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COUNTRY REPORT

Genetic services and testing in South Africa

Jennifer G. R. Kromberg · Elaine B. Sizer · Arnold L. Christianson

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Abstract South Africa is a developing middle-income country with a population of over 49 million people. It has a health system, based on national, provincial and private health programmes, which is in transition. There are well organised but small genetic services, based mostly in academic centres, provincial health departments and the National Health Laboratory Service. Trained medical geneticists, genetic counsellors and medical scientists are available to deliver the service. Funding for this service is limited, due partly to the extensive demands made by the rampant HIV/AIDS epidemic (which has lead to a falling life expectancy, and increasing maternal, child and infant mortality rates) and partly due to some ignorance, among both health professionals and the public, concerning the benefits of genetic counselling and testing in affected families. There are four academic human genetics departments across the country providing counselling (7,313 cases were counselled in 2008), testing services (16,073 genetic tests were performed in 2008) and professional training. They also undertake research. Only one tenth of the required staff, according to the WHO recommendations, is available at present to provide these services, and further employment opportunities are urgently required. However, training of professionals continues, comprehensive genetic testing facilities are available, research on many of the genetic conditions of specific concern to the country has been and is being undertaken, and patients from all over Southern and Central Africa make use of these services.

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J. G. R. Kromberg (✉) · E. B. Sizer · A. L. Christianson
Division of Human Genetics, National Health Laboratory Service and University of the Witwatersrand,
PO Box 1038, Johannesburg 2000, South Africa
e-mail: Jennifer.kromberg@nhls.ac.za

A. L. Christianson
e-mail: Arnold.Christianson@nhls.ac.za

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Introduction

South Africa is a developing middle-income country and it is one of the most advanced and successful countries on the African continent. Since 1994, it has had a fully democratic system of government, reversed much of the previously discriminatory legislation, built more than a million houses and many new clinics, and increased access to clean drinking water, telephones and electricity (Benatar 2004). However, the narrowing of the disparities in health care between rich and poor is a formidable challenge for the new government, especially in the context of the pandemic of the human immunodeficiency virus (HIV) and acquired immune deficiency syndrome (AIDS), as well as the increasing prevalence of tuberculosis (TB). South Africa has one of the highest incidence rates of HIV/AIDS in the world, and this is impeding the development of the country on many levels, affecting efficiency, the maintenance of health and other services, as well as the provision of medical genetic services.

This paper presents the background data on basic demographic statistics, health indicators and health expenditure, and information on the genetic services and genetic testing available in the country.

Demography and health indicators

The total population size of South Africa was 49.32 million people at the last estimate from Statistics South Africa (StatsSA) in 2009. The population is distributed throughout the country in nine provinces as shown in Fig. 1. The original population, many centuries ago, consisted of hunter–gatherers, such as the San, and the pastoralist Khoikhoi; then the Bantu-

Fig. 1 Map of South Africa showing the provinces, major cities, and neighbouring countries (adapted from the Wikipedia map of South Africa 2011)

speaking peoples migrated into the northern parts of the country from equatorial Africa, and the Portuguese, Dutch and English established shipping supply stations and gradually settled in the south of the country. When diamonds were found in the Kimberley area and gold on the Witwatersrand in the late 1800s, there were many more migrants from Europe. At the same time, Asians, mostly from India, came to work, initially on sugar plantations, in the eastern part of the country. Also, around the turn of the century, there was an influx of Jewish immigrants. There are now many different ethnic groups and 11 official languages, but English, which is taught in all schools, is the most commonly used language in the urban areas.

The annual population growth rate for 2007–2008 was reported by Statistics SA (2009) as 0.82% and the percentage of immigrants in the population was estimated at 2.6% in 2005 (Human Development Report [HDR] 2009). The majority (60.7%) of the population were residing in urban areas in 2008 (World Bank [WB] 2010). Most (78%) of the births, between 2000 and 2007, were registered (World Health Organisation [WHO] 2009a), and the total fertility rate was 2.38 children per woman in 2009 (Stats SA 2009). Approximately 15.1% of all live births were to women over the age of 34 years.

South Africa is currently experiencing negative epidemiological transition. The average life expectancy of South Africans was estimated, in 2000, to be 58 years but this decreased to 51 years in 2008 (WB 2010a). Women have a slightly longer life expectancy of 53 years compared to 50 years for men (WB 2010a). Correspondingly, the adult mortality rate (dying between 15 and 60 years) shows an increasing trend over the last two decades, with women having a lower mortality rate (484/1,000) than men (557/1,000) (WHO 2009a). Similarly, the maternal mortality rate has increased significantly (to 40/10,000 in 2005; WHO 2009a), and it has been shown to be nearly ten times higher in HIV-positive than in HIV-negative mothers (Patrick and Stephen 2007). Consequently, South Africa is not meeting the Millennium Development Goal (MDG) for a reduction in maternal mortality (Hogan et al. 2010).

The neonatal and infant mortality rates were 20 and 48 per 1,000, respectively, in 2008 (WHO 2009a), with males having higher rates than females. The under-5 mortality rate rose from 59 per 1,000 live births in 2007 (WHO 2010a) to 67 per 1,000 in 2008 (WB World Health Organization 2010a). South Africa is one of only 12 countries in which mortality rates for children under 5 years of age have increased since the MDGs were adopted in 1990 (Chopra et al. 2009).

Although poverty and the AIDS epidemic are contributing factors to the poor mortality figures, suboptimum implementation of the necessary interventions have resulted in avoidable health system factors contributing to deaths. Moreover, many neonatal, child and maternal deaths could be avoided, particularly if HIV/AIDS-related deaths could be reduced (Chopra et al. 2009). According to Coovadia et al. (2009), South Africa has good policies in place to improve health care in South Africa. However, poor implementation

and monitoring of these policies and poor management, has resulted in variable quality of care within the public health system.

Health expenditure and financing

South Africa had a gross national income per capita of US $5,820 and International $9,790 in 2008 (WB 2010a). The per capita total expenditure on health in 2007 was given as US $490 (WB 2010a). The per capita public expenditure on health, in 2006, accounted for approximately 9.1% of the total government expenditure (WHO 2006), and this increased to 11.6% in 2010, according to the South African Institute for Race Relations Survey (SAIRR 2009). As a percentage of the gross domestic product (GDP), total health expenditure (public and private) accounted for 8.0% in 2006 (WHO 2009b). Only about 16% of the total South African population had private medical coverage in 2006 (Econex 2010). Overall, the type of expenditure that would best describe the funding of health services in South Africa is a mix between private and public funding. Approximately 57% of the population use public medical services and 43% utilise private facilities, to a greater or lesser extent (Econex 2010), but about 80% of health expenditure occurs in the private sector.

Congenital and genetic disease burden

No national surveys on serious congenital disorders (defined as structural and functional disorders and abnormalities, present from birth, which can cause death or disability, WHO 2006) and genetic disorders have been performed and no registries on these disorders are maintained in South Africa. Only data for neural tube defects have been collected systematically from 15 hospitals in the country (International Clearinghouse for Birth Defects 2005).

The total birth prevalence of serious genetic congenital disorders in South Africa has been estimated as 53.4 per 1,000 live births (Christianson et al. 2006). However, studies have been carried out on some of the commoner inherited disorders found in South Africa and the findings are presented in Table 1. In this table, the Afrikaner group refers to the members of a sub-group of the Caucasian (white) population, most of whom emigrated from the Netherlands, Germany and France, about 3 centuries ago, and speak Afrikaans (a unique language derived from Dutch). A comprehensive table showing inherited conditions of unusual prevalence among some southern African populations can be found in the report of Jenkins (1990), and inherited disorders in the black population have been described in a series of papers by Beighton and Botha (1986). More recently, founder mutations have been demonstrated in most of these conditions in South African laboratories (A. Krause, personal communication 2011).

The prevalence of some of the commoner birth defects has been studied in 29,600 black infants (Kromberg and Jenkins 1982b). This study showed that the most common abnormality was polydactly (10.4 per 1,000 births), followed by talipes (1.55/1,000), hydrocephalus (1.3/1,000), neural tube defects (1.1/1,000) and clefting (0.3/1,000). Prevalence studies on certain common chromosomal disorders showed that Down syndrome occurs in approximately one in 525 births in the whole population (A. Christianson, personal communication 2010), fragile X in one in 4,000 males (Goldman et al. 1997), trisomy 18 in one in 10,000

Table 1 Prevalence of some common autosomal dominant (AD) and recessive (AR) disorders

Disorder (inheritance)	Population prevalence	Ethnic group most commonly affected	Reference
Hypercholesterolemia (AD)	10 per 1,000	Afrikaners	Delport 2009
Porphyria (AD)	3 per 10,000	Afrikaners	Hift and Meissner 2005
Sickle cell anaemia (AR)	1 in 100 to 1 in 500	Black immigrants	A. Krause (personal communication 2011)
Gaucher disease (AR)	1 in 1600	Ashkenazi Jewish	Morar and Lane 1996
Spinal muscular atrophy (AR)	1 in 2,000[a]	Black	Labrum et al. 2007
Cystic fibrosis (AR)	1 in 3,000	White	Padoa et al. 1999
Tay Sachs disease (AR)	1 in 3,000	Ashkenazi Jewish	Jenkins et al. 1977
Oculocutaneous albinism (AR)	1 in 3,900	Black	Kromberg and Jenkins 1982a
Thalassaemia (AR)	>1 in 500 >1 in 5,000	Greek (β), Indian (α and β)	A Krause (personal communication 2011)
Galactosaemia (AR)	1 in 18,000	Black	Manga et al. 1999
Polycystic kidney disease (AR type)	1 in 26,000	Afrikaners	Lombard et al. 1989
Fanconi anaemia (AR)	1 in 26,000	Afrikaners, Black	Rosendorff et al. 1987
	1 in 40,000		Morgan et al. 2005

[a]Birth incidence

births and trisomy 13 in one in 25,000 births (Parrott 1997). For the late-onset disorders with a genetic component few data are available. However, incidence estimates for breast cancer suggest that it occurs in about one in 12 white, one in 19 mixed ancestry, one in 18 Asian and one in 50 black women (Mqoqi et al. 2004).

The various factors that increase the risk of a child being born with a birth defect include: advanced maternal age, consanguineous marriage, teratogens, unplanned pregnancy and possibly teenage pregnancies. Alcohol is a common teratogen, and about 5% of children born in the Cape mixed ancestry community have fetal alcohol syndrome (May et al. 2008). Other teratogens, associated with birth defects, include syphilis, rubella infection, warfarin, and epilepsy medications. Immigration has resulted in an increase in conditions such as porphyria variegata (from the Netherlands) (Howson et al. 2008), thalassemia (from Greece and India), and sickle cell anaemia (from West and Central Africa) (Christianson et al. 2006).

Availability of genetic services

The development of genetic services started in the 1950s. Cytogenetics laboratories were initially set up in that decade and informal genetic counselling services were initiated in Johannesburg and Cape Town in the early 1960s (for a detailed review of this early history, see Jenkins 1990). Further development occurred in 1971 when the Department of Health and Welfare appointed a Director of Genetic Services to set up community oriented genetic services and the first two academic chairs of Human Genetics were established, in 1972 in Cape Town and 1974 in Johannesburg.

In 1977, the Minister of Health announced that Genetic Services were an integral part of the health system financed by the State, under the Health Act (Act 63 of 1977). The laboratory services were then expanded, cytogenetic and biochemical testing became more readily available, demand for prenatal genetic diagnosis increased rapidly (Kromberg et al. 1989) and community screening for conditions such as Tay Sachs disease was initiated (Jenkins et al. 1977). The State introduced community education and training programmes for the health sector, particularly the nursing profession, and circulated supporting material (reviewed by Opt'Hof 1985). At the same time the academic centres in Johannesburg and Cape Town developed further and staffing, the training of appropriate health professionals and research related activities increased. New services were set up at the universities in Pretoria (in the late 1980s, although this department was closed down in 2001), Stellenbosch and Bloemfontein (in the early 1990s).

The Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and disabilities were finalised and published by the Department of Health in 2001. An additional 70 medical geneticists, 300 genetic counsellors and 300 laboratory scientists, were required to meet the needs of the population. These calculations were made using the guidelines regarding the health personnel required for an adequate medical genetics service, in first world countries, together with South African population statistics (Department of Health South Africa 2001). The number of posts supported by the National Health Laboratory Service (NHLS) and provincial governments increased in the late 1990s and early 2000s. However, the debate regarding whether Genetic Services were a function of the National Department of Health and/or co-ordinated by them or through the NHLS, or delegated to the nine provincial Health Departments, has continued and led to the slowing down of development of these services.

Key genetic services are available in South Africa for different health purposes and in different settings. These services, provided at tertiary care level, include: prenatal genetic diagnosis; diagnostic, predictive and carrier testing; and genetic counselling services. Regulated abortion services can be requested at secondary and tertiary levels. Neonatal screening for metabolic conditions is only available from a few private laboratories, pre-implantation genetic diagnosis from one private laboratory, and pharmacogenetic testing, although not generally available, is beginning to be introduced for the assessment of TB drugs. No genetic services are available in the rural areas, apart from the few outreach clinics provided by the academic centres, but referrals to the tertiary care level can be made from the secondary and primary levels. However, genetic testing is available for many of the commoner genetic disorders; a complete detailed list of these conditions and further information appears in Diagnostic Genetic Tests, South Africa (Department of Health 2007).

Although family planning clinics and contraception are provided throughout the country, there is very little genetic preconception care. However, couples at risk due to consanguineous marriages, or a genetic disorder in the family, or their population group of origin, can seek genetic counselling. Some special care facilities have been established for children with congenital disorders, mainly in the urban areas. Also, disability grants are provided for parents caring for severely affected children at home, and a few special education and rehabilitation facilities are available, mostly in the cities.

There were five genetic service facilities in South Africa in 2008. The largest was in Johannesburg (at the NHLS/University of the Witwatersrand), and the others were at the Universities of Cape Town, Stellenbosch, Free State (in Bloemfontein) and KwaZulu Natal (in Durban). Additionally, informal genetics clinics are held by paediatricians with a special interest in human genetics at the Universities of Pretoria and Limpopo (in Polokwane). The major academic departments also provide outreach services to some of the

smaller cities and towns in the rural areas. Most of the genetic counselling services available are provided in the tertiary level public health care domain (88%), while the remainder are provided in private healthcare settings (Kromberg et al. 2009).

Laboratory services are offered mainly by the NHLS, which is responsible for the provision of pathology services, including medical genetic laboratory services, for the public health sector. The private health sector can also utilise these services. The NHLS runs 265 laboratories in cities and towns across the country (National Health Laboratory Services 2009), but most genetic tests are performed at the laboratories attached to academic centres. Six different private commercial companies, operating laboratories in four of the nine South African provinces, offer a limited number of genetic tests. While some South African laboratories provide some genetic testing services, mainly for sickle cell anaemia and chromosomal analysis, for patients in other African countries, such as Central African Republic, Uganda, Kenya, Zimbabwe, Botswana and Namibia, where the necessary expertise is generally unavailable.

Access to genetic services

There is no compulsory health insurance system in South Africa. However, all pregnant and breastfeeding mothers, as well as children under the age of 6 years, are entitled to free health care through public hospitals. Other patients have to pay a fee which is determined by a means test. The same applies to genetic services, which are accessible through tertiary hospitals. Those few patients, who access genetic services in private practice, generally have medical insurance and are reimbursed (depending on scheme and plan).

The various barriers that reduce access to genetic services include: geographical barriers and long travelling distances, together with poor public transport to the cities; financial barriers, particularly where a genetic counselling session and/or tests are too costly for the patient; the lack of awareness and ignorance of both health professionals and the public regarding the available genetic services and their value; and cultural barriers, particularly related to fatalistic attitudes to health (Kromberg and Jenkins 1997). Also, many patients use both Western medical and traditional healer services and in the process may reject genetic services. However, there are no legal constraints which reduce the availability of genetic services and the abortion law is quite a liberal one so that prenatal diagnosis (PND) can be performed at any time during pregnancy, if necessary.

Access is also limited due to the shortage of posts and staff to provide the service. Financial support from public funds for genetic services is poor partly due to the Department of Health having other health priorities, particularly the HIV/AIDS and emerging TB epidemics, and probably also due to an element of ignorance regarding genetic services, their cost-effectiveness and benefits for affected individuals, their families and community (McAllister et al. 2010). Many at risk people, who are not aware of their risks and therefore not demanding the service, could benefit from them were services more widely marketed, better staffed and accessible.

State of genetic services

Human resources and training

The latest estimate, regarding physicians, is that only about 24,147 doctors, general practitioners and specialists, were active in the country in 2009 (Econex 2010), and 34.9% of posts for medical practitioners in the public health service were vacant in 2008 (Health Systems Trust [HST] 2008). Furthermore, there were only 104,571 nurses who were active (Econex 2010) and 40.3% of the posts in the public health sector were vacant in 2008 (HST 2008).

Presently, four medical schools in the country have medical genetics professionals on their staff and medical genetics is integrated into the student curricula to a varying extent. Medical students are also trained at three other universities, but medical genetics teaching at these universities is limited and often falls to clinicians in various non-genetic specialities, particularly paediatrics.

Medical genetics was initially recognised as a subspeciality, and in 1999 nine medical geneticists were registered, through a grandfather clause, under this system. Subsequently nine more medical specialists (mostly paediatricians) undertook the newly introduced 2-year medical genetics training and were registered as medical geneticists. However, in 2007, medical genetics was recognised as a primary speciality in medicine in South Africa. Specialist training (over 4 years) towards a post-graduate MMed degree and Fellowship of the College of Medical Genetics is currently offered at four universities. At present, there are 11 medical geneticists, registered with the Health Professions Council of South Africa (HPCSA), of which ten are in academic practice, and seven registrars are in training. This number means that there is one medical geneticist for 4,450,000 individuals, compared with the estimates of one per 3,700,000 for other developing countries and one in 222,000 in Western developed nations (Wertz et al. 1995).

Genetic counselling in South Africa is a recognised and registered health profession with formal post-graduate training at the Master's degree level (requiring 2 years of full-time formal teaching and clinical training, a research project, and a 2-year internship) and registration with the HPCSA. According to the HPCSA (2009), there were 15 registered genetic counsellors in South Africa in 2008. Most were in full-time genetic counselling posts in Johannesburg, but a

few were in part-time provincial or university posts, with NGOs, private laboratories, or in private practice.

Medical scientists can be trained in human genetics if they complete a BSc (Honours) degree in a biological science and a 2-year internship, in a recognised Human Genetics setting, and become registered by the HPCSA (HPCSA 2010). Similarly, medical technologists need to complete a 3-year National Diploma or BSc degree in Biomedical Technology and a 1-year internship, and then register with the HPCSA.

Informal ad hoc genetic training of various health professionals, particularly nurses, has been offered by academic genetic professionals since the early 1980s. However, since about 2003, the Southern African Inherited Disorders Association (initially with funding from the March of Dimes, USA), based at the University of the Witwatersrand, has supported and provided medical genetics education for selected nurses and doctors working in primary health care. This Medical Genetics Education Programme (MGEP), involving distance learning as well as tutorials, was later approved and then financed by the Department of Health, and, at present, it is offered in seven provinces.

Workload

There are now four academic medical genetics departments (previously five in 2008), which are regionally distributed throughout the country, and three universities with no such department but offering a limited service for children with congenital disorders, using paediatricians with an interest in the field. The largest academic department is the Division of Human Genetics, NHLS and University of the Witwatersrand in Johannesburg, Gauteng Province. In 2008, the clinical unit staff included five medical geneticists, two experienced medical officers, and seven genetic counsellors. There were also molecular, serogenetic and cytogenetic service laboratories with about 15 registered medical scientists, three medical technologists and eight medical scientist and technologist interns working on diagnostic genetic testing for many different genetic conditions. Six medical scientists were involved purely in research. The other universities provide for similar, but smaller, clinical and laboratory services.

The National Department of Health has a small section, in the Division of Maternal and Child Health, responsible for Genetic Services. The four staff members have a limited budget to undertake community education, produce educational material and set up policies for Genetic Services, and they work in collaboration with the academic departments of Human Genetics.

The workload in most academic units is heavy, as all are short of qualified staff. In total, in 2008, there were ten medical geneticists, ten genetic counsellors and 42 medical scientist/technologists to provide services to 49 million people. This is totally inadequate, according to the standards set up by the WHO (2006). The number of genetic consultations per health care professional is difficult to estimate retrospectively, due to the variety of academic duties (which include teaching, research and administration, as well as consultations, which may range from screening children for diagnostic purposes at special schools to in-depth hour-long genetic counselling sessions) undertaken by genetics staff members. If the consultations of the four academic units (medical geneticists and genetic counsellors full and part time) are summed, for the year 2008, the total is 7,313 consultations performed by approximately 18.5 clinical staff giving an estimated workload of 395 consultations per professional. However, genetic consultation workloads vary greatly in the different universities. Genetic counsellors undertake approximately 50% of the genetic consultations in Johannesburg and Cape Town, where they are available (Kromberg et al. 2009).

Genetic testing

There are 12 main laboratories, with academic affiliations, performing genetic testing, on a service basis, for chromosome, biochemical and/or molecular diagnoses. The full range of medical genetic tests offered in the academic and public domain is documented in Diagnostic Genetic Tests, South Africa (2007). There are also various private laboratories, where some genetic tests are offered (cytogenetic, molecular and metabolic testing and postnatal metabolic screening), located around the country, mostly in the provinces of Gauteng, KwaZulu Natal, and the Western Cape. These services are used mainly by private patients covered by medical insurance. One such example is the private cytogenetics laboratory run by Lancet Laboratories in Johannesburg, which completed 3,292 tests in 2008 (1,522 were amniotic fluid samples) with three medical scientist staff and three medical technologists (Rosendorff, personal communication, 2010). However, no data are available regarding the genetic tests undertaken in other private laboratories.

The number of genetic tests performed in the country is difficult to estimate, since testing is not centralised and many laboratories in various academic departments, as well as in public and private settings, offer testing for various genetic conditions. However, in the four academic Human Genetics departments with genetic diagnostic laboratories 16,073 tests were performed in 2008.

For example, in 2008, at the Division of Human Genetics laboratory at the NHLS and University of the Witwatersrand, Johannesburg (the biggest genetics laboratory in the country), 12,359 tests were carried out: 7,420 in the biochemical and molecular laboratory (paternities, molecular tests for common chromosome aneuploidies, using Quantitative Fluorescent-Polymerase Chain Reaction (QF-PCR) techniques, and biochemical tests, e.g., screening for disorders common in the Jewish population); 2,870 in the

cytogenetics laboratory; 1,716 in the molecular laboratory (e.g., for fragile X, cystic fibrosis, spinal muscular atrophy); and 353 for genetic ancestry testing in the Human Genome Diversity and Disease Research Unit. These laboratories are referral centres and samples for the rarer genetic disorders, and for tests not offered elsewhere, are received from all over the country. About 22% of the molecular tests were tests which were set up only once for a rare and specific condition. The other academically based laboratories across the country perform fewer tests for fewer diagnoses, but several have expertise in specific conditions. One such laboratory at the University of Cape Town has a special interest in testing for spinocerebellar ataxias and a research interest in retinal diseases and colorectal cancers, and another specialises in testing for porphyria variegata.

Some quality assessment schemes are available and the academic departments of human genetics are exposed to regular peer reviews and voluntarily undertake external quality assessment by organisations such as the College of American Pathologists. The NHLS laboratories, doing genetic testing in academic and tertiary settings, are also required to be accredited by the South African National Accreditation System (SANAS).

The number and demand for tests, particularly on the molecular, but also on the cytogenetic level, is increasing. However, expansion and new developments are being retarded by the training of too few medical scientist and technology students to meet the need (especially in cytogenetics laboratories across the country), by the freezing of vacated posts, by the resulting lack of experienced staff, poor job opportunities and career paths. The lack of maintenance and updating of equipment is also problematic, and has resulted in an inability to offer state of the art testing, such as array based cytogenetic analysis.

Prevention of congenital and genetic disorders

Some attempts have been made towards prevention of congenital/genetic disorders, and due to concerted lobbying, South Africa has had fortification of basic foods, such as bread and maize meal, with folic acid, since October 2003. Consequently, the prevalence of neural tube defects has fallen by 30% (Sayed et al. 2008). Also, salt has been iodised for many years, preventing most cases of postnatal iodine deficiency disorders and goitre (such disorders were never very common in South Africa).

Cultural and social issues pertaining to medical genetic services in South Africa

There are a several specific cultural and social issues relating to genetic services in South Africa (documented by Kromberg and Jenkins 1997). The first issue is associated with systems of thought, prevailing fatalistic attitudes, communal decision-making, the indistinct line between life and death, and belief in the power of ancestral spirits. The second concerns the beliefs and myths about the causes of genetic disorders. The third issue is the tendency, in the majority of people, to consult both Western medical health professionals and traditional healers. The fourth concerns the custom of consanguineous marriage, common practices and taboos, while the fifth issue is about language and communication, since in most local languages there are no terms for words such as genes and chromosomes. All these issues can affect the ways in which genetic services are delivered and received, the communication and interactions in genetic counselling sessions and the choices people make.

National policies and legal frameworks

The National Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (Department of Health 2001) document provides recommendations for the provision of genetic services. It also has sections on general ethical guidelines for medical genetics (modified from the WHO Hereditary Disease Programme 1995, document) and on ethical principles for genetic professionals (from Baumiller et al. 1996). Ethical guidelines for genetic research purposes are also available (MRC, South Africa 2002)

In the existing National Policy Guidelines the priority medical genetic services are described (Department of Health 2001). These include services offered prior to conception, during pregnancy, at birth, in infancy and childhood, and in adolescence and adulthood. The way in which these services could be delivered at various levels from primary to tertiary health systems, is covered. The education of learners at schools and the training of genetic health professionals is also addressed. Recommendations that medical geneticists' and genetic counsellors' posts should be provided, urgently, in every province in the country, in order to offer the services, have not yet been acted upon, and most provinces still have no posts at all.

Interventions are described, including strategies for prevention, such as genetic counselling, preconception and prenatal methods of prevention (e.g., PND), postnatal diagnosis and population screening. There are recommendations regarding: the integration of medical genetics laboratory services into the NHLS, which has been partially achieved, although co-ordination is still poor and duplications and inconsistencies still exist; the composition and functions of a Medical Genetics Advisory Board (it was suggested medical genetics professionals as well as a lawyer should be included); and the evaluation of human genetics programmes.

International Conventions and Directives, such as the European Convention on Human Rights, acknowledge that there are basic human rights for patients with genetic

conditions and that everyone is entitled to basic health care (Convention on Human Rights and Biomedicine of the Council of Europe, 1997). In line with these international standards the South African Constitution of 1996 provides not only for fundamental rights such as the right to life (Section 11 of the constitution), to equality (Section 9), dignity (Section 10) and privacy (Section 14) but also provides, in Section 27, that everyone has the right to have access to health care, sufficient food, water and social security.

In terms of the National Health Act (NHA) of 2003 the State is obliged to provide free health care services to pregnant and lactating women and children under the age of 6 years. Furthermore, free primary health care services must be provided to all those who are not members of medical aid schemes. Section 4 of the NHA also provides authority for women to have access to free termination of pregnancy, subject to the Choice on Termination of Pregnancy Act 92 of 1996. This Act provides the conditions and procedures to be followed for a person to obtain a termination of pregnancy. It states that a woman may obtain a termination upon request in the first 12 weeks, and thereafter, in consultation with a medical practitioner, where the health of the mother or the fetus may be at risk.

The NHA (2003) has clear provisions in Section 7 for consent to medical treatment and, with a few exceptions, a health care service may not be provided without informed consent. Further, Section 8 of the NHA deals with the control of the use of blood products, tissue, and gametes and zygotes in humans and prevents the reproductive cloning of human beings.

Assessment of the attention given to medical genetic services by the national government/policy makers as compared to other health issues

The national policy regarding medical genetic services, which was set out in the National Policy Guidelines (Department of Health 2001), was developed before the impact of the HIV/AIDS pandemic became apparent. That epidemic together with other problems, including the increasing incidence of TB and difficulties in health service delivery, appears to have resulted in medical genetic services having lesser priority recently than in the 1990s. It is hoped that this situation will improve consequent on the recognition, by the WHO, that birth defects present major health problems, and, in 2010, their recommendation that services for the care and prevention of congenital defects in developing countries should be prioritised (WHO 2010b).

Research priorities in genetics/genomics

There are no policies specifically covering funding for research in human or medical genetics/genomics. However, the government finances some medical research, including research in the field of human genetics/genomics, through the *SA Medical Research Council (SAMRC), the National Research Foundation (NRF), the National Department of Science and Technology (NDST), and the Research Trust of the NHLS. The NDST, through its biotechnology strategy, has funded two Biotechnology Regional Innovation Centres (BRICS), which are high-throughput genomics laboratories, the National Bioinformatics Network and, recently, the planning phase, of a National Human Genome Initiative.

Research funding is received from various private sources, e.g., the Cancer Association of South Africa, for short term projects (generally) on an ad hoc or regular basis. Local universities also fund selected research projects undertaken by their staff. Further, South African researchers in human genetics collaborate with international research teams and have received international funding from various bodies such as the National Institute of Health (NIH, USA), Fogarty Foundation, WHO, Wellcome Trust, US Aid, Center for Disease Control (CDC), the March of Dimes (a USA non-governmental organisation), the European Union through the Platform Frameworks, and the Genographic Project of the National Geographic Society.

There are two initiatives which are noteworthy at a national and international level. The South African Human Genome project has been initiated by the NDST to develop research capacity in the area of human genomics. The scope of the project is currently being discussed by stakeholders nationally. This programme, timeously, has the potential to interact with a commitment of the National Institute of Health in the USA and the Wellcome Trust (UK) who have invested in the recently launched H3Africa (Human Heredity and Health: Africa) initiative (http://www.nih.gov/news/health/jun2010/nhgri-22.htm). The latter programme is an effort to provide the basis for a network of research activities, across the African continent, focussed on the issues of human health, environment and human genome variation. This is an unprecedented commitment by international funding agencies to work with African and non-African researchers, in genomics and public health, towards understanding predisposition to disease, and potential targeted interventions, for the long-term benefit of the people of Africa.

Patient organisations and public education in genetics

Parent/patient organisations and support groups, structured as non-governmental organisations, for people with many different genetic disorders are available in South Africa. The Southern African Inherited Disorders Association (SAIDA) is

an umbrella body for such groups. SAIDA is hosted by the Division of Human Genetics, University of the Witwatersrand in Johannesburg and financed by donations. The objectives of the Association are to: raise awareness about genetic conditions; offer support to affected families; stimulate and support research; train support parents; make appropriate referrals; advocate on behalf of members and lobby government members for better services; and network across the country and internationally.

Public education, covering recognition and prevention of congenital disorders, takes place at many levels. Lay public groups, such as Rotary clubs, parent and women's groups, have talks from human genetics professionals when they request them. Medical geneticists and genetic counsellors give educational lectures to various health professional groups, to registrars in other medical specialities (e.g., paediatrics) and lay groups (Kromberg et al. 2009). They also compile leaflets for distribution on a number of common disorders (e.g., genetics of breast cancer), as well as on prenatal genetic diagnosis and genetic counselling services. SAIDA distributes educational leaflets and puts out an annual educational newsletter, which is sent to many lay groups as well as professionals.

Conclusion

South Africa is a country in transition. One of the greatest challenges it faces is the control of the concomitant HIV and TB epidemics. In 2007, the country, with 0.7% of the world's population, had 17% of the global burden of HIV infection, and one of the world's worst TB epidemics, compounded by rising drug resistance and HIV co-infection (*Lancet*, paper 3 abstract, 2009). South Africa is currently underperforming in its efforts to control HIV and, although it has the resources and capability to rise to these challenges, it has not been able to deliver on the four priorities listed in the Strategic Plan for South Africa for HIV/AIDS (*Lancet*, paper 3, 2009). Since medical genetics services are, presently, being developed and delivered against this background, it is not surprising that the recommendations in the Policy Guidelines for the Management and Prevention of Genetic Disorders (Department of Health 2001) have not been met.

However, a medical genetics service was developed in the country prior to the onset of the HIV/AIDS and TB epidemics. At present, this service provides an excellent base on which a more comprehensive service can be built in the future. There are four academic departments of human genetics which have already set up clinical services, compatible with any in developed countries, as well as laboratory services, which have the expertise to offer genetic testing services to the country and to the rest of Africa. They have also developed a research capacity so that local genetic disorders can be investigated on many different levels. Two of these universities, in collaboration with the NHLS, have established training for all the categories of expert staff required to run a sophisticated genetic service. However, the aging of laboratory equipment and lack of financial support for the introduction of new technology are making it difficult for these capable scientists to keep up to date with new developments in the field.

The commitment to provide clinical genetic services in South Africa has declined over the last few years. There is continued discussion as to whether these services should be centrally co-ordinated from the National Department of Health and provided by the provincial Departments of Health, as occurs in the Western Cape and Free State, or whether they should be all be provided by the NHLS, as occurs in Gauteng. Until this issue is resolved or another solution found, clinical genetics services will not improve and are at risk of deterioration. Laboratory genetic services are also at risk from increasing demand for tests in the face of reducing staff availability. These issues will be considered by the newly appointed NHLS Expert Committee for Medical Genetics Services, and it is hoped that solutions to the situation will be found.

Although expansion is difficult in the current circumstances, members of the South African human genetics community are networking with interested people in the rest of Africa and, in March 2011, the first combined congress was organized by SA Society of Human Genetics (SASHG), in conjunction with the African Society of Human Genetics (AfrSHG), and held in Cape Town. At this meeting there were international experts, as well as those from Africa, sharing their expertise, insights and needs, which will both benefit and stimulate the field of human genetics on the African continent in future. At this stage there is some hope that the investment by international and local agencies in, for example, the SA Human Genome project and the H3Africa programme, may raise the profile of Medical Genetic Services and contribute to their upgrading.

Development of medical genetic services, in the near future, will depend partly on increasing the awareness of genetic disorders, partly on lobbying the decision-makers in the Health Departments and NHLS, partly on the control and lessening of the HIV/AIDS epidemic, and mostly on the provision of more employment opportunities for qualified professionals. At present, the available staff can only meet about 10% of the country's genetics needs (based on a rough calculation of the genetic burden of disease). Further technological development (together with purchase of the necessary laboratory equipment) should be planned for, so that South Africa can approach the level of developed countries. Both political will and financial commitment are required to move this enterprise forward. Pressure is being brought to bear on key members of the National Department of Health and its provincial subsidiaries by Medical Genetics professionals, as

well as by genetic support group representatives, to respond to the basic genetic needs of South Africa and make an adequate and appropriate Medical Genetic Service available.

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