



# SAIDA

**Southern African Inherited Disorders Association**

Reg. No. 001 029 NPO

29 May 2015

To: MEC Sibongiseni Dhlomo  
1<sup>st</sup> Floor Natalia Building,  
330 Langalibalele Street,  
Pietermaritzburg, 3201

CC: L. Simelane; L. Magwaza; Z. Zondi; S. Cheattle; B. Kayonga

Dear MEC Dhlomo,

### **Letter of Support for Medical Genetic Services in KwaZulu Natal**

The Southern African Inherited Disorders Association (SAIDA) supports the plans put forward for medical genetic services in KwaZulu Natal (KZN) for the care and prevention of those affected by congenital disorders (CDs). Representing over 30 patient support groups in South Africa (SA), including thousands of children and persons living with disability, SAIDA urges the KZN Department of Health to implement these measures immediately to ensure these vulnerable groups receive relevant, accessible and effective health care as is their constitutional and legal right.

The proposed genetic services for KZN to be implemented through a network of individual units, each comprising a medical geneticist, genetic counsellor and genetic nurse, will provide a point of care for patients who desperately require diagnosis, counselling, and ongoing care and treatment. With no genetic services currently available outside of Inkosi Albert Luthuli Central Hospital in Durban, a huge percentage of the province are being denied access to essential and basic health care services.

The decentralised approach that has been approved for progressive implementation of genetic services is ideal for rectifying this shortfall. With 20% of the population living in KZN, the Provincial Department of Health has an opportunity to lead South Africa with a new model for genetic services providing an example for other provinces to follow. Initiating genetic services in KZN, regardless of where or how humble these beginnings are, will be life changing for those they are reaching and contribute to the Department's vision of 'a long and healthy life for *all* South Africans'.

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We urge you to complete this process and to appoint key individuals identified to take up the challenge of implementing these services, whether in Pietermaritzburg or Durban – to build upon the investment made to date and ensure that KZN maintains its status as a provincial front runner.

To further support our case, and to update you on the current situation of congenital disorders (CDs) in South Africa, the remainder of this letter highlights facts about genetic services for their care and prevention.

#### *Why are genetic services needed?*

Genetic services ensure that people with CDs or at reproductive risk of having children with CDs can live and reproduce as normally as possible and are key in reducing the contribution of CDs to the burden of disease. According to the *Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities* of 2001, genetic services should provide the ‘best possible patient care’ in the prevailing circumstances for those affected or at risk of CDs.

One in every 15 babies born (just under 7% of live births) in SA are affected by a CD, which totals 70,000 a year. However, up to 70% of CDs can be prevented, treated or ameliorated through relevant interventions, which do not have to be costly or high tech. Neural tube defects have been reduced by 30% in SA by folate fortification of maize, demonstrating the huge socioeconomic impact potential of a relatively minimal interventions.

Genetic services for CDs have been overshadowed in the past decade by the competing health priorities of HIV/AIDS and TB. As a result, the full extent of the health burden of CDs, including their contribution to stillbirths and child mortality, has not yet been seen in SA. However, as overall mortality is decreasing with communicable diseases being controlled, the *proportion* of deaths from CDs is increasing, revealing this previously hidden burden of disease.

#### *Child mortality and genetic services*

Child mortality has been drastically reduced in SA since the comprehensive roll out of HIV/AIDS interventions. However, since 2012 child mortality rates in SA have stagnated without significant further reductions and perinatal conditions now contribute a higher proportion of deaths. This indicates that health issues *other* than those currently being addressed require prioritisation. The contribution of CDs to neonatal deaths and stillbirths was recognised by the World Health Assembly (Resolution 63.17) in 2010 and called for their prioritisation as a health care priority.

#### *When should genetic services be implemented?*

Countries usually recognise the need for genetic services when the Infant Mortality Rate (IMR) reaches 40/1000 live births since further reductions in child mortality cannot be achieved without such measures. SA is well beyond this point with an IMR of 33/1000 (2013), and need to act before

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CDs emerge as the leading cause of death in children, which occurs when the IMR drops below 20/1000 live births. SA is following the pattern of industrialised countries where CDs emerged and remain today as the leading cause of death in children, accounting for 20-25% of all child mortality. Those that survive severe CDs are disabled for life.

This trend is already emerging in SA. In the 9<sup>th</sup> Saving Babies report using data from 2012-13 published by the Perinatal Problem Identification Programme (PPIP), congenital abnormalities were ranked as the third cause of early neonatal deaths after immaturity and hypoxia, replacing infections which dropped to 4<sup>th</sup> place. The percentage of children under 5 dying from CDs, underestimated as 4% in 2008, is already rising.

*Congenital disorders are a non-communicable disease*

Although rarely contextualised as such, CDs are the first non-communicable disease experienced by people, present from birth although some only manifest later in life. Many non-communicable diseases, including types of cancer, diabetes and cardiovascular diseases are complex congenital disorders – where individuals are genetically predisposed to develop these diseases. With non-communicable diseases accounting for 43% of deaths in SA, this growing burden of disease cannot be ignored. To prepare for this next generation of genetic services, we first need to ensure the basic services are provided.

*Genetic services: a constitutional right and a legislative obligation*

Genetic services are both a constitutional right and a legislative requirement, as stipulated in chapter 3 of the National Health Act (Act 61 of 2003), specifically section 21 (2)vii. Without such services, the human dignity and human rights of those affected with CDs cannot be upheld, as is their fundamental human and constitutional right which ensures no discrimination upon basis of disability.

Please receive this letter of support in the spirit in which is intended – by working together we can make a better life for all South Africans, including those with congenital disorders.

Yours sincerely,



Helen Malherbe

National Chair: Southern African Inherited Disorders Association (SAIDA)

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