

# Psychiatric Genetic Counseling

## Next Steps

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### Abstract

The world's first specialist psychiatric genetic counseling clinic opened in 2012. Despite ample evidence that psychiatric genetic counseling produces excellent outcomes for patients, even in the absence of any genetic testing, this service is still not widely available clinically despite efforts to train practitioners in the delivery of this intervention. Patients could benefit *now* from the delivery of psychiatric genetic counseling (even in the absence of testing), and we have an ethical duty to consider how to ensure that the infrastructure is in place to ensure optimal outcomes for patients. This is particularly important as we move closer toward the clinical application of genetic testing in the context of psychiatric conditions. It is important to consider *how* such testing might be deployed and to ensure that any testing is delivered according to the established practices for psychiatric genetic counseling that produce the best patient outcomes. This chapter reviews evidence that patients benefit from psychiatric genetic counseling and discusses the barriers to its broader implementation.

### Introduction

The potential for benefits to patients from psychiatric genetic counseling has been discussed in the literature for decades, and studies examining its application and outcomes have recently emerged. Without exception, these studies demonstrate positive, meaningful outcomes for patients after receiving this service. The world's first specialist psychiatric genetic counseling service opened its doors in February 2012 and has continued to examine patient outcomes of this service in a naturalistic setting. Such studies are important for examining the effects of an intervention in a real-world setting, rather than in the tightly controlled environment of a randomized controlled trial (RCT) or hypothetical scenarios. Data from the clinic continue to demonstrate positive, meaningful patient outcomes, including large effect size increases in empowerment,

increases in self-efficacy (Inglis et al. 2015), knowledge, improvements in accuracy of risk perception (Hippman et al. 2016), and potential impacts on behavior change (Semaka and Austin 2019) and mental health outcomes (Morris et al. 2021).

Although there is as much (or more) data demonstrating the value of genetic counseling for psychiatric conditions as there is for many other types of genetic counseling already widely available, psychiatric genetic counseling is underutilized and not widely available to patients (Moldovan et al. 2019). This situation is problematic. Not only do we have data demonstrating that patients could benefit now from a service that already exists, we need to consider how to ensure that the infrastructure is in place to ensure optimal outcomes for patients. This is particularly important as we implement genetic testing in the context of psychiatric settings. Specifically, we need to consider not just if but *how* such testing might be deployed and to ensure that any testing is delivered according to the established practices for psychiatric genetic counseling that produce the best patient outcomes.

In this chapter, I present the concept of genetic counseling as it relates to psychiatric conditions. Thereafter, I discuss barriers to its widespread implementation in an effort to lay the groundwork for overcoming these impediments to patient benefit from this service.

### **Genetic Counseling: History, Models, and Theories**

Genetic counseling emerged as a concept in the 1940s (Resta 1997) and as a specialist healthcare discipline in the 1960s. Initially, applied to situations involving family planning (e.g., genetic syndromes) and genetic testing (e.g., hereditary breast and/or ovarian cancer), genetic counseling aims to support people in making informed, autonomous decisions in line with their values, and it does not necessitate or require genetic testing or a family planning trigger. Genetic counseling is best conceptualized as “a process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease” (Resta et al. 2006). In the context of etiologically complex conditions like psychiatric conditions, genetic counseling involves holistic discussion of both the genetic and environmental contributors to the condition.

Today, in Canada, the United States, Australia, and the United Kingdom, genetic counselors are board-certified, MSc-level specialists (Resta 2006) who use a person-centered approach and a model of reciprocal engagement (Veach et al. 2007) to help people make meaning about the causes of a condition in a family, to understand chances for illness in relatives, and to consider options for intervention (Biesecker 2001; Resta 2006; Resta et al. 2006). Genetic counseling is neither a purely education/information-based nor counseling-based service; it is a hybrid (Austin et al. 2014) that seeks to increase empowerment

(McAllister et al. 2010) and facilitate behavioral change to improve health (Austin 2015; Zierhut et al. 2016). Genetic counseling is informed by the *common-sense model of illness representation*, which posits that people gather and integrate information from multiple sources to appraise and develop representations of health that inform self-management and coping (Diefenbach and Leventhal 2010).

## Psychiatric Genetic Counseling

As a discipline, genetic counseling has existed for around sixty years, and its application in psychiatric contexts has been extensively discussed (Austin and Honer 2004; DeLisi and Bertisch 2006; Finn and Smoller 2006; Hodgkinson et al. 2001; Kumar 1968; Lyus 2007; Reveley 1985; Stancer and Wagener 1984). Still, it was not until the 2000s that empirical investigations began (Austin and Honer 2008). Since then, many studies on the utility of genetic counseling have been published (Costain et al. 2012, 2014), including RCTs (Hippman et al. 2016) along with qualitative (Semaka and Austin 2019), naturalistic (Bodnar and Wisner 2005; Gerrard et al. 2020; Inglis et al. 2015), and meta-analytic (Moldovan et al. 2017) studies.

Manuals for psychiatric genetic counseling have been developed (Austin 2019). Typically, counseling involves an initial 1–1.5 hour session (Uhlmann et al. 2010), followed by a check-in one month later; information is gathered from and provided to the patient, along with support (Austin et al. 2006, 2008; Peay and Austin 2011). The information-gathering component of psychiatric genetic counseling entails uncovering the patient’s existing explanation for the cause of illness and eliciting and documenting a detailed family history. The information-provision component involves the following:

- Research-based information about the factors associated with the indicated condition are related to the participant’s family history and their existing explanation for cause of illness—in lay language and with visual aids to facilitate comprehension.
- Information about factors that contribute to the development of illness is used as a framework to discuss evidence-based strategies for protecting mental health; for example, through sleep, nutrition, and exercise (Baglioni et al. 2011; Bodnar and Wisner 2005; Harvey et al. 2018; Lakhan and Vieira 2008).
- Family history and empiric data are used to discuss chance for self as well as chance for family members (e.g., children) to develop the indicated condition, if relevant and desired (Austin et al. 2006, 2008; Peay and Austin 2011).

The genetic counselor also works to uncover and address emotional ramifications (e.g., guilt, shame, stigma, fear) that may be evoked (Veach et al. 2007).

Psychiatric genetic counseling typically involves the provision of written material to patients related to the content of the sessions. For further details of this process and the visual aids, see Austin (2019).

Based on data showing that psychiatric genetic counseling improves patient outcomes, the world's first specialist psychiatric genetic counseling clinic was established, and outcomes continue to be studied (Borle et al. 2018; Gerrard et al. 2020; Inglis et al. 2015; Semaka and Austin 2019). In summary, data show that:

- Psychiatric genetic counseling increases empowerment and self-efficacy (Inglis et al. 2015), both of which are necessary for engaging people in behavioral change to reduce their risk for mental illness (Holloway and Watson 2002).
- People report changing their behavior after psychiatric genetic counseling (e.g., sleep, nutrition, exercise) to reduce their risk for mental illness (Semaka and Austin 2019).

Data accumulated thus far and new, unpublished work suggest a rationale for examining the impact of psychiatric genetic counseling on mental health outcomes. Both suggest that mental health outcomes might be positively influenced by the provision of psychiatric genetic counseling.

Even without any of the more distal outcomes of psychiatric genetic counseling, the proximal outcome of increasing empowerment is deeply meaningful. Empowerment has been conceptualized as the opposite of internalized stigma (Livingston and Boyd 2010), and internalized stigma is a profoundly important issue in the context of psychiatric conditions. For individuals with psychiatric conditions, such as schizophrenia, the effects of stigma can actually outweigh symptoms associated with the condition itself, even those that dramatically influence language, thought, affect, perception, and sense of self (Hinshaw and Stier 2008). Self-stigma, which arises from experiences and perceptions of discrimination (Livingston and Boyd 2010), has been postulated to be central to the psychological harm caused by stigma (Boyd Ritsher and Phelan 2005; Corrigan and Watson 2002; Livingston and Boyd 2010). Therefore, increasing empowerment through genetic counseling is a deeply meaningful outcome in its own right.

### **Genetic Testing for Psychiatric Conditions**

Though they are not the same thing, genetic counseling and genetic testing are often conflated as concepts. As discussed above, there are meaningful and substantive benefits of genetic counseling for psychiatric conditions even in the absence of providing any genetic testing. Given that discourse in psychiatric genetics is increasingly considering the possibility of implementing

genetic testing clinically, this issue deserves special consideration in relation to genetic counseling.

Psychiatric conditions are complex and heterogeneous, and currently there are no genetic tests with which to confirm, refine, or establish a psychiatric diagnosis. Both common variants of small effect (single nucleotide polymorphisms) and rare variants of larger effect (copy number variations, CNVs) can contribute to the etiology of these conditions.

### **Polygenic Risk Scores**

To measure genetic risk, the collective risk due to the total of an individual's common variation can be summarized into a single variable: a polygenic risk score (PRS) (Wray et al. 2018a, 2021). Public interest in genetic information has led an increasing number of people to upload their raw data (e.g., from 23andme.com, ancestry.com) to third party websites (e.g., impute.me) and to generate their own PRS information (Janssens 2019).

Psychiatric conditions are among the conditions for which PRSs are most frequently sought on these platforms (Folkersen et al. 2020). Relatives of people with psychiatric illness worry about and are interested in understanding/mitigating their own risk for developing these conditions (Austin et al. 2006; DeLisi and Bertisch 2006; Erickson et al. 2014; Lyus 2007; Meiser et al. 2005, 2008; Quaid et al. 2001; Quinn et al. 2014; Wilhelm et al. 2009). Data show, however, that ~60% of people who access their own PRS for any complex condition online (where only information is provided, but no support) have some negative reaction (e.g., sad/anxious), and ~5% may even experience PTSD (Peck et al. 2022).

### **Copy Number Variations**

For autism spectrum disorder, testing for CNVs is considered a first-tier test, yet there are important caveats concerning how the diagnostic criteria related to autism have evolved over time, as laid out by Morris et al. (2022).

While CNV testing is an established practice in pediatric settings when a patient experiences developmental delay or intellectual disabilities, its benefits are less clear when these features are not present, and its application in these circumstances is actually off-label under current FDA approval parameters. Many of the potential benefits and harms associated with CNV testing are detailed in relation to different contextual factors by Morris et al. (2022). CNV testing is *not* an established practice in psychiatric settings. Its consideration as a first-tier test in the context of autism has led to the proposal of justice-based arguments in support of CNV testing within psychiatric populations. Careful ethical analysis, however, reveals differences in the potential for benefits and harms associated with CNV testing in different populations (Morris et al. 2022). While many of the harms could potentially be mitigated through

testing in the context of psychiatric genetic counseling, it is critical to consider the timing of testing in relation to whether there are immediate consequences for care delivery dependent on the test and the individual's capacity to consent. If there are no immediate care consequences dependent on the test results, testing should be delayed for children until they are able to consent (Botkin et al. 2015).

### **Insights into the Outcomes of Genetic Testing for Psychiatric Conditions**

Though outcomes of other types of genetic testing in *non*-psychiatric contexts are generally benign (see Bloss et al. 2011; Collins et al. 2010; Green et al. 2009 but also Lineweaver et al. 2014; Turnwald et al. 2019), genetic risk testing for *psychiatric*-related conditions may have some negative outcomes. Especially for those whose results indicate higher risk, potential negative outcomes include increases in negative affect and distress up to three months after the test (Dar-Nimrod et al. 2012; Lebowitz and Ahn 2017; Wilhelm et al. 2009).

Although clinicians tend to focus almost exclusively on *what* information from genetic testing should be communicated, it is increasingly becoming clear that *how* this information is communicated matters at least as much. In sum, any genetic testing implemented clinically for psychiatric conditions should be delivered in the context of the current evidence-based gold standard intervention that exists to help people who are concerned about psychiatric risk, which is psychiatric genetic counseling.

### **Barriers to Adoption and Implementation of Psychiatric Genetic Counseling**

Although studies have shown that large proportions of people with psychiatric conditions and their family members would like genetic counseling, less than 5% have received it (DeLisi and Bertisch 2006; Kalb et al. 2017; Lyus 2007; Michael et al. 2020; Quaid et al. 2001). Given the evidence of meaningful benefits of genetic counseling for people with psychiatric conditions, together with evidence of interest in genetic counseling within this population, it is important to examine the barriers to the more widespread adoption and implementation of psychiatric genetic counseling.

### **Conflation of Genetic Counseling with Genetic Testing**

There is a tendency to think that meaningful and valuable genetic counseling is not possible without genetic testing. Those of us within the psychiatric and clinical genetics communities tend to have an implicit assumption that for our

knowledge about the genetic contributions to conditions to be clinically useful, we need to have a very detailed and specific level of understanding and ideally a genetic test to offer. Accordingly, the value of genetic counseling is typically thought of as providing information about genetic testing. Therefore, since there is currently no genetic testing being routinely clinically implemented for psychiatric conditions, the perception is that there is no reason to consider providing genetic counseling yet. As discussed above, this is a misunderstanding: evidence shows that psychiatric genetic counseling can provide meaningful positive outcomes for people, even without the provision of genetic testing. Indeed, genetic counseling as a profession began at a time when there was very little to offer in terms of genetic testing for many conditions. Thus, the emphasis was on counseling to help people adapt to the condition or risk of condition in their families. Psychiatric genetic counseling is, in many ways, a return to the very well-established roots of the profession.

### **Counseling without Genetic Testing Is Not Valued**

A closely related concept worth mentioning is the fact that in medical practice and healthcare research, the activity of counseling is at an axiological disadvantage relative to genetic testing. As a society, we place more value on information and technological solutions (e.g., genetic testing) than on care-based activities (e.g., counseling). Our focus tends to be on generating information that we might ultimately deliver (developing genetic testing) and thinking about what information to deliver, rather than on considering how to deliver it. At every level, this results in difficulties in advancing the psychiatric genetic counseling agenda: from securing funding for research to establish best practices and outcomes, to implementing new clinical services. This devaluing of care-based activities is at odds with data that demonstrate that if we want to reap the benefits of precision medicine, by helping people to change their behavior to reduce their risk of common complex conditions, then simply providing information is not enough (Marteau et al. 2010). *Counseling* about genetic information, however, holds the potential to help people change behavior. It overcomes some of the fundamental problems, such as addressing emotions that act as barriers to behavior change and connecting behaviors with genetic information in a coherent manner to provide people a sense of agency (Austin 2015).

### **Turf Wars over Who Should Deliver Psychiatric Genetic Counseling**

If we consider psychiatric genetic counseling to be more about counseling than about genetic testing, then questions arise about who should provide psychiatric genetic counseling. Obvious candidates include genetic counselors and physicians, particularly psychiatrists and family practitioners. Psychiatrists tend to point out, quite rightly, that they have ongoing relationships with patients; thus, they are ideally positioned to provide psychiatric genetic counseling.

Given that psychiatric genetic counseling typically involves uncovering and addressing issues connected to feelings of guilt, shame, blame, fear, and stigma that accompany the explanations people have about their condition, psychiatrists refer (entirely appropriately) to their expertise in addressing these emotions. Family practitioners have similar long-standing relationships with their patients. Theoretically, as primary care providers, they would also be ideally placed to address these issues, at least from an accessibility standpoint. In reality, many physicians lack the time (due to being consumed with crisis intervention and medication management), the confidence, or the expertise in issues related to genetics (Finn et al. 2005; Hoop et al. 2008; Hoop and Salva 2010) to have these conversations with their patients. It is important to point out that psychiatric genetic counseling is *not* a brief or group-based intervention (like psychoeducation can often be). Therefore, simply using visual aids to explain to a patient “your condition is caused by genes and environment acting together” does not constitute useful psychiatric genetic counseling. To be effective, counseling must be personalized and involve a two-way exchange of information together with the provision of counseling support. According to research, a typical session takes ~90 minutes. Psychiatrists sometimes point out the similarities between psychiatric genetic counseling and *personalized* psychoeducation interventions. When the latter is available to patients, it may address the issues addressed by psychiatric genetic counseling. (Note: group-based psychoeducation is quite different and will not meet personalized needs in the same way). However, in practice, personalized psychoeducation is inconsistently provided at best.

Ultimately, although physicians may feel it is their role to provide psychiatric genetic counseling, in practice they are rarely able to deliver it, and if patients are not referred for genetic counseling, they will not benefit from what the intervention has to offer. Anyone with clinical expertise in both genetics and psychiatric conditions could reasonably provide psychiatric genetic counseling. In different jurisdictions, different types of healthcare professionals may be best placed to deliver psychiatric genetic counseling. Importantly, what matters is not *who* delivers psychiatric genetic counseling—or even whether it is called “individual psychoeducation” or psychiatric genetic counseling—but *how* it is delivered. It should be delivered in an evidence-based manner. It is obviously important to consider issues that impact cost: physicians’ time is more expensive than that of genetic counselors, and there is a need for all healthcare practitioners to work to the top of their scope of practice (i.e., they should engage with tasks that only they can do, and that cannot be delegated to someone less senior). This highlights another barrier to psychiatric genetic counseling.

### **The Perceived Shortage of Genetic Counselors**

There is no doubt that genetic counselors are in high demand. Although the popular assumption is that there are not enough genetic counselors, the



profession has reacted to growing demand by developing, for example, practice models to aid healthcare professionals through genetic counseling assistants, use of decision aids, and chatbots to facilitate information provision. In North America, there has been a rapid expansion in the number of training programs and available positions within existing training programs (Hoskovec et al. 2018). In several European countries, where genetic counseling does not yet exist as a profession, efforts are underway to develop this specialty (Ormond et al. 2018). The number of genetic tests available is increasing with incredible speed. Given that genetic counseling is often considered to be something that becomes relevant once genetic testing is available, this creates a situation in which the creation of genetic counseling positions is prioritized according to demand, which in turn is typically driven by genetic testing. The genetic counseling service system constantly reacts to external pressures rather than proactively strategizing to create new positions. Therefore, because genetic counselors do not see referral pressure of patients with psychiatric conditions, positions in this specialty are not created (Chanouha et al. 2022). Referral demand creates genetic counseling position supply; if specialty positions in psychiatric genetic counseling were created, research shows that genetic counselors would want to fill them (Van den Adel et al. 2022).

### **Stigma Associated with Psychiatric Conditions and with Genetic Counseling**

Stigma plays out in relation to psychiatric genetic counseling in different ways. First, psychiatric conditions themselves are, of course, stigmatized. Therefore, within the genetic counseling profession there is a degree of lack of comfort with serving this population (Booke et al. 2020). Interventions have been developed to decrease stigmatizing attitudes toward psychiatric conditions among genetic counselors (Anderson and Austin 2012). Specialized training workshops have been offered (Dillon et al. 2022) and found effective in increasing genetic counselors' comfort with delivering psychiatric genetic counseling. The lack of referrals for psychiatric genetic counseling services means, however, that those who have been through these workshops have little chance to practice their skills and very limited clinical exposure to counsel people with psychiatric conditions.

Second, the term genetic counseling seems to be associated with a degree of stigma from the perspective of psychiatrists. Specifically, when the first specialist psychiatric genetic counseling clinic was opened, psychiatrists who could potentially refer their patients expressed a level of discomfort associated with referring a patient to something called "genetic counseling." They explained that they were concerned that their patients might think that a referral to genetic counseling implied a perception that they should not have children. Although psychiatrists valued the psychiatric genetic counseling service and the outcomes they witnessed, they were reluctant to refer patients because of

this fear. Through conversation and with their guidance, the decision was taken to rename the service to the “Adapt Clinic,” and this was an effective remedy for this barrier.

The term genetic counseling can also inhibit referrals by physicians because of misperceptions that it only involves a discussion of the role of genetics in the etiology of the condition. If this were the case, this would indeed be problematic, as physicians appreciate and discuss the roles of stressors in contributing to the development of psychiatric conditions with their patients. Referring to a service that uses a different conceptual model of etiology would be difficult to justify. As discussed above, genetic counseling for psychiatric conditions involves discussing both the genetic and environmental contributions to these conditions in a holistic manner; it does not involve advising people against having children. In fact, promoting and supporting patient autonomy is central to the ethos of the genetic counseling practice (Veach et al. 2007).

To address this issue, I have considered alternative names for the intervention over time. All are inadequate for a variety of reasons. “Nido-genetic counseling”—in Italian, *nido* refers to the nest, the environmental element of the counseling—would be meaningless to most people. “Attributional therapy” or “etiological counseling” are accurately descriptive, but again unclear. Critically, it is important to remember that regardless of name, the content of the intervention is founded on a genetic counseling practice model. Moving away from the genetic counseling label brings the risk of the intervention being perceived as atheoretical or lacking foundation.

Finally, stigma is relevant as a barrier to broader implementation of psychiatric genetic counseling because research shows that physicians primarily consider referring to this service only after patients ask about it explicitly (Leach et al. 2016). Unfortunately, many patients who could benefit most from psychiatric genetic counseling are the same people who are least likely to inquire about it. People often fear being told by an expert that they are somehow responsible for their illness, that there is nothing they can do about it, that they now have to accept being in a state of poor mental health for the rest of their lives, or that any children they might have will certainly develop the same condition. These ideas are antithetical to the content and values of genetic counseling practice in the context of psychiatric conditions.

### **Psychiatric Genetic Counseling May Be a Cost Saver, Not a Revenue Generator**

In nonpublic healthcare settings, services are prioritized, at least in part, according to the potential for revenue generation. Genetic testing is a revenue generator: When a genetic condition has been identified in one individual, interventions can sometimes be offered to that individual (e.g., prophylactic mastectomies in the context of BRCA testing). Testing can also be offered to relatives, which enhances revenue generation. Psychiatric genetic counseling

does not demonstrate such obvious routes for revenue generation, though it is possible that a genetic counselor providing this service could be considered a “physician extender,” allowing the physician to bill for more patients by providing oversight to the genetic counselor.

If psychiatric genetic counseling does indeed have an influence on mental health outcomes, the expectation would be that it would involve cost savings for the healthcare system because people would be better equipped to self-manage their own risk for psychiatric illness. This is an appealing concept from a public health perspective and would potentially be attractive to publicly funded healthcare systems, but it potentially fits less easily within the context of private healthcare systems.

### **Literature Silos**

Many researchers are interested in and publishing papers on various aspects of the potential applications of genetic testing in the context of psychiatric conditions. Though genetic counseling is not yet firmly established as an academic discipline, a large body of data is emerging from this community about the process and outcomes of genetic counseling in the context of various kinds of conditions and in relation to different kinds of genetic testing. To date, however, there is little cross referencing of the genetic counseling literature in the psychology, psychiatry, or psychiatric genetics literature. This perpetuates the misconceptions about genetic counseling (Gershon and Alliey-Rodriguez 2013; Moreno-De-Luca et al. 2018) and could ultimately act as a barrier to its implementation. As a potentially related issue, the genetic counseling community is female dominated, and accordingly, papers published in this community tend to be by female authors. Data shows that female-authored papers are less cited than male authored papers (Dworkin et al. 2020).

### **Summary**

Genetic counseling for psychiatric conditions is of interest to families and is associated with meaningful positive outcomes for patients even in the absence of any genetic testing being provided. Despite these advantages and ongoing efforts to provide training, it remains limited in its availability to patients worldwide. With this article, I hope to further discussion about the barriers to implementation of psychiatric genetic counseling and how they might be overcome. This work is growing increasingly urgent, given the increasing interest in the application of various forms of genetic testing in psychiatry. This urgency is further underscored by the many ethical issues around implementation of genetic testing, and the fact that many of these ethical issues can be readily addressed in the context of evidence-based genetic counseling.

