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Comments: White Paper on National Health Insurance
Government Gazette No. 39506 (11 December 2015)

Genetic Alliance South Africa (GA-SA) is a non-profit membership organisation representing patient support groups and other stakeholders of the medical genetics community. GA-SA's mission is to improve the lives of the those affected by congenital disorders (CDs) through advocacy, education, research and support.

CDs or birth defects are abnormalities in structure or function present from birth, although they may only manifest later in life.[1] An estimated 3.3 million children under five years of age die from CDs every year and a further 3.2 million survive severe CDs with lifelong disabilities.[2] Over 90% of CDs occur in middle and low income countries, including South Africa (SA), contributing to 95% of CD related deaths worldwide. However, the true contribution of CDs to the disease burden is masked by other issues, preventing an accurate assessment of mortality and morbidity from CDs.

Specific comments on the White Paper on National Health Insurance (NHI):

1. ***Issue of progressive realisation (points 3,106, 111)***: Children's rights are fundamental human rights and thus their right to access healthcare cannot be subject to progressive realisation.[3] While it is stated that NHI will not "provide everything for everyone" as outlined in point 125, not providing health services for children affected by CDs is a violation of their fundamental human rights.

2. ***Lack of prioritisation of CDs as a health care issue***: Point 17 specifies that "vulnerable groups such as women, orphans, the aged, adolescents, and people with disabilities, women and rural communities will be prioritised". Many of these are children affected by serious CDs resulting in lifelong mental, physical, auditory or visual disability. Many CDs are misdiagnosed or remain undiagnosed, and the cause of death is incorrectly reported.[4] As a result, the true contribution of CDs to the burden of disease is underestimated and inaccurately assessed in SA. Genetic services for the care and prevention of CDs have not been developed or prioritised. It is essential that this is rectified and within the context of

[1] World Health Organization. Management of Birth Defects and Haemoglobin Disorders. Report of a Joint WHO-March of Dimes Meeting. Geneva, Switzerland, 17-19 May 2006. Human genetics programme, WHO. 2006:1-27.
[2] Christianson A, Howson CP, Modell B. March of Dimes: global report on birth defects, the hidden toll of dying and disabled children. White Plains, NY: March of Dimes, 2005.
[3] Malherbe HL, Christianson AL, Aldous C, Christianson M. Constitutional, legal and regulatory imperatives for the renewed care and prevention of congenital disorders in South Africa. S Afr J BL. 2016;9(1 MAY):11-7. doi:7196/SAJBL.429.
[4] Christianson A, Modell B. Medical Genetics in Developing Countries. Annu Rev Genomics Hum Genet. 2004;5:219–65.

point 118 that those affected or at risk of CDs are considered a "vulnerable group" in "greatest need" and "experiencing the greatest difficulty in obtaining care."

3. **Capacity building:** It is imperative that capacity building efforts as part of the NHI include medical genetics (point 20, 79). The efforts outlined in points 34 and 35 have not benefited the medical genetic services sector, which has declined as a result of competing health priorities over the past decades. The status of capacity in the sector in 2015 is compared with 2001 in Table 1.[3] A total of 27 Medical geneticists (1 per 2 million) and 95 genetic counsellors (1 per 580 000) are required today.

   While it is agreed that nurses are the backbone of the South African health system (point 37) it is essential that nursing curricula are standardised and updated, including relevant content to take advantage of genetic discoveries to improve healthcare, especially in primary health care settings where genetics training is currently inadequate. Specialised training is also required to develop genetic nurses and genetic nurse counsellors to supplement the shortfall of medical geneticists and genetic counsellors, as recommended in the 2001 Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities.

   In order for the comprehensive package of health services to be delivered as outlined in point 131, mandatory training is required to increase capacity to ensure these services can be implemented as intended.

Table 1. A comparison of medical genetics services capacity in 2001 and 2015[3]

	Recommended·	2001‡		2015	
Category	**Number/ratio (Pop=46 13m)†**	**Number**	**Ratio (Pop=44 82m)†**	**Number**	**Ratio (Pop=54 96m)§**
Medical geneticists	**20/1 per 2m**	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	9ΠΠ	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, February 11, 2016.
ΠΠ Figure increased to 9 in April 2016 plus 6 in private practice. Personal communication: T. Wessels, February 25, 2016.
** NHLS academic medical scientists only. Personal communication: H. Soodyall, July 27, 2015.

4. **Referral services:** The shortage of key health care professionals (point 79) require key specialists for the medical genetic services sector to be trained and appointed in order for the referral services outlined in points 128 and 160 to function – especially with the complete lack of medical geneticists and genetic counsellors in six provinces. Based on current capacity, "planned patient transportation between the levels of care" (point 160) will be required to transport referred patients with CDs from these other provinces to genetic services in the Free State, Gauteng and Western Cape. However, these transport costs are likely to be prohibitive, making it more cost effective to develop genetic services in other provinces for these vulnerable groups.

5. **CDs are a non-communicable disease** (NCD): CDs are the first non-communicable disease (NCD) experienced by people[5] and need to be classified as such within the context of NHI (points 96, 99). Many of the "diseases of lifestyle" referred to in point 188 are actually multifactorial CDs i.e. they have a genetic predisposition, again highlighting the need for genetic services and integrating genetics into health promotion and disease prevention.

6. **Care and prevention of CDs**: Medical genetic services need to include care (diagnosis, treatment and counselling) as well as prevention of CDs at the primary, secondary and tertiary levels (e.g. population and newborn screening). Point 393a(i) needs to ensure that those living with CDs, often disabled as a result, are not excluded. Up to 70% of CDs can be prevented or ameliorated[6] – of which 40% can be corrected through interventions (e.g. surgery of cleft lip/palate, club feet, congenital heart defects) but without these interventions they can lead to death or severe disability.[4] In addition, a further 30% can live with reduced disability as a result of early diagnosis and rehabilitation (e.g. physiotherapy).[4] Point 52 highlights the feature of "universal access" to all South Africans for promotive, preventative, curative, rehabilitative and palliative care" via continuum of care as outlined in point 129 – also applicable to those with CDs.

7. ***Diagnostic Services:*** Early and accurate diagnosis of CDs is vital for the initiation of effective treatment and disease management as well as from a cost-coding perspective. The current public health situation does not allow for efficient, timeous, accurate and cost effective testing resulting in incorrect diagnosis, or late diagnosis leading to progression of disease beyond the point where suitable initiation of medication and treatment is considered cost effective with reduced efficacy. Many CDs, especially rare diseases, are progressive, requiring early diagnosis and immediate initiation of intervention and medical management. Human resource shortages as well as a lack of skills and training coupled with unreliable and faulty technical equipment make the case for ineffective current services.

8. **Burden of disease:** The NHI White Paper excludes the contribution of CDs to the burden of disease**.** It must be noted that:

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[5] World Health Organization, Hereditary Diseases Programme. Guidelines for the development of national programmes for monitoring birth defects. Rome, Italy: The International Centre for Birth Defects of the International Clearing House for Birth Defects Monitoring Systems; 1993.

[6] Czeizel A, Intôdy Z, Modell B. What proportion of congenital abnormalities can be prevented? BMJ. 1993;306:499-503.

- The quadruple burden of disease outlined in point 96 includes child mortality and lists several NCDs but fails to specify CDs or acknowledge the genetic component of these NCDs.
- In point 98 the reduction in neonatal, infant and child (under 5) mortality is outlined but the fact that is has stagnated with no further reductions since 2011 is excluded.[7] Although SA is back in positive epidemiological transition and the Infant Mortality Rate (IMR) and Under-5 Mortality rate (U5MR) are now lower than prior to the HIV/AIDS and TB epidemics, to achieve further significant reductions in child mortality, non-HIV causes of death and disability need to be addressed.[7,8,9,10]
- The previously hidden disease burden of CDs is beginning to emerge.[10] As communicable diseases are controlled (point 40) and overall child deaths are reducing, the proportion of infant and child deaths from CDs is increasing. The 2012/2013 data from the Perinatal Problem Identification Programme (PPIP) indicated that congenital abnormalities (a sub-set of CDs) have overtaken infection as the third leading cause of death during the first week of life in neonates.[11] This proportion will continue to increase, following the trend in industrialized countries where CDs are the leading cause of death in infants and children, accounting for up to 28% of deaths in the under-5's.[12]
- Governments usually recognise the importance of developing comprehensive genetic services when the IMR reaches 30-40 deaths per 1000 live births as a means to achieve further significant reductions in child mortality. With an IMR of 28/1000 live births,[7] SA is well beyond this point.
- When the IMR reaches 20 per 1000 live births as per the target in point 43, CDs will be the leading cause of death in SA. It is essential that CDs are prioritised as a health care issue as called for in World Health Assembly (WHA) Resolution 63.17 of 2010[13], to which SA is yet to respond comprehensively.

9. **Surveillance and monitoring of CDs:** The NHI information systems (points 361-363) need to integrate data for the surveillance and monitoring of CDs. The current system administered by the DOH is underreporting by more than 97% and needs to be improved substantially as part of NHI to record cases more accurately.[9,14] This needs to be a part of SA's response to WHA Resolution 673.17 of 2010 and will also contribute to achieving NHI aim in point 107(i), "tailoring the health service to respond to local needs" by highlighting geographical clustering of CDs.

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[7] Dorrington R, Bradshaw D, Laubscher, R, Nannan N. Rapid mortality surveillance report 2014. Cape Town, South Africa: Burden of Disease Research Unit, South African Medical Research Council; 2015.
[8] Kerber KJ LJ, Johnson LF, Mahy M, Dorrington RE, Phillips H, Bradshaw D, Nannan N, Msemburi W, Oestergaard MZ, Walker NP, Sanders D, Jackson D. South African child deaths 1990–2011: have HIV services reversed the trend enough to meet Millennium Development Goal 4? AIDS. 2013;27(16):2637–48.
[9] Malherbe H, Aldous C, Woods D, Christianson A. The contribution of congenital disorders to child mortality in South Africa. In: Padarath A, King J, Mackie E, Casciola J, editors. South African Health Review 2016. ISSN 1025-1715 ed. Cape Town: Health Systems Trust; 2016. p. 137-52.
[10] Malherbe HL, Christianson AL, Aldous C. Need for services for the care and prevention of congenital disorders in South Africa as the country's epidemiological transition evolves. S Afr Med J. 2015;105(3):186–88. doi:10.7196/SAMJ.9136.
[11] Pattison R, Rhoda N. Saving Babies 2012–2013: Ninth Report on perinatal care in South Africa. Pretoria: Tshepesa Press; 2014:35.
[12] World Health Organization. World Health Statistics 2015. Geneva: World Health Organization; 2015.
[13] World Health Organization. Sixty-Third World Health Assembly. Resolution 63.17. Birth Defects. Geneva: WHO, 2010.
[14] Mtyongwe V. National birth defects data: 2006–2012.Presentation at: 15th Southern African Human Genetics Congress, Sandton, Johannesburg, 6–9 October 2013.

10. **Genetic testing:** Laboratory testing services are an essential tool in the diagnosis of CDs and are required to underpin medical genetic and PHC services. However, budget cuts resulting in inadequate staff and equipment have resulted in these services being severely compromised in recent years.[9,10] Health technology assessment in the NHI needs to ensure that adequate resources (financial and human) are allocated to ensure relevant, efficient and prompt diagnostic services can be provided.

11. **Collaboration with civil society**: Optimum collaboration between government and non-government sectors is outlined (points 188 and 328) through the establishment of the National Health Commission. However, no further details are provided on how the National Health Commission will be established and implemented to ensure this multi sectoral collaboration involving civil society. It will be vital to include the NGOs from the CD community in this consultative process to represent the interests of those affected by or at risk of CDs. More details and clarity on how this will be undertaken is required.

12. **Access to medications:** The Essential Drug List (EDL) outlined in points 337-342 relates to "80% of the most prevalent conditions in SA" (point 340). Approximately 30% of CDs are caused by single gene ("inherited") disorders. While individually these disorders are rare (hence the term "rare diseases"), collectively they are common. Those that are affected by rare diseases and other CDs that receive continuous treatment (eg haemophilia, epilepsy) require access to lifesaving medications. Clarity is required on how medicines on the EDL list are selected, qualifying criteria for entry onto the EDL, and the list of conditions that will be treated. Some conditions require treatment with a combination of different class drugs, where there may not always be cheap alternatives. In the past, entire classes of drugs have been removed based on cost criteria reducing treatment to first-line or monotherapy. This is ineffective and ultimately a false economy, since inadequate and irrelevant medication lead to an increased number of/and extended hospital stays. Consultation and further discussion is required to clarify issues of cost and demand numbers around the availability of medicines (high demand versus singular patients).

13. ***Prescribed Minimum Benefits (PMB) criteria (points 73, 74,132):*** Further clarification on how the chronic disease list will be determined is required. Currently, the PMB List covers many chronic conditions and requires updating to ensure coverage of other un-listed conditions which meet the need for chronic, continuous based care. However, it would be illogical to remove diseases currently included on this list that have been previously determined to qualify for this level of treatment and intervention.

Thank you for this opportunity to comment on the NHI White Paper.

Kind regards,

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