



30 November 2015

To: Ms Malebona Precious Matsoso  
Director-General  
National Department of Health  
Civitas Building  
Cnr Thabo Sehume and Struben Streets  
Pretoria

CC: P. Holele, M. Makua, V. Mtyongwe, K. Sivannan, D. Tshikedi

Dear Ms Matsoso,

### **Request to establish Ministerial Committee on Congenital Disorders**

We, the Governing Board and members of Genetic Alliance South Africa (GA-SA), representing the congenital disorders patient community of South Africa (SA), call upon the National Department of Health (NDOH) to establish a Ministerial Committee on Congenital Disorders (CDs).

CDs or birth defects, are abnormalities of structure or function, including metabolism, present from birth. Some are evident at birth while others only manifest in childhood, adolescence or later in life. The majority (80-85%) are caused by genetics (pre-conception) and the remainder by environmental factors or a combination of the two (multifactorial). Multifactorial or complex CDs include certain types of cancers, cardiovascular disease and other non-communicable diseases (NCD) where there is a genetic prevalence to developing specific disorders or diseases. Serious CDs may be life threatening or particularly in the absence of appropriate treatment, lead to life long physical, auditory, visual, mental or intellectual disability.

Each year globally, 3.3 million children under the age of five die and a further 3.2 million may be disabled for life as a result of serious CDs. Over 90% occur in middle- and low-income countries (MLIC) where 95% of those affected die as a result. In South Africa (SA) it is estimated that one in every 15 babies, or 70 000 live births a year are affected. Of these, 14 000 (20%) are due to Fetal Alcohol Syndrome (FAS) for which SA has the highest documented rate globally, but which are completely preventable.

In 2010, World Health Assembly (WHA) Resolution 63.17 urged member countries to prioritise CDs as a health care issue, recognizing their importance as a cause of stillbirths, and their contribution to neonatal and under-5 mortality. 'Accelerated progress in reducing neonatal mortality including the prevention and management of birth defects' was required to attain Millennium Development Goal (MDG) 4, which was not achieved in SA. As the first NCD experienced by people, CDs are encompassed in Sustainable Development Goal 3 calling, by 2030, for a reduction by one-third in premature

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mortality from NCDs through prevention and treatment and the promotion of mental health and well-being. It is now an imperative that CDs are prioritized in SA in order for this goal to be met and for human rights and dignity to be upheld so no that child is left behind.

We urge the government to establish a ministerial committee dedicated to prioritizing the health issue of CDs in order to achieve the goals of the Committee on Morbidity and Mortality in Children Under 5 Years (CoMMiC) and the Campaign on the Accelerated Reduction of Maternal and Child Mortality in Africa (CARMMA). Further issues for consideration include:

- **Estimating the true burden:** CDs are often not diagnosed or are misdiagnosed, due to a lack of awareness and the inability of attending clinicians to make appropriate diagnoses. The cause of death is often wrongly attributed, contributing to an underestimate of CDs in SA (underreporting is estimated as 88%). This lack of accurate data leads to an inaccurate assessment of the true burden of CDs. It is imperative that improved registration and surveillance systems are developed and implemented as required by WHA 63.17.
- **The proportion of deaths and disability caused by CDs is increasing:** SA is following the epidemiological trend of industrialized countries where CDs emerged and remain as the leading cause of infant and child death and disability today, accounting for 20-25% of child deaths. In SA in 2013, congenital anomalies overtook infections as the 3<sup>rd</sup> leading cause of death in neonates. When the the Infant Mortality Rate (IMR) falls below 20/1 000 live births, CDs will emerge as the leading cause of infant death in SA.
- **The prioritization of CDs to reduce child mortality further:** Rapid declines in childhood mortality stopped abruptly in SA in 2011. Further declines can only be achieved by prioritizing health issues not currently being addressed, first of which is CDs. Significant further reductions in child and infant mortality require implementing accessible, effective, relevant medical genetic services for the care and prevention of CDs. The need for such services is usually recognized when the IMR falls below 40/1 000 live births - SA is well beyond this point with an IMR of 32.8/1 000 live births in 2013.
- **Up to 70% of CDs could be prevented or ameliorated:** If a correct diagnosis and appropriate treatment were delivered, deaths and disabilities could be reduced and in some cases, prevented by 70%. Folate fortification of staple crops in SA has reduced the incidence of Neural Tube Defects by 30% and is an example of a low cost, low tech intervention reaping benefits.
- **Counting the economic cost:** The lack of early detection and intervention comes at a considerable cost. In 2012, R139.2m per month in care dependency grants was paid to 116 000 caregivers of children over one with severe disabilities or disabling chronic illnesses requiring permanent homecare. This excludes disability grants for adults living with CD caused disability and many living with CDs, such as Haemophilia and Huntington' Disease who are not considered applicable, due to lack of knowledge amongst decision makers.
- **Laws and policy framework are lost in translation:** A comprehensive legislative and regulatory framework exists in SA for the provision of medical genetic services, including National Health Act Section 21 (2) (b) (vii). However, implementation has been fragmented and unsustainable, and existing medical genetic services have deteriorated over the past decade due to competing health priorities. This shortfall in services is unacceptable and is preventing the constitutional human rights and dignity of those affected from being upheld, including the most vulnerable.
- **Policy revision cannot occur in a vacuum:** Revision of the 2001 Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities is underway. However, successful implementation will require commitment at the highest level to ensure relevant resource allocation to rebuild and develop the required services and human capacity required. The limited focus on CDs and human genetics within the Directorate for Women's

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Health and Genetics at the NDoH is inadequate for the task at hand due to competing issues (family planning, sexual assault and family planning) taking precedence.

The mission of the NDOH is to improve health status of South Africans through the prevention of illness, disease and the promotion of healthy lifestyles, and to consistently improve the health care delivery system by focusing on access, equity, efficiency, quality and sustainability. This mandate includes a commitment to reducing child mortality and morbidity, as well as the burden of CDs upon the individual, family and society in general. We believe this can only be achieved by the establishment of a Ministerial Committee dedicated to addressing CDs and thus preventing many neonatal, infant and child deaths and morbidity.

As a non-profit organization uniting patient support groups and other stakeholders relevant to the care and prevention of CDs, GA-SA is dedicated to partnering with government in such an initiative. Originally established in 1973 as the Southern African Inherited Disorders Association (SAIDA), GA-SA aims to provide support to those affected by CDs and advocate on their behalf, educate medical professionals/public to improve diagnosis and care, and facilitate relevant research.

We hope this request will be favourably received and look forward to hearing from you soon, so that together we can work towards improving the health of **ALL** South Africans. Also attached is a recent paper outlining the epidemiology in SA, published in the South African Medical Journal highlighting the emerging health crisis of CDs for your consideration.

Your sincerely,



Helen Malherbe

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