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GENETIC COUNSELING IN SOUTH AFRICA

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INTRODUCTION—BACKGROUND

South Africa, although classified as a middle-income country, is the richest country in Africa. It is a country with many peoples of different origins, some ancient and some modern, many cultures, and a multitude of different natural resources. It has a democratically elected government, several large, thriving cities, with extensive urbanization, a large rural population, and much migration between rural and urban communities. It is a country in transition at all levels, including the healthcare systems and services: "South African society is fluid and undergoing rapid social changes unparalleled in the rest of Africa" (Binka, 2007). There are gaps in health itself, in health research funding, and in access to information concerning health conditions (Wall, 2007), which are typical of developing countries. Fifteen years after the political liberation of South Africa, the country is struggling to live up to the ideals articulated by those with a vision, including Nobel laureates Nelson Mandela and Desmond Tutu (Tollman & Kahn, 2007). There are many difficulties in building a stable system of government and a social environment responsive to the needs of the whole community.

There are 49.32 million people living in South Africa (estimate as of mid-2009: www.statssa.gov.za). The majority (79%) of the population is Black, and the major ethnic groups include the Nguni (e.g., Zulu and Xhosa) and Sotho-Tswana. A further 9.6% of the population is White, mainly of Dutch or English origin, and the remainder (11.4%) is of mixed ancestry, and/or Indian or Asian. There are 11 different official languages and many different religions. About 14% of the people have no education, but 32% have completed their schooling and have a Grade 12 certificate. There are many disparities in access to healthcare, especially between the private and a state health sectors, and staff shortages are exacerbating the problem. South Africa also has one of the highest HIV-positive rates in the world, and the HIV/AIDS epidemic places a significant burden on the country's health budget and the provision of healthcare services (Doherty & Colvin, 2004). According to official estimates, as of mid-2009 (www.statssa.gov.za), there are 5.2 million people living with HIV/AIDS in South Africa and the overall HIV positive prevalence rate is 10.6% of the population. Life expectancy is 53.5years for males and 57.2 for females, while infant mortality is 45.7 per 1000 live births.

There is still much poverty in the country and, in 1994, 6.3% of the population were living on less than US$1 per day, while in 2005 this number had risen to 11% (Editorial, *Lancet*, 2008). In addition, rates of under-5 mortality have increased since 1990. Many of these childhood deaths are due to the high prevalence of HIV-positive individuals in South Africa. Almost one-third of all pregnant women are HIV positive and, in 2003, HIV-related infection caused more than half the deaths in children aged below 5 years. Another major problem is that one in seven nurses and nursing students are HIV+ and there are more nurses with full-blown AIDS than are being trained (Bateman, 2008), which could shortly cripple the health system. However, some progress is being made and in 2001 almost 21% of 15–49-year-old people were HIV+, whereas in 2005 this figure was about 19%. Life expectancy is dropping and morbidity increasing, due to the epidemic. Nevertheless, access to safe water and sanitation has improved, school enrollment is high, and the difference in literacy rates for boys and girls is negligible (Editorial, *Lancet*, 2008). Also, the number of Internet users increased from 0.83% of the population in 1996 to 10.75% in 2005 and the number of mobile phone users has risen astronomically. These are encouraging steps in South Africa's development.

South Africa is bordered on the northwest, north, and northeast by several countries. These include Namibia, Botswana, Zimbabwe and Mozambique, while Swaziland and Lesotho are landlocked and surrounded by South Africa. These countries and their citizens often seek help and expertise in healthcare (particularly in the field of human genetics) from South African experts, and migration to the country, particularly from Zimbabwe due to its present crisis situation, is extensive.

EPIDEMIOLOGY OF COMMON GENETIC CONDITIONS

It is widely recognized that in the world population, about 6% of all infants born have some degree of congenital or birth defect, and that these defects are responsible for many neonatal deaths (Christianson & Modell, 2004). Furthermore, it is estimated that one person in ten will experience a multifactorial disease in her/his lifetime. The types of genetic disorders in a community depend on the genetic profile of that community, some being common to all the ethnic groups in the community and some occurring at much higher rates in specific ethnic groups (Beighton & Botha, 1986a,b,c).

In South Africa the more serious common monogenic or single-gene disorders that occur in all groups include Huntington disease (Hayden, 1981; Krause & Greenberg, 2008), Marfan syndrome, myotonic dystrophy, neurofibromatosis (Beighton et al., 1991), polycystic kidneys, tuberous sclerosis, and osteogenesis imperfecta, among others (Beighton, 1976). In addition, those that occur at higher rates than usual in specific groups include: oculocutaneous albinism in the Black population (Kromberg & Jenkins, 1982a); cystic fibrosis in the White groups (Denter et al., 1992); Tay Sachs and Gaucher disease in the Jewish population (Jenkins et al., 1977; Goldblatt & Beighton, 1979); familial hypercholesterolemia and porphyria in the Afrikaans (of Dutch origin) population (Jenkins, 1990); beta thalassemias in the Greek, and various thalassemias in the Indian populations (Krause, 1994). The X-linked disorders, such as hemophilia, Duchenne muscular dystrophy, fragile X syndrome, and some inherited forms of retinal degenerative disease (Greenberg et al., 1994) appear to occur in all populations at more or less the same rates as elsewhere.

The multifactorial or polygenic disorders have also been found in all the population groups at varying rates. These disorders include the neural tube disorders, talipes equinovarus, isolated hydrocephalus, facial clefts (Kromberg & Jenkins, 1982b), epilepsy (Christianson et al., 2000), and intellectual disability (Kromberg et al., 2008). Fetal alcohol syndrome, however, occurs at unusually high rates in specific areas of the country where the predisposing environmental factors predominate. For example, it is found in 6%–7% of children in the Western Cape, and at similar levels in the Northern Cape province (Viljoen et al., 2005).

Chromosome disorders are found at varying rates in different ethnic groups. Down syndrome (Smart, 1981; Kromberg et al., 1992) and trisomies 18 and 13 occur more frequently, as expected, in the infants of older women in all groups, as has been observed worldwide. Sex chromosome abnormalities, such as Klinefelter (XXY) and Turner (XO) syndrome, as well as triple-X syndrome also occur.

While knowledge of the inherited cancer syndromes is increasing in South Africa, few epidemiological data are yet available. However, cases of inherited colorectal cancer —such as hereditary nonpolyposis colon cancer (HNPCC) and familial adenomatous polyposis (FAP); Stupart et al., 2008; Anderson et al., 2007; Goldberg et al., 2000; Ramesar et al., 2000) and breast cancer—have been observed in all groups. Data on a variety of cancers are collected by the National Cancer Registry of South Africa, and in the case of breast cancer (the commonest cancer in women) in general, the incidence is highest in White women (lifetime risk 1/12) followed by Asian women (risk 1/18), those of mixed ancestry (risk 1/18), and Black (risk 1/49) women (Mqoqi et al., 2004). The incidence of colorectal cancer in the Black population appears to be reduced, but at least two cases (one Zulu and one Xhosa family) have been reported (Grobbelaar et al., 2002).

LOCAL BELIEFS ABOUT GENETIC DISORDERS

Cultural transformation has been taking place in the country for centuries, particularly since the influx of foreign traders and explorers in the 1400s. However, when modern health services are offered to developing communities, who, despite adopting some of the ways of the foreign culture still retain many of their own cultural ways, many problems may ensue. It is therefore essential for the providers of the new services to have a good understanding of the cultural influences on the perception of such services—in this case genetic services. Some of these influences include: ideas about disease causation; common taboos and practices; myths and superstitions; use of traditional healers; systems of thought; and language issues (Kromberg & Jenkins, 1997).

Causes of disease are not easily understood in any developing population with limited scientific insight. Ideas about causation are, therefore, often associated with traditions and myths and old wives tales. Ancestor worship is common in various South African ethnic groups, and transgression of the rules is believed to lead to ill-health in the transgressors and/or their child. Such ill-health can include congenital abnormalities in the offspring. For example, 51% off the rural traditional healers interviewed by Kromberg et al. (2008) considered the cause of childhood disabilities to be the breaking of taboos.

The different local ethnic groups have differing taboos about abortion, but in general, especially previously, they were totally opposed to it. One proverb stated that "to force out the womb is grievous, and the knot of the cradle skin is a flower"—i.e., women derive great pleasure from carrying a baby slung on the back (Schapera, 1940). This taboo, however, did not apply to newborn infants, and infanticide and euthanasia were practiced, historically, with regard to defective children and sometimes one or

both twins (Jeffreys, 1953). Livingstone (1857) described the case of a mother who was excommunicated from her village with her albino child until she agreed to put him to death; she was then allowed to return. Generally, the midwife smothered or did away with the abnormal child and told the father it was stillborn, and if the chief became aware of the case it was never prosecuted (Schapera, 1940). However, these attitudes and taboos are changing and moving toward acceptance; abortion in certain circumstances (such as a severe genetic disorder in the fetus) is becoming acceptable, and if infanticide is still occurring, it is carried out in secret.

Another common practice with relevance to genetic services is that associated with mate selection. In several groups, the preferred mating was with a first cousin, such as a mother's brother's daughter (Krige, 1937). Such marriages might be motivated by the keeping of the bride-wealth (often cattle) or dowry in the family, keeping the power in the family, the gaining of political benefits, or greater family cohesion or solidarity (Bittles, 2008). However, locally these cultural practices affect the rates of the common recessive conditions such as oculocutaneous albinism, and this condition is found at higher rates in groups that favor consanguineous marriages than in those for whom this practice is taboo (Kromberg & Jenkins, 1982b). Polygamy and polyandry are also practiced locally, with both advantages and disadvantages. Polygamy might alter the sex ratio, with more females being born (Miller, 1993) and sequential polyandry might mean that a woman would seldom have two children with the same recessive disorder.

Myths and superstitions have been encountered with regard to genetic disorders, and one of the most prevalent is the widely held belief that albinos do not die (Kromberg, 1992). This myth affects the acceptance of people with albinism in the community, and contributes to the anxieties of mothers who give birth to infants with albinism (Kromberg et al., 1987). The genetic counselor needs to be aware of such myths if the genetic counseling is to be culturally relevant, sensitive, and appropriate for the client (Kromberg & Jenkins, 1984)

Traditional healers are still powerful in the local community, and many people use the services of both Western medical doctors and traditional healers (Freeman et al., 1994). These healers believe that childhood disabilities are caused by breaking taboos (51%) or fate (12%), using contraception (4%), inheritance (4%), or consanguinity (3%) (Kromberg et al., 2008). However, they do influence the way in which people see the causes of disease and seek treatment. Their influence needs to be taken into consideration by those offering services, since, for example, facts given in genetic counseling can be disregarded by the patient if the traditional healer gives a different opinion.

Systems of thought in the Black population appear to favor collective rather than individual thinking (Hammond-Tooke, 1989). This system may be accompanied by an external locus of control, and the social schema of the body may predominate over the individual schema. A balance between the individual and sociological schemata of the body is essential for competent and self-steering behavior (Manganyi, 1973). If there is imbalance, slow decision-making processes may occur and individuals may be reluctant to make any decisions or to act without consulting another person.

Another philosophy related to this worldview is that of fatalism. One of the major components of culture involves the complex of beliefs which surround the relation of man to nature, man to his fellow man, and man to the supernatural powers that are believed to control the universe (Read, 1966). If this control is believed to be absolute, then individuals cannot change what happens to them. Kuppermann et al. (2006) reported that African-American women have higher levels of fatalism than women from other ethnic groups. Furthermore, in another U.S. study, higher scores on a fatalism scale were found to be associated with lower willingness to request termination of pregnancy when the fetus had a severe abnormality (Learman et al., 2005). This philosophy has many implications for genetic counseling, since, if patients do not make decisions or act on the genetic information given, for fatalistic reasons, they may not benefit fully from the knowledge and choices available to them.

INTERNATIONAL DEVELOPMENT OF GENETIC COUNSELING

Genetic counseling is a young and growing healthcare profession available mostly in first-world countries. It has emerged as the Human Genome Project gained publicity, and as the genetic causes of many disorders became understood and recognized such that their prevention and, in some cases, even treatment, becomes a possibility.

The term *genetic counseling* is broadly defined as a dynamic psychoeducational process centered on genetic information (Biesecker & Peters, 2001). The goal of this process is to facilitate the client's ability to use genetic information in a personally meaningful way, so that choices become available, psychological distress is minimized, and personal control is maximized. Genetic counseling aims to preserve client autonomy, self-directedness, competence, and feelings of worth, and to be client-centered and nondirective (Kessler, 1979).

The profession emerged partly in response to the fact that people were living in the community with a genetic disorder in the family, but a specific informed health professional was not available to counsel them. Also, it was recognized that about 1 in 10 people will have a genetic disorder in their lifetime, and many of these people will request and make use of accessible genetic services.

Genetic advice about inherited traits was offered as early as 1906, soon after Bateson suggested that the study of human heredity be called "genetics" (Walker, 1998). The first people who provided genetic advice were generally physicians and PhD geneticists, and the first genetic clinic was set up by Dr. Dight in Michigan in 1940. However, in 1947, Sheldon Reed coined the phrase "genetic counseling" and explained that it was a kind of genetic social work, without eugenic connotations and with concern for the nonmedical impact of genetic diseases (Reed, 1955). The necessity to educate the family through the decision-making process was recognized. The family history was assessed, and the risk of recurrence of their family condition was estimated. Reed (1980) correctly predicted that the demand for genetic counseling would soon exceed the supply of genetic counselors.

The first genetic clinic in the United Kingdom was set up in 1946 at the Hospital for Sick Children, Great Ormond Street, London, by Professor Cedric Carter, assisted by his social worker, Kathleen Evans (J. Kromberg, personal communication). By 1955 there were dozens of clinics in the United States. In the 1960s and 1970s, the prime motivation of medical practitioners was the elimination of genetic disorders and achieving a significant reduction in the healthcare burden incurred by such disorders. In 1972 the World Health Organization (WHO) stated that the role of the genetic counselor (usually, at that time, understood to be medically trained) should be to assist the physician in diagnosis, in estimating recurrence risks, in interpreting the information for the clients in meaningful terms, and in helping them to reach and act upon an appropriate decision.

The training of nonmedical genetic counselors began in 1969 at the Sarah Lawrence College in New York (McCarthy Veach et al., 2003). The program was organized over two years, and by 1973 four more programs had developed. In 1989 the first Asilomar Conference was held in California, and heads of genetic counseling training programs were invited to discuss the syllabus, selection of students, practical work, ethical issues, roles of counselors, definitions of terms, and other components of the program. Professor Jennifer Kromberg, from Johannesburg, South Africa, was invited to participate. She had set up the first local training program early in 1989 at the University of the Witwatersrand, Johannesburg.

RECENT INTERNATIONAL DEVELOPMENTS AND THE TRANSNATIONAL ALLIANCE FOR GENETIC COUNSELING (TAGC)

Professor Janice Edwards, director of the genetic counseling program at the University of South Carolina, USA, was the Jane Engelberg Fellowship awardee of the National Society of Genetic Counselors (NSGC) for 2004/5. In 2004, she invited five international leaders in the genetic counseling profession from Australia, Japan, South Africa, the United States, and the United Kingdom, who shared an interest in genetic counseling education worldwide, to join together and organize an international meeting of genetic counselors. Professor Jacquie Greenberg from Cape Town, South Africa, was invited to participate, as she had just established the training program at the University of Cape Town in 2004. (http://tagc.med.sc.edu/).

The first meeting of the group was held in Manchester, UK, in May 2006, and was attended by 72 delegates from 15 countries and the Transnational Alliance for Genetic Counseling (TAGC) was formed. The aim of the meeting was to create an interactive opportunity for international genetic counseling program directors, professional organizations, as well as credentialing bodies that represent genetic counselors, to consider the profession in its rapidly evolving international context and to create sharing that would enhance all the educational programs and foster the transnational development of the genetic counseling profession.

The group met again in Barcelona in 2008 to develop the TAGC mission and vision. The TAGC Standing Committee was created, and one appointee from each of the fifteen countries represented at the Manchester meeting in 2006 was nominated and elected. The Alliance is now an independent and autonomous organization, and the principal office is located in the United States, in the County of Richland, state of South Carolina. Issues such as international student/faculty exchange, teaching collaborations, research collaborations, and core competencies/credentialing are among the organization's priorities.

TAGC strives to foster communication and collaboration among the international genetic counseling community and enhance genetic counseling education transnationally. The general consensus from the Barcelona meeting was that, with regard to core competencies/credentialing, there is agreement that reciprocal registration arrangements would be of great value and could be on a country-by-country basis. In addition, the establishing of core competencies between countries would be helpful. At the Barcelona meeting, a working document from the United Kingdom entitled "Registration of Overseas Genetic Counsellors Working Group, on behalf of the Genetic Counsellor Registration Board UK and Eire and the Association of Genetic Nurses and Counsellors UK and Eire Committee" was presented and discussed. The UK working group had been in contact with members of each registration/certification board from the United States, Canada, Australia (which includes New Zealand and is jointly called Australasia), and South Africa, and they reported that they had identified that currently, no reciprocal agreements exist for registration across countries.

The website at http://tagc.med.sc.edu/ serves as the hub for the TAGC, continuing and building upon the global connections and is a resource site for the greater genetic counseling community. There is an open invitation to explore the site and provide input; international perspectives are most welcome and strongly encouraged.

After the first meeting of this TAGC group in 2006, Janice Edwards, Jacquie Greenberg, and Margaret Sahhar compiled the following data, based on personal communication with members of TAGC, which summarizes the evolution of genetic counseling in developing and developed countries. Since the creation of Master's level education for genetic counseling in the United States in 1969, thirty-two U.S. programs have emerged. Canada has been training genetic counselors for over twenty years, the first program having been established in 1985; three others are currently training, and another two are proposed. South Africa was the third country to initiate postgraduate training for genetic counselors, and its first program was introduced in 1989 with a second program being added in 2004. Europe's first program was established in Manchester, UK, in 1992, and an additional training site for the United Kingdom was founded in Cardiff in 2000. Cuba has trained over 500 genetic counselors since 1995 in a unique model, providing a Master's degree in genetic counseling to family physicians, who then serve in community health centers throughout the country. The Dutch also have had a national training program since 1996. Four educational programs emerged in Australia between 1996 and 2000 as one-year post-graduate certificate programs requiring on-the-job training to achieve full recognition as a genetic counselor. Two of these programs are currently shifting toward a two-year Master's degree curriculum. Israel has trained over 40 genetic counselors since 1997. Japan saw the creation of seven programs between 2002 and 2005. Taiwan created their first program in 2003, and Norway soon thereafter. The French government decreed genetic counseling as a new health profession with the initiation of their first training program in 2004; twenty-three genetic counselors have formed the French association of genetic counselors. Saudi Arabia formed a program in 2005 and has educated four Saudi nationals at home, and sent three abroad for education, bringing the trained cohort of counselors to seven in a very short time. Spain joined the ranks of established genetic counseling programs when their first students graduated in January 2008. Other programs are envisioned for Finland, India, and China, as well as in other countries who hope to develop the capacity to bring the genetic counseling profession into their genetic services delivery. However, at present, apart from South Africa, no other genetic counselor training programs are being developed in Africa, and very few in other developing countries. (http://precedings.nature.com/documents/1574/version/1)

DEVELOPMENT OF THE GENETIC COUNSELING PROFESSION IN SOUTH AFRICA

In the 1950s Professor Phillip Tobias (anatomy), Professor Lewis Hurst (psychiatry), and Dr. I. Anderson started providing heredity counseling at the University of the Witwatersrand Medical School in Johannesburg (Jenkins, 1990). Around the same time, a specialist physician in Cape Town, Dr. Hymie Gordon, also started to see patients requiring genetic advice. In 1972 the University of Cape Town established a Chair in Human Genetics, and Professor Peter Beighton took the position and set up genetic counseling clinics at Groote Schuur Hospital, the Red Cross Children's Hospital, and Princess Alice Hospital. Also in 1972, Professor Trefor Jenkins (together with Professor J. Hansen, Pediatrics Department) set up a formal genetic counseling clinic in Johannesburg (Jenkins et al., 1973) and in 1975 Professor Jenkins was offered a Chair in Human Genetics at the University of the Witwatersrand. These two universities are presently the only two with such Chairs. Both professors established genetic counseling clinics and began counseling people with many different conditions—for example, 241 patients were seen in a six-month period in 1982 in Johannesburg (Kromberg & Berkowitz, 1986)—and conducting research projects on a variety of genetic disorders.

In the early 1980s it was decided that it was unnecessary for the medically trained staff at the clinic to see all the patients and that graduates, initially graduate social workers, could be trained to take selected cases at the University of the Witwatersrand. One such experienced staff member with a PhD, Professor Kromberg, had been involved in counseling since the service was first offered formally in Johannesburg in 1972. She was asked not only to provide genetic counseling services to selected clients, but also to develop a training course for genetic counselors at the Master's level, in 1988. She consulted widely and visited the courses at Sarah Lawrence College in New York and at the University of California, Los Angeles. The course was then set up in the Department of Human Genetics at the University of the Witwatersrand, Johannesburg. The first two students were accepted in 1989, and both were graduates with BSc (Hons) degrees in genetics. They completed the coursework in principles and practice of genetic counseling and medical genetics over a two-year period, and also wrote up a research project, before being awarded the MSc degree. The University of Cape Town started offering a similar MSc program, by course work and dissertation, in 2004.

In 1975 the South African Medical and Dental Council registered two cytogeneticists (one working at UCT and one at Stellenbosch University) under the category of genetic counselors. These were the first genetic counselors in South Africa who were not registered medical doctors. Their role mostly concerned chromosome

disorders, and patients were mostly seen either in the laboratory or at a pregnancy counseling clinic. In 1992, the Health Professions Council of South Africa (HPCSA) was approached to set up a register of genetic counselors. This was completed in due course, and in 1996 a dedicated category was introduced by the HPCSA and a total of 10 individuals were placed on this new register, initially through a grandfather clause (as they had PhD or MSc degrees, many years of experience working in various fields of human genetics, and offered some genetic counseling, but they had little or no relevant training) has recently been excluded, as most of these counselors are no longer active and the majority of the counselors now on the register are fully qualified, with the relevant Master's degree. At present there are 23 people on the register, and graduates must be officially registered with the HPCSA and must maintain that registration throughout their careers, if they are to be employed as genetic counselors in South Africa.

In the early days, graduates in South Africa had to take jobs in genetics laboratories and/or in research programs, or they emigrated to countries where jobs were available (and several graduates are now working in the United States, United Kingdom, and Australia). Job creation was, and still is, a serious problem, and it was only in 2000 that the new professor of human genetics in Johannesburg, Professor Denis Viljoen, was successful in motivating the creation of full-time permanent jobs for genetic counselors in the National Health Department. These posts now fall under the National Health Laboratory Service (NHLS) in Johannesburg. There are four junior posts and one senior post available at present in Johannesburg, and these are all filled with fully qualified graduates. There are no official permanent posts in the other provinces, although there is one intern post and one 6/8ths genetic counseling post at UCT/NHLS in Cape Town. Human genetics staff members at the University of Cape Town (UCT) are strongly advocating the creation of permanent full-time posts in the city, and at present, two qualified HPCSA registered genetic counselors are employed in part-time research posts.

Genetic counselors are currently all required to be registered with the HPCSA in order to practice (HPCSA Regulations, 2009) and a genetic counselor representative sits on the Committee of Medical Scientists at the HPCSA. This committee recently embarked on a process to revise the scope of the profession of medical scientists (including genetic counselors) to ensure that these professionals perform acts for which they are appropriately trained. The legislation has been passed (Government Gazette, May 22, 2009) and since 2010, genetic counselors are required to pass a national assessment of competency to be registered with the HPCSA. In the past this registration was automatic. As genetic counselors are required to do a two-year internship training (one year concurrent with the second academic year, and one year post academic qualification), drawing up of national intern training guidelines is part of the standardization process and it is currently underway. At the same time, the South African Qualifications Authority (SAQA) embarked on a process of establishing generic academic qualifications. This, together with the HPCSA initiatives, will provide appropriate training that is in line with that of other countries such as the United States, Canada, Britain, and Australia.

The South African Society of Human Genetics (SASHG) was established in 1983, when a group of human geneticists broke away from the South African Genetics Society (set up in 1956) to form their own group. Genetic counselors have been active members of the SASHG from the beginning. The main function of the Society is to hold biennial congresses for everyone working in the field. The recent 13th congress was held in Stellenbosch in 2009, and genetic counselors played an active part. They invited an expert international speaker from Australia, and had a full afternoon session dedicated to genetic counseling issues. They also had a national genetic counselors' meeting to discuss current issues, future plans, and a proposal to form a subcommittee of genetic counselors under the auspices of the SASHG. Several counselors have also attended international congresses of the Psychosocial Issues in Genetics group, the National Society of Genetic Counselors, USA, the British, American, Australian, European, and international societies of human genetics, international birth defects congresses, where they have presented some of their research work, and both meetings of the recently established Transnational Alliance for Genetic Counseling (TAGC). In this way genetic counselors in South Africa keep up-to-date with what is happening in the field, even though they are somewhat isolated geographically, being at the tip of the African continent.

ORGANIZATION OF GENETIC COUNSELING SERVICES IN SOUTH AFRICA

The governing of South Africa is organized through a national and provincial system. There are nine provinces (Mpumalanga, North West, Limpopo, Kwazulu/Natal, Gauteng, Free State, Western Cape, Northern Cape and Eastern Cape). Health services are governed by the Minister of Health, with a Director General at the national level. The provinces are autonomous and, although the role of national government is to advise and guide, implementation of programs is undertaken at the discretion of the provincial heads, taking the needs of each specific province into account. This has been a major challenge for the country. The key legislation governing the health sector in South Africa is the National Health Act. Implementation of this has been slow, but progress has been made in some areas (Gray & Jack, 2008).

There are many disparities in health and access to healthcare in South Africa (Benatar, 2004). The government has, since 1994, attempted to address these inequalities by establishing a district-based system of primary healthcare, nationalization of health laboratory services, regulation of healthcare professionals, and reforming health legislation. However, staff shortages and allocation of resources remain a challenge. Social inequalities, such as poverty, negative attitudes toward women and girls, racial and other discrimination, disempowerment, and the HIV/AIDS pandemic are some of the issues that the country still faces. Furthermore, health services are funded through state organizations and the private sector. State health services are provided to the majority of the population by hospitals (quaternary, tertiary, and secondary) and clinics. The private health sector, which services a minority of the population, is privately funded by medical aid societies to which members belong by paying monthly fees. Professionals offering services in private practice are paid by the patient's medical aid society. As a result, there are many disparities between services offered in the private and state sectors.

The "Human Genetics Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities" of 2001 provides a framework for the planning of genetic service delivery nationally. It constitutes a "hub and spoke model" whereby the main centers are the hubs, providing outreach services to more distant parts of the country. Genetic counseling services are offered, at present, in the capital cities of four of the nine provinces: Gauteng, Western Cape, Free State, and Kwazulu/Natal. Only two of these provinces (Gauteng at the University of the Witwatersrand (Wits) and Western Cape at the University of Cape Town (UCT) have trained genetic counselors with Master's degrees and offer a fully integrated service including molecular, cytogenetics (and serogenetics at Wits), laboratory, and clinical support. All four of the provinces, however, provide outreach clinics to other regions within their province and occasionally to other provinces.

The Division of Human Genetics of the National Health Laboratory Service (NHLS, previously the South Africa Institute for Medical Research – SAIMR) and the University of the Witwatersrand's first genetic counseling clinic was established at the Transvaal Memorial Hospital (TMI) in 1972 (Jenkins et al., 1973). This clinic, due to its siting, mainly provided a service to the White population of Johannesburg. Later, in 1987, a genetic counseling clinic was set up at Chris Hani Baragwanath Hospital (CHB) (Kromberg & Jenkins, 1988) to provide an accessible service to the Black population from Soweto and other peri-urban areas. The Division of Human Genetics at Wits now offers a genetic counseling service at all three state-funded tertiary teaching hospitals and at a private hospital in Johannesburg (Gauteng), as well as regular national outreach clinics. The service is provided to patients from all the population groups. On average, about 80% of the patients seen are from the state healthcare sector. The services are provided to antenatal, pediatric, and adult patients with many different diagnoses, from common chromosomal disorders to rare single-gene conditions, as well as multifactorial and mitochondrial conditions. Altogether, a total of 1837 patients received genetic counseling in the year 2008, and 1975 in 2007. Diagnostic testing is performed in the Division for a wide range of conditions, by DNA, chromosome and enzyme based investigations, and/or for prenatal genetic diagnosis, diagnostic confirmations, and predictive testing. A cancer genetics service was introduced in 2006, and the number of patients seen is increasing annually. The genetic counseling service is provided by HPCSA registered professionals. The division has ten clinicians (three in training), nine genetic counselors (two in training), and one genetic nurse counselor.

In the Western Cape (WC), medical services to state patients are provided by the University of Cape Town (UCT) and the University of Stellenbosch (US). Medical genetic services in the WC work on the recommended "hub and spoke" model with the two academic hospitals: Groote Schuur Hospital (GSH)/UCT and Tygerberg Hospital/US (funded from the provincial budget), constituting the "hub" of the model, with spokes extending to regional and district services via outreach and support to regional specialists and general practitioners. The different responsibilities of the clinic team relate to service provision, treatment protocols, skills development, service planning for specific conditions, along with teaching, training, and research. Screening clinics at LSEN schools (for learners with special educational needs) and institutions (approximately 20 clinics per year), as well as genetic outreach programs in the WC (two outreach weeks per year), are undertaken by the various teams. These teams can comprise medical geneticists, genetic counselors, nurses, and scientists. The primary role of the team is to provide genetic services within the WC provincial health system, and a small private-sector genetic service is also offered.

The Division of Human Genetics at UCT has five clinicians (one part-time and one in training) and five HPCSA-registered genetic counselors, two of whom are primarily involved with research programs. There are also two genetic counselors-in-training and two full-time genetic nursing sisters. The genetic service includes antenatal, pediatric, and adult services, as well as outreach programs. Within the genetic service provided by UCT in the western Cape (WC), genetic counseling is offered by medical geneticists, genetic counselors, and genetic nurses. The five medical geneticists are medical doctors with a special interest in the diagnosis, care, and prevention of genetic conditions and birth defects.

For diagnosed conditions, genetic counseling is frequently provided by the genetic counselors or genetic nurses. Three of the genetic counselors have a Master's level qualification and relevant experience in genetic counseling. The remaining two are PhD graduates with many years of experience in human genetics. The two genetic nurses have a long track record and extensive counseling experience within the public service. At the University of Cape Town's various genetics clinics, approximately 5000 individuals/families were seen and counseled from 1972–1988, and then from 1989–2008 approximately 2000 patients/family members received genetic counseling every year (e.g., 2024 in 2006, 2141 in 2007, and 2249 in 2008, as reflected in the annual reports of the relevant years).

The two academic hospitals (provincially funded) in the WC therefore provide outreach services ("spokes") to a number of sites within their drainage areas, as well as to schools. For these genetic outreach programs, as well as the school visits, the "clinical" group works as a team. Therefore, a good number of new patient children and adults, and school and outreach cases seen by clinicians, have their initial family/clinical history taken by a genetic nurse or counselor. Similarly, a number of nurse and counselor cases will receive input from clinicians during these outreach programs.

CURRENT TRAINING OF GENETIC COUNSELORS IN SOUTH AFRICA

TRAINING OF GENETIC COUNSELORS AT THE UNIVERSITY OF THE WITWATERSRAND (WITS)

http://www.wits.ac.za/pathology/humangenetics/9225/home.html

The Division of Human Genetics of the National Health Laboratory Service (NHLS) first offered a Master's degree in genetic counseling in 1989. Postgraduate students from a variety of backgrounds, including genetics (the majority of students), social work, psychology, and medicine have been enrolled. There is much competition for places and only about 10% of the applicants succeed in entering the program. A total of 16 students have successfully completed the coursework and research report, and obtained the degree. The course extends over two years, during which the students learn the principles and applications of medical genetics and genetic counseling. There is a major coursework component comprising tutorials, essays, case reports, and experiential practical exercises. The students learn to apply the information and skills through providing a genetic counseling service to patients and through clinical work under supervision by a clinical psychologist, as well as the more experienced genetic counselors. They also learn to work in multidisciplinary teams and to interact with many other academics, health professionals, social science professionals, and international exchange students who seek genetic counseling work experience in Johannesburg.

A supervised research project is carried out and written up in the second year. This requires the students to decide on a feasible project, study the relevant literature, write a protocol and see it through the university higher degrees committee, apply for ethics clearance, assess the funding requirements, collect the data, computerize the data, use statistical tests where necessary, analyze the findings, discuss results, draw conclusions, and write up an acceptable research report (assessed by external examiners). Furthermore, students should produce a paper for publication on their project. Projects that have been completed in this way in the division have contributed to the local understanding of people with genetic disorders and their problems and needs. Topics have included expectations of genetic counseling, perceptions of chorionic villus sampling, psychosocial issues and Fragile X syndrome, trisomies 13,18, and 21 in an African population, breast cancer in the Jewish community, spina bifida in Gauteng, Huntington disease and adolescents, fetal abnormalities, and termination of pregnancy, among others.

In addition, experience is gained by attendance at departmental meetings, journal club presentations and meetings, seminars and conferences, educational meetings, and community visits. Students are also encouraged to present their research work at the biennial human genetics conference of the SASHG, to get involved with local genetic support groups, and to give talks on genetic conditions to the lay public when requested.

The students who complete the course successfully have to undertake an internship year before they can be registered as a genetic counselor. On completion of this three-year program, the students are well honed and experienced, higher-degree graduates with unique research credentials, as well as both the psychosocial and genetic skills to provide a comprehensive genetic counseling service within a variety of health settings.

TRAINING OF GENETIC COUNSELORS AT THE UNIVERSITY OF CAPE TOWN (UCT)

http://web.uct.ac.za/depts/genetics/counselling/

The Human Genome Project, by facilitating the identification of actual disease-causing genes, as well as predisposing genes for many human disorders, has led to a dramatic transformation in the practice of medicine and primary healthcare. The impetus for the establishment of a Master's program in genetic counseling at UCT was the urgent need in the Cape Province and other parts of southern Africa for genetic counselors that are adequately educated to provide genetic-oriented and supportive counseling to families and individuals with genetic

conditions. In addition, it was recognized that there is a need for genetic counseling researchers who will broaden the scope of this new profession in southern Africa by generating research literature from and for the continent of Africa. The overall benefit of such a postgraduate program will be to fulfill the need for genetic health education and service development for the people of Africa who suffer from a genetic condition.

Students were first admitted onto the UCT program in 2004 and five have graduated, completed their internship, and are registered with the Health Professionals Council of South Africa (HPCSA) to practice professionally in South Africa (although one is presently working as a genetic counselor in the United Kingdom).

The purpose of the MSc in Genetic Counseling program at UCT is to provide students with a wide range of knowledge and skills unique to genetic counseling, which will enable them to register with the HPCSA and to work in the public or private sectors in communities, hospitals, clinics, special schools, and laboratories. There is an explicit commitment to a primary healthcare approach of client management throughout the program.

The second purpose of the qualification is to equip the students with management, leadership, and research skills that will enable them to participate in national healthcare development and policymaking, and to contribute to the international body of knowledge in genetic counseling. The third purpose of the UCT qualification is to endow South Africa with a nucleus of genetic counselors to provide effective, efficient, and accessible genetic counseling services while continuing to improve their knowledge and skills. In addition, it also affords the UCT MSc graduates in genetic counseling the opportunity of proceeding to a PhD degree.

The program provides students with genetic counseling knowledge, attitudes, and skills to manage genetic diseases in southern Africa and to contribute to the international body of research in genetic conditions. Interdisciplinary collaboration occurs by means of lectures and workshops, in the Medical Genetics and Principles of Genetic Counseling modules, where students from the MMed program in palliative medicine and MMed in medical genetics participate as well. Students are exposed to and work with other health disciplines in various clinical areas during their clinical practice. In order to progress into the second year of study, all courses completed in the first year have to be passed and a research proposal has to be completed and approved by the Clinical Laboratory Sciences Research Committee and the Faculty of Health Sciences Ethics Committee. The UCT program requires students to be competent in analytical and critical thinking, problem-solving, numeracy, computer literacy, appropriate writing and verbal language proficiency, and to demonstrate an enthusiasm for learning. They need to be able to search for new knowledge and research and, given the prevalence of HIV/AIDS in the country, they are also involved with HIV/AIDS-related issues during the course of the program.

The Division of Human Genetics at UCT also accommodates elective students from other countries, who work with the UCT students in the clinics, thereby increasing their awareness of genetic counseling programs and practice in other countries. Inter-institutional collaboration also occurs with the Department of Genetics and Obstetrics and Gynecology at Tygerberg Hospital (U.S.) so that students from UCT gain part of their counseling experience at other hospitals, as well as in various clinics.

In summary, the UCT program aims to produce healthcare professionals equipped with psychosocial and communication skills to relay medical, genetic, and scientific information to patients and their families, as well as to other healthcare professionals and the wider community. They are trained to provide support and management for all patients and their families with a history of genetic disease, as part of a primary healthcare approach of client management in South Africa (http://web.uct.ac.za/depts/genetics/counselling/).

ROLES AND FUNCTIONS OF GENETIC COUNSELORS IN SOUTH AFRICA

The roles and functions of genetic counselors in South Africa are developing and emerging out of insights gained from both appropriate international contacts and a deeper relevant knowledge and understanding of the local situation. Kessler (1980) described the roles, in general, as including: healthcare professional providing supportive care; education; resources and referrals; advocacy; grief counseling; and research. However, roles in specific situations are determined by professional responsibilities, employers' requirements, service site, needs of the surrounding communities, genetic disorders found in those communities, and the experience and vision of the counselors themselves. Roles also vary and change as counselors adjust to the insights they receive from patients in genetic counseling interactions, from supervision discussions, and from other learning experiences.

A study on roles of genetic counselors in South Africa showed that roles fall into five basic fields: counseling, teaching, research, marketing, and administration (Kromberg et al., 2009). Regarding the counseling role, genetic counselors are being trained to counsel people with a diverse range of genetic conditions, and these increase every year as the counselors gain experience. In the two-year period from 2007 to 2008, for example, counselors counseled 2554 cases in South Africa, and they presented with 44 different diagnoses (Kromberg, personal communication). In comparison, in Queensland, Australia, a more experienced team of counselors counseled for 79 different diagnoses (Kromberg et al., 2006). Advanced maternal age

and Down syndrome were still the commonest indications for genetic counseling in the country, but abnormal ultrasound, and/or fetal abnormalities, was the third most common indication. Although South Africa has had a sophisticated prenatal diagnosis service for many years (Kromberg et al., 1989) the new advances in ultrasound technology mean that fetal abnormalities can be detected more reliably and more specifically in utero, and skilled genetic counseling is required. Other disorders, such as retinal disorders (http://web.uct.ac.za/depts/genetics/retina) and cancer (particularly colorectal cancers) (http://web.uct.ac.za/depts/genetics/cancer), were seen more often by Cape Town counselors, due to the research interests of scientists in their university departments, (http://web.uct.ac.za/depts/genetics/neuro) while hemophilia and cystic fibrosis were seen more commonly in Johannesburg due to the attachment of counselors to the large local clinics treating those conditions (Macaulay, 2008). The patients being counseled were mostly from the Black population (65%) in Johannesburg, while those from the Mixed Ancestry group (63%) were counseled more frequently in the Cape. This ethnic distribution of patients was appropriate for the population structure found in the two different areas. The vast majority of patients were seen in the state hospital system (80%), which is the primary responsibility of the counselors; the remainder were seen either in private practice (12%) or in rural outreach clinics (8%).

The teaching role of counselors is increasing, and all are involved in teaching trainees (both medical geneticist and genetic counselor trainees), medical and other students, health professionals, learners in schools, and the lay public. The counselors' teaching role includes a supervision role, in which counselors supervise the learning experiences of students and new counselors, who are developing their interviewing skills and expertise in practice. In South Africa there are no genetic counselors who have specialized, and there are no specialization courses; also, there are no counselors in full-time education roles (such as are found in Australia), and all are generalists. Some counselors, however, have become particularly interested in a specific field, often due to having carried out their Master's research project in that field—for example, cancer genetic counseling—and may do more counseling for and teaching on the condition for which they have developed expertise (Greenberg, 1989, 2008; Greenberg et al., 2009; Loubser et al., 2009).

Research has always been an integral part of the local MSc in genetic counseling program, unlike the equivalent degree gained in Australia and the United States. Every qualified counselor has completed a research project and research report, to the satisfaction of external examiners, and a few have proceeded to a PhD (one is presently involved in a PhD degree, partly due to the fact that there is no counseling job available for her at present). Altogether, 69% of counselors are presently involved in research (mostly as a member of a research team) and 37% are published authors. Recent projects have included: general practitioners' (GP) knowledge of cancer genetics, cystic fibrosis testing in relatives of an affected child, an investigation into the level of genetic knowledge and family communication about genetic risk in parents of children with cystic fibrosis, decisions made after the diagnosis of a fetal abnormality, utilization of the Huntington disease (HD) genetic service, the use of genetic tests by the life insurance industry, and Duchenne and Becker muscular dystrophy and the implications for at-risk individuals, as well as another study regarding the level of genetic knowledge of parents of sons with Duchenne muscular dystrophy (Loggenberg et al., 2007). In addition, the perceptions of patients with inherited forms of retinal disorders while participating in a genetic research program was examined (Basson et al., 2007), as well as an investigation for a PhD on the HD predictive testing service in the WC over the past 11 years (Futter et al., 2009).

Counselors also have a responsibility to market the genetic counseling service, and they all receive training in community genetics and the public health aspects of human genetics. Generally, marketing is carried out by distributing pamphlets and articles on, for example, genetic counseling, prenatal diagnosis, and specific genetic disorders; giving talks to the public or on radio or TV; writing articles for newspapers, popular magazines, or local professional or medical journals (such as Greenberg et al., 1996; Krause & Craig, 2001); and holding meetings for specific purposes (such as providing feedback to GPs from a research project on their knowledge of cancer genetics). All the patient information documents on the UCT web page have been compiled by genetic counselors during the course of their training (http://www.uct.ac.za/depts/genetics).

Administration duties are involved in every genetic counseling job. These duties include client-related activities (mainly the compiling, keeping, and organizing of client records), clinic coordination duties, being on call and/or on receiving office, and organizing meetings and/or in-service seminars.

Counselors therefore contribute a great deal to the provision of genetic services in South Africa. They counsel approximately one-third of all the cases seen for genetic counseling, and the numbers are increasing, and they play many other roles that are essential to the smooth and efficient running of the service.

Essentially, genetic counselors in genetic counseling sessions elicit a relevant family and medical history, construct an accurate pedigree, and then calculate risk status of the individual and family and offer choices for dealing with this risk. In the process they need to be able to obtain an appropriate social and psychosocial history of the individual and family. They are also expected

to be able to interpret special investigations associated with the genetic condition, and apply theoretical scientific knowledge, sensitively. As part of this process, they also need to be able to adapt to change and tolerate uncertainty. They need to plan and execute a range of interviewing and counseling techniques, and then also manage the clients, patients, and families with empathy, caring, compassion, sensitivity, acceptance of diversity, and respect for their dignity, privacy, and confidentiality. Genetic counselors are trained to observe and interpret interactions between family members and counselors and inform patients of local and international resources and services. They also critically evaluate genetic counseling techniques used in practice, identify their own personal limitations, and recognize the need to seek help when necessary. It is essential that they are able to identify the limitations of the scope of genetic counseling and refer to other health practitioners appropriately and in a timely way. In addition, they need to know how to identify relevant ethical and legal aspects associated with clients and their families, and then to refer patients/families to specific genetic support groups and nongovernmental organizations (NGOs) where relevant. In South Africa, they have to be in a position to apply knowledge of human rights as referred to in the constitution, and recognize the cultural, social, religious, and historical contexts of their clients and families.

In summary, genetic counseling has several broad goals in the South African setting that are generally similar to those in other countries:

- Firstly, once a clinical diagnosis has been made, it is essential to assist the family in addressing their concerns relating to the development or transmission of their genetic disorder, to assess their risks and give choices, and to facilitate informed decision making that promotes patient autonomy as well as quality of life for the family.
- Secondly, genetic counselors should help individuals and their families adjust to difficult situations in a way that involves a realistic review of the positive and negative aspects of possible outcomes, promotes competence and mobilizes support, and is consistent with the family's beliefs, values, and culture. Where necessary, genetic counselors should ensure that informed consent, based on an adequate understanding of the technical information and its implications for the affected individual and family members, is promoted.
- Lastly, genetic counselors should aim to develop and promote a relationship of trust, understanding, and support that encourages the family to continue utilizing the counseling service, as well as the services of other appropriate healthcare professionals.

CHALLENGES FOR THE GENETIC COUNSELING PROFESSION IN SOUTH AFRICA

One of the biggest challenges genetic counselors face in South Africa today is to find ways to make genetic counseling more understandable and accessible to the local people. Many languages spoken in the country have no words to describe genetic concepts, and there is as yet no African language-speaking registered genetic counselor. In many communities there are myths and stigmas surrounding birth defects that are being explored, but their relevance to genetic concepts is not well understood by genetics health professionals. In the Third World and developing countries, like South Africa, underdevelopment causes marked social differences, and among people of lower socioeconomic strata there is less dissemination and understanding of scientific concepts. Partly due to the persistence of traditional medicine in many developing countries, and often due to a lack of written records, a large number of people with a genetic disorder in the family do not know how to explain what has happened in their family (De Pina-Neto & Petean, 1999). Individuals often draw on other belief systems which may be held in addition to scientific explanations. The genetic explanation may be accepted on one level, but the personal meaning of the situation comes in large part from other beliefs (Weil, 2000). Research in the South African context is currently being undertaken to investigate individual beliefs regarding genetic disorders, as well as aspects relating to communication in multicultural genetic counseling settings (research currently being undertaken by Wessels et al., 2009).

When a genetic condition is diagnosed it has a profound impact on the family both emotionally and practically. South Africans who are affected by a genetic condition are often confronted with unique circumstances and barriers, including poverty and minimal access to resources. Affected individuals and families may not be afforded information and interventions to address basic health issues, let alone to maximize their quality of life. Members of rural communities may have to travel hundreds of kilometers to tertiary hospital settings to obtain comprehensive genetic services. Genetic counseling often has to take place through a translator with no training in genetics or counseling, leading to possible misunderstanding between the patient and the counselor. In a developing country like South Africa, genetic counseling will not only be influenced by psychosocial issues within a family but by problems related to poverty, low level of education, unemployment, and inaccessibile services.

Disability creates different burdens for each family. Maslow's (1970) complete hierarchy of needs should be considered when examining family strengths and weaknesses. A family cannot be expected to follow up on recommendations for therapeutic treatment for their child,

or to attend clinic regularly, if, for example, they are not able to provide food or heat for the family. The healthcare professionals involved with these families should be familiar with local resources or services that are available to assist families, where necessary, with food, clothing, shelter, and financial assistance (Ziolko, 1991)

Participants with little formal education may benefit from additional counseling. Although it is possible that some may still not understand genetic concepts after several sessions, it is nevertheless the counselor's obligation to try and find ways of communicating the relevant complex information (Somer et al., 1988). In order for family members to incorporate risk information, they need an opportunity to discuss how new information about inheritance fits into, or supersedes, previously held beliefs. This type of discussion has been identified as a useful method for helping individuals internalize information about inheritance and reproductive risk (James et al., 2006).

In a recent research project, local genetic counselors, themselves, suggested areas for role expansion, in an attempt to meet these challenges. These areas include raising the visibility of the profession, increasing the numbers and variety of suitable referrals, and making the service more widely known and accessible (Kromberg et al., 2009).

Clearly, more education of the medical and lay public is needed. If the health professionals were better informed, they would refer a wider range of patients, more people would benefit from the counseling services, and counselors could expand their expertise to a broader range of genetic conditions. Furthermore, if the general public becomes more knowledgeable about genetic conditions they will understand their relevant family history better and will demand an appropriate and more accessible service. It is recognized, too, that genetic testing is being done, often in private practices, without informed genetic counseling pre- and post-testing—regardless of the international injunction that such testing may be debatable, the results may be complex, and skilled genetic counseling is essential if the service is to be used appropriately (Kromberg, 1993). Patients are still having genetic testing, even prenatal genetic diagnosis testing, without understanding what the results might show and what the implications of the results could be, while genetic counselors are trained to provide the service associated with this scenario.

Employment for genetic counselors remains a huge challenge; currently the NHLS provides some employment, and there no genetic counselors employed by the state-funded hospitals. As in many other countries, such as Australia (Kromberg et al., 2006), most of the genetic services are based in the big cities and the rural populations have little access to them. The challenge for South African counselors is to increase their outreach services, and/or to train and place counselors in outlying towns (as happens in Queensland, Australia; see Kromberg et al., 2006), so that more at-risk people can have their needs met. Similarly, many of the local population who have private medical aids do not attend the state hospitals, and therefore do not come into contact with the genetic clinics; they may not be referred, and may not make use of genetic services. For this reason, private practice may need to increase and become more widely offered, in settings such as private laboratories where genetic testing is carried out, or infertility clinics, or cancer clinics (which are all settings for genetic counselors in other countries) so that this section of the population has more access to services. Another model would be to have genetic counselors attached to groups of general practitioners, so that patient management improves for those with a history of genetic disorders. Genetic counselors should also become more involved with genetic support groups in educational and supportive roles, as well as to provide information for members regarding the available genetic services.

Cancer genetic counseling is a growing field in South Africa, as it is in Queensland, Australia (Kromberg et al., 2006) and other countries. The number of referrals has increased in Johannesburg since the completion of two cancer related research projects. However, cancer testing facilities are not yet fully developed in the country, and these are required if a functional testing and counseling service is to be developed. In the meantime, genetic counselors are counseling more cases every year, and this part of their service is likely to be much in demand when the laboratory service is up and running.

Offering genetic counseling services within the local health system has many challenges, not the least of which is the selection, training, and integrating into the health system of the staff required to run this new and developing service. The report from the United Kingdom on "Clinical Genetic Services into the 21st Century" recommended that two full-time clinicians and eight genetic counselors are required per 1 million people of a population (Royal College of Physicians, 1996). By this standard (which was supported by the World Health Organization) approximately 98 clinicians and 392 genetic counselors are required to provide an adequate service in South Africa. With the number of practicing medical geneticists and genetic counselors (some in training) currently being about the same and, importantly, less than 20 for each profession, the country has a very serious shortage of fully trained staff. Both professions are therefore attempting to attract students, train them, find employment for them, and retain them in South Africa once they are qualified and experienced. This situation requires the urgent attention of the National Health Department (in terms of job creation), the provincial health departments, and academic institutions (who can train more counselors if jobs become available).

In general, these are the challenges of any new profession in a young and developing country, and especially in one overwhelmed by an AIDS epidemic that demands a large slice of the health budget. However, the numbers of genetic counselors are increasing in other countries, and if the need is to be met they need to increase much more quickly in South Africa. The recent Law Reform Commission in Australia (ALRC, 2003) inquired into genetic privacy, a problem that will have to be faced in the future locally, and recommended that increased resources for genetic counseling services were a priority if problems such as invasion of privacy were to be prevented.

THE FUTURE OF THE PROFESSION IN SOUTH AFRICA

The future of the genetic counseling profession in South Africa will be determined by the way the challenges currently presented are faced and managed. One of the most important tasks is ensuring that more genetic counselors are trained, and that the WHO-recommended ratio of counselors to population size becomes a more attainable goal. The genetic counseling profession in South Africa has modeled itself partly on genetic counseling services in First World countries such as the United States, United Kingdom, and Australia. However, South Africa is a developing country with a very diverse population, and the challenges faced by local genetic counselors will differ somewhat from those of other countries. It is therefore acknowledged that specific attention needs to be paid to local diversity and cultural issues during training and when seeking areas of potential employment for genetic counselors. Due to some of the day-to-day challenges genetic counselors in South Africa face—for example, dealing with and being dependent on interpreters and/or counseling individuals with little or no education—the workload may be somewhat greater than in developed countries. Local clients may need more sessions to ensure their understanding of the necessary information. For these and many other reasons it is essential that the number of genetic counselors in South Africa is increased. Ensuring employment opportunities for such counselors is a vital element that will influence both the future of the profession and the availability of genetic services in South Africa.

Apart from the required extra positions which need to be made available, primarily by the Department of Health, a secondary practical model that has been proposed to increase employment opportunities is that trained genetic counselors are attached to groups of general practitioners (GPs) or specialist clinics (e.g., in-vitro fertilization [IVF] clinics) and are then jointly paid by public and private funding. This could maximize the access of both the GPs/specialists and the clients to current information and genetic services, while at the same time providing ongoing genetic management and follow-up. The added advantage of this model would be that it could bring current information on the new genetic tests and technologies to the GPs in the community and rural areas, thereby facilitating and improving the outcome of the primary healthcare model in South Africa.

CONCLUSION

One of the other developing countries in the process of establishing genetic services in Africa is Tunisia. Chaabouni-Bouhamed (2008) states that there is no national program in Tunisia dedicated to hereditary disorders, and even the national program on maternal and child healthcare does not include these conditions. However, genetic counseling and prenatal diagnosis seem to be the methods of choice for the prevention of genetic diseases, and therefore such services should be prioritized and developed, despite the financial and emotional costs of such programs. He adds that genetic services need support, and community genetic services need initiating. In particular, the following is required:

> a comprehensive national program to develop basic genetic services; the strengthening of human resources in the practice sector (clinical geneticists, social workers and trained laboratory personnel); training courses for primary care worker so that they can recognize at-risk families; population screening programs and a prenatal screening program based on ultrasonology; and a national registry for congenital malformations. Support for such programs should come mainly from public funds, but also from the private sector.

Chaabouni-Bouhamed concludes that as genetic disorders are prevalent in the region, and as they have literature on these conditions in Tunisia, the country should benefit from modern scientific advances in the field in the same way that industrialized countries do. Basic medical genetic services should be implemented and supported to capitalize on these benefits. South Africa, at the other end of the continent from Tunisia, is some way along this path. However, it is imperative that further support comes from government, if a comprehensive program as described by Chaabouni-Bouhamed)—but including the new professionals, genetic counselors, who play a great part in providing services in developed countries—is to be strengthened and offered more equitably and with accessibility, locally.

Genetics and genomics are integral components of medical practice today. No longer confined to

dysmorphology and Mendelian genetic disorders, it is anticipated that genetic understanding will play a crucial role in the management of common illnesses such as cancer, diabetes, psychiatric disorders, hypertension, and infectious diseases in the future. Genetic specialists have a central role to play in education and research in order to relate these advances in developing countries in a way that will maximize impact on healthcare. Professional genetic counselors are an integral part of any holistic genetic service and it is their duty to provide the communication interface between the patient and the clinical and laboratory teams. This new complex science should be made more accessible to everyone, and it is essential that the understanding and ethical practice of genetic and genomic medicine be promoted in all developing countries today. In addition, academic research and teaching in genetics needs to be promoted in a manner that will benefit all nations. Education needs to be provided to professionals and laypersons to support the integration of genetics into the primary healthcare services. Today's research should provide a sound scientific foundation on which to develop a practical and realistic genetic service for tomorrow, for and in Africa and the developing world.

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