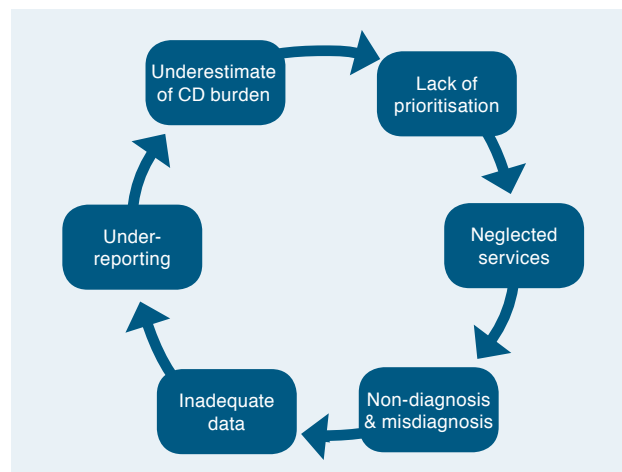
Data modelling of CDs

There is a lack of birth prevalence data for CDs in South Africa, as is the case for many MLICs.^{14,16} The overriding factors for this are the limited facilities available and the lack of skilled clinicians to identify and diagnose CDs.^{14,16,17} This is exacerbated by the incompleteness of vital registration data and inadequacies of other mortality and morbidity data sources, especially for infants and children.^{3,8} To fill this gap, country-specific prevalence estimates are generated using a combination of local data for specific indicators combined with known prevalence rates from more well-resourced countries.^{14,16} These modelled data¹⁴ of genetic causes and an estimate of teratogenic causes^f indicate that a minimum of 6.8% of South African births are affected by CDs.⁹ Of these, 80.5% are caused by genetic factors and 19.5% by teratogens. This translates to one in every 15 live births in South Africa being affected by a CD.⁵ The proportion of teratogenic CDs is more than the 10–15% expected in MLICs and is one of the highest documented prevalence rates in the world due to the high prevalence of FAS, which is entirely preventable.¹⁴

Reliable surveillance data provide a vital information tool for policy-makers to plan, implement, monitor and evaluate policy accordingly to prevent adverse health conditions and improve public health. With 6.8% of live births affected by a CD in South Africa, a total of 83 118 live births would have been affected in 2012, based on vital registration data.³⁴ However, only 2 174 cases were reported via the Birth Defects Collection Tool (BDCT) administered by the Department of Health in 2012.³⁵ With only 2.6% of the expected CDs being reported, this indicates under-reporting by 97.4% for 2012. When taking into account that only 26% of CDs are diagnosable during the early neonatal period,³⁶ under-reporting of 90% is still unacceptably high. This is hampering the recognition of CDs as a key contributor to the burden of disease.

The lack of empirical data as evidence for CDs in South Africa prevents policy-makers from accurately assessing the contribution of CDs to the disease burden.^{37,38} Figure 4 outlines the cycle caused as a result of this underestimation. Underestimation leads to a lack of prioritisation of CDs, and to CD prevention and care services being neglected. The lack of CD diagnosis due to poor services leads to under-reporting and poor data. Under-reporting contributes to an underestimate of CD deaths and disability. CDs are not considered to be a healthcare priority and the cycle resumes. Modelling is used as a tool to highlight the gap between the expected health need and current services available.

Figure 4: The cycle caused by the underestimation of CDs



Emerging data

Despite the lack of evidence-based data, available mortality data are beginning to reveal the hidden disease burden of CDs. Under-reporting of births and deaths leading to incomplete vital registration data, especially for children, makes it an unsuitable source for monitoring and evaluating child mortality. District Health Information System (DHIS) data, which record deaths in public sector hospitals, also tend to be inadequate, as outlined by McKerrow in 2010.³ The PPIP is the most detailed source of information on factors contributing to perinatal death and hence child mortality.³ The 2012/13 PPIP data,²⁷ representing 75.6% of all DHIS-recorded births in South Africa, indicated that congenital abnormalities^h have overtaken infection as the third leading cause of death during the first week of life in neonates after deaths from immaturity and hypoxia. Congenital abnormalities accounted for 11.24% and 8.20% of early neonatal deaths in infants weighing >1 000g and >500g compared with 8.84% and 7.44% of deaths respectively from infection.²⁷ This shift, combined with the stagnation of neonatal mortality since 2009, and of infant and under-five mortality since 2011,⁸ speaks to the epidemiological transition in South Africa.

This trend continued in the Western Cape Province (WC) in 2014, with congenital abnormalities ranked as the third cause of early neonatal death in infants weighing >500g and >1 000g.ⁱ At 10%, deaths in the under-fives from congenital abnormalities in the province are double that recorded in other provinces and nationally.^{4,39} The WC neonatal mortality rate is also half the national rate of 11 per 1 000 live births,⁴⁰ making the province a good example of what will occur in other provinces in the coming decade as healthcare services improve and CDs are revealed.

Following on from the PPIP programme is the Child Healthcare Problem Identification Program (CHILD PIP), which audits child mortality from 28 days to 18 years. In 2005, deaths from congenital abnormalitiesⁱ were mentioned in only 0.3% of cases from the 15 hospitals across six provinces participating in CHILD PIP.⁴¹ CHILD PIP acknowledged this as an underestimate due to the lack of access

f Personal communication: A Christianson, 15 August 2013.

g These modelled estimates are based on national figures published in Appendix B of the 2006 March of Dimes Report.¹⁴ These figures are currently under revision.

h No formal definition is given by the PPIP programme for congenital abnormalities; however, it is understood to refer to obvious structural abnormalities only.

i Personal communication: N. Rhoda, 25 February 2016.

j Although no formal definition of congenital abnormalities is given, the diagnoses made include single gene, chromosomal and multifactorial disorders and post-conception disorders.

to post-mortems and lack of paediatricians to assess (diagnose) the cases.⁴¹ Between 2005 and 2014, mortality data were submitted to CHILD PIP by 198 hospitals across 49 districts. Of the 44 854 deaths recorded,^k 2.2% cited congenital abnormalities as either the main cause or underlying cause of death. Although higher than the 2005 figure, this too was reported as an underestimate due to the lack of identification of CDs by health workers conducting the audit, and limitations in the cause-of-death categorisation in the current CHILD PIP programme, which is now being rectified by the development of a new data form.^l

As the U5MR has decreased, the proportion of deaths in the perinatal period has risen.⁴² In South Africa, just over 40% of deaths among under-fives occur during the neonatal period.⁴³ Globally, the contribution of neonatal deaths to under-five mortality is projected to rise from 45% to 52% by 2030.⁶ The contribution of CDs to these deaths should be recognised and addressed if child mortality, particularly neonatal mortality, is to be reduced further.

Since available data sets reported only include sub-groups of CDs, the true contribution of CDs to the burden of disease is likely to be higher than estimated.

Congenital disorders and disability

CDs are not merely a cause of child mortality but also of morbidity. For every child who dies as a result of a CD, many survive serious CDs and sustain lifelong mental, physical, auditory or visual disability.¹⁴ The economic cost as a result of this morbidity is considerable. In 2012, 116 000 beneficiaries – caregivers of children older than one year with severe disabilities or disabling chronic illnesses requiring permanent home-based care, including those affected by CDs, acquired conditions and injuries – received means-tested care dependency grants of R1 200 per month.⁴⁴ This totals R1.6 billion annually, excluding inflation, adult disability grants and other costs.

In lower-resourced countries, the majority of children born with serious CDs (3.3 million annually) die due to a lack of appropriate care, and a further 3.2 million who survive are disabled for life.¹⁴ Early intervention and relevant care can save the life of the child from a life-threatening serious CD, and cure or ameliorate the degree of long-term disability. Many of these interventions, including one-off surgeries for congenital malformations, are relatively inexpensive compared with the cost of ongoing chronic care for untreated CDs. Many community-based preventative measures are both inexpensive and 'low-tech'.¹⁴ Where appropriate services for the care and prevention of CDs are available, 30% of CD deaths in the first year cannot be prevented. However, 40% of the cases can be cured, mainly by surgery, and 30% survive with disability.^{45,46} In South Africa, specialised surgery capacity is limited and unavailable in some provinces, preventing widespread access to such intervention. Strengthening surgical capacity in all areas is required, as both general and specialised surgery could help to alleviate this shortfall.

Interventions must incorporate both prevention and care. As outlined in the 2006 March of Dimes Report,¹⁴ there are three types of prevention: primary prevention, in which CDs are avoided prior to conception through basic reproductive health approaches, including folic acid supplementation; secondary prevention, which aims to

reduce the number of babies born with CDs through screening, prenatal diagnosis, avoidance of potentially teratogenic substances during pregnancy and the option of termination of pregnancy; and tertiary prevention, being the early detection, diagnosis, cure and mitigation of CDs after the child is born, including surgical interventions and palliative care. Tertiary prevention is equivalent to care that constitutes diagnosis, treatment, counselling and psychosocial support of those affected by CDs.

In lower-resourced countries, prevention tends to be overemphasised to the detriment of care due to the misplaced myth that care is expensive.^{14,16} This may cause those affected by CDs and consequently living with a disability to be marginalised, which undermines their human dignity and human rights. Christianson's mantra, "Care is an absolute. Prevention is the ideal" was coined in 2000.⁴⁷ This emphasises both care and prevention (at all levels) as integral components of medical genetic services, and one cannot be neglected at the expense of the other.

In many MLICs, comprehensive services for the care and prevention of CDs are not implemented. In addition to the immense loss of life and suffering of those affected, there is a significant economic cost. Implementation of key interventions can reduce this cost and save lives, as outlined in Table 2. Congenital malformations^m are the most common type of CD and also the most treatable. Almost half of the congenital malformations such as cleft lip and/or palate and congenital heart defects can be cured through paediatric surgery, but without this intervention, the result is death or permanent disability.

To date, the folate fortification of staple food (maize meal and bread) to reduce neural tube defects (NTDs) – an example of primary prevention – is the only preventative intervention being comprehensively implemented in South Africa. Since fortification began in 2003, a 30.5% reduction in NTDs has been seen.⁴⁸ In addition to the obvious reductions in mortality and morbidity, there is also a considerable cost benefit. According to Sayed et al.,⁴⁸ with the average estimated cost of treatment being R100 000 per NTD case for the first three years of life, averting 406 cases every year would render a minimum annual saving of R40.6m, offset by the minimal cost of R1.4m per year for the 2% fortification.

If all other interventions were implemented as outlined in Table 2, the burden of genetically determined CDs could be reduced by almost 70% and would generate sizable economic benefits due to the gain of almost three years of healthy life per head of the population.^{14,16,46} However, the current static child mortality rates in South Africa are an indication that current interventions being implemented fall far below this level of potential.

k 87.6% of these deaths occurred in children aged five and younger. Personal Communication: M. Patrick, 17 March 2016.

l Personal Communication: M. Patrick, 17 March 2016.

m A malformation due to multifactorial inheritance.

Table 2: Summary of estimated potential effects of interventions for preventing genetically determined CDs

Type of CD	Birth prevalence per 1 000 live births	Intervention	Maximum postnatal lives saved (per 1 000 live births)	Maximum reduction (%)	Estimated average increase in longevity per head of population (years)
Congenital malformations ⁿ	36.5	Paediatric surgery (Tertiary Prevention/care)	17.70	48.5	1.24
		Folic Acid Supplement (Primary Prevention)	11.50	31.5	0.81
		Prenatal diagnosis (Secondary Prevention)	3.50	9.6	0.25
		Total congenital malformations	32.70	89.6	2.30
Chromosomal disorders ^o	3.8	Family planning (Primary Prevention)	0.75	19.7	0.05
		Prenatal diagnosis (Secondary Prevention)	0.5	13.2	0.04
		Total Chromosomal disorders	1.25	32.9	0.09
Genetic risk factors ^p	2.4	Routine antenatal & neonatal care (Tertiary Prevention/care)	2.40	100	0.17
Inherited disorders (severe, early onset) ^q	11.5	Genetic counselling (Primary Prevention)	1.73	15	0.12
		Neonatal screening (Tertiary Prevention/care)	0.7	6.1	0.05
		Prenatal diagnosis (Secondary Prevention)	1.15	10	0.08
		Total inherited disorders	3.60	31.1	0.25
Total	54.2		39.90	73.7	2.80

Source: Christianson and Modell, 2004;¹⁶ Christianson et al., 2006.¹⁴

Medical genetic services

Interventions to prevent, detect and care for CDs are collectively known as medical genetic services. The aim of genetic services is two-fold: to reduce suffering by offering care to those affected, and to improve health by preventing CDs.¹⁴ These services are key in reducing the contribution of CDs to the burden of disease. By providing the 'best possible patient care' in the prevailing circumstances for those affected by or at risk of CDs, medical genetic services ensure that people with CDs, or those at reproductive risk of having children with CDs, 'can live and reproduce as normally as possible'.^{17,49,50}

In many lower-resourced countries, the development of medical genetic services has been driven by epidemiological transition. Governments begin to see CDs as an important public health issue when the IMR falls below 40–50 per 1 000 live births and only limited reductions in child mortality can be achieved by addressing other health issues.^{12,16,17,50} With an IMR of 28 per 1 000 live births in 2014,⁸ South Africa is well past this threshold when medical genetic services should be comprehensively implemented. CDs should be prioritised alongside other NCDs and ongoing interventions against communicable diseases in order to further reduce child mortality and to provide better care for those who are disabled as a result of CDs.⁵ When the IMR reaches 20 per 1 000 live births (the MDG 4 target for South Africa), CDs will emerge as a leading cause of death in infants.¹²

n Obvious structural abnormalities/malformations due to multifactorial inheritance.

o A structural or numerical abnormality of the chromosomes.

p G6PD deficiency and Rhesus haemolytic disease of the newborn.¹⁴

q Single gene disorders, including autosomal-dominant, autosomal-recessive, sex-linked inheritance and mitochondrial disorders.

Medical genetic services in South Africa

Since the early 1970s, the mainstay of medical genetic services in South Africa has been the work of human genetics departments at major academic centres and medical schools in urban areas, starting in Johannesburg and Cape Town.⁵¹ Access to these services was limited mainly to urban areas, with some outreach into rural areas being conducted by the academic centres.

Services improved following the publication of the Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities in 2001.³¹ The number of posts supported by the National Health Laboratory Service increased in response to the policy.⁵¹ However, implementation continued through the established framework of academic centres, rather than through the integration of services into primary health care and the extension of clinical genetic services beyond urban areas, as was recommended in the 2001 Guidelines.

As services for HIV and AIDS developed over the past decade to combat the epidemic, tertiary medical genetic services were neglected.⁵ By severely limiting the implementation of the 2001 policy guidelines, the lack of investment in medical genetic services has resulted in insufficient trained personnel, inadequate capacity at all levels, and severely compromised laboratory services.^{5,51} In 2013, South Africa was reported as the only country of eight emerging economies evaluated where positive development in improving medical genetic service structures had ceased, and indeed retrogressed.³⁷

The framework of medical genetic services across the continuum of health care in South Africa is shown in Table 3. This demonstrates how interventions to prevent and care for CDs may be integrated into maternal and child care (e.g. comprehensive antenatal care) and highlights current gaps in implementation.

Table 3: Overview of medical genetic services across health care in South Africa (X=service in place in SA; S= service required in SA)

Type of Prevention/Care	Intervention	1° Health Care			2° Health Care	3° Health Care	
		Clinic	Community Health Centre	District Hospital	Regional Hospital	Provincial Tertiary Hospital	Academic Hospital
Primary Prevention: Pre-conception care ensuring individuals born free of CDs and not damaged in early embryonic period	Family planning	X	X	X	X ^r	X ^r	X ^r
	Optimising women's diet:^s • Folic acid supplementation (5mg daily) ^t • Iron (200mg daily) • Alcohol/Smoking (education)	X	X	X	X	X	X
	Pre-conception screening	S ^u	S ^u	S ^u	X	X	X
	Maternal infections:^v Detection and treatment (primarily syphilis)	X	X	X	X	X	X
	Treating health conditions:						
	• Diabetes mellitus		X	X	X	X	X
	• Epilepsy		X	X	X	X	X
	• DVT or cardiac conditions (Wafarin)			X	X	X	X
Genetic counselling (if required)		S ^w	S ^w	X	X	X	
Secondary Prevention: Reducing the number of children born with CDs	Screening:						
	Ultrasound		S	S	X	X	X
	Advanced maternal age screening	S	S	S	S	X	X
	Pre-natal diagnosis (amniocentesis, chorionic villus sampling, cordocentesis)				X	X	X
	Genetic counselling and psychosocial support		S ^w	S ^w	S ^w	X	X
	Options: • Therapeutic • Termination of pregnancy		S	X	X	X	X
Tertiary Prevention/Care: Early detection, cure, amelioration and care once a child is born with a CD	Early diagnosis:						
	• Examination of every newborn by trained observer (top to toe)		S	S	X	X	X
	• Biochemical screening (Hypothyroidism)		S	S	S	S	S
	Care interventions:						
	• Medical/therapeutic		X	X	X	X	X
	• Surgery			X ^x	X ^x	X ^x	X ^x
	• Habilitation ^y			X	X	X	X
• Palliative care			X	X	X	X	
• Genetic counselling and psychosocial support		S ^w	S ^w	X ^w	X	X	

r Family planning is associated with specialist services in these secondary and tertiary hospitals although it is not listed as an official service.

s Mandatory iodisation of table salt (40–60ppm) was introduced in South Africa in 1995 to prevent iodine-deficiency disorders.

t Mealie meal and wheat flour have been fortified with folic acid in South Africa since 2003.

u Pre-conception screening at the level of community health centres and district hospitals should include screening for advanced maternal age, taking a family history and relevant referral.

v The rubella (German measles) vaccine is available only in the private sector in South Africa.

w Genetic counselling for common disorders such as Down syndrome and spina bifida.

x This may be limited to general surgery in some provinces where specialised surgery (e.g. cardiac, craniofacial, etc.) is currently unavailable. Some CDs may require only general surgery (e.g. Meckel's diverticulum), whilst others require more specialised surgical intervention (e.g. cleft lip and/or palate).

y For example, occupational, speech and physiotherapies.

Table 4: A comparison of medical genetics services capacity in 2001 and 2015

Category	Recommended 2003 ³²		2001 ³¹		2015	
	Number	Ratio (Pop=46 13m) ⁵²	Number ²	Ratio (Pop=44 82m) ⁵²	Number	Ratio (Pop=54 96m) ³⁴
Medical geneticists	20	1 per 2m	4	1 per 11.2m	12 ^z	1 per 4.9m
Genetic counsellors	80	1 per 580 000	<20	1 per 2.2m	8 ^{aa}	1 per 8.4m
Medical scientists/technologists	100	1 per 450 000	50	1 per 900 000	26 ^{ab}	1 per 2.1m

Medical genetic services are now at a lower base than in 2001, as outlined in Table 4. The 2003 recommended human capacity requirements³² to be trained and in-post by 2010 remain unfulfilled. Today there are 12 medical geneticists, compared with four in 2001 and the 20 recommended by 2010.

Of the 14 genetic counsellors practising today, only eight practise in State services, compared with 20 in 2001 and the 80 recommended. Since existing posts were frozen and no new posts were created to accommodate newly qualifying genetic counsellors, many have been forced to leave the service or the country, or to work in private practice. No provision was made for genetic counsellors in the Occupational Specific Dispensation (OSD), a government initiative aimed at attracting and retaining skilled employees through improved remuneration. Budget cuts have also reduced diagnostic laboratory personnel numbers to unsustainable levels and equipment has not been upgraded or maintained.^{5,51} Many practitioners have left, retired or emigrated due to a high service workload, limited opportunity to undertake research, and the inability to perform their tasks satisfactorily due to inadequate medical genetic laboratory services. To reverse this, adequate staffing and modern equipment are required along with the necessary training to ensure the translation of this technology is appropriate to the country's needs and circumstances.

The lack of access to health workers is possibly the greatest constraint across the health system in terms of South Africa achieving health goals,²⁶ including the MDGs. Rectifying the shortfalls in specialist healthcare professionals in the medical genetic services sector is critical.

The Medical Genetics Education Programme (MGEP) is a distance-education postgraduate training course for nurses and rural medical officers. This equips in-post healthcare professionals with basic knowledge and skills to identify and diagnose common disorders, counsel patients and to refer those affected by CDs appropriately, providing an additional supporting capacity for medical geneticists and genetic counsellors. Between 2004 and 2013, over 1 000 healthcare providers, mainly labour ward nurses, were trained through the MGEP course held countrywide.^{ac} Today, fewer than 100 remain in services for the care and prevention of CDs. The lack of continued government support for this training, including financial, trainee contact and uptake of these skills by facility management, has forced trainees to discontinue their genetic nursing role and move to other fields. This has had a direct negative impact on the national surveillance of CDs via the BDCT, as many MGEP-

trained labour ward nurses, together with midwives, obstetricians and paediatricians, are the frontline healthcare professionals encountering CDs in the continuum of care.

Legislative and regulatory framework

The World Health Assembly (WHA) call in 2010¹³ to prioritise CDs as a healthcare issue through Resolution 63.17 was fundamental for medical genetic services worldwide. WHA 63.17¹³ recognised the importance of CDs as a cause of stillbirths and neonatal deaths, and their contribution to under-five mortality, and it is recognised that for MDG 4 to be achieved, "accelerated progress in reducing neonatal mortality including the prevention and management of birth defects" was required. Although South Africa is yet to respond to WHA63.17,¹³ a number of other international treaties and protocols of relevance to CDs have led to the development of equivalent national legislation. Table 5 lists international treaties and conventions that included content of relevance to CDs, many of which are foundational for national legislation.

South Africa is well placed to respond to WHA 63.17¹³ since a comprehensive, national legislative framework already exists for the provision of medical genetic services for the care and prevention of CDs. Relevant national legislation is outlined in Table 5. The Constitution of the Republic of South Africa⁵³ provides for fundamental rights to life, equality, dignity, freedom and security of the person, and education. The socio-economic right for all to access healthcare services, including reproductive health care, is provided subject to the concept of progressive realisation.^{ad} Every child – including those with CDs and disabled as a result – has the right to 'basic nutrition, shelter, basic healthcare services and social services' (section 28, 1c), but these rights are not subject to progressive realisation.

The provision of medical genetic services is specified in the National Health Act (NHA) 61 of 2003 through a clear directive in Chapter 3, under "Main functions of the National Department" in section 21 (2) (b) (vii) as follows:

The Director-General must, in accordance with national health policy, issue and promote adherence to, and norms and standards on health matters including genetic services.⁵⁴

Also of relevance to CDs in the NHA are epidemiological surveillance, management, prevention and control of NCDs, and the health needs of vulnerable groups including children and the disabled.⁵⁴ Other key national legislative instruments of relevance to CDs are outlined in Table 6.

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

aa Of these eight genetic counsellors, three are employed full-time directly by the State, three full-time by tertiary institutions, and four are employed part-time, plus six in private practice. Personal communication: T. Wessels, 25 February 2016.

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

ac Personal communication: D. Tshikedi, 2 October 2013.

ad Defined as recognising that economic and social rights can only be achieved over time, subject to the availability of resources.

Table 5: International treaties, conventions, declarations and protocols of relevance to medical genetic services

Document	Article/ Rule/Overview
World Programme of Action Concerning the Disabled (1982)	Prevention, rehabilitation and equalisation of opportunities
Standard Rules on the Equalization of Opportunities for Persons with Disabilities (1993)	<ul style="list-style-type: none"> 1 Awareness-raising 2 Medical care 3 Rehabilitation 4 Support service 5 Accessibility
United Nations Convention on the Rights of the Child (signed 1989 and ratified 1995)	<ul style="list-style-type: none"> 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)	12 Physical and Mental Health
United Nations Millennium Declaration (signed 2000) ¹	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)	<ul style="list-style-type: none"> 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)	<ul style="list-style-type: none"> 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)	Healthcare provision and delivery
African Youth Charter (signed and ratified 2009)	<ul style="list-style-type: none"> 16 Health 23 Girls and young women 24 Mentally/physically challenged youth
World Health Assembly Resolution 63.17 (signed and ratified 2010) ¹¹	Urges member states to address CDs as a healthcare issue through specific actions

Table 6: Key national legislation of relevance to medical genetic services

Title	Overview	Sections relevant to CD
Constitution of the Republic of South Africa (108 of 1996) ⁵³	Chapter 2 – Bill of Rights	<ul style="list-style-type: none"> • 9 Equality • 10 Human dignity • 11 Life • 27(1)(a) Access to healthcare services, including reproductive healthcare • 28 (1)(c) Every child has the right to basic healthcare services
Health Professions Act (56 of 1974)	Regulates the health professions through the Health Professions Council of South Africa	
National Health Act (61 of 2003) ⁵⁴	Framework for a structured and quality uniform health system	<ul style="list-style-type: none"> • 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) and 25(2)(w) Management, prevention and control of communicable and non-communicable diseases • 23(1)(a)(ix) and 27(1)(a)(ix) Epidemiological surveillance and monitoring of national and provincial trends • 21, 23, 25, 27 Implementation of national/provincial policy and compliance • 39(2)(a)&(d), 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)	Law related to abortion	<ul style="list-style-type: none"> • 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 or mental abnormality • 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)	Laboratory services for the public health sector	<ul style="list-style-type: none"> • 4 and 5(1) Cost-effective and efficient health laboratory services including training
Mental Health Care Act (17 of 2002)	A legal framework for mental health in South Africa with an emphasis on human rights	
The Nursing Act (33 of 2005)	Regulates the nursing profession through the South African Nursing Council	
Children's Act (38 of 2005)	Protection of children and their rights	<ul style="list-style-type: none"> • 11 Children with disability or chronic illness • 156 (1)(G) Care and protection
Social Assistance Act (13 of 2004)	Rendering of social assistance	<ul style="list-style-type: none"> • 7 Care dependency grants • 9 Disability grants

National policy

The National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities³¹ and the National Guidelines for the Care and Prevention of the Most Common Genetic Disorders, Birth Defects and Disabilities³³ are the only two policy documents that focus solely on CDs. In 2014, a process of revision of the 2001 Policy Guidelines was initiated and is due for completion in 2016.

A new edition of the Guidelines for Maternity Care in South Africa was published in 2015,⁵⁵ replacing the 2007 edition. This contains relevant content on the identification of mothers at high risk of having babies with a CD, as well as essential information for primary prevention of CDs and referral to genetic services.

Implementation of the 2001 Policy Guidelines,³¹ and therefore of the underpinning legislation, has been fragmented, especially in the past decade. The lack of recognition for the contribution of CDs to the disease burden has resulted in the exclusion of comprehensive interventions from national strategies. The lack of integration of key interventions is noticeable in the National Department of Health's

Strategic Plan for Maternal, Newborn, Child and Women's Health and Nutrition in South Africa 2012–2016.⁵⁶ While CDs are mentioned as a cause of neonatal death and as contributing to 15–20% of children affected by long-term/chronic health conditions "not receiving the care they require", no responding interventions are outlined.⁵⁶ In other documents, individual CDs or categories such as mental health disorders are recognised in the National Department of Health's Strategic Plan 2014/15–2018/19,⁵⁷ but with no acknowledgement of the genetic predisposition of such conditions.

CDs are the first NCDs experienced by people – infants and children.¹² However, they are not contextualised as such in national strategies and interventions to address the growing NCD disease burden. In the 2nd Triennial Report of the Committee on Mortality and Morbidity in Children (CoMMiC),²⁶ CDs are included under long-term health conditions in children together with acquired childhood conditions resulting from infections. This precludes the use of the term 'NCD', effectively burying CDs beneath communicable disease once again.

Conclusions and recommendations

CDs are currently not recognised as a healthcare issue in South Africa. Thus, their contribution to the disease burden is currently underestimated, and the impact of interventions for their prevention and care is not considered. Instead, those born with and dying from CDs are largely overlooked and those surviving with disability are largely ignored, as care is not being provided to this most vulnerable group. Despite the lack of data due to poor national surveillance, CDs are beginning to emerge as a significant cause of mortality in children. Data from child mortality audit programmes indicate the growing contribution of CDs to neonatal death and to deaths in children under five years of age. Following the epidemiological trend of industrialised countries, the contribution of CDs to the disease burden will continue to increase in South Africa as the country develops, until they eventually constitute the leading cause of child death and disability. By not addressing this issue comprehensively now, many lives will be lost unnecessarily and others will survive with lifelong disability as the result of serious CDs. There is also a significant economic cost associated with CDs. Up to 70% of the deaths and the disability caused by CDs can be prevented or mitigated through relevant interventions.^{45,46}

South Africa has already passed the point at which other nations have identified the need to develop comprehensive medical genetic services in order to reduce child mortality further. This is largely a consequence of the abating HIV and AIDS and TB epidemics which have buried CDs as a health issue. While the control of these epidemics must continue, it cannot be at the expense of other child healthcare needs if child mortality, including neonatal deaths, is to be further reduced.^{5,7}

While a good legislative framework provides for genetic services in South Africa, the significant shortfall in implementation indicates that this intention has been lost in translation. Medical genetic services currently available are inadequate in terms of capacity and infrastructure at all levels and far from the seamless continuum of care required. While the implementation of accessible and relevant medical genetic services for the care and prevention of CDs is essential to contribute to reducing static mortality rates further in South Africa, such interventions must also plan for morbidity and ongoing treatment of those affected. Such interventions have major economic implications for South Africa, and the CD contribution to the burden of disease should be addressed holistically.

If South Africa is to meet the Sustainable Development Goal (SDG) 3⁵⁸ target to end preventable deaths in newborns and children, and to reduce the U5MR to at least 25 per 1 000 by 2030, CDs must be comprehensively addressed. Reducing premature mortality from NCDs by 2030 will require CDs to be contextualised as a NCD in South Africa, in alignment with the international definition of NCDs. The SDG goal for universal health coverage and access to quality essential healthcare services can only be achieved if relevant medical genetic services for the care and prevention of CDs are made available for all South Africans.⁵⁸

The re-engineering of the healthcare service and the NHI initiative⁵⁹ provide opportunities for the rebirth of medical genetic services, and for rectification of their currently compromised state. Their integration as part of services for women's, maternal and child health would allow medical genetic services to develop throughout the continuum of care in all appropriate stages of life. Such services are vital to

uphold the dignity and constitutionally and legally enshrined rights of those affected by CDs.^{5,60}

Priority actions include:

- Increased political will and financial commitment – This should be accompanied by appropriate CD-related expertise on ministerial and government committees dealing with the neonatal/infant/child mortality and the development of women, maternal and child health services.
- Improvement of national surveillance, patient registries and monitoring of CDs – Linked to existing systems for sustainability, these should be accompanied by ongoing training to increase coverage and accuracy of CD data identification and documentation.
- Capacity-building – The education and training of healthcare professionals and the creation of related posts is required at all levels, especially for staff in primary health care facilities.
- Increased community education and awareness – Such programmes are required to ensure awareness, understanding and knowledge of available services and how to use them.
- Role of lay advocacy/patient support groups – These need to be recognised, supported and strengthened to partner with government and the medical genetics community.

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