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Exploring Genetic Counselors' Perceptions of and Attitudes Towards Schizophrenia

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Abstract

Schizophrenia is a common complex condition, for which no genetic testing is yet clinically available. Genetic counseling for psychiatric disorders is viewed by genetic counselors as a growth area, and to meet any increase in demand it is important to understand existing context. Thus, we surveyed general practice members of the National Society of Genetic Counselors, to examine perceptions and attitudes relating to schizophrenia. A total of 136 genetic counselors completed the survey, of whom 50% were engaged in general practice roles and therefore eligible to participate. Of these 40% reported “rarely” or “never” asking about psychiatric illness when taking a family history. Some respondents expressed concern that discussing genetics of schizophrenia and providing risk assessment with families may be more confusing or worrisome than helpful. Many counselors reported that patients feel frustrated with the inability of genetic counselors to provide individual risk calculations. It appears that genetic counselors are reluctant to ask patients about psychiatric illness, and are concerned that their services might not be helpful in the context of schizophrenia.

Keywords

genetic counseling; psychiatric; schizophrenia; risk assessment; family history

INTRODUCTION

Schizophrenia is a complex disorder that affects approximately 1% of the population, with recent research indicating that it affects males slightly more frequently than females. The condition is characterized by positive symptoms (gain of function) like hallucinations and delusions, negative symptoms (loss of function) like avolition (lack of motivation), alogia (poverty of speech) and anhedonia (loss of the ability to experience pleasure), and disorganization. Schizophrenia is associated with significant economic burden to society, and emotional burden for affected individuals and their families. Complex diseases like schizophrenia, arise as a result of the combined effects of both genetic and environmental influences, and tend to occur with an increased frequency amongst the family members of an affected individual but do not display a consistent Mendelian pattern of inheritance [1]. Although the exact mechanism of pathogenesis in the development of psychiatric disorders is not yet completely understood, the development of complex genetic disorders is influenced by susceptibility genes, with which other factors (such as other genes and/or environmental stressors) must interact for the disease to present [1].

With respect to schizophrenia specifically, over recent years, a number of genes have been identified (including: Neuregulin (NRG1), Dysbindin (DTNBP1), Regulator of G protein signaling 4 (RGS4), D amino acid oxidase (DAAO)/G72, Catechol-O-methyl transferase (COMT), DRD3/HT2a, and DISC1) which have been linked to pathogenesis. Most of these genes play a role in glutamate neurotransmission, which suggests that the primary defect in schizophrenia may be dysfunctional glutamate neurotransmission [2].

Environmental factors that seem to increase vulnerability to schizophrenia include: stress, living in a city, low socioeconomic status, immigration, winter birth, cannabis use, and obstetric complications [3]. Some findings show that stress (an environmental factor) has stronger relationships with psychosis in patients with a positive family history of schizophrenia than for those with no family history of psychiatric illness, consistent with an interaction between genetic predisposition and environmental factors [4].

Genetic counseling is a specialist service which aims to provide information and support to families affected by disorders that have a genetic contribution. While genetic counseling can certainly be applied to psychiatric illnesses like schizophrenia it has traditionally been viewed by the broader public and healthcare system as being primarily aimed at genetic disorders with Mendelian inheritance patterns. As the public becomes more aware of the genetic contribution to schizophrenia, it has been postulated that referrals for genetic counseling and genetic services for this condition may increase [5]. Indeed, psychiatric genetic counseling was seen as an area of growth by 96% of the members of the National Society of Genetic Counselors, who responded to a 1994 survey [6]. As more is learned, genetic testing for schizophrenia may eventually become clinically available, and several studies have now demonstrated that individuals with psychiatric disorders and their family members are generally interested in this option [7–9].

Even in the absence of genetic testing, genetic counseling seems to be of great interest to a large proportion of affected individuals and their families [10–12]. Families often want an explanation as to why their family member became ill, may be interested in risks for other family members to become similarly affected, or may have questions regarding their own risk that can be addressed by using empiric data as a basis for individualized risk assessment [13].

Although some work has been done with regards to determining the needs of affected individuals and their family members relating to genetic issues, considerably less research has addressed genetic counselors' practices and perceptions surrounding psychiatric illnesses like schizophrenia. One pilot study examined the needs of practicing genetic counselors when discussing psychiatric illness, and found that respondents felt unprepared to raise the issue of psychiatric disease with patients, or to answer questions regarding psychiatric illness raised by clients and 22% reported raising issues related to psychiatric illness "seldom to never." [14].

This exploratory study sought to expand upon these themes, by investigating genetic counselors': 1) approaches to genetic counseling for schizophrenia, and 2) investigating perceptions and attitudes towards providing genetic counseling for schizophrenia. We

specifically selected schizophrenia for investigation because we felt it was the psychiatric illness that genetic counselors might be best able to address, as compared to other psychiatric illnesses, there is more information available about genetic/environmental contributors, and recurrence risks for relatives. This exploratory, descriptive study included questions regarding how genetic counselors feel about: gathering information about family history of psychiatric illness; and providing information about etiology of and risk assessment for schizophrenia.

METHODS

Recruitment

We designed an internet-based questionnaire to be completed by genetic counselors. The survey was posted on a website (Survey Monkey), and an electronic waiver of written informed consent was provided. Study participants were recruited between October and December 2006 by means of an invitation to complete the survey that was posted on the National Society of Genetic Counselors (NSGC) list serve. The study was approved by the Arcadia University IRB.

The survey was open for all genetic counselors to complete, but we selected only those responses that were submitted by genetic counselors engaged in general practice genetic counseling. The responses from those engaged in providing specialty services (e.g. cancer counseling) were excluded, as counselors in such positions focus on providing a service related to specific conditions, and may have less opportunity to ask about or address a family history of psychiatric illness.

Questionnaire

The questionnaire contained several demographic items, including respondents' age, sex, number of years practicing as a genetic counselor, and practice specialty. As well, we collected information about genetic counselors' practices relating to schizophrenia, to help us to contextualize the data of interest (see Table 1). These items were answered using a Likert scale that included the following options: never, rarely, sometimes, often, and always. Questions relating to perceptions of and attitudes towards schizophrenia could be answered by selecting a response from a 5-point Likert scale that included the following options: strongly agree, agree, neither agree nor disagree, disagree, strongly disagree (see Table 1).

Data Analysis

Data were entered into an SPSS database, and were analyzed using descriptive statistics.

RESULTS

Demographics

A total of 136 genetic counselors completed the questionnaire of whom 65 (48%) were general practice counselors, and were therefore eligible to participate in the study. Ages ranged from 24 to 64 years (average 32.5 years), and years of experience ranged from <1 to 34 years (average: 6.7 years), 98% of respondents were female.

Current practices relating to Schizophrenia

Of the 65 general practice genetic counselors, 40% “rarely” or “never” ask patients about family history of psychiatric illness themselves, only 31% said that they “always” or “often” ask. Most genetic counselors (83%) reported seeing patients who are referred specifically to discuss psychiatric illness “rarely” or “never”. When presented with a significant family history of schizophrenia, 40% “rarely” or “never” discuss environmental risk factors, and 75% “rarely” or “never” discuss early warning signs of mental illness. Further, regarding the provision of information about the pattern of inheritance and current understanding of the genetics of schizophrenia, 48% provide “as little as possible”, “very little” or “some detail”, 14% provide “as much as possible with the available literature”, and 38% reported being “as general as possible at first, then more if requested”. When providing risk assessment information, 44% were “as general as possible” or “slightly patient specific”, 27% were “as general as possible at first, the more specific if the patient requests,” and 29% said “as specific as possible with the available literature”, none reported that they “do not provide” specific risk assessment information.

Genetic Counselors’ perceptions and attitudes towards Schizophrenia

Most respondents (71%) reported no discomfort when asking patients about family history of psychiatric illness, but 13% did report feeling uncomfortable in this situation, and 16% were uncertain. A third (33%) felt that patients were uncomfortable when asked about psychiatric illness, 38% did not think that patients were uncomfortable, and 29% were uncertain.

Data regarding genetic counselors perceptions of the degree to which discussing with patients the known genetics of schizophrenia and specific risk assessment is helpful. In total, just over ¼ of respondents (26%) agreed or strongly agreed that discussing the known genetics of schizophrenia and its pattern of inheritance is more confusing than it is helpful. 14% agreed or strongly agreed that discussing specific risk assessment in high risk families is more worrisome than it is useful, and just under half of respondents (47%) agreed or strongly agreed that patients often feel frustrated with the inability of genetic counselors to provide an individual risk calculation for schizophrenia.

Interestingly, 76% of genetic counselors surveyed agreed/strongly agreed that they would be more willing to discuss a family history of psychiatric illness if genetic testing was available and 80% agreed/strongly agreed they would be more willing if individualized risk assessment could be provided.

DISCUSSION

In this exploratory, descriptive study, a large proportion of genetic counselors reported that they do not ask about psychiatric illness when taking a family history. Thirteen percent of genetic counselors reported feeling uncomfortable about asking patients about family history of psychiatric illness, and a greater proportion (33%) felt that patients are uncomfortable being asked. While these proportions represent the minority of genetic counselors, the origins of the discomfort (e.g. do genetic counselors hold stigmatizing attitudes towards

individuals with schizophrenia?) are worthy of future investigation. In particular, it would be interesting to determine whether patients actually do feel uncomfortable to be asked about family history of psychiatric illness, or whether the discomfort actually arises from the genetic counselor, indicating countertransference. Further research into such questions is warranted, as the implications – including exacerbation of stigma relating to mental illness – of the discomfort expressed are largely negative.

When a family history of schizophrenia arises, many genetic counselors in this study reported providing little detail about the pattern of inheritance and current understanding of the genetics of the illness. This may be due to lack of knowledge, time constraints, or belief that it may not be beneficial to discuss a disorder for which there is no genetic testing. From these data, it is not possible to infer which (if any) of these hypotheses may be correct, but this would be important to investigate in future studies.

The fact that many genetic counselors in this study would be more willing to discuss schizophrenia if genetic testing were available is of interest. Despite ever-increasing knowledge regarding genetics of schizophrenia, the genetic variants that have been identified to date that seem to play a role in illness susceptibility only have small effects on risk. The effects of interactions of these variants on risk remains unknown, and because of the important role of the environment, genetic testing will never discriminate absolutely between individuals who will and will not develop schizophrenia. Thus, at least for the foreseeable future, any variants for which testing becomes available will each only allow modest modifications to risk, and thus will be unlikely to impact the practice of genetic counseling in the same way that the advent of the availability of BRCA1/2 testing impacted the profession. Currently, family history is a better predictor of risk for schizophrenia than would be genetic testing for any of the genetic variants that have been identified to date [15]. It would appear that perhaps, in the absence of being able to provide access to genetic testing, some genetic counselors may be uncertain of the value of their service.

In this study, some genetic counselors felt that discussing risk assessment in high-risk families is more worrisome than useful. Many respondents reported very modest attempts to provide patient-specific risk assessment, and accordingly, almost half reported feeling that patients are often frustrated by the inability of genetic counselors to provide individualized risk assessment. Many reported a greater willingness to discuss a family history of schizophrenia in more detail if an individual risk calculation could be provided. Together, these findings indicate that there seems to be a systemic problem with regard to risk assessment for schizophrenia in genetic counseling.

Previous commentaries on genetic counseling process have emphasized the importance of facilitating accurate risk perception [16–18]. This is equally true in the case of psychiatric illnesses – healthy relatives of affected individuals overestimate risks and this influences reproductive decisions [11, 12, 19]. Respondents in this study reported being more willing to discuss a family history of schizophrenia if an individual risk calculation could be provided, thus implying that respondents felt that this is currently not possible. Although it can be challenging, individualized risk assessment for psychiatric illnesses can be performed: empiric risks are available and can be modified according to factors that have been detailed

elsewhere [13]. Thus, perhaps genetic counselors' practice might be enhanced by further education in this area (for example, see Peay et al 2008 and Austin et al 2008).

Further, in order to improve genetic counselors' comfort with schizophrenia and willingness to address family histories of psychiatric illnesses in clinical practice more broadly than simply addressing recurrence risks, it seems that there may be a need for a greater emphasis on exposure to and discussion of these conditions, starting at the graduate program level.

Limitations

This study was designed to be exploratory, and primarily descriptive in nature, thus limiting our ability to draw conclusions about possible associations within the data. Second, demographic data were illustrative not exhaustive. Further, only one invitation to participate in the study was sent, follow up invitation may have improved the response rate and increased sample size. In addition, the choice of anchors on some of the questions that involved Likert scale based responses may not have been optimal. For example, the anchor that was selected to reflect uncertainty to a particular item was "neither agree nor disagree", but respondents could have selected this option to reflect ambivalence. We feel however, that the visual layout of the scale (left to right arrangement of responses reflecting strong disagreement to strong agreement) provides some robustness. Despite being anonymous, this type of study is vulnerable to respondents wanting to give the "right answer", based on assumptions of how a "good" genetic counselor thinks and acts. Last, we cannot exclude the possibility of ascertainment bias - perhaps genetic counselors that participated in this study were more likely to have an interest in psychiatric illness. Despite these limitations, this study is important in that it begins to shed some light into current practices relating to psychiatric genetic counseling, and as such it can inform future research into this important area of practice that genetic counselors perceive to be growing.

Future Research

There is a need for further study in several related areas. First, it would be interesting to investigate how individuals referred for genetic counseling respond to being asked about psychiatric illness as part of the family history. In the event that patients were comfortable with these questions, perhaps this would allay genetic counselors' fears about raising the issue. Second, for the same reason, it would be interesting to assess reactions of individuals who are referred for psychiatric genetic counseling to uncertain risk figures. Third, there is also a need for research into the reasons for which genetic counselors provide little detail about the pattern of inheritance and current understanding of the genetics of schizophrenia (e.g. lack of knowledge, time constraints, or belief that it may not be beneficial to discuss a disorder for which there is no genetic testing), to inform the design and provision of constructive solutions.

Conclusions

Genetic counseling for psychiatric illnesses may have important effects, and affected individuals and their families are keen to receive information and support that genetic counselors can provide. Learning about genetic counselors' perceptions of providing services

for psychiatric illness is integral to improving our understanding of factors that hinder or facilitate this type of genetic care, and thus improving service. This study showed that genetic counselors: infrequently ask patients about psychiatric illness, seem to lack confidence in their ability to address family histories of psychiatric illness, and may have somewhat unrealistic expectations regarding the utility of genetic testing for schizophrenia. There appears to be a need for educational interventions for genetic counselors aimed at increasing knowledge and confidence in helping families with these disorders.

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Table 1

Survey questions

Questions about genetic counselors' practices relating to schizophrenia	Do you routinely ask about a family history of psychiatric illness? [#]
	When discussing a significant family history of schizophrenia, how much detail would you provide to the patient about the pattern of inheritance and current understanding of the genetics of schizophrenia? ^a
	When discussing a significant family history of schizophrenia, how often do you discuss the environmental risk factors that are known to be associated with schizophrenia?
	When you counsel a patient with a significant family history of schizophrenia, how often would you discuss the early warning signs with your patient?
	When discussing a significant family history of schizophrenia, how do you provide specific risk assessment information? ^b
Questions relating to genetic counselors' perceptions and attitudes towards schizophrenia	It is uncomfortable to ask patients if there is a history of psychiatric illness in the family. [#]
	Patients seem uncomfortable when asked about a history of psychiatric illness. [#]
	Discussing the known genetics of schizophrenia and its pattern of inheritance for patients at high risk is more confusing than it is helpful.
	Discussing patient specific risk assessment in families at high risk for schizophrenia is more worrisome than it is useful.
	Genetic counselors would discuss a family history of schizophrenia in more detail if genetic testing were available.
	Genetic counselors would discuss a family history of schizophrenia in more detail if an individual risk calculation could be provided.
	Patients often feel frustrated with the inability of genetic counselors to provide an individual risk calculation for schizophrenia.

[#] The term "psychiatric illness" was used, rather than "schizophrenia". These questions were constructed to reflect genetic counselors practice, which would usually involve inquiring about whether there was a family history of schizophrenia only after asking about mental or psychiatric illness more broadly.

^a Answer options that were available for this question included: "As little as possible", "Very little detail", "Some detail", "As general as possible at first, then more specific if the patient requests", "As much detail as possible with the available literature".

^b Answer options that were available for this question included: "As general as possible", "Slightly specific (patient specific)", "As specific as possible with the available literature", "As general as possible at first, then more specific if patient requests", and "Don't provide".