Quality Assessment of Genetic Counseling Process in the Context of Presymptomatic Testing for Late-Onset Disorders: A Thematic Analysis of Three Review Articles

Milena Paneque1,2, Jorge Sequeiros1, and Heather Skirton2

Presymptomatic testing (PST) is available for a range of late-onset disorders. Health practitioners generally follow guidelines regarding appropriate number of counseling sessions, involvement of multidisciplinary teams, topics for pretest discussion, and follow-up sessions; however, more understanding is needed about what helps consultands effectively and the impact of amount and quality of genetic counseling on the psychosocial sequelae of PST for late-onset disorders. We conducted a thematic analysis of three review articles on quality of the genetic counseling process, aiming at (1) exploring current evidence; (2) identifying quality assessment indicators; and (3) making recommendations for genetic counseling practice in late-onset disorders. We undertook a systematic search of 6 relevant databases: 38 articles were identified and 3 fitted our inclusion criteria; after quality appraisal, all were included in the review. The number of sessions, time spent, consultation environment, follow-up, and multidisciplinarity were identified as variables for quality assessment. Research on counseling in the context of genetic testing in familial cancer tends to be related to outcomes and indicators for quality assessment, while research concerning other late-onset diseases is mainly focused on the psychological impact of the test results. The quality and content of the overall process in noncancer late-onset disorders is insufficiently articulated. Despite the fact that PST for Huntington disease and other degenerative conditions has been offered for more than 20 years, good methodological approaches to assess quality of genetic counseling in that context remain elusive. This restricts improvement of the protocols for genetic services and, in general, healthcare for the at-risk population.

Introduction

Genetic counseling is an area of clinical healthcare focused on the needs of individuals and families affected by or at risk for a genetic condition. Its aims include helping them to understand the relevant aspects of the disorder and/or the risks to develop it. Psychological support to help the family adjust to the diagnosis, make important life decisions, and cope with the psychological sequelae of living with the disease or the at-risk status are also integral to genetic counseling (Godard et al., 2003). Unfortunately, little is known about how genetic counseling helps consultands prepare effectively for genetic testing or to live with an increased risk of a genetic disorder (Coviello et al., 2007; Rantanen et al., 2008a, 2008b), and as yet there is no agreement about the best way to assess effectiveness of this process.

The need to establish guidelines for quality management and assessment in genetic counseling is recognized (Rantanen et al., 2008b) and may be increasingly important as new genetic technologies are utilized by both genetics services and primary care (Rantanen et al., 2008a). As knowledge of the underlying genetic causes of disease improves, presymptomatic testing (PST) in adult-onset disorders is becoming increasingly feasible and more widely available. While in some cases, such as the hereditary cancer syndromes, PST may enhance opportunities for screening and preventive treatment (Cooper et al., 2010; Jasperson et al., 2010; Tinelli et al., 2010), in other adult-onset conditions, including neurodegenerative disorders, the possibility of predicting the onset of symptoms many years in advance has not been matched by similar progress in prevention or treatment (Rolim et al., 2006; Coviello et al., 2007; Rantanen et al., 2008a, 2008b). Although they may have different meanings for some professionals (e.g., predictive testing being used to describe any genetic test performed on a healthy person or, on the other hand, to refer only to susceptibility testing in

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multifactorial diseases), the terms presymptomatic and predictive testing are often used interchangeably (Kääriäinen et al., 2008). For brevity, in this article, we will use the term PST throughout, defined here as a molecular genetic analysis to detect a specific mutation before the onset of significant signs and symptoms. PST is used for conditions with 100% penetrance (such as Huntington disease) and those with reduced penetrance (such as familial breast/ovarian cancer).

There are a number of professional guidelines for genetic testing and counseling (Coviello et al., 2007; Tibben 2007; Rantanen et al., 2008a, 2008b). As protocols were initially developed for PST in Huntington disease, this condition was used as a paradigm for development of protocols in other conditions (Erez et al., 2010). Continuously assessing practice, measuring endpoints, and improving genetic counseling remains a great challenge (Sequeiros et al., 2006; Rantanen et al., 2008b). A Working Group on quality assessment of genetic counseling was able to produce a checklist of quality indicators, but these relate very much to logistical factors (such as location of clinics and training of staff) rather than the impact of counseling on clients (EuroGentest, 2008). In general, the literature examining outcome criteria for genetic counseling practice is problematic (Clark et al., 2000; Skirton, 2001; Wang et al., 2004; McAllister et al., 2006, 2007; Payne et al., 2008). Some aspects have been proposed as important indicators, such as the counseling model used, the ascertainment of the consultand’s agenda, and length of the genetic counseling process, but these have not been clearly operationalized (Wang et al., 2004). In addition, outcome criteria, often specified in advancement of goals and objectives of genetic testing counseling, can limit the eventual findings of any research study (McAllister et al., 2006).

Preliminary studies suggest that some domains could provide a good starting point toward a core set of outcome measures for clinical genetic services (Clark et al., 2000; Wang et al., 2004; Payne et al., 2008). Aspects such as knowledge about the condition, fulfillment of the counselee expectations, ability to cope, quality of life, perceived personal control, empowerment, decision making, and diagnostic accuracy all seem to be relevant concepts (Payne et al., 2008). Other studies have focused on risk comprehension or recall, counselee expectation, motivation and satisfaction, or reproductive decision making (Bernhardt et al., 2000; Clark et al., 2000; Wang et al., 2004; Skirton et al., 2005). Patient’s anxiety levels have also been included as indicators of counseling effectiveness (Skirton, 2001). Specifically in the context of PST, the majority of the studies have focused on psychological reactions to testing and family communication (Rolim et al., 2006; Paneque et al., 2007, Tibben, 2007; Forrest et al., 2008).

Some methodological limitations of previous studies, such as the excessive use of cross-sectional studies design and the lack of global applicability due to bias in selection of participants, have been described (McAllister et al., 2006). The most commonly used methods for assessing outcomes are systematic reviews, Delphi surveys, and qualitative research (Clark et al., 2000; Wang et al., 2004; McAllister et al., 2006, 2007). Unfortunately, longitudinal studies are less frequent. A recent study used a triangulation of these methods to identify validated outcome measures that could be used specifically for genetics; as a result, a model of empowerment, describing the patient benefits from using clinical genetics services, was developed (McAllister et al., 2006) (Table 1).

### Table 1. Model of Empowerment Describing Patient Benefits from Using Clinical Genetic Services

<table>
<thead>
<tr>
<th>Dimension</th>
<th>Definition</th>
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<tr>
<td>1. Decision making</td>
<td>The perception that one can make important life decisions in an informed way</td>
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<td>2. Knowledge and understanding</td>
<td>The perception that one has sufficient information about the disease, including risk to oneself and one’s relatives, and any treatment, prevention, and support available</td>
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<td>3. Instrumentality</td>
<td>The perception that one can make effective use of the health and social care systems for the benefit of the whole family</td>
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<tr>
<td>4. Future orientation</td>
<td>The perception that one can look to the future having hope for the fulfilling of family life, for oneself, one’s family, and/or one’s future descendents</td>
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*Adapted from McAllister et al. (2008).

Due to the variety of published data regarding the clinical settings of genetic counseling practice, a wide range of conceptions about the nature and goals of genetic counseling and the multiple foci on genetic counseling (i.e., process and/or outcomes), we decided to conduct the first phase of our study by focusing on the latest relevant reviews on the topic. This is cited as an initial step in determining the need for further reviews or empirical studies (CRD, 2008).

### Aim and Objectives

The aim of this systematic review was to identify and critically review the evidence available on quality assessment of genetic counseling in PST. The objectives were to (1) to identify key reviews related to the quality of genetic counseling in a systematic search of the relevant literature, (2) appraise the existing evidence, (3) perform a thematic analysis to synthesize the findings of the reviews, and (4) identify evidence gaps and make recommendations for further research.

### Methods

#### Design

In accordance with the features of a systematic review described by the Centre for Reviews and Dissemination and the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) statement (CRD, 2008; Moher et al., 2009), we set objectives, and inclusion and exclusion criteria for this review. An initial search of the literature indicated that there were over 200 individual research articles that could have been included in a review of primary research related to this topic. This would have made a review unwieldy; we therefore decided to focus upon review articles. This approach was methodologically sound, as systematic reviews are acknowledged as rigorous sources of evidence for practice, whereas individual studies may be flawed or provide less reliable sources of evidence (CRD, 2008). Undertaking thematic analysis of the available review articles enabled us to systematically examine and synthesize the work already undertaken in...
this field, to determine the level of attention given to quality assessment of genetic counseling generally and ascertain the level of research work undertaken in the neurodegenerative disorders, in comparison to other conditions.

**Search strategies**

We searched the Medline, PsychINFO, Web of Science, SCOPUS, and CINAHLPlus databases for relevant studies, published between January 1997 (after direct mutation testing became available and PST counseling protocols were generally adopted) (Tibben, 2007) and December 2009. In addition, we manually searched copies of the *European Journal of Human Genetics* and the *American Journal of Human Genetics*, as well as undertook searches of the reference lists for potentially useful articles. Databases were searched during January 2010. Figure 1 shows a summary of the key search terms, which were based on terms relevant to quality assessment studies of the genetic counseling process in the context of PST for late-onset disorders.

**Inclusion and exclusion criteria**

We included reviews focused on quality of genetic counseling in PST. Reviews of qualitative studies, quantitative studies, or those that utilized mixed methods were eligible. Only articles in English were included, but there were no limits on the country in which studies were conducted. Articles describing guidelines or recommendations or ethical/legal impact of genetic testing/services were excluded, as they did not include any specific studies about the topic. Two reviewers (M.P. and H.S.) independently screened abstracts of identified studies. Articles were rejected if both reviewers determined that the article did not meet the inclusion criteria. Any uncertainties led to re-evaluation of the full text.

**Search outcomes**

Using this refined search strategy, we identified 53 abstracts, but 15 were duplicate articles. Studies cited and other articles by the same authors were also searched for potential inclusion. Of the 38 abstracts, 31 were excluded: 12 related to clinical trials or medical interventions; another 12 related to psychological assessment after predictive testing; 3 were focused on preimplantation and prenatal diagnosis and four dealt with ethical and legal issues. Seven full articles were studied by both authors (M.P. and H.S.); one related solely to patient information (Lewis *et al.*, 2007), two were reviews of potential psychometric tools for quality assessment (Kasparian *et al.*, 2007; Payne *et al.*, 2008), and a fourth reported guidelines, rather than assessment of counseling (Rantanen *et al.*, 2008b). Finally, three review articles were left for quality appraisal (Fig. 2).

**Quality appraisal**

These articles were assessed using the tool suggested by Dixon-Woods *et al.* (2006). This method is suitable when the review includes studies with different methodologies and involves five focused questions, relating to clarity in reporting aims and objectives, research methods and analytical procedures, as well as whether conclusions appear to be justified. Although there were variations in their quality (Table 2), none of these articles was excluded.

**Data abstraction**

Each article was read multiple times and discussed in depth by the reviewers. Table 2 summarizes the key aspects of the articles selected, including its aims, methods of analysis, main findings, and quality issues. The findings were then analyzed and synthesized as described before (CRD, 2008), to identify over-arching themes.

**Results**

**Studies included in the review**

The three reviews selected were carried out in 2007 and 2008 by multidisciplinary teams of medical geneticists, genetic counselors, nurses, psychologists, and other healthcare professionals.

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**FIG. 1.** Key terms used for database search.

**FIG. 2.** Results of electronic search identifying 38 potentially relevant review articles retrieved by electronic search.
<table>
<thead>
<tr>
<th>Reference</th>
<th>Title</th>
<th>Purpose of study</th>
<th>Methodology</th>
<th>Sample and size</th>
<th>Analysis</th>
<th>Disease at risk</th>
<th>Main findings</th>
<th>Quality issues</th>
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<tr>
<td>Kosonen et al.  (2008)</td>
<td>“Have patients opinions been asked? A review of 102 studies from the years 1997 to 2007 on patient perspective of genetic counseling services in genetic testing situations.”</td>
<td>Review: Search of PubMed, Cinhall, and PsyclINFO databases, from 1997 to June 2007, using the terms: genetic counseling/ counseling in combination with counsellor/counselor, expectation(s), perception(s), needs, satisfaction, and knowledge.</td>
<td>102 studies</td>
<td>SPSS, descriptive statistics to report the types of conditions and issues studied.</td>
<td>Several settings 72 studies in cancer context; 3 in cystic fibrosis; 2 in chromosome disorders; 1 in Huntington disease; 1 in hemochromatosis; 1 in Duchenne muscular dystrophy.</td>
<td>(1) Presymptomatic testing for monogenic adult onset was more established than cancer predisposition test and patient perspective of late onset neurodegenerative disorders (LONDS) had been investigated earlier. (2) Many of the genetic testing situations, especially those predicting future illness, are so complex that comprehensive genetic counseling is needed. (3) Studies performed revealed high satisfaction. (4) Almost all studies have focused on pretest or the initial genetic counseling session, with a need for more research on a range of potentially different types of interactions, including follow-up sessions. (5) More research is needed on large and representative samples of providers and clients. (6) Biomedical rather than psychosocial communication exchange is still the prevalent.</td>
<td>No systematic analysis of results. Little information is provided on the method of analyses and data abstraction. Reports the types of topics studied, rather than an analysis of the findings of the articles. The authors did not conduct any quality appraisal of articles included.</td>
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<td>Meiser et al.  (2008)</td>
<td>“Assessment of the content and process of genetic counseling: a critical review of empirical studies.”</td>
<td>Assessment of the content and process of genetic counseling.</td>
<td>Critical Review: MEDLINE, PsychLIT, and EMBASE were searched for articles published between 1978 and November 2007 using the following terms: genetic counseling, interaction analysis, audiorecording, videorecording, audiotaping, doctor/patient communication, communication research, and communication behaviors. Searches were conducted for key authors who published in this field. Manual search of the journals Patient Education and Counseling and Journal of Genetic Counseling.</td>
<td>18 studies, in 34 articles.</td>
<td>Systematic search of the literature and analysis under the themes: mixed samples, hereditary cancer, and reproductive decisions.</td>
<td>Several settings (only one on Huntington disease).</td>
<td>(1) A substantial set of findings are single projects published in multiple articles. (2) Most studies reviewed assessed a relatively narrow range of goals of counseling process and outcomes. (3) Most communication analyses have been conducted in cancer genetic counseling. (4) Almost all studies have focused on pretest or the initial genetic counseling session, with a need for more research on a range of potentially different types of interactions, including follow-up sessions. (5) More research is needed on large and representative samples of providers and clients. (6) Biomedical rather than psychosocial communication exchange is still the prevalent.</td>
<td>There was no evidence given for quality appraisal of the articles included. No genuine meta-synthesis or meta-analysis of findings. Inadequate description of data extraction and analysis process.</td>
</tr>
<tr>
<td>Reference</td>
<td>Title</td>
<td>Purpose of study</td>
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<td>Sivell et al. (2008)</td>
<td>“How risk is perceived, constructed and interpreted by clients in Clinical Genetics, and the effects on decision making: systematic review.”</td>
<td>Determine the ways in which individuals perceive, construct, and interpret their risk and the subsequent effects these have such as the use of services and tests.</td>
<td>Review: Six electronic databases (Medline, CINAHL, Cochrane Library, EMBASE, PsycInfo, and National Research Register) were searched, from 1985 to January 2006, using the following terms: risk and communication, genetics, predisposition, screening, or counseling, service delivery, health services, organization; trial design filters. Manual searches of journals and examining reference list of included articles. Narrative synthesis of the data was undertaken.</td>
<td>59 studies (4 Huntington disease).</td>
<td>A narrative synthesis of the data was undertaken and organized according to the three study aims: assessment of perceived risk; construction and comprehension of risk; effects of perceived risk.</td>
<td>Several settings: 36 focused on breast/ovarian cancer; 5 on other cancers; 6 on cystic fibrosis; 4 on Down syndrome and neural tube defects; 4 on Huntington disease; 2 on Alzheimer disease; 1 on ataxia-telangectasia; 1 on Duchenne muscular dystrophy.</td>
<td>(1) Research has focused on four keys areas: psychological well-being, use of services, health-related behaviors, and reproductive intentions. (2) The majority of studies focused on cancer genetics. (3) A clear impact of genetic counseling on risk perception and psychological outcomes was not identified, partly due to the heterogeneous nature of research.</td>
<td>Full description of methods used. No mention to quality appraisal of articles included in the review.</td>
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The first was a systematic review of 102 studies (Kosonen et al., 2008), performed in the context of the EuroGentest Network of Excellence (www.eurogentest.org). It focused on patient perspectives on genetic counseling. Little information is provided on the methods of analysis and data abstraction. The second study was a critical review of empirical studies on assessment of the content and process of genetic counseling (Meiser et al., 2008), using studies in which genetic counseling sessions were audi-taped or videotaped. The authors submitted a total of 18 studies (in 34 articles) to a communication analysis. Eight (44.4%) were performed in mixed samples (where patients attended with a variety of issues). Five assessed genetic counseling sessions for familial cancer testing, mainly for BRCA1/2, but also colorectal cancer (27.7%), whereas four (22.2%) focused on reproductive genetic counseling. One was a study of PST for a late-onset disorder. Narrative synthesis was the focus of the third selected review (Sivell et al., 2008); 59 studies presented data on the ways in which individuals perceive, construct, and interpret their risk, as well as subsequent effects.

A total of 179 studies were included in these 3 systematic reviews; they were conducted between 1978 and 2007, in various settings of genetic counseling: they comprised 75 empirical studies, 58 quantitative studies, 11 treatment/outcome clinical trials, 9 case studies, and 6 systematic reviews. Only five (2.7%) focused on PST for a late-onset condition (Huntington disease), whereas 113 (63.2%) were conducted in the context of familial cancer testing; other conditions under study were cystic fibrosis (n=9, 5.0%), Down syndrome and neural tube defect (n=4, 2.2%, each), Duchenne muscular dystrophy, and hemochromatosis (n=2, 1.1%, for each).

As a result of the in-depth analysis of the reviews, three overarching themes were identified: (1) consultand’s agenda issues, (2) understanding of risk, and (3) communication.

### Theme 1: Consultand’s Agenda Issues

Of the 102 studies in the review by Kosonen et al. (2008), only 40 (39%) focused on patients’ expectations of genetic counseling, relating to information content (e.g., general information and information about specific issues), quality (e.g., quality of information, atmosphere, communication style, and procedural aspects), procedures (e.g., access to genetic testing and examination by doctors), advice (e.g., advice about different matters and decision-making preferences), benefit to others (e.g., to help family members and to help scientific research), and topics about psychologically positive outcomes (e.g., reassurance and emotional aspects). The authors suggested that, as none of the included studies originated from patient organizations, some issues that could be very significant may not have been properly addressed. Meiser et al. (2008) presented a critical review of empirical studies assessing the content and process of genetic counseling. The authors analyzed 18 studies (published in 34 articles) involving consultands at increased risk for hereditary cancer, a range of other conditions, and studies involving reproductive decisions and genetic counseling. In only one of the studies appraised did the researchers establish the counselee’s expectations at the beginning of the consultation, by asking why the patients had been referred and what they expected (Hollowell et al., 1997). Meiser et al. (2008) also presented another study regarding directiveness of the counselor, patient satisfaction, and the extent to which expectations were met, concluding that there was no association between these variables (Michie et al., 1997). In another relevant study, the authors reported that the client’s agenda was elicited in almost all sessions analyzed (Pieterse et al., 2005). The client’s agenda was not specifically addressed in the third review (Sivell et al., 2008).

### Theme 2: Consultand’s Understanding of Risk

Kosonen et al. (2008) referred the percentage of the studies included in their review that assessed patient’s risk knowledge after genetic counseling. These include questions about own risk, family members’ risk and population risk, and risk factors. A relevant study (Michie et al., 2005) included in the second review (Meiser et al., 2008) related to the provision of recurrence rates, factual information, and consultant perceptions. Skeptical attitudes (by patients) toward usefulness of risk information were evident, whereas probabilities and risk information did not appear to alleviate patient uncertainty during the follow-up sessions. Sivell et al. (2008) specifically analyzed risk perception, construction, and interpretation related to the effects on decision making. Assessment of perceived risk varied conceptually and in the way it was measured. Risk perception was assessed as a percentage (or number out of 100), odds ratio, ordinal/categorical scales, and/or visual analogue scales. Accuracy of risk perception was commonly analyzed by comparing the numerical estimates of patients with the estimates calculated by their clinicians. Evidence also suggested that individuals experience difficulties in understanding a numerical risk. Most of the studies related to perceived risk were done in a familial cancer testing setting (Brain et al., 2000; Hopwood, 2000; Lobb et al., 2003; Andrews et al., 2004). Information about availability of prenatal diagnosis and knowledge of risk numbers were significant factors in deciding to have more children, in 94 families with a child with neural tube defect (Swerts, 1987). A few studies provided data on the effects of risk perception on psychological well-being (Decruyenaere et al., 1999; Appleton et al., 2000; Smith et al., 2002; Bowen et al., 2004), use of services (Binedell et al., 1998; Morris et al., 2001; Lim et al., 2004), uptake of health-related behaviors (Ken et al., 2003; Roberts et al., 2005), and reproductive intentions (Wertz et al., 1992; Fanos and Gatti, 1999). Results were not consistent either within or among different conditions. Nevertheless, the authors suggest that those studies offered some evidence that risk perception (and usually high levels of perceived risk) is often influenced by the individuals’ own personal experience of illness and their experiences of others’ illness, despite advice from their clinicians not to make such comparisons. Studies included in Sivell et al.’s review (2008) showed a trend toward a decrease in the risk perceived and more accurate perception after genetic counseling. However, the patient’s ability to accurately recall the risk provided fails to demonstrate whether or not they understood their risk and what meaning they attributed to it (Sivell et al., 2008).

### Theme 3: Communication in the Genetic Counseling Process

One of the factors that could be considered as a measure of effective counseling is appropriate bidirectional communication between consultands and health professionals. Kosonen...
et al. (2008) showed that the patient’s understanding of the issues discussed was explored in 47% (n = 48) of the assessed studies. In addition to general knowledge of genetics, understanding of specific issues, such as risk of developing the disease, population risk, family member’s risk, inheritance, procedures, test results, treatment, and family implications, were assessed. The authors suggested that patient comprehension may be affected when the information is provided by healthcare professionals from fields of medicine other than genetics. Further, they highlighted the inadequate volume of research about consultands’ understanding of the information given in diagnostic situations. Nevertheless, the authors did not present any reference to the genetic counseling models that were used in the studies reviewed or the contents of the counseling sessions.

Some interesting findings presented in the review by Meiser et al. (2008) were related to contents, models, and consultand’s understanding. For example, situations in which a diagnosis cannot be confirmed were characterized by negotiation and animated discussion between the genetic counselor and consultand (Brookes-Howell, 2006). Sensitive and empathic counseling seemed to be a key component for patients living with diagnostic uncertainty. Also, the need to examine alternative styles in genetic counseling was emphasized (Ellington et al., 2005). Some difficulties understanding medical terms, regardless of educational background, suggest the need for careful explanation to reduce potential fear and confusion among patients (Chapple et al., 1995). Additionally, few studies explored how comprehension of the information can be checked, through confirmation of understanding and disconfirmation of incorrect perceptions. The need to find a balance between ensuring correct information and the respect for the expertise of others was presented as a possible dilemma of clinical geneticists (Lehtinen and Kaariainen, 2005). Another study (Michie and Marteau, 1996) examined the influence of input variables (the genetic condition and consultand’s anxiety, coping style, main concerns, and expectations) and process variables (length of consultation and content). The authors of this study concluded that, while input variables influenced consultand satisfaction after counseling, less evidence was found about significant predictors of patient’s outcome (Michie and Marteau, 1996). Additional factors studied in the same review (Meiser et al., 2008) were consultand’s recall, finding that patients valued information related to family implications, whereas counselors placed greater importance on information about the test, diagnosis, and prognosis. Only in 25% of occasions, did clinicians assess comprehension of the information (Michie et al., 2005). In a Roter Interaction Analysis, adapted for a genetic counseling communication (Roter et al., 2006), the average length of consultation was described as 47 min, with verbal dominance by the counselor, who averaged five statements for each patient statement. Both teaching and counseling models of genetic counseling emerged. The teaching pattern included clinical teaching (31% of video-recorded sessions) and psycho-educational teaching (27%), whereas the counseling pattern included supportive counseling (33%) and psychosocial counseling (9%). Psychosocial counseling obtained the highest rating and resulted in less verbal dominance of the counselor and higher levels of facilitation (e.g., asking for patient opinion and understanding). These findings are in accordance with two other studies included in the review, showing an association between facilitating understanding behaviors and lower levels of patient depression (Duric et al., 2003; Pieterse et al., 2007). Finally, the only study related to the PST context assessed specifically the use of counselors’ “reflective frames” (Sarangi et al., 2004). As part of this study, 24 pretest and follow-up counseling sessions were audiorecorded and analyzed through discourse analysis, aiming at assessing encouragement of clients to discuss their feelings and copying styles. Although they found wide differences in methodology and design of the articles in their review, Meiser et al. (2008) identified as a common issue that providers generally speak more than consultands do. Regarding the contents of the consult, they suggest that genetic counselors need to take more time discussing the implications of family history and talking didactically when providing information about genetics (Meiser et al., 2008).

Their findings indicated that a large proportion of the communication in genetic counseling is biomedical or educational, rather than psychosocial, but again nothing was specified for PST of late-onset disorders. In addition, as a result of their analyses, Sivell et al. (2008) discussed some theoretical frameworks, such as the Common Sense Model of Illness perceptions (CSM) (Leventhal et al., 1980) and the Social Learning Theory (Bringle and Antley, 1980), which can provide useful models for the practice of genetic counseling.

Discussion

While there are a number of evidence-based outcomes and process indicators for quality assessment in genetic counseling for familial cancer syndromes, these are not currently available for late-onset neurological disorders. The review by Meiser et al. (2008) showed that most communication analyses have been conducted in cancer genetic counseling; however, Kosonen et al. (2008) claimed that many genetic testing situations, especially those predicting future illness, are so complex that comprehensive genetic counseling is needed. The same authors highlight differences when comparing cancer risk and neurodegenerative risk, because of follow-up programs and prophylactic options. However, only 5 studies out of 179 included in the 3 selected reviews related to PST in neurodegenerative disorders. Kosonen et al. (2008) claimed that PST of monogenic adult-onset diseases (e.g., Huntington disease) is already more established and patients’ perspectives had been investigated earlier; however, we found little evidence to substantiate that argument. On the contrary, research concerning testing for other late-onset diseases is focused on the psychological impact of the results, and little is articulated regarding quality and content of the process, that is, how these results are achieved. Rantanen et al. (2008b) confirmed the complexity of the genetic counseling process and the current profusion of guidelines available internationally for genetic counseling practice; however, while the guidelines may have been based on evidence of outcomes, evaluation of the process and the contribution of counseling to those outcomes has not been sufficiently undertaken.

These findings reflect an imbalance between research and/or practice focused on familial cancer and that focused on late-onset neurological disorders. They are consistent with those of Lewis et al. (2007), who found that patient information for those with familial cancers was much more likely to include psychosocial information (to support dealing with the disease...
or with risk) than information offered to patients with other late-onset conditions. While there may be no or few differences in the genetic counseling requirements of those for whom treatment and prophylactic measures exist and those for whom these measures are not available, this has not been satisfactorily evaluated. This may be increasingly important as genome-wide studies enhance identification of susceptibility for a wider range of conditions.

Health professionals have a responsibility to assess and improve their practice (Rolim et al., 2006; Rantanen et al., 2008b). There is evidence that the experience of PST does not, in general, adversely alter the psychological well-being of the patient (Tibben, 2007); however, this should not induce practitioners to believe that the services being provided are as effective in terms of quality as they could be. This is much more complex if we consider that perhaps the consultation process could have a minimal effect upon patient outcomes and also that the counseling interaction between counselee and counselors has not been explored enough (Michie et al., 1996; Meiser et al., 2008).

As previously stated, a system for quality assessment of genetic counseling (EuroGentest, 2008) has been developed that includes consultation environment, staff education, use of appropriate language, and multidisciplinary team working. While these factors are important and may provide an indication of the organizational characteristics of the service, they do not specifically address the experience of the patient or the contents, behaviors, and relationships that constitute the genetic counseling interaction during PST.

The study by Michie et al (1996) highlighted that attention is needed not only on the process of consultation and outcome variables, but also on patient and counselor variables before the consultation. In that sense, future studies on what makes the process of genetic counseling effective in predictive testing should include preinterview patient and counselor expectations and preinterview patient concerns as well as post-test psychological measures. In general, more research is needed to address different types of interaction, according to the genetic context, including pretest and follow-up sessions after PST.

Strengths and limitations

We focused on review articles in this systematic review. While it is possible that this may have resulted in omission of some relevant articles, our original search included all original articles and reviews and we were not aware of any highly relevant articles that were omitted from the reviews. Using this strategy enabled us to synthesize a vast number of articles and effectively identify gaps in the current literature, before our planned empirical study. The conclusions of this review are novel and make a substantive contribution to our understanding of the state of research into quality aspects of the genetic counseling process across a range of different settings and conditions. In particular, it has enabled us to identify the paucity of research in neurological conditions when compared with other inherited diseases.

Conclusions

Despite the fact that PST for disorders such as Huntington disease, hereditary ataxias, and other degenerative conditions has been offered for more than 20 years, good methodological approaches to assess quality of the genetic counseling process in the context of PST remain elusive. This restricts improvement of the protocols for genetic services and in general healthcare for the at-risk population. We therefore propose that rigorous research, including longitudinal studies of the consultand’s experience of this process, is required.

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Disclosure Statement

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References


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