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.

Disclosure: No significant relationships.
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.

Disclosure: No significant relationships.
Keywords: molecular diagnosis; rare diseases; genetics

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

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the detection of all size range and types of genetic variation including CNVs, trinucleotide repeats and translocations. All this led to an impressive change in interpreting genomic variants that need to be strictly linked to clinical information before it can be used by clinicians to improve diagnosis or care. Bioinformatic tools to annotate variants, predict their effects and select the genes and genomic regions of interest are needed to guide the clinical work followed with careful evaluation of the prioritized variants based on the clinical knowledge (https://www.cost.eu/actions/CA17130/#tabs|Name:overview).

Disclosure: No significant relationships.
Keywords: exome/genome analysis; Copy Number Variation (CNV); genetic testing; testing methods

W0018

How to do genetic counseling in psychiatry?

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Genetic counselling has been defined as the process of helping people "understand and adapt to medical, psychosocial, and familial aspects of genetic conditions." It can also help patients and families deal with stigma and understand the significance of possible genetic findings. Psychiatric genetic counselling (PGC) is an emerging field aimed to help people with a personal or family history of psychiatric illnesses such as schizophrenia, bipolar disorder, or neuropsychiatric conditions, to understand genetic etiological mechanisms as a critical component. Counselling strategies are used to identify and adapt to psychological and familial consequences of the conditions and to reduce stigma surrounding the psychiatric illness. A recent survey showed that PGC is still not routinely offered and usually only discussed at the initiative of the patient, e.g. if they ask about the possibility of "hereditary" illness, or if a caregiver during a session for another indication, identifies the family history. If a monogenetic or chromosomal cause is identified, the genetic counselling follows a more traditional path, but if, on the other hand, the cause is complex, the counselling will not be as clearcut. It will then focus on explaining risk for disease with quite uncertain riskscores as no causative genetic change is identified. Although genetic testing most often cannot be offered and individual risk scores based on genetic markers cannot be given, there is still great value for patients and their relatives in PGC. Studies have shown that the effect of PGC is an increase of empowerment and a reduction of stigma.

Disclosure: No significant relationships.
Keywords: Genetic; Counselling; schizophrénia; bipolar

Clinical/Therapeutic

Recently proposed trans-diagnostic criteria for apathy: Commonalities and differences with the avolition/apathy domain of schizophrenia

W0019

Apathy in schizophrenia: assessment in clinical settings and overlap with other dimensions of impairment

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Negative symptoms are considered a core feature of schizophrenia. They are present since the prodromal phase and tend to persist more than other psychopathological dimensions in the chronic stages. The domain of apathy has attracted research efforts for the strong association with poor functional outcome. This negative symptom domain is observed in a number of neuropsychiatric disorders and might have both overlapping and distinct pathophysiological mechanisms. In schizophrenia it can be secondary to other aspects of the disorder, such as positive symptoms and depression, to drug side effects and/or social isolation, often observed in affected subjects. When primary to schizophrenia, apathy is conceptualized in terms of a reduction of the voluntary activity due to a lack of interest and motivation for goal-directed behavior initiation and persistence. In a percentage of subjects, apathy tend to persist and do not respond to available pharmacological and psychosocial treatments. The assessment of this domain in patients with schizophrenia using internationally recognized criteria for its definition, as were recently developed in other neuropsychiatric disorders, might help disentangle the different pathophysiological mechanisms. In the presentation, studies of apathy in schizophrenia will be illustrated to highlight the relationships with cognitive dysfunction, other psychopathological dimensions and functional outcome using state of the art instruments to assess the construct in schizophrenia.

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Keywords: Avolition; negative symptoms; apathy; Primary negative symptoms

W0021

Is apathy a true trans-diagnostic construct? preliminary findings of the european study on apathy in schizophrenia

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Apathy is a quantitive reduction of goal-directed activity either in behavioural, cognitive, emotional or social dimension in comparison to the person's previous level of functioning in these areas.

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