The Genetic Counsellors of South Africa (GC-SA) is a focus group of the Southern African Society of Human Genetics (SASHG).

Genetic counsellors are registered medical professionals that help people understand and adapt to the impact that a genetic condition has on their health and wellbeing, and that of their family, and offer information regarding prevention, management and testing (NSGC, 2006).

A genetic condition refers to a condition that results from a change or mutation in a gene(s). These are normally rare conditions and often present at birth, although many can develop in adulthood. Genetic conditions impact about 5% of the general population (Christianson et al, 2005). Complex disorders are more common and result from the combination of the interaction of genes with gene and/or the environment.

The understanding of the role of genetics in the development of disease is expanding rapidly and becoming central to disease management to prevent birth defects, provide effective treatment options, decrease the burden of diseases such as cancer and diabetes, and for prevention.

Barriers to the delivery of medical genetics include awareness, training, knowledge and genetic based services. Lack of utilisation of these services is due to a lack of knowledge of the value and availability of the support.

The knowledge of medical genetics promotes high quality primary care particularly for women and children within the reproductive continuum (Kemper A.R. et al., 2010), which would include preconception, prenatal and perinatal/newborn screening. Medical genetics is extending beyond this continuum into all primary care. With an increased understanding of the basis of genetics in disease will allow for improved detection. Early detection can improve the overall outcome through prevention and effective management (optimal medical treatment) (Whitt K.J. et al., 2016).

‘…genetics has had a large role in the healthcare of a few patients and a small role in the healthcare of many. We have recently entered a transition period in which specific genetic knowledge is becoming critical to the delivery of health care for everyone.’ (Guttmacher and Collins, 2002)

Introduction of advances in medical genetics into healthcare is complex in terms of the testing requirements, relationships between genes, environment and health, and also other aspects that relate to ethics, law and social constructs (Shelton and Whitcomb, 2015)
The role of genetic counselling includes the delivery of clinical genetic counselling services to the individual but also to health professionals and support groups, and includes other aspects such as education, teaching, research, marketing and public engagement (Kromberg J., Wessels T and Krause A, 2013)

Genetic counsellors form part of a larger clinical genetics service, which include medical geneticists, genetic nurses and genetic testing laboratories.

The inclusion of clinical genetic services is pivotal in establishing quality healthcare services for all South Africans based on their health needs.

**Comment on the he NHI White Paper:**

**Quality Healthcare Services**
The NHI has been established to ensure access to quality personal health services, which includes reproductive health. Clinical genetic services form the basis of the ‘reproductive continuum’ from preconception, prenatal, perinatal and postnatal healthcare, through appropriate counselling and screening options. Earlier diagnosis increases the possibility for improved outcome aligning with the need to detect “barriers to learning” before children attend school.

Current limited access to clinical geneticists, genetic counsellors and genetic testing options in the ‘reproductive continuum’ can directly impacts the burden of disease and mother and child mortality. (Verma IC. and Puri RD, 2015). Clinical genetic services should be included in quality healthcare services provided through the NHI.

**Comprehensive Package**
Clinical genetic services has a role across the continuum of services that form part of the “comprehensive package of health services” of the NHI Organisation, including but not limited to:
- Preventive, community outreach and promotion services
- Reproductive health services
- Maternal health services
- Paediatric and child health services to
- Health counselling and testing services
- Chronic disease management services
- Mental health services including substance abuse
- Diagnostic radiology and pathology services

Clinical genetic services, including genetic counselling, is not bound to a particular tier in the organisational structure of the NHI. The value of clinical genetic services across all tiers should be considered in the establishment of NHI healthcare structure.

**Access to Resources**
The barrier to clinical genetic services is access to resources, including medical geneticists, genetic counsellors, genetic nurses, genetic testing options and the location of these various services (Malherbe et al, 2016) Lack of resources is attributed to reduced funding for clinical genetic counselling services due to competing health needs such as the burden of HIV/AIDS. Lack of funding resulted in the decline of the number of clinical genetic professionals in practice due to limited job opportunities, and this directly impacted training opportunities reducing the number of newly qualified professionals. Migration of professionals, particularly genetic counsellors, into private resulted from necessity for employment and
not competing salary options. A similar pattern has been noted for medical scientists and employ through private laboratories. Resources are required to establish clinical genetic services.

**Disease Prevention, Health Education and Promotion**
The role of genetic counsellors includes education through outreach programmes and clinics, and training for healthcare professionals, patients and patient groups. Providing for the “continuum of care from community outreach, health promotion and prevention”. Clinical genetic services in South Africa have offered the continuum of care through community-specific programmes based on the underlying risk for genetic conditions for the particular community, such as albinism (Kromberg JR., 1997) and fetal alcohol syndrome (Viljoen et al., 2005) initiatives. Prevention is possible with knowledge of risk. Genetic knowledge of disease and assessment for disease risk can be used for both the individual and a community to ameliorate risk.

**Laboratory Services**
Laboratory services form the foundation to delivering optimal clinical genetic services. Laboratory expenditure is defined as a ‘cost-driver’ in the current healthcare system. The impact of limited access to genetic testing resulted in a reduction in genetic test opportunities and a direct impact on clinical genetic services. Establishment of new technology will improve the output of genetic testing laboratories and enhance overall clinical services. Consideration into the upgrade and establishment of clinical genetics laboratory and new testing technologies is required.

**Patient Support and Advocacy**
Prescribed minimum benefits (PMB) provide a foundation for people with rare genetic conditions to access costly and effective treatment. Orphan diseases and treatment options need to remain a priority particularly as they provide support and improve health outcomes for individuals’ that form part of what is defined as the ‘vulnerable group’ in the South African population.

**Clinical Genetic Services and Burden of Disease**
Access to appropriate clinical genetic services has a direct impact on managing the burden of disease, particularly in relation to maternal and child mortality (Christianson et al., 2005). Genetic counselling services include patient education, primary healthcare education, facilitation of genetic testing, interpretation of results and management options which optimise health outcomes, reduce the burden of disease and create prevention opportunities.

Thank you for your consideration and the opportunity to comment on the NHI White Paper.

Yours sincerely,

Noelene Kisnley
Genetic Counsellor MSc(Med)
Chair: GC-SA

Mobile no.: 082 547 5720
Email: noelene@greenmail.co.za
References:


