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## S19.04

Psychiatric epistemology in Spain: ideas and models

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Keeping in mind the constrictions and limitations that have marked the development of Spanish psychiatry during a good part of the 20th century, it is not surprising that its contributions to the epistemological and methodological foundations of psychopathological knowledge have been relatively scarce if compared with other countries and national traditions. Nevertheless, the writings of some outstanding authors include valuable reflections and theoretical insights that go beyond the mere reception of foreign ideas. Apart from the intense concern for anthropological questions or for the problems of existential analysis which were so typical during the central decades of the century, there have been a series of notable contributions related to the concept of understanding in psychiatry, to the development of psychopathology as an objective hermeneutics, to the historical and empirical calibration of the mental symptom and to the theoretical implications stemming from the nature of the psychiatric experience which will be presented and reassessed in the course of this presentation.

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## S20. Symposium: THE PHENOTYPIC SPECTRUM OF AUTISM CHALLENGED BY GENETIC STUDIES

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### S20.01

Autism: a molecular plasticity disorder

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**Background and Aims:** Autism (MIM#209850) is a complex neurodevelopmental affection that is largely genetic psychiatric disorder. Several genes have been found associated with autism but their expression levels and neuropathological effects remain unknown in autistic brain.

**Methods:** We compare the level of expression of autism candidate genes in post-mortem brain region samples between controls and patients. We studied Brodmann area (BA) 46 and the granule cells of the cerebellum lobule 6, for which neuropathological findings and functional abnormalities have been reported in autism.

**Results:** Different levels of transcription for SLC25A12/AGC1, EN2 and Nr-CAM genes are observed in the cortex and granule cells. Difference of expression are observed between patients and controls. We focused on SLC25A12 for which polymorphisms have been associated to autism in various studies. SLC25A12 encodes the mitochondrial aspartate/glutamate carrier and its function is requested to produce energy in neurons. By hybridation in situ, we analysed the expression pattern of SLC25A12 in human development and we studied the effects of SLC25A12 over-expression on mouse embryonic cortical neurons.

**Conclusions:** Convergent evidence suggest that level of expression of candidate genes may be involved in autism pathophysiology by modifying neuronal networks and molecular plasticity in specific brain subregions at both pre- and postnatal stages.

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### S20.02

From mental retardation to autism: common aspects, common genes

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**Background and Aims:** Autism and mental retardation (MR) represent an etiologic enigma for clinicians and scientists. It is however considered that these conditions are closely related and are also associated with genetic alterations. The aim of this presentation is to provide an update of findings indicating that MR and autism share some common genetic causes, and to address questions of the cognitive functions involved in these brain disorders.

**Methods:** Various genetic strategies have shown that autism and MR are associated with identical mutations, raising the hypothesis of common genetic causes. Particularly, the characterization of chromosomal abnormalities has led to define some genomic territories encompassing candidate genes. Furthermore, the study of individuals or families with X-linked MR indicated a significant number of patients with both MR and autism.

**Results:** Interestingly, many genes involved in autism and MR disorders encode proteins of the postsynaptic density proteome network. Mouse genomic studies have shown specific cognitive abnormalities indicating that the postsynaptic proteome seems to be crucial for the establishment and/or maintenance of the normal cognitive function.

**Conclusions:** A close relationship exists between MR and autism since 75% of people with autism suffer from MR of varying degree, and 20-30% of people with severe MR exhibit some autistic features. Accumulating data also provides evidence that similar neurobiological pathways would affect both MR and autism. The study of syndromic forms of autism associated with MR should provide a powerful basis for the identification and the understanding of the pathophysiological pathways underlying these two conditions.

### S20.03

Do autism and ocd have shared genetic vulnerability?

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Obsessive compulsive disorder (OCD) is observed at increased rates in first-degree relatives of probands with autism spectrum disorders (ASDs). In addition, OCD-like traits are observed in autism, and in Asperger syndrome. Furthermore, subjects with OCD may have traits that overlap with some aspects of higher functioning ASDs. These observations suggest that OCD and ASDs may share some genetic risk factors. In support of this, it has recently been suggested that both common and rare functional variants in the serotonin transporter (SLC6A4) may increase risk for OCD and/or ASD. We will review our large-scale analysis of common and rare functional variants SLC6A4 in ASDs and relate these results to studies of OCD. In parallel studies, we have carried our linkage analysis in families with ASDs, focusing on those with more severe OCD-like traits. These families demonstrated increased

evidence for linkage to chromosomes 1, 6, and 19. Evidence for linkage to chromosomes 6 and 19 have been observed in other studies, which we will summarize. Finally, we have examined the evidence that common variants in the NRCAM, TPH1 and TPH2 genes are associated with ASDs, particularly in patients with more severe OCD-like traits, and these results will be summarized.

### S20.04

Do ADHD and autism have related traits considering the genes involved?

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ADHD and autism spectrum disorders (ASD) are neurodevelopmental disorders with a complex presentation and etiology. They are both considered to be heritable conditions with a widely accepted genetic component to the etiology and efforts to find susceptibility genes underlying these disorders are gradually yielding interesting findings (Faraone, 2006, Grice, 2006). The challenge for genetics studies for the individual disorders will be elucidation of the relationships between susceptibility genes and the complex phenotype.

Given the genetic underpinnings of both disorders it is not unreasonable to ask the question as to whether there is shared genetic vulnerability. Genetic overlap is not widely reported in the literature and there is a relative dearth of direct investigation of this question. Evidence suggests a role for genes involved in the dopaminergic and serotonergic systems in both ADHD and autism but it is not clear if genetic variation in genes involved in these systems is similar. Further investigation is warranted to directly investigate potential overlap and subsequently to address the complex task of understanding the relationship between genetic susceptibility, possible common endophenotypes and the clinical phenotype. It is likely that multiple mechanisms, both genetic and environmental may be at play.

A discussion of the existing literature with respect a putative overlap in genetic vulnerability and the clinical phenotype is presented. Furthermore the specific challenges for research in assessing these factors in future studies is discussed.

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## S21. Symposium: SPECIFIC ASPECTS OF SUBSTANCE USE DISORDERS AMONG MIGRANTS (Organised by AEP Sections on Cultural Psychiatry and Alcoholism and Addiction)

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### S21.01

Concepts of illness among addicted migrants

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Cultural and social barriers often prevent migrants in Germany from frequenting centers for information, counseling and treatment of psychiatric disorders. To verify existing hypothesis and to discover further reasons for the low accessibility rate of health care system in a first step

we conducted 16 qualitative interviews with professionals of the drug treatment facilities in Germany, 15 with opiate dependent Turkish patients, 3 with alcohol addicted Germans and 3 with alcohol addicted ethnic Germans from the former Soviet Union. Also we examined cultural differences in the explanatory models of psychiatric disorders including addictive behavior among Turkish and German adolescents as well as among ethnic German adolescents who migrated to Germany from the former Soviet Union. The statistical device ANTHROPAC was applied to map the semantic space of concepts associated with problems of addiction and psychiatric disorders.

Relevant barriers which prevent migrants from frequenting drug treatment facilities were found out. Frustration, missing of integration and loss of perspectives, which have a big influence on the maintaining of addiction in general.

Ethnic German migrants from the former Soviet Union and native German youths used addiction concepts in a rather similar way.

Preventive information programs may profit from considering these differences and need to use concepts that are accepted and clearly associated with psychiatric disorders by immigrant populations.

### S21.02

Addiction among Russian and Turkish migrants in Germany: developing prevention strategies

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Although about 20% of German population are migrants not enough knowledge exists about health status, alcohol and tobacco addiction and preventive health behaviors with respect to addiction in migrants. The largest populations of migrants in Germany are Turks and Russians. The goal of the project presented in this paper is to investigate the relationship between health status, alcohol and tobacco addiction and preventive health behaviors of Russian and Turkish speaking migrants in Hannover, Germany. Additionally, the project deals with the question of possible barriers migrants experience on the way to the health service system in Germany. For example, cultural differences in understanding of health and disease concepts, language difficulties, lack of knowledge about possible preventive measures and lack of information in mother tongue could be possible barriers that make the access to a healthy way of living more difficult for migrants. To investigate possible barriers migrants experience we apply the mother tongue mediator approach that allows better access to the community of migrants, helps to overcome cultural differences in understanding of health related concepts, and makes the information on preventive behaviors in community of migrants more acceptable. Results of a pilot study in Russian speaking migrants (N=18) are in line with hypotheses. 28.6% of the sample report alcohol addiction, high level of psychological distress and depression, low level of perceived social support and low level of physical health. Language difficulties and lack of bilingual information on health topics are most frequent barriers experienced in this sample.

### S21.03

Substance use and schizophrenia among south Asian migrants to the UK

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