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21st-Century Genetics in Psychiatric Residency Training:

How Do We Get There?

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Our expanding knowledge of how variation in the human genome contributes to complex diseases has propelled many recent advances in medicine. In oncology, understanding the genetic basis of cancer predisposition, tumor growth, and treatment response has become an essential part of the field and its training. Although current diagnoses in psychiatry are based on clinical observations, we are approaching a time when our genetic and molecular understanding of mental illness may play an important role in etiological understanding and treatment.[1]

It is now well established that different types of genetic variations can increase one's risk for psychiatric disorders. For example, highly penetrant rare variants in hundreds of genes or genomic regions have been strongly associated with autism spectrum disorders (ASD), intellectual disability, and developmental delay, while common variants in more than 100 locations in the human genome have been statistically linked to schizophrenia.[1] The translation of these research findings is starting to permeate psychiatric clinical practice in particular areas. Genetic testing, specifically chromosomal microarray and Fragile X testing, is now recommended for all patients with ASD, intellectual disabilities, and developmental delay.[1] Highly penetrant rare variants are found on chromosomal microarray in 10% to 30% of individuals with these disorders and can have implications on clinical management (Table).[1] Despite these recommendations, genetic testing is occurring only for a minority of these patients. Conversely, pharmacogenetic testing, which can provide information about a patient's drug metabolism rates, response, and risk for adverse events, is widely available

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through direct-to-consumer tests and frequently used despite a significant need for additional research demonstrating improved clinical outcomes for psychiatric patients.[2]

As a field, we must be able to confidently assess and critically appraise the evidence supporting these and other genetic tests. We must understand the concepts that drive these recommendations to properly use this information to enhance patient care. However, current education about genetics across psychiatry residencies and fellowships is heterogeneous and sparse.[3] Psychiatry residents generally leave training programs with low levels of genetics literacy, which may lead to an apprehension about using genetic tools and integrating genetic principles into clinical care.[4] This "education gap" will only widen as the understanding of psychiatric genetics and our ability to translate this knowledge into the clinic expands. Efforts by organizations, such as the American College of Graduate Medical Education and the American Board of Psychiatry and Neurology, to increase their emphasis on the emerging role of genetics in psychiatry will be important to bring attention to this issue. Closing this gap will also require modernized and standardized residency and fellowship education curricula for psychiatric genetics. Efforts to address these issues are currently underway.

The International Society of Psychiatric Genetics Committee on Resident Education (CRE) was created in 2015 with the goals of (1) identifying specific areas in genetics and genomics that are essential for all psychiatrists to understand and (2) collaborating with other organizations to create training material that builds on these defined key knowledge areas. Through a review of the relevant literature and consensus, the CRE has produced a full description of the learning objectives.[5] Briefly, the objectives cover the basic principles of genetics, common genetic variants, rare genetic variants, pharmacogenetics, epigenetics/gene expression, ethical and social issues in genetics, and principles of risk communication. The CRE is currently working with partners to create educational modules that are based on these recommendations, with a focus on creating material that is free, universally available, frequently updated to reflect novel discoveries, based on principles of adult learning, clinically grounded, and potentially administered without depending on local expert faculty.[6] However, further research is needed on the effectiveness of nonexperts teaching psychiatric genetics with these supportive tools.

One of our partners is the National Neuroscience Curriculum Initiative,[6] which creates freely available online learning activities on various neuroscience topics (http://www.nncionline.org) and uses many of the key education principles discussed previously. There are currently modules on genetic testing for rare variants in ASD and pharmacogenetic testing in bipolar disorder. The 2 modules serve different but important purposes. The ASD module encourages clinicians to order diagnostic testing and helps them understand and interpret results and their implications for clinical management. The pharmacogenetics module encourages residents to think critically about using testing given the limited evidence supporting its clinical benefits.

Psychiatry is not the only medical specialty that is seeking to improve genetics education. The Inter-Society Coordinating Committee for Practitioner Education in Genomics was formed in 2013 by the National Human Genome Institute to "improve genomic literacy

of physicians and other practitioners and to enhance the practice of genomic medicine through sharing of educational approaches and joint identification of educational needs."[7] It includes 56 current member organizations representing all fields of medicine (http://www.genome.gov/27554614) (including the International Society of Psychiatric Genetics). The Inter-Society Coordinating Committee for Practitioner Education in Genomics recently published "The Universal Genomics Instructor Handbook and Toolkit," which is a new, free educational resource designed to educate clinicians in all medical specialties in genomic medicine (http://www.pathologylearning.org/trig/resources). The customizable nature of this tool allows many different clinicians with different levels of genetics knowledge to benefit from it. It can also be easily updated as our knowledge advances.

Looking to the future, we must be prepared as a field to tackle the challenge of creating high-quality, interactive, and accessible training materials that communicate increasingly complicated concepts in genetics. For example, psychiatry residents will need to understand the concepts of polygenicity and pleiotropy and learn how multiple genetic risk factors may in the future be combined to predict disease or treatment outcomes (eg, polygenic risk score). They will need to learn to integrate genetic information with other biological, psychological, and social information to optimize clinical care for patients. We encourage psychiatry training programs to consider the increasing role that genetics and precision medicine are playing in our field and tailor curricula accordingly. Programs can make use of new, free, and publicly available materials regardless of levels of expertise among the faculty. With a greater understanding of psychiatric genetics, 21st century psychiatrists will feel empowered to use modern genetic tools to better serve patients.

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Table.

Clinical Benefits of Genetic Testing and Counseling in Psychiatry

Clinical Benefit	Example	Implication
Improved etiologic understanding of the disorder	A de novo deletion of 16p11.2 is identified in a patient with ASD	Parents understand that the etiology of their child's condition is due to a new genetic change and not due to a deficiency in their parenting (https://rarediseases.info.nih.gov/diseases/10740/16p112-deletion-syndrome)
Community support for the patient and family	A patient with developmental delay receives a diagnosis of 15q11-q13 duplication syndrome	The patient and family can find resources, advocacy opportunities, and other families affected by this syndrome through the Dup15q alliance (http://www.dup15q.org)
Reproductive counseling and family planning	The parents of a patient with childhood-onset schizophrenia and a de novo 22q11.2 deletion are considering having a second child	The parents are counseled that the chance of having a second child with 22q11.2 deletion syndrome is slightly increased compared with the general population based on studies of siblings of patients with ASD[1] (https://ghr.nlm.nih.gov/condition/22q112-deletion-syndrome)
Ongoing surveillance for known comorbidities	A patient with ASD receives a diagnosis of PTEN deletion	The patient has a known elevated risk of cancer. Ongoing cancer monitoring is provided to the patient
Experimental treatment referrals	A patient with ASD receives a diagnosis of 22q13 deletion syndrome	Possible referral to a clinical trial for IGF-1 therapy (https://www.mountsinai.org/clinical-trials/pilot-treatment-study-of-insulin-like-growth-factor-1igf-1-in-autism-spectrum-disorder)
Reduce risk of severe medication adverse effects	A patient of Han Chinese ethnicity with bipolar disorder carries the *HLA-B*15:02* variant	The patient would not be administered carbamazepine because of their significantly elevated risk of Stevens-Johnson syndrome[2] (https://www.pharmgkb.org/guideline/PA166105008)

Abbreviations: ASD, autism spectrum disorders; IGF-1, insulin-like growth factor1; PTEN, phosphatase and tensin homolog.

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