Challenges for providing genetic counselling in Colombian genetic clinics: the viewpoint of the physicians providing genetic consultations

Clemencia Rodas-Pérez¹ · Angus Clarke² · John Powell³ · Margaret Thorogood¹

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Abstract This study explores the provision of clinical genetic services in Colombia, in order to promote improvements in these services. We carried out semi-structured interviews with 20 doctors working in genetic clinics, and we report the challenges in providing genetic counselling that they have identified. Education and training in genetics in Colombian medical schools were described as inadequate, and interviewees found that many of their medical colleagues knew too little to be able to refer appropriately to a genetics service. The doctors interviewed recognised their own limitations, including lack of knowledge, absence of training in communication skills and the strong influence of religious beliefs on the consultations of some colleagues. There may also be communication problems during the consultation: patients may fail to understand the explanations given or to accept the incurable nature of genetic conditions. There are also problems with access, because genetic services are not covered by the health insurance system in Colombia. More training in genetics is required at medical schools in Colombia, at the undergraduate and postgraduate level. There was a consensus that most medical specialities are limited in their knowledge, awareness and understanding of genetics. Furthermore, many medical geneticists did not believe that other health professionals should work as genetic counsellors. These findings may assist in the

development of improved genetic counselling services in Colombia and in the establishment of an enhanced academic curriculum of basic and clinical genetics at Colombian universities.

Keywords Colombia · Genetic counselling · Doctors' views

Introduction

Although developing countries such as Colombia have started to apply genetic knowledge within health care, this is usually approached from the perspective of clinical diagnosis, with not enough attention to genetic counselling issues or to access to laboratory genetic investigations.

Genetic services were first established in Colombia in 1964, predominantly in association with research projects. Since then, regular genetic services have started in ten Colombian cities, predominantly based in university hospitals and private institutions located in the central regions of the country. The main private and public universities have their own genetic clinics and laboratories, and some engage in collaborative research with institutions in Europe, USA and other Latin American countries.

Laboratory investigations for genetic disorders have recently become more widespread in Colombia. When last reviewed, however, there was no access to such services through the public health system (Penchaszadeh 2004). Access to genetic services is restricted by health-care reimbursement laws, which do not cover genetic services or genetic tests (Giraldo 2004). Unfortunately, neither the compulsory health service plan (POS) nor the private medical insurance plans cover cytogenetics or DNA testing. In 2012, however, the Orphan Diseases Law was adopted and, currently, geneticists are working on the application of this law through

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✉ Clemencia Rodas-Pérez
clemencia.rodas-perez@warwick.ac.uk

¹ University of Warwick, Gibbet Hill Road, Coventry, UK CV4 7AL

² Cardiff University, Cardiff, UK

³ University of Oxford, Oxford, UK

regulations in the context of genetic diseases (H Mateus, personal communication; Federación Colombiana de Enfermedades Raras (FECOER) 2012). Currently, only some biochemical tests are covered for certain inborn errors of metabolism. For this reason, laboratory investigations for genetic disorders are very limited.

Colombia started to develop a research programme in molecular genetics in 1993. A number of research projects in clinical genetics are performed at Colombian universities, supported by the Colombian Institute for Science and Technology Development (COLCIENCIAS).

At present, the main research projects focus on the best-recognised genetic diseases in Colombia, including cystic fibrosis, Duchenne muscular dystrophy, Marfan syndrome, Huntington disease, myotonic dystrophy and the hereditary ataxias.

All faculties of medicine in Colombia have undergraduate courses in basic genetics, usually as part of general biology, and most public and private medical schools have courses in clinical genetics (Giraldo 2004; Penchaszadeh 2002). Though the teaching of basic genetics in medical schools is mandatory, their quality varies and there is uncertainty whether genetic knowledge and information is interpreted accurately and used appropriately (Giraldo 2004). Furthermore, there is no agreement about the content of clinical genetic courses in medical schools; thus, training and knowledge of physicians varies a great deal. There are three postgraduate training programmes in human genetics, but just one in clinical genetics. Although there is a formal genetic training requirement for medical geneticists employed in health institutions, there is no official recognition of the speciality of clinical genetics.

Genetic counselling in Colombia (Giraldo 2004) is conducted exclusively by clinical geneticists, as is the case throughout Latin America (Penchaszadeh 2002). These doctors are not only in charge of diagnosis, but they also perform genetic counselling; what makes it more difficult is its access for patients (Penchaszadeh 2004). There are no teaching programmes for genetic counsellors or assistants. Even nurses who take daily care of patients with genetic disorders do not have the necessary training to help patients with their treatment or disabilities (Giraldo 2004).

The aim of this study is to explore the provision of clinical genetic services in Colombia and to promote improvement in these services. Its results may assist in the development of improved genetic counselling in Colombia and an enhanced academic curriculum of basic and clinical genetics.

Methods

We conducted 20 interviews with doctors practising in clinical genetics to explore, in some depth, their opinions and attitudes concerning clinical genetics, genetic counselling and genetic

services in Colombia. We used semi-structured interviews, because every doctor interviewed was a ‘key’ informant, and we wished to gather and compare the understandings, insights and opinions of each one (Silverman 2001; Green and Thorogood 2004).

Data collection for the study was conducted in two parts: a pilot study, between December 2006 and January 2007, and the main part which was completed in July–August 2007. This project was evaluated and approved by the appropriate research ethics committees for research projects.

We approached 32 clinicians trained and practising in genetic consultation and counselling, in the major cities in Colombia, by email or telephone. We received a positive answer from 20 physicians (of the 35 clinical geneticists in Colombia) and then agreed an occasion to interview each of them. The interviews were performed at the doctors’ convenience, in a private room at their place of work, and were audio recorded. Its duration varied from 10 to 45 min. Informed consent was obtained from all participants in the study.

Of the 20 interviewees, 14 worked in Bogotá and 6 worked in other cities; 9 were women, and 11 were men. The age range of doctors interviewed is described in Table 1.

The interviewed doctors work at universities and carry out consultations at private, public and university hospitals. Not all hold a degree or formal accreditation in genetics, but all have an important link with academia, being based at the main universities in Colombia. A few interviewees also have their own private practice in clinical genetics.

Most doctors work with all aspects of clinical genetics and are tutors in human and clinical genetics in the principal Colombian universities. Some combine their work in general genetic clinics with specialised clinics for particular genetic conditions and congenital malformations. Those clinics are located at the main universities, and several are funded by the Colombian science research councils and/or from other countries.

The topics chosen for the interviews with doctors are summarised in Table 2, and the topic guide is in Appendix 1.

Audio recordings were listened several times for clarification and to make sure that all the information had been retrieved. Interviews were transcribed and translated from Spanish to English. Transcripts were then analysed systematically, by simple thematic coding, based on interpreting the content of the talk and grouping it into categories. The coding and preliminary results were regularly discussed among three of the authors. The results are presented as the compiled

Table 1 Age range of the 20 doctors interviewed

Age (years)	Number of doctors
25–35	9
36–45	4
>46	7

Table 2 Interviews with doctors—topics covered

1. Tasks of physicians in respect to clinical genetics
2. Education and training of physicians in clinical genetics and genetic counselling
3. Risk assessment and delivering risk information in Colombian genetic clinics
4. Physicians' level of knowledge and training in clinical genetics and counselling
5. How difficult is it to provide genetic counselling in Colombia?
6. Genetic counselling and the new legislation on abortion in Colombia
7. Differences between clinical genetic consultation and genetic counselling
8. Influence of Colombian physicians' own beliefs on genetic counselling
9. Enabling other health professionals to perform genetic counselling in Colombia

summaries of the views of the informants, with some data given as illustrative examples.

Results

Issues in training and education for clinical genetics and genetic counselling

Most of the interviewees agreed that education in basic genetics and clinical genetics is deficient at the undergraduate level and, indeed, very heterogeneous among the different medical schools. Genetic counselling is entirely absent from the curriculum. In contrast, their views concerning postgraduate medical education varied. While half of those interviewed reported training in clinical genetics at university, none had formal training in genetic counselling. Interviewees judged training in clinical genetics to be insufficient, only through the observation of clinical practice. Several interviewees, however, considered it to be of a higher standard than in previous years, and some believed that some Colombian universities provide a better education in medical genetics than other Latin American countries.

There was a widespread opinion that most doctors working in genetics have learned more from colleagues or their own study and experience, than from formal training.

Doctor 1: "...even in my master's degree, I didn't have enough tools to be able to do a consultation and/or counselling...I have received priceless knowledge and experience from my tutors and colleagues...and, of course, I have read many books and papers..."

Some interviewees took a clinical genetics course when pursuing their master's degree. There is a general feeling, however, that something was missing from that training. In

particular, the absence of courses is related to communication skills and counselling.

Doctors explained that most courses emphasise knowledge on the 'facts' relevant to a wide range of genetic conditions and their risk assessment, without specifically studying the most common genetic diseases in Colombia. Several doctors mentioned that the extensive ethnic diversity in Colombia required the study of all genetic diseases to ensure appropriate risk assessment.

Doctor 2: "...The ideal situation would be to have Colombian data (incidence and prevalence of genetic diseases), but as we don't have it, we have to prepare for genetic counselling with our knowledge of the disease, inheritance patterns and inheritance mechanisms. The risk is then assessed with incidence and prevalence data from other countries..."

The doctors interviewed believed that there is a lack of experience with some clinical topics related to genetics. They need time to keep up to date with the published literature and for their own professional development, on a daily basis, because genetics is an area of rapid and continuous change and it is impossible to cover it all in university courses.

Doctors who studied medicine in the 1970s and 1980s explained that they found it difficult to access training in genetics in Colombia: most studied abroad and several had training in clinical genetics and counselling when visiting foreign institutions. It is common practice to contact and visit genetic centres in the USA, Canada, Mexico, Chile and Brazil. The USA and Canada are particularly attractive, as they are closer than Europe and run well-established genetic services.

Despite the limitations described, most doctors reported that the situation has improved in recent years; universities have been pressured to teach more genetics and remain updated in the basic sciences and genetics, for their courses to be recognised nationally and internationally.

There was also a consensus regarding training in genetic counselling; doctors recognised that training by observation is insufficient and it should be a priority to deliver a formal programme of genetic counselling for clinical genetics students. This has already been established in other countries, so there are already recommendations for training curricula for people working in the provision of genetic diagnosis and community genetics education (Parker 1994; Van Langen et al. 2003; Skirton et al. 2010; NHS National Genetics and Genomics Education Centre (2014).

Delivering genetic and risk information—a challenge of knowledge and comprehension

When delivering information to patients, interviewees stated that they used many resources to explain the characteristics

and evolution of the disorder under consideration, its severity, inheritance patterns and associated risks. The use of numbers, percentages and proportions to explain the risk of transmission is a common practice (Weil 2000; Rose and Lucassen 1999; Skirton et al. 2005; Parsons and Atkinson 1993; Henneman et al. 2008) and was reported by all interviewees.

Interviewees stressed that delivering genetic information in a way that can be understood by patients can be a difficult task, especially when risks, percentages and probabilities are involved. Risk is an especially difficult concept, as it integrates probability of an event with the degree of adversity in an inevitably subjective manner:

Doctor 3: "...I know as well that patients' interpretation of risk is very subjective, for example, for some patients 25 % of risk to get the disease doesn't mean much, but for others 25 % is too much to take the risk..."

Three doctors explained that patients' comprehension, interpretation and recollection of the data are affected by their emotional situation and their coping ability. Doctors reported cases where patients felt overwhelmed and simply could not pay attention to the explanations, as their feelings and thoughts did not allow them to concentrate.

Nine doctors reported that many patients did not understand their explanations, even when patients say they do. This is worse when only one member of a couple is present in consultation. There is no guarantee that the matters discussed will be reported in an appropriate way to other members of the family.

Doctor 4: "...Sometimes it's difficult when...for example, only the patient's mother comes to see me...of course the theory says that the geneticist needs to give genetic counselling to the couple...but you know, that here, in Colombia, you can sit waiting for a whole year and you will never see them come together to the consultation. The problem is that you are not sure how accurately the information will be transmitted between the couple and the family..."

However, several interviewees suggested that doctors could help patients understand some genetic concepts; for that, it is necessary to explore the education level of the patient, before starting explaining the genetic condition.

Doctor 5: "...Generally, I try to define the educational status of my patient and then I decide what the best way is to give the information and risk assessment..."

Doctors agreed that it is very important to use lay language. Professionals providing genetic counselling should know the power of language in the process of communication and the

importance of integrating it with other factors, such as culture and beliefs (Middleton 2005; Middleton and Wiggins 2013; Chapple et al. 1997).

For many doctors, the nature of genetic conditions (which can often be managed or ameliorated, but not cured) makes it problematic to help people understanding genetic concepts and adopting a more realistic attitude. Consequently, they have to support patients facing difficult decisions and they also need to be able to give bad news and, then, support patients as they adjust to them.

One doctor expressed the opinion that the main cause of the cautious attitude of patients towards genetics is their lack of knowledge about why they have been referred to the genetics clinic. He believed that, before delivering any information to patients, it is essential to first introduce them to the subject of genetics and the reasons for referral:

Doctor 6: "...The genetic consultation requires an introduction. It starts from the moment I explain to my patients what the reason for the consultation is, what the general purpose of genetics is, and why they have been referred to this clinic...most of my patients don't know why they need a genetic consultation..."

Ten doctors expressed the opinion that the delivery of genetic information requires more than one session, particularly to allow the patient to discuss the matter at home, and then patients would be able to prepare questions and comments for the next consultation. Young doctors said that they had a preference for making several appointments with patients. They explained that it allows them to read more about the condition and to extend their knowledge about it:

Doctor 7: "...For uncommon genetic diseases, I use the information from journal publications. Sometimes, I don't have 'fresh' information at the consultation, so I ask my patients to come back...and then I can review the current literature, in order to prepare a complete report for my patient for our next appointment..."

Many interviewees talked about the importance of identifying the most appropriate way to deliver information and whether the patient has understood it. Other challenges discussed were how to help people understand why it is important to know the reason of their referral to the genetics clinic and to know about the condition they have, why the requested tests are necessary and why they need to come back to the clinic.

The level of experience that patients have of the disease was a very important issue for some doctors. Many patients know their condition very well, because they have other relatives affected: doctors find it easy to orient them during the consultation; however, for the patient who learns about the

disease for the first time, this may cause problems for everybody: doctor, patient and relatives. Doctors feel that they have to deal not only with the psychological impact of the bad news on patients and their families but also to provide them all the information and understanding they do need.

Professional status

For most doctors, the very uneven knowledge on genetics among different health professionals—general practitioners, specialists and others—can create problems with the diagnosis and referral of potential patients. Several interviewees feel that other doctors do not value their knowledge, and they find it challenging to convince other clinical specialists of the importance of medical genetics. They think there is a lack of knowledge of genetics and a general failure to appreciate its applicability to health care.

Doctor 6: “...So, if genetics is not relevant for general physicians and other specialists, it will be much less so for patients. I think doctors should work very closely with paediatricians and gynaecologists...but it’s difficult; it requires professional education...”

This situation is not exclusive to Colombia. A study of McCahon et al. (2009) exploring the attitudes of UK primary care doctors (GPs) towards having a role in the provision of routine genetic services, illustrated that a significant number of GPs are untrained or unwilling to provide that support. Also, the research of Kyne et al. (2011) which aimed to determine the quality of referral letters to a cancer genetic clinic showed that, despite finding that many referrals were reasonable and appropriate, there is still the need to consider targeting medical education and other resources to improve compliance of referrals with the criteria set.

Regarding professional issues, another topic discussed by the interviewees was the possibility of training other health professionals to deliver genetic counselling. The doctors interviewed considered it inappropriate in Colombia, because (they feel) only doctors should be allowed to provide genetic counselling.

Doctor 8: “...I disagree; doctors are doctors because of their dedication and experience of years of study and practice. That cannot be improvised for other professionals, even though they may be health professionals...”

Some interviewees said that, if other health professionals were allowed to provide genetic counselling, it would divide something which should be together and would threaten the trust long established between doctor and patient. Another problem described was that patients might regard it as inappropriate and strange for non-medical health professionals to perform genetic counselling.

In contrast, other interviewees (all <35 years) reported that having other health professionals trained as genetic counsellors would be a good idea. As they have noticed that the request for genetic consultations has increased in the last few years, they considered that having non-MDs trained in genetic counselling will become a need, because medical doctors have insufficient time, as genetic counselling is a very demanding activity and it is only done by those who think it is an important issue.

Even if there were suitably trained professionals available, it is uncertain whether the health system would be ready to hire them: the doctors interviewed thought that it would be very difficult to find opportunities to employ these new professionals.

Doctor 9: “...In my opinion, it would be excellent to have other health professionals working in genetic counselling. I know that in other countries nurses doing genetic counselling are very capable (even more capable than many doctors)...I think that they will do it very well, as they will be trained specifically to do that. However, I would be concerned for the demand of those counsellors...I don’t think there will be space for them at the present”.

The use of health professionals other than doctors to deliver genetic counselling has been successfully implemented in many countries (Andrews 2001; Ormond et al. 2003; Weil 2000; Penchaszadeh 2004; Cordier et al. 2012); however, it is important to determine how this training should be appropriately delivered, especially for professionals without a background in health sciences (Greendale and Pyritz 2001).

Resources—time and institutional service

The challenge of limited resources was also discussed by the interviewees. One important resource is time. Interviewees reported that genetic counselling at public hospitals and some private institutions is affected by the time limit for consultations, as non-medical administrators provide doctors instructions as to the number of consultations required in a given time. Some doctors working for such clinics reported that they are constrained by this institutional pressure. In contrast, in the remaining genetics clinics and in the university hospitals, the time allocated for consultations was considered sufficient.

Doctors considered that the health system does not take into account the needs of individual patients with genetic conditions. Additionally, the law excludes all congenital defects from coverage because they are considered to be costly and these are not the responsibility of the POS (Compulsory Colombian Health Insurance Plan). All doctors interviewed felt frustrated because their clinical assessment of these patients is constrained by the inability of most patients to pay

for their health care. Doctors do not know whether to order tests and therapies, as they know that patients often cannot afford them.

Doctor 6: "...we don't have the support of the POS as we are not paid for all the time we spend with our patients. The POS doesn't cover the cost of specialised genetic tests, so we are in a serious bioethical problem where the government doesn't give us the necessary tools to support and help our patients".

The doctor–patient relationship

Seven doctors considered that, given how difficult it can be for them to deliver information to their patients and given the gaps in provision within the present system of Colombian health care, it is imperative for them to establish a doctor–patient relationship, based on communication and trust, if their genetic counselling sessions are to have any value. This reliance on the traditional, pre-established relationship between doctor and patient is considered by most of the interviewees to be the most important element. As Colombian culture is still very respectful and faithful to doctors' opinions, the trust between doctor and patient is one of the most important aspects in genetic counselling in the country. The environment of the consulting room is often highly charged with emotions and uncertainty during genetic counselling; for this reason, establishing a good doctor–patient relationship was also seen by some as a challenge. They considered that the doctor's empathy for the patient is important and beneficial for giving information and delivering good and bad news, particularly when patients are expecting a positive outcome from the consultation.

Meaning of genetic consultation and genetic counselling

The perception of the difference between a genetic consultation and genetic counselling was variable among interviewees. Two doctors regarded these concepts as complementary and that they cannot be considered separately. In general, they defined genetic consultation as a very clinical approach to the condition, where doctors do the physical examination, fill in a clinical history and request general information regarding the symptoms and evolution of the genetic pathology.

Doctor 11: "...Basically, both are complementary. I cannot do a genetic consultation without genetic counselling..."

Doctor 10: "Err...there shouldn't exist a difference between both terms. I think genetic counselling must be the last part of a genetic consultation. During consultation I learn about the patient's situation and I give information on the disorder...and with all that information I

do genetic counselling. All genetic consultations should include genetic counselling".

A third group recognised that there is a difference between a genetic consultation—with or without any specifically counselling dimension—and the more standardised clinical aspects, such as physical examination, filling in a clinical history and requesting general information regarding symptoms and evolution of the disease. They consider genetic counselling to be the process where a doctor gives risk information, patients and relatives are supported and reassured about their problem, and some possible actions to follow are explained, so that the patient may make a decision about the condition. Interestingly, one of the interviewees suggested that all specialities should have a counselling approach:

Doctor 6: "...Genetic consultation is basically a medical concept and it's no different to the consultation of other specialities. But genetic counselling requires the diagnosis and knowledge of the condition and additionally has implications for not only the patient, but the whole family. I think this kind of encounter shouldn't be limited to the genetics field. At the end of the day, all specialities could have an occasional counselling meeting...it is all about the patient's health and it involves the whole family...it shouldn't be exclusive for genetics conditions".

For most doctors, the main objective of genetic counselling is to deliver genetic information and provide risk assessment to patients and relatives in a clear and proper manner. Interestingly, two of the most important aspects related to the process of genetic counselling were rarely considered by many doctors: the patients' psychosocial needs and their concerns about the future.

Some interviewees, although they work at genetic clinics, explained that genetic counselling is not a compulsory practice. It is not considered as an essential part of the process of seeing genetic patients and their families.

The variation in these responses was unexpected, in particular those that considered genetic counselling as a secondary and non-essential process. As Rubinstein (2008) argues, in addition to the expected clinical expertise, it is important that doctors know that a skilled genetic counselling approach is required to guide patients and their relatives through an overwhelming amount of technical and medical information and also to deal with the emotional burden of the decision-making process. The genetic counselling process cannot be considered optional.

New legislation on abortion in Colombia and the genetic counselling process

Legislation on abortion has been a controversial issue in many countries, whether developed or developing (WHO and World

Health 2006). In most Latin American countries, the practice of abortion is illegal and, consequently, it is not considered for genetic diseases or congenital malformations. In Colombia, abortion was illegal until May 2006, when the constitutional court ruled it to be legal in cases such as (i) where a woman's life or health is in danger, (ii) where pregnancy was the result of rape, and (iii) where the foetus has malformations incompatible with life outside the womb. Abortion is, thus, now permissible under the law (MinSalud 2006), for those particular circumstances; however, it is still being rejected and, consequently, prenatal diagnosis is being stigmatised, particularly by the Catholic Church and conservative parties.

As González Vélez (2005) pointed out, before 2006, Colombia was within the 0.4 % of the world population where abortion was completely prohibited. It was estimated that nearly 350,000 clandestine abortions were performed each year. Recent statistics, between 2006 and 2009, illustrated that foetal malformations were the reason mentioned in 62 % of abortions performed in Colombian clinics; 20 % were due to a risk to the life of the woman, while rape or incest was cited in over 17 % of cases (WHO and World Health 2011).

Regarding this issue, most doctors said that the law is difficult to apply because it goes against the country's predominant Catholic religion, values and precepts:

Doctor 11: "...It is a good option for many people and many families. However, and again, religious beliefs don't allow having this alternative in consideration".

Doctor 12: "...The voice of the church is very loud and clear about genetic manipulation, abortion and birth planning...there is nothing we can do about that".

The new law is unclear particularly regarding its scientific approach. Eight years after its establishment, it is already subject to much criticism and is being followed only by a few doctors and in a few institutions.

Opinions were divided about the question on how the new legislation would affect the genetic counselling process. Some doctors think that the new law may be problematic instead of helpful:

Doctor 10: "...Accepting abortion means that we are intolerant of genetic diversity. I think that society should accept people with genetic diseases in an adequate way. I think, in the past, abortion was a transitional measure to deal with genetic disease...which sadly is still current. Abortion solves the problem only in the immediate, short term..."

Some interviewees find that this legislation goes against the beliefs of patients and it should not be considered as an option for them. Contrarily to this, other doctors found this new legislation to be an acceptable alternative for their patients; they thought it

might be useful and applicable in many cases and specifically in those where there are severe genetic malformations.

Doctor 1: "...I think [the new legislation] affects genetic counselling in a positive way, as we can offer our patients another option...It widens the horizon of our patients...and offers them the possibility to have an abortion in safe conditions".

Although opinions are divided, all the interviewees agreed that the new law is unclear in many respects and needs to be revised. They agreed that the classification of genetic conditions, which count as an acceptable indication for abortion, is confusing. Some considered that the concept of viability is very difficult to define. Additionally, some said that the revision of the law needs to be a process where doctors, other health professionals and the legislature work together to produce more realistic and more precisely framed legislation.

Doctors' beliefs and genetic counselling

Regarding the issue of the influence of doctors' beliefs during genetic counselling, most doctors consider themselves capable of putting to one side their own moral precepts during the consultation. Some comments were made regarding the difficulty of maintaining a neutral position; however, most felt that they knew how to manage the situation. Interestingly, some interviewees talked about the influence of other colleagues' beliefs on their counselling practice.

Doctor 13: "...Sadly, the religious factor also affects the ability of some colleagues to provide proper and unbiased genetic counselling".

A concern was expressed by some about the attitude of certain doctors who take their own religious beliefs into the genetic consultation, particularly in the setting of a prenatal consultation:

Doctor 14: "...There are lecturers who are very religious and they take those beliefs into the genetic counselling. They distort genetic counselling completely, particularly in those cases related to prenatal diagnosis. This is something that worries me very much".

Some interviewees also reported that many doctors are compelled to observe the precepts of the institutions where they work, particularly for those working in Catholic institutions.

Discussion and conclusions

In Colombia, medical education in genetics is rather variable and considered to have been neglected in some universities.

This problem has also been described in other developing countries (Penchaszadeh 2004; Alwan and Modell 2003). The lack of consistent national standards for education in genetics at medical schools is problematic, both for undergraduate medical students and for residents specialising in medical genetics, because it creates a disparity in the ability of doctors to play an active role in the provision of genetic services. A clear understanding of clinical genetics and genetic counselling for all medical students is crucial for the success of specialised clinical genetics services (Weil 2003; Rose and Lucassen 1999; Harper 2010). Just as it is important for a doctor to have the necessary knowledge to be able to diagnose and treat a disease, it is also critical that during the doctor–patient encounter, genetic knowledge and information is properly delivered to the patient as well as the support and guidance the doctor can provide (Emery and Hayflick 2001; Greendale and Pyeritz 2001; Finkler et al. 2003). Furthermore, recent advances in genetics are having a great impact on the education and training of all health-care professionals. In countries like the USA, Canada, UK and Germany, among others, a new curriculum for medical students and nurses has already been developed (Fears and Weatherall 1999; Weil 2000; Kirklin 2003; Kroese et al. 2007). There are also studies regarding genetic test evaluations for clinicians (Burke and Atkins 2002; Fears and Weatherall 1999; Kroese et al. 2007) and the needs of non-specialist health professionals for education in the more recent, genomic aspects of medicine (Skirton et al. 2010; Feero and Green 2011; Tabor et al. 2011; Foundation 2011a, b; Maniolo 2013).

Developing skills in genetic counselling should be a priority for doctors training in clinical genetics. It was the view of the professionals interviewed that GPs and other health professionals should have a good awareness of these issues, in order to ensure they can give the appropriate information to their patients in a helpful, non-directive manner and so that they can make appropriate referrals. A study in the UK (Watson et al. 1999) showed that GPs recognised they lack knowledge and skills related to genetics and do not feel confident in managing patients with genetic conditions. Given the pace of change in genetics, this situation is unlikely to have improved.

The process of learning about genetic counselling should not be limited, as at present, to the passive observation of practice. Medical schools should design a formal programme with genetic counselling as an important topic in clinical genetics, as in other countries (Lantigua et al. 2007; Marcheco 2009; Novoa and Burnham 2011).

In Colombia, the medical school curricula could usefully be standardised, with specific modules or courses related only to genetics and with a requirement that all medical schools have a minimum number of hours for teaching genetics. Proper training in genetics should be considered for students of other health-care professions, such as nursing. This would ensure that all doctors and other health-care professionals

would have an acceptable minimum level of knowledge and understanding of genetics. This would counter the present widespread inconsistencies in professional awareness and competences described by all doctors interviewed. Additionally, a similar approach should be considered for postgraduate studies in clinical genetics.

Explicit attention to communication skills should be required for all students of medicine and of the other health-care professions, in particular for those who are going to work in a genetics clinic. Universities and health institutions should offer additional development courses for doctors and nurses to keep them updated in genetics. Despite the problems experienced when integrating communication with other clinical skills, many countries have accepted that training communication skills is an essential component of the education of doctors and other health professionals (Skirton et al. 2010, 1998; Kurtz et al. 2003; Mathews 1983).

Among the main challenges encountered in the practice of genetic counselling, reported by the doctors interviewed, were the delivery of genetic information and risk and the wide range of diagnoses with which doctors must be familiar. Most talked about the need for exploring the patient's level of education, personal beliefs, previous experience with the condition diagnosed and the social circumstances of the patient and their relatives, in order to define the most appropriate way to deliver genetic information to them. One key aspect to consider is that patients' comprehension might be conditioned by the way the knowledge is transmitted by doctors, which is closely related to their competence in communication. However, it is not always possible to establish trust in a doctor–patient relationship, especially when patients' expectations are unrealistic or inappropriate (Michie et al. 1999). In general, doctors considered that trust, empathy and respect are the main characteristics that allow the creation of a good relationship between doctors and their patients.

Doctors were asked about the meaning for them of a clinical genetics consultation and of genetic counselling and what approach they used in their consultations. Many responses were very elaborate and similar to the general definitions found in textbooks. While some were brief and the concepts of clinical genetics and genetic counselling overlapped, for others, there was no difference between these two concepts. The relationship between the concepts required elaboration from all interviewees, who invariably hesitated before responding: they had to take time to reflect first. Three doctors mentioned the patient's psychosocial needs and concerns as factors to be considered during counselling. Several issues were addressed and discussed in defining an acceptable and appropriate response, but relating the account given in interviews with the details of professional practice in the clinic would require a separate, additional study.

Doctors often expressed dissatisfaction with the quality of genetic counselling provided by colleagues working in

different institutions. Doctors at other universities were unhappy about the genetic counselling provided at the Catholic universities: the Catholic Church is very influential in Colombia, which limits the application of prenatal and pre-conception genetic testing, contraception and abortion. Some doctors said that they felt uncomfortable discussing these issues during genetic consultations, while others were less explicit regarding their own beliefs; however, some indicated that they have to follow the rules of the Catholic institutions within which they worked. For some, this may be a combination of their own beliefs and the institutional precepts, while for others, the issue is predominantly the institutional pressure.

According to Oyama and Koenig (1998) patients are generally more involved in religious beliefs and practices than doctors. It is important for religious patients that they know their physicians' beliefs and share their own beliefs with them. An appropriate training in genetic counselling and communication skills should help doctors to address difficult topics during consultation (Stewart 1995; Ferguson and Candib 2002), develop an appropriate approach to requests from patients for self-disclosure and manage issues that are in disagreement with their own beliefs. Continuing support for qualified genetics professionals can be obtained through ongoing psychological supervision (AGNC Supervision Working Group et al. 2007).

The question of whether it would be helpful and appropriate for non-medical professionals to provide genetic counselling was not met with enthusiasm. Five doctors found the idea plausible, but still thought it would be difficult to introduce other health professionals into the health-care system in Colombia, because of budgetary and institutional constraints. Fifteen disagreed with the idea. Their reasoning is related not only to establishing the counsellors' professional role but also to the same budgetary constraints and difficulties. This negative response is in tension with the view, expressed by some, that there are insufficient geneticists in Colombia to meet the demand for clinical genetic consultations and counselling.

It is possible that some doctors feel threatened by the idea of having other professionals working in their area. There is also the traditionally conservative nature of the medical profession; they find it inappropriate that non-MDs practise as genetic counsellors, and they add that it would be unacceptable to their patients as well. For many generations, doctors have had control of all medical decisions and advice in Colombia, with other health professionals acting only in support of their decisions (Goffman 1956; Elliott and Hein 1978). As in many other areas, there is a hierarchy in health care, which has persisted because of what is at stake: this can sometimes be life and death. At present, there is no overt conflict in status among health-care professionals in Colombia, where doctors are still at the top, whereas in some other countries, nurses have become much more involved in decision-making

for diagnosis and treatment. This may happen because of the heavy demand for services in hospitals and health centres (Snelgrove and Hughes 2000).

The same situation has been encountered in other countries in relation to genetic counselling. As described by Greendale (2001), the perceived lack of medical genetics professionals has led to the involvement of primary health-care providers in the delivery of genetics services. However, to achieve this, it was essential to provide the appropriate training to these professionals and for them to work as a team with clinical geneticists (Andrews 2001; Ormond et al. 2003; Weil 2000).

Having other health professionals providing genetic counselling in Colombia might be very useful, not only because it would meet a pressing need to support doctors in the genetic clinics but also because it could allow doctors to do a more thorough clinical consultation, as they will not need to do all the counselling themselves. In some situations, this will shorten the waiting list for genetic consultations, as doctors will have more time to organise their appointments. Additionally, the involvement of other health professionals in genetic counselling would clearly require some clarity in the definition of the two components of a genetics consultation: clinical genetics and genetic counselling.

Regarding the legislation on abortion, there was consensus among interviewees that, before applying the law, this would need to be thoroughly revised by a broadly based panel of professionals from different disciplines. For the interviewees, a law which is not grounded on a firm understanding of both science and clinical reality cannot effectively be implemented. This is an issue, the resolution of which is limited by the influence of the Catholic Church on the lives of Colombian people. It is similarly problematic for doctors working within Catholic institutions to deal with the institutional pressure and to comply with their own ethics. The responses of all the doctors interviewed were complex and centred on an analysis of the law and its flaws. Only two doctors talked openly about its benefits for their patients.

The crisis of this law peaked in September 2010, when the new Colombian General Attorney, an outspoken Catholic, announced that he was preparing a law to put before the Colombian Congress to allow doctors to object to perform an abortion on conscientious grounds. Currently, all clinics are required by law to safeguard the right to abortion, by referring patients to another institution if they lack doctors willing to perform the procedure, but this is not always enforced.

When facing the future, it is important to consider how changes should be introduced in Colombia in a manner consistent with the social, cultural and economic realities of the country, the status of the health system and its existing policies, the level of people's education and the current level of knowledge of genetics among health professionals. It is crucial to define appropriate and realistic approaches that can

improve effective delivery of genetic services. Future research in this area might evaluate whether the changes implemented are working and determine what further adjustments would be appropriate and helpful.

The findings of this interview-based study are likely to be broadly representative of the views of clinical geneticists in Colombia, as a majority of this professional group within Colombia participated. There may have been, however, a subgroup of professionals who were more reluctant to participate and who hold different views. There may also have been some reluctance among some participants to express fully their views, where these depart from traditional, Roman Catholic beliefs, by fear that confidentiality of their answers might be breached. The other principal limitation of this study is that it was based on clinicians' accounts of their practice, without a detailed study of how this relates to their actual behaviour in the clinical setting. Observing and assessing the relation between descriptions and real clinical practice is challenging but would be important and rewarding for clinicians and their patients.

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Compliance with ethical standards All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. Informed consent was obtained from all individual participants included in the study.

Appendix 1 Topic guide of interviews with doctors

Job of physicians in respect to clinical genetics

Background/first contact with genetic topics

Workplace/clinical/universities/other institutions

Professional status

Education and training of physicians in clinical genetics and genetic counselling

Personal learning experiences in clinical genetics and counselling

Voids on training and education in genetics at medical schools

Genetic knowledge and training of GPs and other health professionals

Importance and status of genetic counselling in Colombia

Risk assessment and delivering risk information in Colombian genetic clinics

Approaches for delivering genetic and risk information

Patients understanding of doctors' explanations

Communication of genetic information among families

Physicians' level of knowledge and training in clinical genetics and counselling

Differences of doctors training depending of the universities

Awareness of the meaning and importance of genetic counselling

Views of training and education in genetics of other colleagues doing genetic consultation

How difficult is it to give genetic counselling in Colombia?

Universities' and health institutions' perception of genetics

Challenges faced during consultation

Problems of knowledge and comprehension (doctors and patients)

Doctor–patient relationship

Resources (budget, time, professionals)

Health insurance cover of genetic conditions

Lay beliefs and public knowledge of genetics

General understanding of the importance of genetic counselling

Genetic counselling and the new legislation on abortion in Colombia

Opinions and views

How this law affects clinical genetics

Abortion and the Catholic Church

Differences between genetic consultation and genetic counselling

Influence of Colombian physicians' own beliefs during counselling

Personal and religious beliefs

Enabling other health professionals to give genetic counselling in Colombia

Opinions and views

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