

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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doi: 10.1192/j.eurpsy.2021.158

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; schizophrenia; 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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doi: 10.1192/j.eurpsy.2021.159

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.

Disclosure: Prof. Mucci has been a consultant and/or advisor to or has received honoraria from Gedeon Richter Bulgaria, Janssen Pharmaceuticals, Lundbeck, Otsuka, Pfizer and Pierre Fabre.

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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doi: 10.1192/j.eurpsy.2021.160

Apathy is a quantitative 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.