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GENETIC COUNSELING: A GLOBAL PERSPECTIVE

Roles of Genetic Counselors in South Africa

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Abstract Genetic counseling is a growing health profession in South Africa. Training (set up in 1988) and registration are in place, but job creation remains challenging. The aim of the present study was to investigate the roles played by genetic counselors in the provision of genetic services, in South Africa. A questionnaire comprising items on the types of roles they performed was constructed and counselors were asked to make the log-books, in which they recorded their daily counseling activities, available. A check list was drawn up so that relevant information could be collected systematically from these log-books. Then departmental statistics were accessed from the two universities providing genetic services and genetic counselor training. Structured interviews were conducted with the genetic counselors (16 of 23 participated), and data were collected from their completed questionnaires, log-books and the departmental statistics, for the years 2007 and 2008. These data were analyzed and the findings showed that the counselors counseled about one third (39 %) of all the cases seen at genetic clinics per annum, and the total numbers were increasing. They counseled for 57 different genetic disorders, and their clients represented the range of local ethnic groups. They also had educational, research, marketing and administrative roles. They expected to expand these roles and advance the profession in future. Genetic counselors are versatile, playing several significant roles. As these become better recognized, demand for their services should increase, jobs should be created and the service expanded.

Keywords Genetic counseling · Genetic counselors · Training · South Africa · Roles

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Introduction

The genetic counseling profession is a young, dynamic and developing health care profession in South Africa. It is inextricably intertwined with the rapidly advancing field of human genetics, the new findings of molecular genetics, and their applications to the well-being of human populations. Counseling skills, in-depth knowledge of human genetics and genetic testing, and the ability to communicate complex genetic information to patients, form the basis of genetic counseling practice.

Internationally, the profession emerged as a result of the fact that 5 % of children are born with some congenital abnormality and/or a serious physical or mental handicap (Harper 2004) and affected families and individuals can benefit from genetic counseling and preventative services (McAllister et al. 2008). Furthermore, rapid advances are being made in the understanding of genetic disorders and specialized training is required for individuals imparting information about these advances to people affected by or at risk for these conditions. In South Africa, formal genetic counseling clinics, available to all patients, have been provided in the public health care sector (mostly at tertiary care hospitals) and in limited private practice, since the 1970s (Jenkins 1990). However, in the early 1980s it was recognized that the medical social workers assisting at the clinics could receive in-service training and manage selected cases. One such staff member was one of the authors of the present article, Professor J Kromberg, who was working at the Department of Human Genetics, University of the Witwatersrand (Wits) and the National Health Laboratory Service (NHLS), in Johannesburg. She proceeded to counsel, initially, prenatal diagnosis cases (Kromberg et al. 1989) and then a variety of other cases, until the late 1980's. Then, following completion of her doctoral training (Kromberg 1986), she and the head of Department, Professor T Jenkins, initiated a Master's level training program for genetic counselors, at the request of a Human Genetics student, Brynn Levy. A curriculum was developed based on ideas gained from on-site visits to the Sarah Lawrence program in New York and the University of California program in Los Angeles, but structured to meet the demands of the local university. In addition to the course-work (in Medical Genetics and the Principles and Practice of Genetic Counselling), a research project and research report were required. The first genetic counselor training program was then set up in 1988 (one of the first to be established outside the USA), as an MSc(Med) in Genetic Counselling degree with a 2 year internship (1 year overlapping with the second year of the degree), in the Department of Human Genetics at the University of the Witwatersrand, Johannesburg. Later, in 2004, the University of Cape Town (UCT), in Cape Town, initiated a similar training course. Enrolled students can apply for university funded Merit Awards which cover their tuition fees and a stipend.

Applicants, usually with an undergraduate Honors degree, entered the genetic counseling training program from a variety of backgrounds, including human genetics, nursing, psychology, public health and social work, and the numbers of qualified genetic counselors increased slowly. In 1992 Genetic Counseling was recognized as a health profession by the Health Professionals Council of South Africa (HPCSA) and registration for Genetic Counselors was initiated. In the early years ten counselors registered under the grandfather clause; they had either a PhD or MSc degree, and/or were post-graduates with many years working, and offering some counseling informally, in various fields of human genetics. The first MSc(Med) qualified counselor immigrated to the USA, passed the American Board of Genetic Counselors certification examinations there and has worked for many years as a genetic counselor in New York. Although counselors were being trained in South Africa job creation did not follow until 2000, when eight new jobs were created in the Department of Human Genetics, NHLS, in partnership with the Faculty of Medicine, University of the Witwatersrand in Johannesburg, Gauteng province. At the same time great difficulty was being experienced with job creation in the Cape Province and no jobs were forthcoming. One of the problems was, and still is, the nature and financing of the health care system, which is a mix of public funding (at national and provincial level) used by the majority, and private funding (through medical aid plans), used by the minority of the population. More recently, in 2012, another province, Kwazulu Natal, has initiated genetic services and the first genetic counseling job has been created and filled. However, recently, some of the posts, including intern posts, in Johannesburg, have been frozen, to the severe detriment of the service and the profession (Kromberg et al. 2012). As a result, qualified counselors are beginning to set up private practices, with supervision from the staff of the academic departments of Human Genetics. In 2011, South African Genetic Counselling qualifications were accepted by Australia and the UK and a reciprocal employment agreement was reached. Four South African counselors are now working in these countries.

The South African training is based on the philosophies and principles described internationally. The definition most commonly used is that genetic counseling is a “dynamic psycho-educational process centered on genetic information” (Biesecker and Peters 2001 p.194) and the goal is to empower the client to use genetic information in a personally meaningful way, minimizing psychological distress and increasing personal control. Genetic counseling, in South Africa, is based on “client-centered” and “non-directive” principles and it aims to preserve client autonomy, self-directedness, feelings of worth and competence (as specified by Kessler 1997). A good genetic counselor should be a good communicator, well trained in client-centered counseling and able to explore, encourage, offer choices, and be empathic, non-judgmental and respectful of client’s needs and values (Weil 2000). To support and promote these principles a professional group, Genetic Counsellors–South Africa (GC–SA), was founded in 2009, and through this group the necessary professional documentation is being developed. New regulations regarding scope of the profession, training, internship, registration and continuing education for qualified counselors were published in the Government Gazette of May 2009 (www.hpcsa.co.za) at the same time the training syllabus was approved by the SA Qualifications Authority (www.saqa.org.za) and, presently, further policy documents are being prepared.

Qualified counselors in the clinics are expected not only to counsel patients, but to participate in genetic support group activities, contribute to medical student, other health professionals and medical scientists training on human genetics, as well as give talks to lay groups, organize other community activities, and undertake relevant research. In addition, due to the cultural mix of the ethnic groups living in South Africa, counselors are trained to be aware of cultural issues (Kromberg and Jenkins 1997) and should be able to work with a variety of patients, by taking into account their specific background and the possible related issues that might arise.

South Africa is situated at the southernmost tip of the African continent, it has a population of about 51million (80 % being black Africans, 9 % white, 9 % of mixed ancestry, and 2 % Indian and Asian) (Statistics South Africa 2011). The majority (60.7 %) live in urban areas (World Bank 2010). There are 11 official languages, but English is most often used in education and business. The total fertility rate was 2.38 in 2009 (Statistics South Africa 2009). There are several cultural practices and beliefs that impact on genetic counseling practice and these include systems of thought, favoring collective rather than individual thinking, an external locus of control and fatalistic philosophy (Kromberg and Jenkins 1997). Also, traditional beliefs about disease causation and reproduction, and the common use of both Western medical services and traditional healers, should be taken into account.

Although the National Policy for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities, published by the Department of Health in 2001, provided recommendations for the provision of genetic services, these have not been followed up and the country has a dearth of trained staff. There are only 11 medical geneticists (1 per 4,500,000 people), 10 genetic counselors (1 per 5,000,000) and 42 medical scientists in human genetics, practicing at present. However, a wide range of services are provided by these staff including prenatal diagnosis, pediatric genetics, cancer genetics (services provided by both medical geneticists and genetic counselors), some metabolic genetics (provided by a medical geneticist) and very little reproductive genetics. Some specialty clinics, such as the Cystic Fibrosis, Hemophilia, Breast Cancer and Neurogenetic clinic, have had the services of a genetic counselor. A wide range of genetic tests can be performed in the genetics laboratories in the major cities. These services have been described in detail in the article on Genetic services and testing in South Africa (Kromberg et al. 2012).

General factors that need to be considered in South Africa include the unique black African population, whose genetic disease burden is still being researched, and whose specific mutations for common disorders are being identified. For example, founder and unusual mutations have been identified for Fanconi anaemia (Morgan et al. 2005), spinal muscular atrophy (Labrum et al. 2007) breast cancer (Van der Merwe et al. 2012), and Huntington's disease (Magasi et al. 2008) in the black population. However, those for other common genetic conditions still need to be understood. Also, the high consanguinity rates in some ethnic groups (e.g. in the parents of people with oculocutaneous albinism it was 41.7 % in the Tswana speaking group, Kromberg and Jenkins 1982) need to be remembered. Apart from albinism in the black population several other genetic conditions are common in various South African ethnic groups and these have been documented in Kromberg et al. (2012).

South Africa is a developing middle income country, with a fully democratic system of government but the development of a comprehensive health care service, which should include a genetic service, has been hampered by the HIV/AIDS pandemic and the increasing prevalence of tuberculosis. Due to this pandemic there are over 5 million people living with HIV/AIDS, about 10 % of the population carry the virus, and the average life expectancy, estimated at 58 years in 2000, had fallen to 51 years in 2008 (World Bank 2010). The country has adequate policies in place to improve health care, but poor implementation, management and monitoring of these policies has resulted in varying quality of care within the public health system (Coovadia et al. 2009).

Genetic services have been developing slowly within this context. The academic training (including clinical and research training) of medical geneticists, genetic counselors and medical scientists is in place, initially promoted by two of the top universities, and subsidized by government funding. However, internships (previously supported by the National Health Laboratory Service) are being reduced which results in incomplete training and the inability of individuals to register as fully qualified. The supply of staff for the future, therefore, is seriously compromised. Although the training is very closely integrated with the genetic services, where all the clinical work is undertaken, this does not translate into job opportunities. This situation is retarding development of the profession in the country.

In general, it is internationally recognised that the key role of genetic counselors is to provide a genetic counseling service related to genetic information, usually in a multidisciplinary team, to the people in their community. However, the roles of genetic counselors might be expected to vary in different settings and the nature of these roles appears to be determined by the professional responsibilities, employer's and team requirements, service site (metropolitan or regional), community needs, training, background, individual abilities, experience and vision (as shown in an Australian study by Kromberg et al. 2006). Genetic counselors possess a multitude of skills, from counseling, teaching, research, to management and more (McCarthy Veach et al. 2003). However, the roles and functions of South African counselors, and the skills they have developed, have not been explored. The aim of the present exploratory study, therefore, was to investigate the roles genetic counselors play in the provision of genetic services in South Africa.

Methods

Study Population and Procedures

The sample for the study included all those genetic counselors (23 at the time of the study) who were registered with the HPCSA, as independent practitioners, and who worked either in the Johannesburg/Pretoria Northern region (Gauteng Province) or in the Cape Town/Stellenbosch Southern region (Western Cape Province) of the country. Their names, addresses and telephone numbers were obtained from their departments or from the HPCSA register and they were approached (by telephone or in person) and asked to participate in the study. If they agreed, an information sheet and consent form was sent to them electronically. Two declined to participate: one was a general practitioner who had worked in the field of human genetics part-time for many years; and the other was a cytogeneticist who had done some counseling in the past but was no longer seeing any clients. Both were registered as genetic counselors under the grandfather clause, in the early days of the registration process. A third registered counselor had left to work in the UK and did not respond to email messages.

The remaining 20 counselors were contacted again and, where possible, an appointment was arranged, so that the information sheet and consent forms could be delivered and signed and the questionnaire administered face-to-face. Most could be interviewed in Johannesburg, but a visit was also made to Cape Town, where two counselors were interviewed. The others, who lived too far away to come for an interview, were sent the questionnaire by post. Altogether, 16/20 (75 %) counselors completed questionnaires, 12 in face-to-face interviews (with JGRK who transcribed their responses on to the questionnaire) and four by post, and another four did not return the questionnaire, despite telephonic follow-up.

Quantitative data were obtained (with each participant's consent) from the counselors' log-books, that are required for HPCSA registration. In these log-books they recorded, daily, their various genetic counseling related activities.

In order to obtain perspective on their contribution to genetic services, data on the numbers of clients seen by counselors and medical geneticists were collected from the departmental statistics kept by the two universities providing training programs, as well as genetic services, namely the Universities of the Witwatersrand (in Johannesburg) and Cape Town. These staff members sometimes counseled cases together, but the case was then allocated to the professional who took the final responsibility for the case and the statistics are presented as such.

Statistical data were collected retrospectively for the 2 year time period, January 2007 to December 2008, and the counselor interviews were conducted in 2008–2009.

The project was approved unconditionally by the Human Research Ethics Committee (Medical) of the University of the Witwatersrand, on 21 June 2008.

Instrumentation

A questionnaire was constructed for use in the interviews, and a project information and consent sheet for participants was written (these documents are available from the authors). The questionnaire comprised a section for demographic details and a section of closed and open-ended questions. These questions covered the types of roles that could be performed, including counseling, educational, research, marketing, administrative and supervision roles. They also covered any support group and conference activities undertaken. There were three open-ended questions on the future of the profession, and what new roles could be carried out in South Africa, and the nature of role expansion ("How do you see your role expanding?"). The questionnaire was used in structured interviews, but where distance was a problem it was sent by post, completed by the participant and returned in the post.

A check list was constructed to collect information from the counselors' log-books on the dates of their counseling sessions, the diagnoses, types (public or private) and ethnic group of patients seen, the counseling site, and any other relevant activities undertaken. The participants were then asked to make their log-books available to the researchers. However, only 13/16 log-books were found to be complete in every detail, for the 2007–2008 period. These 13 could be used to extract the diagnoses (which were counted and ordered in terms of most to least frequently seen) of the patients and the other details required for the completion of the check list.

Data Analysis

The analysis of the data was undertaken using descriptive statistics and frequencies for most of the responses to the questionnaire, as well as for the data from the log-books and the departmental statistics. However, the responses to the open-ended questions on the completed questionnaires were coded, themes were developed and quotes from participants were selected to support these themes (Smith 2000).

Results

Characteristics of the Sample

The demographic details provided on the completed questionnaires were analyzed and the characteristics of the sample are shown in Table 1. All the counselors were registered with the HPCSA, but three counselors (two with PhDs and one with a BSc (Hons) were registered in the early days of registration under a grandfather clause. Nearly half the group had only 2 years of experience in the field, while four had 7+ (range 7–23 years) years of experience. Most respondents (11, 69 %, of whom three were part-time) were employed as genetic counselors, but five were employed in medical scientist (3) or genetic education (2) posts. The majority (15/16) of counselors were either in university based posts at the University of Cape Town (UCT), in the Cape Province, or in National Health Laboratory Service (NHLS) posts in Johannesburg, linked to the University of the Witwatersrand (Wits), in the Gauteng province, some having formal joint appointments with the NHLS and Wits; however, one worked part-time in a University based post in Pretoria, Gauteng. All the participants undertook some

Table 1 Characteristics of the sample (*N*=16)

Characteristic	No	%
Qualifications		
BSc (Hons)	1	6.5
MSc (Med)	13	81
PhD	2	12.5
Experience		
2 years	7	44
3–6 years	5	31
7+ years	4	25
Registered genetic counsellor	16	100
Time worked		
Full-time	5	31
Part-time	11	69
Region		
Gauteng Province	11	69
Western Cape Province	5	31

counseling, but the focus of their job was counseling for only 12 (75 %) of the respondents, while it was counseling and/or teaching for 7 (44 %), and/or research for 2 (12 %), and/or laboratory work for 2 (12 %); the balance of these roles depended on the requirements of the post. The details of these roles appear in Table 2, and they are explained in more detail below.

Counseling Roles

From the statistical data submitted by the Divisions of Human Genetics at Wits/NHLS and UCT, the counselors saw 2,554 cases between 2007 and 2008 (see Table 3). More cases were seen in Johannesburg, as there were more counselors based there during the period under review. The total number of cases increased in the second year compared with the first year, the increase being recorded specifically in Cape Town. From anecdotal evidence, the decrease in cases at Wits/NHLS was mainly due to the reduced numbers being seen for prenatal genetic counseling at one large hospital, where the obstetricians were overburdened and short staffed and referral had become problematic.

Table 2 Roles of genetic counsellors (*N*=16)

Role	No	%
Counselling	16	100
Counseling focus[a]	12	75
Teaching		
>once a month	5	31
1–6 times a year	11	69
Research		
Involved in research	11	69
Published a paper	6	37
Administration and marketing		
Administration	16	100
Documenting cases	16	100
Clinic co-ordination	12	75
On call duties	12	75
Marketing/public engagement	13	81

[a] See text for explanation

The data collected from departmental statistics for the year 2008, which were complete for both universities, showed that clinical geneticists were counseling the majority of the referred cases (61 %, 2049/3365,) and genetic counselors were seeing over a third (39 %, 1316/3365) (see Table 3).

From the data in the 13 available and complete log-books the diagnoses of the clients counseled could be extracted, counted and then ordered in terms of most to least frequently seen. The nine most common diagnoses recorded differed according to the city where the counseling took place (see Table 4). Altogether, a total of 57 different diagnoses were seen during the study. The individual counselors counseled for between 19 and 44 different diagnoses, depending on their level of experience; the more experienced counselors dealing with more diagnoses than those less experienced. In total, prenatal diagnosis counseling (particularly for advanced maternal age) is still the most common of all indications for referral for genetic counseling, as happens in most developing genetic services.

In order to assess whether there was equitable access to genetic services, the data on ethnic group of the clients attending for genetic counseling were collected from the participants' log-books and the findings were compared with the

Table 3 Cases counseled by genetic counselors and clinical geneticists by year and university

Cases seen by year	Wits/NHLS	UCT	Total	
	No	No	No	%
Genetic counselors				
2007	716	522	1238	
2008	669	647	1316	39 %[a]
Sub-total	1385	1169	2554	
Clinical geneticists				
2007	1259	N/A[b]	N/A	
2008	1168	1557	2049	61 %[a]
Sub-total	2427	N/A	N/A	
Total				
2008 only			3365	100 %

[a] See text for explanation

[b] *N/A* Not available

Table 4 Most common diagnoses seen for counseling in genetic services

Site of counseling	Cape Town	Johannesburg
Diagnosis	Advanced maternal age	Down syndrome
	Abnormal ultrasound in pregnancy	Advanced maternal age
	Down syndrome	Haemophilia
	Retinal disorders	Cystic fibrosis
	Developmental delay	Fetal abnormalities
	Cancer (Hereditary non-polyposis colon cancer, breast cancer)	Trisomy 18
	Sickle cell anemia	Multiple miscarriages
	Neurofibromatosis	Huntington's disease
	Muscular dystrophy	Metabolic disorders

population demographics for the area where these clients received the service. Approximately 28 % of the cases seen in the Cape were from the black ethnic groups, while nearly 65 % of those in the Johannesburg area were from these groups. Furthermore, about 63 % of cases in the Cape were of mixed ancestry (where most of this group lives), while only 7 % in Johannesburg area were from this group.

When the log-book data on counseling sites were collected and analyzed the picture was very similar for the Cape Town and Johannesburg areas. Counselors in both cities saw most patients in local state hospitals (~85 % and 75 % respectively). Private patients were counseled less often in the Cape (~7 %) than in the Johannesburg area (~18 %), where a small private practice had been set up in a local private hospital, and very few patients were seen in outreach clinics (~8 % and 7 %, respectively).

Teaching Roles

The analysis of the interview data on the responses to the questions on teaching roles showed that some of the genetic counselors had extensive roles and all had had some experience. They provided teaching to many different groups including clinical geneticists and genetic counselors in training, registrars, medical, paramedical and other students, general practitioners, and many different lay groups in the community. Altogether 5/16 (31 %) counselors provided teaching more than once a month and 11/16 (69 %) provided it occasionally (1–6 times a year) (Table 2). Nearly half the group (7/16, 44 %) also had a regular supervision and in-service role in the training of student counselors and junior staff.

Research Roles

Some genetic counselors spend more of their time in research than others, depending on their interests, their opportunities to work in research teams and whether they had higher degree requirements. The data from the study interviews showed that 11/16 (69 %) counselors were involved in research, initiated either by themselves or others (Table 2). Further, a third (6/16, 37 %) had published a paper on their research work (either as first of several authors or as a co-author). Research projects ranged from, for example, assessing the impact of offering genetic counseling services at a cystic fibrosis clinic (Macaulay et al. 2012), to investigating mothers' responses to the diagnosis of an abnormality in their fetus in utero (Todd et al. 2010), and general practitioners' knowledge of cancer genetics (van Wyk 2008).

Marketing and Administration

Since genetic counseling is a young and developing profession, counselors were asked in their interviews about their role in marketing the service and in public engagement. The majority (13/16, 81 %) were involved in such activities (Table 2). They had made use of pamphlets on genetic counseling and on some of the most common genetic disorders, articles, talks, radio and television programs and meetings to publicize the genetic counseling service. Where necessary counselors had written patient information leaflets on specific disorders, had them printed and distributed to target groups (for example, on breast cancer and genetic testing).

Administration has a part in every profession and counselors described their administrative roles in their interviews. Most roles were related to clients and documenting their interactions with clients (16/16, 100 %), but counselors also coordinated clinics (12/16, 75 %), were on call regularly to take telephone calls from clients and doctors (12/16, 75 %), and organized departmental and other meetings.

Future Roles and Role Expansion

Counselors were asked open ended questions on the future of the profession, on what new roles they might expect to fulfill, and how they thought their roles should expand in future. The themes derived from an analysis of their responses are detailed below.

Two themes emerged from the analysis of the responses on the future of the profession. The first concerned the role as educator, and the fact that more educational opportunities should be offered to the medical and lay public. As one participant stated:

> “we need to educate the doctors, sell the service and promote the profession” (P15).

This activity should result in an increase in both demand for the service and in appropriate referrals and, consequently, the needs of more people who required counseling should be met. The second theme concerned the feeling that the profession deserved more recognition and one participant stated:

> “the first step in the recognition process would be the establishment of genetic counseling posts in the hospital sector, not only in the academic and laboratory services” (P13).

Another theme (the third) was derived from the responses on possible new roles. This theme concerned the provision of more pre- and post-test counseling for genetic disorders, which should be offered when genetic testing (particularly for predictive purposes and pre-symptomatic diagnosis) was being undertaken. One counselor’s response illustrates this theme:

> “.......private diagnostic laboratories in South Africa should be made aware of the benefit of having counselors as part of their businesses.....”(P16)

The fourth theme was related to the expansion of roles, and participants felt that providing services to clients with a wider range of conditions and, when they are better defined genetically, to clients with common non-Mendelian and polygenic or multifactorial disorders, would be important. As one counselor said:

> “as testing develops roles will change and as management changes more complex diseases should be seen e.g. heart disease, cancers, diabetes (based on research findings)” (P 8).

The counselors were also concerned with playing a more active role in the development of outreach clinic counseling and of private practice (since job creation in the state or provincial sector was so difficult). They felt that both should be encouraged, as should the offering of counseling services to members of support groups for families with genetic conditions.

Discussion

Genetic counselors contribute significantly to the work of genetic service provision in South Africa. Although the profession is young and the trained counselors are just gaining experience they are playing several essential roles. Their counseling role is similar to that described in other countries although they do not yet counsel for all the conditions seen by their more experienced counterparts in Australia (Kromberg et al. 2006). The role also differs from that played in Australia due to the composition of the South African population, the genetic disorders found in this population, and the way in which services are organized and used locally. Cancer genetic counseling is growing rapidly in Australia and it was the second most common reason for referral to clinics in Queensland prior to 2006 (Kromberg et al. 2006). Some Queensland counselors spent 50 % of their time counseling cancer cases, especially where the genes for the condition are known in specific target groups and testing for gene carriers is available. However, after the completion of the present project the authors observed a similar upward trend in genetic counseling for cancer in Johannesburg, South Africa, too, and in 2012 about 20 cases a month were being counseled (Kromberg, personal communication, 2012).

The common diagnoses for which patients were counseled differed according to where they received their genetic service. The inclusion of such conditions as retinal disorders and the two common cancers high on the list of diagnoses in the Cape was due to the research interests there, while the inclusion of hemophilia and cystic fibrosis in Johannesburg was due to the policy of placing genetic counselors in the clinics dealing with those disorders, so that clients might have easier access to them. Since neural tube disorders are common in both these regions it was somewhat surprising that these disorders did not appear on the most common diagnoses list (however, fortification of basic foods with folic acid was introduced in 2003 and, consequently, the incidence of neural tube defects had fallen by 30 %, according to Sayed et al. 2008). Referral of appropriate cases needs to increase in South Africa if the expertise of the trained counselors is to be fully utilized. In Johannesburg, Down syndrome cases are recognized and frequently referred, but cases such as those with a history of, or affected by, neural tube defects, cystic fibrosis, Duchenne muscular dystrophy and those in consanguineous matings, which previously were often referred for genetic services (Kromberg and Berkowitz 1986) are now seldom referred and counseled. The reasons for this situation need to be identified, but they could be indicative of the faltering services.

In the present study genetic counselors were counseling just over one third (39 %) of referred cases, this percentage is similar to that provided by the data for 2012 for Johannesburg which show that counselors saw 42 % (846/2013) of cases, while medical geneticists saw 58 % (1167/2013) (T Wessels, personal communication, 2013). However, in Queensland, Australia, counselors contributed to 80 % of clinical sessions (Kromberg et al. 2006). Although the data could have been recorded differently, this difference, also, could be partly due to the types of cases that are referred in the two settings, partly to the way cases are managed and multidisciplinary teams function. However, the genetic counselor involvement was observed to increase rapidly over time (for example, there was a 30 % increase between 1998 and 1999, Kromberg et al. 2006), in Australia, and this is likely to happen as genetic counselors progress, more jobs become available and the service becomes better used in South Africa. Similarly, the numbers of diagnoses counseled for were more limited in the South African series (57 compared with 79 in Queensland) and these should increase as experience and referrals increase.

The fact that the majority of the clients counseled by genetic counselors **was** from the black population in the Johannesburg area and from the mixed ancestry group in the Cape, was generally consistent with the demographics for these two areas, and therefore it suggests that access to the service for these groups is appropriate. This is an improvement on previous years, when for example, in 1986 in genetic services, it was reported that the majority (over 85 %) of cases seen by the genetic staff, prior to the introduction of genetic counselors, in Johannesburg, were from the minority white population (Kromberg and Berkowitz 1986). However, to meet the problems of access (Bee 2005), further rural outreach clinics still need to be developed, when more staff members are available to pursue this demanding and time consuming side of the service (which although previously offered, in a limited way, is now severely restricted due to staff shortages).

In general, the roles played locally, where genetic counselors are usually generalists, are similar to those described in the USA. A National Society of Genetic Counselors' Professional Status Survey (2010) showed that 83 % of counselors provided direct clinical services, while 17 % indicated non-clinical roles. The specific roles reported by counselors in the US included laboratory support, management, marketing, business development, research, teaching and clinic co-ordination. The main difference in South Africa appears to be the extra emphasis on the research role of counselors. Findings from a recent study in Europe, although addressing slightly different issues to those of the present study, showed that most counselors there were involved in the initial patient contact, in explaining genetic tests, ordering tests, obtaining informed consent, providing psychological support, and, as in South Africa, they were managing cases autonomously (Skirton et al. 2013).

In order for the service of genetic counselors, as a part of the team providing genetic services, to be more fully and appropriately used, and their status and abilities better acknowledged, their educational role needs to be extended, since the education of medical and paramedical practitioners in this regard, is essential. In 1982, in Johannesburg (Kromberg and Berkowitz 1986) and in 1999 in Queensland (Kromberg et al. 2006) general practitioners comprised the second largest referral source of clients. For this reason and because the local service is still building up a referral base, further genetic education of general practitioners would be worthwhile. Such education programs have been requested by these practitioners, for example, as a result of a recent survey in which they acknowledged the gaps in their understanding of cancer genetics (Van Wyk 2008). The multidisciplinary nature of the genetics team means that, when a patient is referred, an assessment can be undertaken by the medical geneticist and the counseling provided by the counselor, where appropriate. Links between primary care workers and genetic services have been receiving attention in the UK too. One study showed that, although overall awareness among GPs was poor, interventions to improve links could lead to an increase in appropriate referrals (Turnbull et al. 2007). Counselees who receive genetic counseling have also been studied in the USA, and the findings show that they are generally highly satisfied with the encounter, find that their needs are addressed and they have received emotional support where necessary (Aalfs et al. 2007). Similar results were obtained in a South African study undertaken by the first locally trained genetic counselor (Levy 1992).

Although the sample for this study was small and not all the available genetic counselors responded, it did cover most of the practicing group. Another limitation was that the study was planned as a preliminary one and therefore issues were not investigated in-depth. Furthermore, the study was undertaken prior to 2009 and the situation may have changed somewhat now. Nevertheless, it provides a good initial insight into the roles genetic counselors play and the various services they offer in South Africa.

Conclusion

The results from the present study show that genetic counselors contribute strongly to the field of genetic counseling in South Africa. They have meaningful roles in counseling, teaching, research, marketing of the service, public engagement and administration. The profession is still young and in a developmental stage, but roles are evolving and becoming appropriate to the local populations and their genetic needs.

The findings have implications for the practice of genetic counseling since they suggest that more marketing of the profession should be undertaken to increase its recognition, more community education should be provided and referrals sought so that more patients can be assisted. Lobbying of the Health Department (and possibly the private diagnostic laboratories) needs attention, so that more job opportunities are created.

It is recommended that, in future, a similar project to the present study could be planned, but with different methodology, more in-depth questions and/or perhaps the use of exploratory focus groups. Also, a follow-up study would be worthwhile to assess the nature of the expansion of the roles and development of the profession, since the present project was completed. Due to the multicultural nature of the population in South Africa, a study to provide information on how counselors alter their counseling with patients from different ethnic groups, would be recommended, with input from anthropologists (a multicultural PhD study, on advanced maternal age patient counseling, is presently being completed by one of us, TW)

The genetic services, at present, are meeting less than 10 % of the local genetic needs (Kromberg et al. 2012) and staffing urgently needs to increase if the balance is to be met, in the future. Both public and private support is necessary in a country where other health needs (such as the overwhelming HIV/AIDS epidemic) tend to dominate. However, demand for the services of genetic counselors should escalate as the medical and lay public begins to realize what their services can offer. Increasing visibility and demand will have implications for job creation, service delivery and training in the field, as well as implications for the many people in the community who need the service and would benefit from receiving it.

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