Taken as a whole, psychiatric disorders are considered as complex genetic disorders. There are clear genetic mutations and susceptibility factors to these disorders. However, these form the full spectrum of impact, frequency, and mutation type. With rare large scale chromosomal rearrangements and copy number mutations of high impact at one end, and common single nucleotide variations of minor impact at the other. This multitude of variation type also means that different epidemiological study designs are needed to test the genetic component of these disorders, from familial forms, to common population level studies. This process has been facilitated by advances in genomic analysis, that enable the measuring of genetic variation at a greater depth in a greater number of individuals and has led to a boom in genetic information. This has given us a greater understanding of the genetic aetiology of psychiatric disorders and how they are biologically related to each other. How this information can be translated to the clinics, can now be considered. Genetic testing in psychiatric disorders, is currently possible for certain disorders and mutation types, but is not universally advised. Much still remains to be understood about population level genetic risk factors before they could conceivably be utilised in the clinic. Whereas genetic testing of high impact mutations could be of use to the clinical programs, and are actively tested for in clinics across Europe.

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Keywords: psychiatry; genomics; genetics

W0016

How genetics can help diagnosis and treatment in psychiatric conditions

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The understanding of the genetic architecture of psychiatric disorders has made significant advances in the last decade and some scientific findings can now be translated into clinical practice. The rise of genetic testing and the awareness of patients and their families motivate psychiatrists to examine this approach. The COST Action EnGagE (CA17130) is promoting these developments in Europe. Whereas the findings of common variants are the domain of research, screening for rare variants at the genome-wide level is already applicable in clinical practice. It is now possible to return meaningful results to the individual to help him/her understanding the disease and the comorbidities, to guide treatment and to perform genetic counseling. In this presentation, we will give meaningful examples for psychiatric practice. For instance, around one-third of the patients diagnosed with autism spectrum disorder can benefit from a molecular diagnosis (fragile X syndrome, SHANK3 deletion…). Microdeletion or microduplication may explain a fraction of schizophrenic cases (e.g. del22q11). Identification of rare variants causing the disease may decrease the stigma and feeling of guilt often reported by patients and families. This could also help to detect and manage other comorbidities. It is expected that treatment guidelines and clinical trials would be developed in the near future for patients carrying a rare variant, opening the way to personalized psychiatry. Finally, this effort has a huge impact on the family, by enhancing genetic counseling in psychiatry. The rise of psychiatric genetics might align our field more closely with the other medical specialties.

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W0017

Essential information on genetic testing methods that each clinician needs to know/understand

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Genetic testing is well established in many areas of clinical medicine, is increasingly used in clinical psychiatry and it becomes increasingly important to understand the scope and limitations of the different genetic tests applied. The recommended genetic work-up of patients with neurodevelopmental disorders (such as intellectual disability or autism spectrum disorders) includes conventional karyotyping (low resolution) able to detect chromosomal rearrangement and structural variants (>5Mb, 5 million-bp), testing for fragile X-Syndrome, screening for deletions and duplications down to 20 Kb by Comparative Genomic Hybridisation (CGH), able to detect Copy Number Variation (CNVs; gain or loss of genetic material compared to the reference genome). Sanger sequencing is used for mapping of single base pair genetic variants in single genes but unable to identify deletions or duplications. The more advanced Next Generation Sequencing (NGS) have enabled to detect variants in panels of 10-100 (or more) genes, or in all coding regions using Whole Exome Sequencing (WES; 23.000 genes). Whole Genome Sequencing (WGS) analysis enables also