Analysis of pre-test interviews in a cohort of Brazilian patients with movement disorders

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Abstract Spinocerebellar ataxias and Huntington disease are heritable, adult onset, neurodegenerative disorders of movement. Both are autosomal dominant and caused by expansions in trinucleotide sequences in several genes. Because these expansions are associated with an almost complete penetrance, genetic tests are available at the diagnostic and predictive level. In this study, we describe the expectations and issues raised during pre-test interviews for genetic counselling for these diseases. Data from pre-test interviews with 97 patients and at-risk relatives for spinocerebellar ataxia (SCA) or Huntington disease was comprised of close-ended questions (demographics, personal and current disease history) and open-ended questions, where individuals were asked to describe their hopes and expectations on the genetic counselling evaluation and also their degree of knowledge about genetics and medical genetics. Amongst the main expectations identified in patients and at-risk relatives, issues related to the aetiological diagnosis and/or disclosure of the at-risk status

were those most frequently mentioned (57 %). Improvement in quality of life was another identified issue (17 %). Interestingly, the issue of inheritance/transmission was identified as the main expectation by a minority of individuals (3 %). Pre-test interviews are valuable tools to identify issues raised by consultands and promote a better communication between the patient, family and the genetic counselling team.

Keywords Genetic counselling · Pre-test interview · Predictive test · Spinocerebellar ataxia · Huntington disease

Introduction

Genetic counselling aims at helping individuals and families to deal with the impact of genetic contribution to a disease (Harper 2008). Information on genetic risks may be necessary in different moments of an individual's lifetime: pre-conception, prenatal, childhood, or genetically determined diseases of adult life, like hereditary cancer and some neurodegenerative diseases. In all these contexts, establishment of a diagnosis and evaluation of family history are integral parts of this process. Education on the genetic/hereditary aspects of the disease, genetic testing and promotion of educated choices in order to better adapt to the disease context are also fundamental to this process (Forrest et al. 2007; Bernhardt et al. 2000). The usefulness of the genetic counselling process is highly dependent on issues related to cultural values, previous experience with the disease, social and financial background and family dynamics. It is well-known that some of these issues may have highly symbolic meaning (and often stigmatising consequences), usually impacting on the subjective impression the patient develops and, consequently, on the patient's ability to fully comprehend the implications of a given diagnosis (Biesecker and Peters 2000). The growing

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availability of genetic (both diagnostic and presymptomatic) testing for monogenic, highly penetrant neurodegenerative disorders of adult life has broadened the scope of genetic counselling for these diseases. In particular, genetic tests for severe late-onset neurodegenerative disorders have become available (Bombard et al. 2011). Currently, there is no cure or few available therapeutic options for these diseases; the process of genetic counselling and presymptomatic testing must follow well-established guidelines (MacLeod et al. 2013). In this study, we describe the expectations and issues raised during pre-test interviews for genetic counselling for two hereditary movement disorders: Huntington disease (HD) and spinocerebellar ataxias (SCA), in a public genetic counselling clinic Rio de Janeiro, Brazil.

Patients

Ninety-seven persons were referred for genetic counselling because of a movement disorder in the family. Most families had ataxia and/or chorea and/or dystonia. These included 3 individual from families with SCA2, 23 from families with Machado-Joseph disease (MJD/SCA3), and 10 from families with SCA7. Additionally, 43 patients presented ataxia without an aetiological diagnosis, and 18 families individuals had either confirmed or suspected HD. Ninety individuals were symptomatic; seven individuals were still asymptomatic and sought presymptomatic testing. The sample was composed of 53 females (*F*) and 44 males (*M*). Their age distribution ranged from 18 to 68 years.

Methods

Every new patient had a previous evaluation with a clinical psychologist, followed by the initial meeting with the genetic counselling team, on the same day. Persons eligible for genetic testing were then offered a subsequent appointment with the psychologist. Follow-up consultations were adapted to their specific needs. The data presented here derive from interviews performed by the psychologist with patients or at-risk relatives, from 2010 to 2014. The interview comprised closed-ended questions on social, economic and demographic aspects, personal history and characterisation of the current disease. It also contained open-ended questions, where patients or relatives at risk were asked to describe their hopes and expectations on the genetic counselling process and their degree of knowledge about genetics, in general, and medical genetics, in particular. Analyses of the interviews were based on both quantitative and qualitative approaches (Becker 1999). Quantitative analysis included social and demographic data. In the qualitative analysis, the content of the interviews (narratives, descriptions) was analysed with the Content

Analysis technique (Bardin 1979). This aimed at identifying and characterising “cores of meaning”, which are part of the communication process, the presence or frequency of appearance which might be relevant in the counselling process. It helped the identification of relevant agendas and issues present, though not yet clearly manifested. It included tests of association of words, to identify stereotypes and connotations. The use of inductive words (stimuli) generated other words (responses), or induced words (Bardin 1979). According to Bardin (1979), tests of association of words help the psychologist to identify blockages or zones of repression in the clinical setting, i.e., issues important to the context, but not clearly stated in the consultand’s speech. Globally, the analysis comprised the following steps: (a) identification of central ideas and agendas (cores of meaning) concerning prior concepts and expectations; (b) comparison between the different cores of meaning observed; (c) identification of thematic axes, around which patients built their concepts; and (d) discussion of observed thematic categories (Bardin 1979).

Results

Most interviews (90) were with symptomatic persons, whilst seven were asymptomatic, at-risk relatives referred for genetic counselling. Their demographic characteristics are shown in Table 1. Analysis of the cores of meaning that were more frequently observed (reason for consultation, concepts on genetics and description of main complaint) identified two major themes: (1) diagnosis and disclosure of the at-risk status and (2) quality of life improvement. Additional issues identified were as follows: (3) information, (4) prevention/cure, (5) inheritance/transmission and (6) social disability income. In seven interviews, no major theme seemed to stand out when compared to the others so we included these under the category “No major expectation identified” (Table 2).

Importance of a definite diagnosis

The need for an aetiological diagnosis was the issue most frequently raised by the interviewed subjects. At enrolment at the genetic counselling clinic, most patients described a previous history of unsuccessful diagnostic investigations. Medical genetics, as a specialty in the clinic, was perceived as a novelty by many patients. In their own words, “*Everything I know of genetics is restricted to what has been taught in school*” (*F*, aged 32; *M*, 23; *M*, 55; *M*, 56). To most of these patients and families, genetics meant finding answers to fill the gaps that generated anxiety in their families for generations and interfered in their current and future life projects: as stated by one patient, a diagnosis that explains “*the death of my father and my sibs*” (*F*, 52). The lack of genetic diagnosis makes characterisation of the disease more difficult,

Table 1 Demographic profile of the studied sample ($n=97$)

	Number	Percent (%)
Civil status ($n=97$)		
Married/stable union	41	42
Single	40	41
Widow/widower	6	6
Divorced	10	10
Offspring ($n=97$)		
With descendants	57	59
Without descendants	40	41
Labour ($n=97$)		
Employed	31	32
Unemployed	14	15
Social security (disability) income	41	42
Never worked	11	11
Education ($n=86$)		
Elementary (incomplete)	7	8
Elementary (complete)	23	27
Intermediate (incomplete)	9	10
Intermediate (complete)	26	30
Superior (incomplete)	2	2
Superior (complete)	19	22
Did not answer	11	11

since many health professionals are not acquainted with them and depend on molecular confirmation of a clinical diagnosis or suspicion. This obstacle is mentioned by one of the patients as: “on a chair we can do many things, drive, and use the computer (...). Our problem is standing up.” (M, 56). In his perspective, as well as many other patients, impairment begins when unbalanced gait/standing is first noted.

At-risk status

The feeling of being at-risk emerged under various forms in the interviews. As stated by a 42-year-old female relative of a proband, who requested presymptomatic testing because she

believed her family saw her as symptomatic, “*I want to get rid of this ghost*”, referring to the risk of carrying the disease. In another family, an asymptomatic 38-year-old at-risk male justified his desire to undergo presymptomatic testing stating “*I am sure I have this disease, I want to organise my life and take decisions about my job and work*”. For many interviewed probands, defensive behaviours are frequently observed, as in the following statements: “*I am here because of a suspicion of chorea*” (F, 33) or “*my doctor suspects I have ataxia*” (M, 37).

Quality of life

Quality of life is considered here as the perception of life according to the individual’s cultural and moral values, goals, expectations and preoccupations. Individuals grouped in this category seek alternative therapies that might ameliorate their life quality. Many lived through the experience of taking care of their sick parents/relatives and expressed hopes for a favourable result in the genetic test. Statements like “*to keep my job*” (M, 33), “*to get social benefits*” (F, 55), or “*I will not take the role of the disabled*” (F, 28) suggest that one is dealing with persons whose social demands (related to their own subsistence) are perceived as more important than their health concerns.

Information is a recurrent theme in interviews, as in “*to know if it is genetic*” (M, 70) or “*to know if there are remedies*” (F, 67); however, the information that there is no cure was mentioned by them in almost all interviews.

Prevention/cure

In most interviews, the idea of prevention is manifested in the following statements, as justification for the genetic test: “*to avoid the disease*” (F, 34), “*to block transmission*” (F, 55), “*to better organise my study and my work*” (M, 28), or “*to have a milder form of disease*” (M, 23). All these statements are somehow linked to the notion of predictive medicine and prevention, a shift from the paradigm of diagnosis and treatment, as reviewed by Zimmern (2014). On the other hand, some patients did manifest expectations about cure, as in “*to know that there is cure*” (F, 23) or “*to get cured*” (F, 49).

Inheritance/transmission

The concept of genetics was intertwined with the idea of inheritance or, as defined by one patient, genetics is “*the transmission of diseases from one generation to the next*” (M, 56). What becomes evident in each case is the notion of inheritance. Genetics is, thus, “*transmission of something, from one generation to the next*”; amongst most interviewed patients, the notion of inheritance was highly associated with the occurrence of the disease in previous generations. On the other

Table 2 Main expectation on the genetic counselling process ($n=97$)

Main expectation before genetic counselling ($n=97$)	Number	Percent (%)
Diagnosis/at-risk status	57	58
Quality of life improvement	17	18
Information	7	7
Prevention/cure	4	4
Inheritance/transmission	3	3
Social disability income	2	2
No major expectation identified	7	7

side, there was an apparent lack of preoccupation concerning transmission to descendants. The fact these patients did not see themselves, at this initial moment of genetic counselling, as someone capable of transmitting the disease to their offspring indicates a psychological defensive mechanism, a denial of the risk for their descendants.

Social disability income

Possible eligibility for a social disability income was manifested as the main expectation by two symptomatic patients.

Discussion

The pre-counselling interview aimed at promoting a better liaison between the patient/consultant and the team and was focused on the situation-problem brought by the patient. Another aim of this interview was to introduce a psychosocial approach into the genetic counselling process, to facilitate the comprehension of the diverse demands these patients present (Kessler 1979).

The day-to-day co-existence with symptoms of a hereditary neurodegenerative disease of unknown aetiology results in an overwhelming emotional experience. The person develops a feeling of vulnerability, loss of appropriate care and exclusion, at a moment when he/she has to deal with the consequences caused by the symptoms that appear without an acceptable explanation, and at the same time, finds the necessary solutions to readapt and reorganise their life with the disease. The lack of diagnosis adds a further degree of difficulty for these patients and families. Initially, affected persons find themselves at a loss and feel their lives have become arrested, not only psychologically, but also in real life. Providing a diagnosis and adequate information about the disease is necessary to allow informed decision-making at the individual, familial and communal level.

Although theoretical risks for transmission to offspring can be accurately estimated, given the monogenic character of these diseases, the subjective perception of the risk seems to have no correlation with the objective risk possibly because of the age-dependent penetrance. The experience of being at risk for Machado-Joseph disease, i.e., the situation of living upon that subtle line between carrier and non-carrier “is experienced as a second disease in the family” (Boutté 1990). The psychological overload imposed to the families with hereditary neurodegenerative diseases precedes diagnosis and reinforces the idea that objective risk information is not capable to determine how the subject will cope with the subjective impact of receiving a diagnosis. These movements establish, a priori, incertitude, in spite of the fact that these patients are symptomatic and carry the cardinal signs of the disease. Such responses

represent defensive movements towards a risk situation, to postpone the confirmation of a disease with no cure.

Because degenerative heritable diseases are usually incapacitating people for work, these patients grow more and more dependent on a caretaker, usually the spouse or a close relative, whose work capability is also impacted; this results in the worsening of the financial situation of the whole family. In this context, work is a real concern to patients, generating potential conflicts, since many patients are not considered eligible for a disability pension, either because the final diagnosis is not defined or initial symptoms are not considered sufficiently incapacitating by social security. Social security in Brazil grades deficiency in three categories: mild, moderate and severe (Brasil 1991). Unfortunately, many of our patients who lose their jobs are not considered eligible for social security, based on disability after the initial evaluation, and eventually do not insist on the process and give up, due to the many bureaucratic steps to have their request reconsidered, and they end up depending on relatives and thereby increasing the financial burden of the family.

During the genetic counselling process, the ideal situation is the one where the individual has access to genetic information and, at the same time, he/she gradually acquires control over the situation of being affected or at risk. Perhaps the most important function of the counselling team is to encourage patients to actively decide on their own benefit. In order to deal with this intense experience, it is important to find ways to tackle with these feelings (frustration, loss, bereavement and sadness) since the first appointment, even before the diagnosis is established. Information also helps families make sense of the disease itself, as exemplified in the case of a 36-year-old woman who was referred by her neurologist for diagnostic investigation of HD. She had been adopted and did not have any information about her biological parents. She began developing behavioural disturbances. In her family, these behavioural problems were attributed to family dynamics, as her brother had recently got engaged and her own marital relationship had ended. When she was diagnosed with HD, not only could the family finally understand the disease course and prognosis but they also find the best way to deal with her symptoms thereafter. Genetic information also has a central role in deciding whether an individual will undertake a diagnostic or presymptomatic testing. Whatever the genetic counselling model adopted, the role of the counsellor is to provide the counsellee with consistent information and correct misconceptions (Kessler 1979). Klitzman (2010) interviewed 64 individuals who underwent presymptomatic testing (for Huntington disease, hereditary breast cancer and alpha-1 antitrypsin deficiency) and observed many misconceptions on genetics and probabilities. Such misunderstandings may hinder comprehension of the risks and emphasise not only the need to address these cognitive facts but also the underlying emotional issues (Klitzman 2010).

Provided genetics is viewed as inheritance; the prevailing view amongst the studied individuals is that genetics is the major if not the only determinant of health and disease. Under this reductionist view, genetic determination becomes overrated and seems to hold the final explanation to the disease, without paying attention to other influences, namely, the environmental ones. The paradigm of causality is transferred to the genome, transforming the (affected) individual in a “prisoner of genes”, and ultimately, does not take responsibilities in the individual, societal and environmental contexts (Penchaszadeh 2004).

Patients with disabilities whose family income is lower than one fourth of the current regional minimum wage *per capita* are entitled to receive an income equivalent to one national minimum wage, even if they have never contributed to social security (informal economy). Thus, only families with very low income *per capita* are actually eligible for this kind of pension.

In spite of the initial desire to get tested, many patients and at-risk relatives are neither fully aware of the long and sometimes complex clinical investigation (that they will have access to information that is not easy to deal and they will be faced with difficult decisions) nor fully conscious that pre-symptomatic testing will provide information on the future health status of an asymptomatic individual, as opposed to most medical exams that provide information on the current status of a disease (Evers-Kiebooms et al. 2000).

Guimarães et al. (2013) interviewed 22 individual that had undergone genetic counselling for presymptomatic testing (for HD, SCA and familial amyloid polyneuropathy) and analysed various aspects of the genetic counselling process. They emphasise the need to apply current existing protocols on a “case by case” basis, in order to address individual issues within families (Guimarães et al. 2013). According to Peters and Petrill (2011), the “client’s background is the primary frame of reference to understand and find meaning in genetic information” (...). These authors developed a tool to assess the background, needs and expectations of consultands in the genetic counselling process (Peters and Petrill 2011).

For families, genetic counselling is the possibility to redefine individuals’ roles, since the disease interferes with family organisation, long before a diagnosis is established. The at-risk state, so familiar to families who co-exist with hereditary degenerative diseases, imposes feelings of fragility and vulnerability. They also experience guilt and shame for the possibility of transmission of the “defective” gene to their descendants (Kessler 1994; de Wit et al. 1998).

Adding to this burden, access of patients with rare diseases to genetic counselling and to diagnostic and predictive testing has become a public health policy in Brazil, only since 2014. Nevertheless, a gap still exists between defining a policy and implementing a network of outpatient clinics and laboratories specialised in the field.

Dealing with the fact that genetic diseases are, for the most part, incurable results in a unique emotional experience to the families. After the initial shock, there is a storm of emotions and a mix of denial, frustration, anger and sadness (Lickleder et al. 2008). Living through such emotions, however, is of great importance to deal with the ambiguities associated with the at-risk status. In fact, many theories of cognitive evaluation indicate that individuals are not as much influenced by the situation itself but rather by their engagement with it (Beck et al. 1985). In this way, the evaluation that at-risk individuals do is focused mostly on ambiguities associated with the genetic context (at-risk status) and not as much in concrete figures about genetic risk, diagnosis or prognosis (Lippman and Frazer 1979).

In this context, psychological support has an important role, as it helps patients and families to reorganise themselves, to better face the demands of the disease, to deal positively with diagnosis and prognosis and, above all, to improve the process of decision-making during genetic counselling.

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