Genetic Counseling in Psychiatry: Preparing for Anticipated Demand

December 01, 2006 | ADHD [1], Bipolar Disorder [2], Schizophrenia [3], Autism [4], Major Depressive Disorder [5], Alcohol Abuse [6]

In a recent article on genetic counseling in psychiatry, Christine Finn, MD, and Jordan W. Smoller, MD, ScD, noted that family and twin studies have documented the familiality and heritability of schizophrenia, bipolar disorder, major depression, anxiety disorders, autism, attention-deficit/hyperactivity disorder (ADHD), and Tourette syndrome, among others, and that molecular genetic studies have begun to identify possible susceptibility loci for several of these disorders, most notably schizophrenia.

"Right now, the clinical applications of genetics to psychiatry are relatively small," Finn told Psychiatric Times, "but I think that is going to change dramatically in a relatively short time as we are able to identify genes that may be important for the development of psychiatric disorders or that may be important for gauging responses to medications or even other forms of treatment."

Finn, who is board certified in both psychiatry and genetics, warned that there may not be enough trained genetics professionals to handle the increasing demand for genetic counseling and testing. What's more, she said, genetic counselors currently do not get much exposure to psychiatry. "Inevitably," she added, "it is going to fall on primary care physicians and other specialties, like psychiatry, to have some facility with genetic information and to be able to use that [information] in their clinical practice to really give patients the highest standard of care."

Unfortunately, genetics and genetic counseling "is not something that a lot of psychiatrists know much about," Finn said. In a survey of 352 psychiatrists, respondents had a median score of 44% on the general genetic knowledge items and 33% on the psychiatric genetic knowledge items. In particular, the surveyed psychiatrists tended to underestimate the genetic contribution to schizophrenia, autism, Alzheimer disease, and ADHD. Despite their knowledge gaps, 83% of the psychiatrists considered it their role to discuss genetic information with patients and families. The need is great and growing. American Psychiatric Association (APA) guidelines have endorsed genetic counseling to aid family planning for individuals with bipolar disorder; the APA also indicated that when genetic counseling is used in the psychiatric evaluation of adults, the relevance of patients' value systems should be taken into consideration.

Genetic information also can guide treatment. For example, a patient undergoing a first episode of depression whose close relatives have bipolar disorder may be better off starting with a mood stabilizer rather than a standard antidepressant. Disorders for which genetic testing may be valuable already available, Finn said, is genetic testing for several highly penetrant single-gene or chromosomal abnormalities that can cause psychiatric symptoms. "The one I am most familiar with and most interested in is velocardiofacial/DiGeorge syndrome [VCFS]," she said. "It is relatively common as genetic disorders go. Other than Down syndrome, it is probably the most common genetic syndrome that we know about. Down syndrome occurs, on average, in about 1 in 700, and [VCFS] occurs, depending on what you read, in between 1 in 2000 and 1 in 4000 live births."

VCFS is caused by a microdeletion in the long arm of chromosome 22. Psychotic symptoms develop in about one fourth of VCFS patients. Conversely, among patients with schizophrenia, Finn said,
about 2% are estimated to have undiagnosed VCFS, and among those with childhood-onset schizophrenia or psychosis, about 6% are estimated to have undiagnosed VCFS.

While features of schizophrenia in patients with VCFS are not readily distinguishable from those of schizophrenia in patients without VCFS, according to Finn, a couple of differences do exist. Individuals with schizophrenia and VCFS are thought to have fewer negative symptoms but more adverse responses to atypical neuroleptics, including increased frequency of seizures.

Some other genetic disorders with psychiatric manifestations that can be tested for include fragile X syndrome, linked to learning disabilities, mood lability, and autistic-like behaviors; Smith-Magenis syndrome, a condition marked by mental retardation, distinctive facial features, sleep disturbances, and such problems as hyperactivity, anxiety, and explosive outbursts; Huntington disease, a progressive disorder that often begins with irritability, depression, small involuntary movements, poor coordination, and trouble learning new information or making decisions; and Wilson disease, the signs and symptoms of which can include clumsiness, trembling, difficulty in walking, speech problems, deteriorating school performance, depression, anxiety, and mood swings.

"When I see children with bipolar disorder, I am often running through the criteria for Smith-Magenis to see [whether] that is something I need to think about," Finn added. While Huntington disease and other disorders are linked to a single gene, most cases of psychiatric illness are believed to be genetically complex, Finn said, so "we may not ever get a yes or no answer from testing."

**Testing and counseling**

Psychiatric disorders in most cases result from "multiple genes interacting with each other and interacting with environmental factors that, for the most part, we may not know about," she said. Consequently, genetic testing in psychiatry may be mostly about susceptibility and probabilistic test results. Patients may be told, for example, based on this combination of 8 genes, that they have a 50% chance of having schizophrenia develop.

Despite the complexity, most research studies suggest that patients, family members, and mental health clinicians are interested in genetic counseling and testing.  

Finn, who is affiliated with Harvard Medical School-Partners Healthcare Center for Genetics and Genomics and Massachusetts General Hospital, provides genetic counseling relevant to psychiatric disorders. She counsels people affected with psychiatric disorders who have questions about passing on illness to their children or who are thinking about using medications during pregnancy. She also counsels individuals who are planning to adopt a child with a family history of mental illness, as well as siblings and other relatives of affected individuals who have questions about their own risk or about risk to their children. Many times, Finn observed, siblings especially have not been able to talk with mental health professionals about risks and their experiences growing up with someone who has a psychiatric disorder. She uses the counseling opportunity to educate them about the disorder, including the typical symptoms, the usual course of illness, and treatment options.

**Training needed**

To prepare for the emerging advances in genetics and heightened demand for genetic counseling, Finn believes there must be increased educational efforts during medical school and residency training to convey genetic information specific to psychiatry, along with an increased emphasis on genetics in psychiatry in continuing medical education (CME) programs. In the survey of psychiatrists, an overwhelming majority (77% to 93%) indicated interest in a variety of educational opportunities about genetics, including CME courses and written and Web-based materials. Finn noted that there is an upcoming article in the Harvard Review of Psychiatry (2007;15[1]) that will discuss education in genetics for residents and propose some curriculum guidelines.

The National Society of Genetic Counselors, according to Finn, is beginning work on practice guidelines for genetic counseling in psychiatry and hopes to collaborate with representatives from the APA on this topic.

In their journal article, Finn and Smoller listed several online resources that provide useful information about the genetic basis of a range of disorders including neuropsychiatric conditions. These include GeneTests (www.genetests.org) and Online Mendelian Inheritance in Man (OMIM) (www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM) Another excellent resource, Finn said, is the Web site of the National Coalition for Health Professional Education in Genetics (NCHPEG) (www.nchpeg.org) of which the APA is a member. Psychiatrists, Finn said, can find on the site an online version of the CD-ROM, "Genetics and Major Psychiatric Disorders," and other educational materials on psychiatric genetics. The tutorial reviews the genetic basis of major psychiatric disorders, describes genetic conditions that may present with psychiatric symptoms, provides recent research updates in the field of psychiatric genetics, and outlines issues in genetic counseling for
psychiatric disorders. NCHPEG's Content and Instruction Working Group also is working on a project focusing on genetics in bipolar disorder.

**Elements of genetic counseling sessions**

Finn and Smoller,¹ in their review article, describe the indications for and suggested elements of psychiatric genetic counseling sessions. At a minimum, the session should include gathering information (purpose of the consultation, individual and family histories, and evaluation of capacity of the client to participate in consultation); conveying information (estimations of recurrence risk based on family history and empiric risk data; general information regarding psychiatric illness, available treatments, and options for support); exploring the consultant's understanding of the information; facilitating decision making and providing support; and following up (in person or in writing).

"This is a complex process and not something you can squeeze into a couple of minutes' conversation with your patient," Finn said, adding that it is a process that deserves a lot of time, thought, preparation, and often collaboration with genetics professionals. These consultations often take her about an hour and a half to 2 hours and another hour of documentation, she said, and reimbursement can be a problem.

"Reimbursement has come up almost every time I have done one of these consultations. If I put it under genetics, it is more straightforward, but if I try to do it as a psychiatrist, there really isn't a good CPT code to reflect what I am doing," she said. The reimbursement issue is even more challenging when the client does not have a diagnosed psychiatric disorder but is at risk for one.

"As we move to a model in general in medicine where we hope to be providing more preventive care, there needs to be a shift in the way we can bill for services," she added.

Patients who have questions beyond the knowledge base of their psychiatrist can be referred to the National Society of Genetic Counselors. (www.nsgc.org), which allows individuals to search for genetic counselors based on location and areas of specialization; to GeneTests; or to a department of genetics at a major medical center.

"Overall," Finn added, "it is an exciting time to be in psychiatry and to have an additional tool to help us understand psychiatric disorders and . . . to help us think about new treatment opportunities and prevention strategies."

**References:**


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