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ORIGINAL ARTICLE

Genetic professionals' views on genetic counsellors: a French survey

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Abstract The genetic counselling profession was established in France in 2004. Eight years later, 122 genetic counsellors have graduated from the unique educational French program which awards the Professional Master Degree of Human Pathology, entitled "Master of Genetic Counselling and Predictive Medicine". As part of a global evaluation of this new profession by health genetic professionals, we undertook a national survey investigating various aspects such as employment, work responsibilities and integration. To our knowledge, this is the first study to investigate the views of genetic professionals on the genetic counsellors' role. Of 422 French professionals invited to take part in this study, 126 participated. The survey underlines that this profession is significantly recognized by physicians practicing within genetics departments. French genetic counsellors are allowed to manage consultations independently, without the necessary presence of a qualified medical geneticist but under his or her responsibility. Genetic counsellors participate in a wide range of consultations. They provide both information for relevant and for genetic testing and sometimes disclose the genetic test result to patient. Eventually, the role of genetic counsellors appears to be directly dependent from the relationship of trust between the two health professions.

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Introduction

The inclusion of non-medical genetic counsellors and/or genetic nurses as part of a health multi-disciplinary team has been shown to be an effective way to deliver genetic counselling services in a number of countries including USA, Australia, Canada and UK (Emery et al. 1999; Godard et al. 2003). In some European countries, genetic nurses and counsellors have been working for at least 35 years. The first European country in which genetic counsellors were practising was the UK in 1980 with the first master's degree to the University of Manchester in 1992 (Cordier et al. 2012). Due to enhanced diagnostic and genetic testing options, the pressure on genetic services has considerably increased. In order to cope with this increase, the Education Committee of the European Society of Human Genetics (ESHG) was collaborating with other groups of expert (such as EuroGentest) to write and develop better strategies to facilitate professionals' education and recognition of all health professionals that work in a genetic setting in Europe (Coviello et al. 2007). In 2010, the first European document on core competences was produced and used as a basis for education concerning health professionals working in the field of the genetics (Skirton et al. 2010).

In France, setting up a new health profession is a challenging process as it requests several ministerial steps. The genetic counselling profession was established in 2004 when the Law no. 2004-806, 9 August 2004, amended the Code of Public Health (Legifrance; Cordier et al. 2013). Several laws and decrees were necessary in order to develop this profession, which is now well organized by ministerial institutions. Today, 122 genetic counsellors have graduated from the specific

educational program which awards the Professional Master Degree of Human Pathology, entitled "Master of Genetic Counselling and Predictive Medicine". This master was evaluated by the genetic counsellors division from the European Board of Medical Genetics, complies fully with the core curriculum and is suitable training programmes for European genetic counsellors (www.eshg.org).

In a recent systematic review, Skirton et al. (2014) investigated the genetic counsellor's role and indicated that when genetic counsellors work in specialist genetic settings, they undertake a significant workload associated with direct patient care in the clinical environment. For instance, they frequently record the family history, draw the family pedigree and discuss the natural history of the condition and the genetic test, and the possibility of testing in that particular patient.

Previous studies (Skirton et al. 2013; Kromberg 2013) have shown that genetic counsellors manage cases related to a wide range of conditions, predominantly where the diagnosis has been clearly established. As a matter of fact, a number of authors have suggested that given the increasing burden on genetic counselling services, there is argument for an increased use of genetic counsellors in countries where they are under-utilized.

Our main objective was to undertake a national survey investigating various aspects related to the genetic counselling profession in France, such as their employment, work responsibilities and integration. To our knowledge, this is the first study to investigate the views that genetic professionals have on the genetic counsellors' role.

Materials and methods

Design

This study was a cross-sectional online survey.

Participants

We identified 422 professionals working in or closely with a genetic service and all were invited to take part in this study. These professionals were clinical and laboratory geneticists. Genetic counsellors were excluded from this survey. Of the 422 potential participants, 46 were excluded (i.e. either the provided email address was erroneous or participants were out of office at the moment of the survey). From the remaining 376 invited participants, a total of 126 professionals took part in this study.

Measures

The questionnaire, designed by two medical geneticists and three genetic counsellors, was submitted to an epidemiologist, for feedback. It included 31 questions of different types, including multiple choice and short answer questions. Demographic questions related to the participants, specialty and area of practice, the role of genetic counsellors, the type of consultation that may be performed by a genetic counsellor independently, the skills required of genetic counsellors and remarks on training.

Procedure

An invitation letter was emailed to participants. This letter introduced a brief history of the genetic counselling profession, the aim of the study and instructions needed to access and complete the survey. The survey was managed online through a secure website, protected by a username and password. A reminder was sent, a month later after sending the initial invitation.

Results

Participants

One hundred and twenty-six professionals working in various genetics services agreed to participate in the survey (participation rate of 33.5 %): 88 were women (70 %) and 38 were men (30 %) with an average age of 49 years and an average seniority of 17 years [1, 43].

The background of professionals was genetics (57 %), biology (14 %), oncology (10 %) and paediatrics (7 %). Participants worked in areas including prenatal diagnosis (20 %), general genetic counselling (18 %), oncogenetics (16 %), paediatrics/syndromology (16 %) and neurogenetics (11 %) (further details in Table 1). Of the total, 83 % (n=126) of participants worked with a genetic counsellor for an average of 4 years and had one to two genetic counsellors in their service, while 17 % of participants did not work with a genetic counsellor as they either believed that the presence of a genetic counsellor is not necessary to their service or they had difficulties in creating a post.

Table 1 Domain of activity

Domains of activity	Numbers	Percentage
Prenatal diagnosis	60	20 %
General genetic counselling	56	18 %
Oncogenetics	49	16 %
Pediatry/syndromology	49	16 %
Neurogenetics	34	11 %
Cardiogenetics	16	5 %
Endocrinology	9	3 %
Hématology	2	1 %
Preimplantatory diagnosis	11	4 %
Others (reference centers)	18	6 %

Genetic counsellors' role

First, our analysis was aimed at investigating whether and to what extent genetic counsellors were involved in participants' consultations. We therefore took into account the responses of the 104 professionals working with one or more genetic counsellors. Data indicate that 35.6 % of participants (n=37) include genetic counsellors in their consultations, whereas 40.4 % of participants (n=42) answered that this was not possible. For the remaining participants, 24 % said that such consultations only occurred sometimes. The inclusion of genetic counsellors' in physicians'consultations was independent of the physician' seniority or specialty.

Specifically, genetic counsellors tend to work closely with physicians not only when in training or under supervision but also when they reached a level of expertise so that their input into the consultation is valuable. The relationship between genetic counsellors and physicians was based on trust and it was, to a great extent, rather dynamic.

Second, in order to clearly assess genetic counsellors' role and tasks, a list of roles was considered by the participants in terms of the extent (i.e. "often", "sometimes", "rarely" or "never") to which genetic counsellors performed these activities during consultations (details in Table 2). Results indicate that genetic counsellors were rather autonomous, as they had a number of responsibilities related to patients and their families. The first contact with the patient was most often an activity undertaken by the genetic counsellor (58 %). Also, genetic counsellors discussed the family history (76 %); as a matter of fact, only rarely (5 %) the family history is collected by the medical geneticist, a research assistant or a student in clinical rotation. Genetic counsellors were reported by 68 % participants to regularly explain to patients and their families the disease characteristics, the inheritance pattern and their genetic testing options, while informed consent was discussed by genetic counsellors in 56 % of the cases. However, there was a significant difference in terms of the proposal of genetic testing. In sessions more focused on genetic counselling, 44 % of physicians were satisfied for the genetic counsellor to offer genetic testing, 29 % occasionally agree to this and 18 % are reluctant to genetic counsellors to propose the test. When genetic testing was part of a diagnostic consultation and testing is approached as a medical procedure, participants' responses were mixed. It is worth noting that a number of professionals (18 %) leave this responsibility to genetic counsellors, particularly in cancer genetics (i.e. the diagnosis is based on personal and family history, histological studies of tumours, age of onset of cancer in the family) and where patients' cases can often be discussed with physicians prior to the consultation which is rather different from consultations more focused on observation and assessment of clinical signs, daily habits and on conducting additional tests.

While 31 participants (29.8 %) allowed genetic counsellors to offer genetic tests as part of diagnostic consultations and 62 (59.6 %) as part of "genetic counselling" consultations, 48 (46.2 %) stated that it was possible for genetic counsellors to deliver genetic test results. This responsibility seems to be dependent of the area of practice of the health professional interviewed. It is noted that the professionals working in the field of cancer genetics (66.7 %) are the most resistant to genetic counsellors undertaking this task.

In addition to the recent introduction in France of follow-up for patients at high risk of developing cancers (National Institut of Cancer 2010), genetic counsellors will be required to validate recommendations for monitoring, assessing cancer risk to write custom monitoring plans, and to review during « dedicated » consultation for this project monitoring, patients wishing to have a more sustained monitoring and seeking additional information. They will be in direct contact with carriers of a genetic defect or non-carrier patients but with a significant risk of developing cancer (or second cancer). This may be due to the fact that the genetic counsellor on oncogenetics is usually the one who performs the initial genetic counselling session for the patient; however, it is necessary that the patient is seen at least once by the clinician, usually at the disclosure of the result.

Finally, when participants were asked whether they were satisfied with genetic counsellors carrying out consultations

Table 2 Role of the genetic counsellors

Questions	Often	Sometimes	Rarely	Never
The GC is the first contact with the patient? (n=91)	53	24	10	4
The GC realizes the pedigree tree? (n=91)	69	12	5	5
The GC explains the genetic test? (n=87)	59	17	5	6
The GC proposes genetic test for consultation of genetic counselling? (n=85)	37	25	8	15
The GC proposes genetic test for consultation with diagnosis? (n=87)	16	15	25	31
The GC obtains the consent of the patient? (n=87)	49	15	10	13
The GC gives the patient's result? (n=87)	23	19	21	24
The GC writes the report of the consultation? (n=89)	41	19	13	16

Table 3 Could a genetic counsellor make a consultation session autonomously (n=126)

Type of consultation	Number (n=126)	Percentage
The risk assessment for autosomal recessive diseases	95	75 %
The risk screening for disease X-linked	71	56 %
The risk assessment for autosomal dominant diseases	81	64 %
Presymptomatic or predictive (non-cancerous) testing of autosomal dominant diseases	27	21 %
Presymptomatic testing for familial cancer	42	33 %
Genetic counselling for consanguinity	83	66 %
Consultations for medically assisted procreation	67	53 %
Consultations for prenatal diagnosis	67	53 %

autonomously (i.e. solo), 79 % indicated that genetic counsellors working in their service could carry out genetic counselling sessions independently.

Specifically, we wanted to explore which consultations/sessions participants believed that genetic counsellors could conduct alone, without a medical geneticist being involved (examples of responses available are shown in Table 3). Data showed that physicians would be willing to let genetic counsellors independently manage consultations focused at risk assessment for autosomal recessive diseases, autosomal dominant diseases and consultations for consanguinity (i.e. consultations where the genetic counsellor would assess risks of a genetic abnormality to patients). Surprisingly, participants did not seem confident in trusting genetic counsellors to manage sessions for presymptomatic testing.

Genetic counsellors' skills

Finally, we wanted to investigate participants' view on genetic counsellors' skills, as they have been previously defined (Sobol et al. 2008; EBMG): (1) accompany the person throughout their care pathway, (2) assess a risk situation in the field of genetics, (3) work in a inter-disciplinary team and contribute to the development of medical diagnosis, (4) integrate ethical, legal and ethical dimensions of professional practice and (5) use of information systems and contribute to research.

Responses from the 126 participants are shown in Table 4. Three skills seem to be "significant": The genetic counsellor must (1) recognize his or her own limits (85 %), (2) have the ability to work in a medical team (79 %) and (3) the ability to establish a trusting relationship with the patient and his family (75 %). Interestingly, "help the patient in the decision to carry out a genetic test" and "ability to identify needs and unspoken of the patient" were rated as irrelevant, whereas these two competences are the definition of genetic counselling (Reed 1955; Fraser 1974).

The vast majority (86 %) of participants were satisfied with genetic counsellors' education and were willing to work with one or more genetic counsellors. However, 11 % of participants were undecided regarding genetic counsellors' education as they were unaware of the content of the master's programme, 3.2 % had no opinion on education and 3.2 % expressed regret that the program was only provided in one university. Only 2.4 % of participants expressed dissatisfaction regarding the courses but did not enter comments in the "comments section" in order to clarify the reason.

Table 4 Competences required for the genetic counsellors (n=126)

Competences	1 Insignificant	2	3	4	5 Significant
Obtain medical information to confirm a family pathology	2 (2 %)	3 (2 %)	15 (12 %)	26 (21 %)	80 (63 %)
Ability to transmit genetic information to the patient	0	2 (2 %)	14 (11 %)	28 (22 %)	82 (65 %)
Consider the ethical and legal aspects	1 (1 %)	1 (1 %)	14 (11 %)	28 (22 %)	91 (72 %)
Demonstrate an ability to organize and prioritize files	0	1 (1 %)	7 (6 %)	37 (29 %)	81 (64 %)
Ability to work in a medical team	0	0	4 (3 %)	22 (17 %)	100 (79 %)
To recognize its own limits	0	0	4 (3 %)	15 (12 %)	107 (85 %)
Ability to establish a relationship of trust with the patient and family	0	0	10 (8 %)	22 (17 %)	94 (75 %)
Take into account the feelings, anxieties, beliefs and expectations of the patient	2 (2 %)	1 (1 %)	11 (9 %)	37 (29 %)	75 (60 %)
Ability to identify needs and unspoken of the patient	1 (1 %)	3 (2 %)	17 (13 %)	47 (37 %)	58 (46 %)
Help the patient in the decision of realize a genetic test	6 (5 %)	7 (6 %)	40 (32 %)	40 (32 %)	33 (26 %)

Discussion and conclusion

Genetic counselling is a profession that has been recognized in France. Yet, it is not completely understood by the medical profession. Genetic counsellors have several roles and skills that they are trained for but medical professional do not yet seem aware of. Also, it is often difficult to create a job opportunity due to several administrative issues (no defined status, lack of proper genetic counsellors' salary grid and no billing of their consultation session) or lack of budget.

In 2000, it has been reported by the UK Human Genetics Commission that "clinics may support and indeed encourage the process of family communication and genetic counsellors may give advice on how such information may be disseminated to those for whom it may be most relevant" (Human Genetics Commission 2000). It seems that this profession is clearly recognized by doctors practicing or working within genetics departments. They are now able to manage consultations independently, without the presence of a qualified medical genetics but under their responsibilities. They can carry out their own consultations, providing both information for relevant pathology and for genetic testing, and sometimes get to announce genetic test result to patient. It seems that the role of genetic counsellors is directly dependent on the relationship of trust between the two health professionals. These new genetic professionals seem to be perfectly integrated into teams, and genetic counsellors are viewed as full members.

Responsibilities entrusted to genetic counsellors are related to their professional experience, human quality and professionalism. However, we must also take into account the ability of physician to delegate tasks, accept such a profession redefining the roles and the relationship of trust that can be established between the two professions.

Finally, with the new sequencing technologies, like whole exome and genome analysis, genetic counsellors would be active part in approaching the new critical issues, like incidental findings (IFs). A Canadian study concerning the genetics professional's perspectives on reporting IF from clinical genome-wide sequencing concluded that geneticists and genetic counsellors need to work with a great collaboration and that the viewpoint of these two health professionals is important (Lohn et al. 2012).

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Compliance with ethics guidelines All procedures followed were in accordance with the ethical standards.

Informed consent was obtained from all participants included in this study.

Conflict of interest Christophe Cordier, Nicolas Taris, Ramona Moldovan, Hagay Sobol and Marie-Antoinette Voelckel declare they have no conflicts of interest.

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