14 - 28.14 Genetic Counseling

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Schmidt NB. Combining psychotherapy and pharmacological service provision for anxiety pathology. J Cogn Psychother. 2005;19(4):307. Szigethy, EM, Friedman, ES. Combined psychotherapy and pharmacology. In: Sadock BJ, Sadock VA, Ruiz P, eds. Kaplan & Sadock's Comprehensive Textbook of Psychiatry. 9th ed. Vol. 2. Philadelphia: Lippincott Williams & Wilkins; 2009:2923. Szuhany KL, Kredlow MA, Otto MW. Combination Psychological and Pharmacological Treatments for Panic Disorder. Int J Cogn Ther . 2014;7(2):122–135. Ver Eecke W. In understanding and treating schizophrenia: A rejoinder to the PORT report's condemnation of psychoanalysis. J Am Acad Psychanal. 2003;31:11–29. 28.14 Genetic Counseling Medical geneticists and specially trained and qualified genetic counselors have traditionally provided genetic counseling to patients in need of such help. Many psychiatrists, however, are also well placed to provide genetic education and counseling because they often have knowledge of their clients' needs and family histories and have ongoing therapeutic relationships. The ideal approach for providing psychiatric genetic counseling is through a multidisciplinary team approach, with collaboration between genetics and mental health professionals. Genetic professionals often seek collaboration with a psychiatrist for those with difficult psychiatric medical or family histories. Genetic professionals also seek collaboration or referral for persons with a psychiatric disorder; those who are having difficulty adapting to a genetic-related diagnosis; those dealing with the death of a family member; or those who are experiencing persistent difficulty with decision making regarding prenatal diagnosis or genetic testing. In turn, genetic professionals can be available for professional consultation regarding risk assessment, the collection and construction of complicated family medical histories, and the availability and limitations of genetic or genomic testing. DEFINITIONS Genetic counseling is the process of helping people to understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. According to the National Society of Genetic Counseling, it integrates three factors: (1) interpretation of family and medical histories to assess the chance of disease occurrence or recurrence; (2) education about inheritance, testing, management, prevention, resources, and research; and (3) counseling to promote informed choices and adaptation to the risk or condition. The process aims to minimize distress and facilitate adaptation, to increase one's feeling of personal control, and to facilitate informed decision making and life planning. Genetic counseling is not limited to considerations of the genetic contributions of disease. Genetic counseling also considers environmental components of the presenting disease along with genetic ones. Table 28.14-1 lists common terminology used in the field of genetic

counseling. Figure 28.14-1 illustrates a complex family medical history

presented in the form of a pedigree. FIGURE 28.14-1 Pedigree of a family with velocardiofacial (VCF) syndrome. ADHD, attentiondeficit/hyperactivity disorder; Dx, diagnosed; MR, mental retardation. (From Sadock BJ, Sadock VA, Ruiz P, eds. Kaplan & Sadock's Comprehensive Textbook of Psychiatry. 9th ed. Philadelphia: Lippincott Williams & Wilkins, 2009, with permission.) Table 28.14-1 Genetic Terminology

GENETICS AND MENTAL HEALTH Disorders can recur in families for many reasons, including the functioning of genes (single genes vs. polygenic) (Table 28.14-2), shared environmental exposures, a combination of genetic and environmental factors (multifactorial), and cultural transmission. Single gene disorders are caused by defects in one particular gene, and they often have simple and predictable inheritance patterns. By contrast, most psychiatric disorders are multifactorial in etiology, influenced by multiple genes as well as environmental factors, making them more difficult to predict. Table 28.14-2 Examples of Psychiatric Disorders Recognized as Having a Genetic Component to Their Etiology

Two phenomena that further complicate genetic counseling include penetrance and expressivity. Penetrance refers to the portion of individuals with a specific genotype who also manifest that genotype at the phenotype level. If all individuals who carry the dominant gene show any phenotype of the gene, the gene is said to be completely penetrant. Currently, only rare examples exist of known genes for mental disorders that demonstrate complete penetrance of symptoms in the presence of a single gene. One such example is early-onset familial Alzheimer's disease resulting from mutations in the amyloid precursor protein (APP) located on the long arm of chromosome 21. In contrast, expressivity refers to the extent to which a genotype is expressed. In the case of variable expressivity, the trait can vary in expression from mild to severe, but is never completely unexpressed in individuals who have the gene. The genes that result in most mental disorders are believed to regulate a wide spectrum of traits demonstrating variability of expression (spectrum disorders). COMPONENTS OF THE GENETIC COUNSELING PROCESS Requests for genetic counseling are often initiated by the client's or relatives' questions about the disorder that is present in the family. In the case of mental illness, the questions are often posed to the treating psychiatrist. The client's questions are most effectively addressed through an interactive process that provides the client, as well as the professional, with information pertinent to the next step in the communication process. The basic components of genetic counseling are outlined in Table 28.14-3. Table 28.14-3 Steps and Process of Genetic Counseling Contracting Contracting is a vital portion of the psychiatric genetic counseling session. Often the goals of the session will vary based on the consultand's histories and reason(s) for concern. The provider should work with the consultand at the beginning of the session to define mutual goals.

Documentation of Diagnosis, Collection, and Review of Family Medical History A family medical history (FMH) is collected, and at least a three-generation pedigree is constructed. The collection of FMH begins with the individual seeking information. The consultand (or client) is the individual seeking information. Proband is the term used to identify the affected person within the family who first brought the family to medical attention. The FMH should be comprehensive and include the following information: ages (or dates of birth) of each family member, the age at which the diagnosis was made for individuals with the disorder, pregnancy losses (including the gestational

length along with the recognized cause, if known), the recognized cause and age of any deceased family members, and ethnic backgrounds (Table 28.14-4). Table 28.14-4 Topics Included in the Family Medical History Confirmation or clarification of the diagnosis is essential to the provision of valid information within the session. This usually requires obtaining medical records to clarify or to confirm the suspected diagnosis in the relatives. Depending on the situation, genetic testing may be available for at-risk members in families with single-gene disorders; but because DNA (deoxyribonucleic acid) testing for most mental disorders is not yet an option, risk assessment is based solely on analysis of the pedigree. The collection and review of the FMH with the patient might elicit or recall intense feelings of sadness, guilt, anxiety, or anger. Furthermore, the graphic presentation of the family history may bring to light a more concrete realization of an individual's risks; therefore, attention to the patient's affect is important throughout the process. Specific issues that may hinder an accurate psychiatric family history and may increase consultand affect related to family history are listed in Table 28.14-5. Table 28.14-5 Issues that Can Hinder an Accurate Psychiatric Family History

Communication of Risk and Decision Making Individuals vary in their level of understanding risks. The provision of risk information is best approached in a balanced and accurate manner that is tailored to the patient as much as possible. There is the temptation to use nonnumeric phrases of probability (e.g., often, rarely, most likely); however, the meaning of these nonnumeric phrases is highly subjective and their use in the genetic counseling session introduces the potential for bias. Ideally, risks should be presented in several different ways, taking clues from interactions with the client that inform the approach. Some examples of approaches to assist the client's understanding of risks include stating numeric risks as percentages (25 percent) and as fractional risks (one-in-four chance). It is important to frame risks from the perspective of a negative and a positive outcome; for example, there is a 1 percent chance that the test will result in a complication and a 99 percent chance that there will be no complication. Owing to the high rate of co-occurring disorders and the wide phenotypic range of psychiatric disorders, patients should be informed of potential risks for disorders other than those that brought them to genetic counseling. An example of this is the risk to first-degree relatives of an individual diagnosed with bipolar disorder. In this situation, the risk for bipolar disorder is increased for first-degree relatives, as are the risks for unipolar disorder, schizoaffective disorder, and cyclothymia. It should be made clear that the risks are determined from populations and not derived from individuals and, therefore, are estimates at best. Table 28.14-6 provides a compilation of recurrence risks from various referenced sources in the literature. Table 28.14-6 Empirical Risks for Selected Mental Disorders

PSYCHOSOCIAL COUNSELING AND SUPPORT Setting the stage for the inclusion of psychological and emotional issues can occur early in the process by verbalizing the intent to provide factual information, as well as fostering a discussion of the client's reaction to the information. Insight into the client's perspective and experiences with the disorder, values, beliefs, and family dynamics can begin to be obtained through asking what brings the client to the genetic counseling session. Eliciting this personal information provides a relational context from which the provider can assess concerns and emotional issues. Collection of the FMH can also provide a backdrop of the client's and family's experiences with the disorder. The

exchange of information that occurs during the collection of the FMH can identify underlying risk and perceptions, family beliefs or myths regarding the disorder, and existing support system within

the family. A couple in their mid-30s with a 10-year history of infertility had been trying to adopt a child for a number of years. Recently, the adoption agency they were working with told them of a baby who was being placed for adoption because the biological mother was affected with bipolar disorder and did not feel that she could provide adequate care for the baby. The FMH collected on the newborn baby did not identify others in his family with mental disorders. The recurrence risk for bipolar disorder to the newborn was, therefore, estimated to be between 5 and 20 percent, with additional risks for other mental disorders. The couple individually reacted quite disparately to the estimated risks. In attempting to help them clarify the factors contributing to their feelings regarding the risks, the husband shared his experience with a childhood neighbor who had “some kind of mental illness” and detailed the “torment and agony” that the child brought to the family. Retorting, the woman shared the fact that her coworker also had bipolar disorder and did “just fine” at work with the help of medication. She therefore did not feel that the risks for mental disorders were of concern. The psychiatrist facilitated the couple’s discussion of the spectrum and meaning of mental illness, along with recurrence risks in the context of a genetic education and counseling session. Although the couple did not come to agreement at that meeting over the potential for adopting the child, they did feel that the information and sharing of experiences and perspectives about mental disorders were beneficial. They agreed to return in 1 week after further considering the issues in an effort to reach a decision regarding the adoption. (Courtesy of Holly L. Peay, M.S., and Donald W. Hadley, M.S.) CHALLENGES POSED BY PRESYMPTOMATIC AND SUSCEPTIBILITY GENETIC TESTING Psychiatrists will be on the front line for receiving requests for genetic counseling and testing because of their established relationship between patients and families with mental disorders. The identification of these risks will most likely occur before the discovery or availability of preventative options. The option of knowing risks without preventative options raises concerns regarding the impact of such knowledge on the individual’s mood, anxiety, distress, self-image, reproductive decisions, career decisions, family relationships, insurability, employment, and, potentially, other areas. A model for the provision of presymptomatic genetic testing is provided through the protocol developed for Huntington’s disease (see the Hereditary Disease Foundation web site at www.hdfoundation.org). This model recommends conducting education, counseling, and evaluative sessions over an extended period of time (3 to 4 months), during which time information is provided, questions are addressed, and counseling is initiated, thus maximizing informed decision

making. The process is most appropriately undertaken in the absence of other stressful events (e.g., death of a family member, diagnosis of the disease in another family member, job loss, and divorce). Studies suggest that most individuals receiving information of their increased risk for the disease in their family experience significantly more anxiety, depression, and psychological distress and have poorer perception of their health over the short term (within 1 month after receiving test results) compared with their baseline levels, but no difference over the long term (as long as 1 year after the receipt of results) compared with pretest levels. Consideration should also be given to the impact of such information on the spouse, because initial studies have suggested that the spouse may experience higher levels of depression related to the presymptomatic diagnosis than the client. Furthermore, partners of gene-positive individuals may experience increased levels of intrusive thoughts, avoidance, and hopelessness over the short and long term compared with baseline levels. ETHICAL, LEGAL, AND SOCIAL CONSIDERATIONS Certain individuals and families may experience significant levels of stigma associated with the identification of a genetic disorder, a situation already familiar to individuals and families with mental illness. The added knowledge of a hereditary component may heighten stigmatization. Conversely, having an

identified, biological basis may supplant current public perceptions that mental illness is somehow a personal or family failure in moral, spiritual, or attitudinal perspectives. Questions frequently arise about the privacy of an individual’s genetic information, the ability of employers or insurers to access such information, and the potential of using the information against them by denying insurance, raising rates to unreasonable levels, or denying jobs, and a host of other possible concerns. Currently, no overarching federal laws comprehensively protect citizens of the United States from the potential of these abuses, although significant efforts are continuing in this regard. The status of existing and proposed state and federal laws can be reviewed through the web site of the National Human Genome Research Institute (www.genome.gov). REFERENCES Aatre RD, Day SM. Psychological issues in genetic testing for inherited cardiovascular diseases. Circ Cardiovasc Genet. 2011;4(1):81. Alcalay RN, Caccappolo E, Mejia-Santana H, Tang MX, Rosado L, Ross BM, Verbitsky M, Kisselev S, Louis ED, Comella C, Colcher A, Jennings D, Nance MA, Bressman SB, Scott WK, Tanner C, Mickel S, Andrews H, Waters C, Fahn S, Cote L, Frucht S, Ford B, Rezak M, Novak K, Friedman JH, Pfeiffer R, Marsh L, Hiner B, Siderowf A, Ottman R, Marder K, Clark LN. Frequency of known mutations in early-onset Parkinson disease: implication for genetic counseling: The consortium on risk for early onset Parkinson disease study. Arch Neurol. 2010;67:1116. Beattie MS, Copeland K, Fehniger J, Cheung E, Joseph G, Lee R, Luce J. Genetic counseling, cancer screening, breast cancer characteristics, and general health among a diverse population of BRCA genetic testers. J Health Care Poor Underserved. 2013;24(3):1150–1166. Costain G, Esplen MJ, Toner B, Hodgkinson KA, Bassett AS. Evaluating genetic counseling for family members of individuals with schizophrenia in the molecular age. Schizophr Bull . 2014;40(1):88–99. Finucane B. Genetic counseling for women with intellectual disabilities. In: LeRoy BS, Veach PM, Bartels DM, eds. Genetic Counseling Practice: Advanced Concepts and Skills. Hoboken, NJ: Wiley; 2010;281.

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