SES17. AEP Section "Mental Retardation": Behavioural phenotypes

Chairs: W.M.A. Verhoeven (NL), S. Tuinier (NL)

SES17.01

PSYCHOPATHOLOGICAL PHENOTYPES

W.M.A. Verhoeven[1,2]*, S. Tuinier[1], L.M.G. Curfs[3]. [1]*Vincent van Gogh Institute for Psychiatry, Venray;* [2]*Erasmus University, Rotterdam;* [3]*Clinical Genetics Centre, Maastricht, PO Box 5, 5800 AA Venray, Netherlands*

Several syndromes with a specific genetic etiology have been reported to be associated with a more or less typical profile of behavioural abnormalities and meet therefore the definition of a behavioural phenotype. Some syndromes, however, are frequently accompanied by psychiatric symptoms, that should be considered as a psychopathological phenotype.

With respect to psychiatric symptomatology, two syndromes are of special interest, Prader-Willi syndrome (PWS) and Velo-Cardio-Facial Syndrome of Shprintzen (VCFS). PWS is after adolescence associated with relapsing psychotic episodes characterized by emotional turmoil, anxiety, confusion, rapid mood swings, hallucinatory experiences, paranoid ideation and increase of obsessive rituals as well as subacute onset, short duration and full recovery upon treatment with mood stabilizing agents. The psychopathological phenotype of VCFS comprises thoughts of reference, paranoid ideation, anxieties, emotional and affective instability, hallucinatory experiences and psychomotor agitation.

Thus, both syndromes are associated with a relapsing psychotic condition with a variable expression, emanating in the context of a known genetic disorder and probably anteceded by a specific psychological and behavioural profile. Although PWS psychoses meet the criteria for cycloid psychosis, it is advocated to advance the PWS-psychiatric syndrome and the VCFS-psychiatric syndrome as separate diagnostic entities

(1) Verhoeven, W.M.A., Tuinier, S., Curfs, L.M.G. (2000). Prader-Willi psychiatric syndrome and Velo-Cardio-Facial Syndrome. Genetic Counseling, in press.

SES17.02

BEHAVIOURAL PHENOTYPES IN DIFFERENT FORMS OF X-LINKED MENTAL RETARDATION

J.P. Fryns[1]*, M. Borghgraef[1], S. Frints[1], J. Steyaert[1,2], L.M.G. Curfs[2]. [1]*Center for Human Genetics, University Hospital of Leuven, B-3000 Leuven, Belgium*
[2] *Clinical Genetics Center, University of Maastricht, Maastricht, The Netherlands*

X-linked mental retardation (XLMR) is the most frequent genetic cause of mental retardation with an incidence of 1 in 1.000 life births. After the clinical delineation of the fragile X syndrome and the discovery of the Fragile X mental retardation gene (FMR-1), the scientific interest in XLMR has increased significantly in the last 10 years. This has resulted in the clinical delineation and molecular characterization of more than 200 other X-linked mental retardation conditions.

In a minority of these, the mental retardation is associated with distinct clinical signs and these XLMR forms are designated as MRXS (syndromic forms of XLMR). In the majority of these conditions, clinical findings are non-specific (MRX - non-specific forms of XLMR).

In young fragile X children the recognition of their distinct behavioural phenotype is an important part in the clinical diagnosis.

At the present time, it becomes evident that also in MRXS (ATRX syndrome - Coffin-Lowry syndrome - MASA syndrome - XLMR with Marfanoid habitus) the behavioural phenotype may be distinct, and associated with specific neurological symptoms (e.g. dystonia, dysarthria).

We present also data on the new MRX genes and the cognitive and behavioural findings in affected individuals.

SES17.03

BEHAVIOURAL PHENOTYPES IN CLINICAL PRACTICE

L.M.G. Curfs[1]*, W.M.A. Verhoeven[2,3], S. Tuinier[2], J.P. Fryns[4]. [1]*Clinical Genetics Center, Maastricht;* [2] *Vincent van Gogh Institute for Psychiatry, Venray;* [3]*Erasmus University, Rotterdam, The Netherlands*
[4]*Center for Human Genetics, Leuven, Belgium*

Behavioural phenotypes are recognizable characteristic patterns of behaviour associated with genetically determined disorders. Great advances have been made in our understanding of these underlying cognitive and behavioural profiles of a number of different syndromes. This presention will provide an overview of review findings on the topic of behavioural phenotypes and addresses distinctive behaviour characteristics for the demarcation of some of the genetically determined syndromes associated with mental retardation.

SES17.04

THE BEHAVIOURAL PHENOTYPES OF GENETIC SYNDROMES

A.J. Holland

No abstract was available at the time of printing.

S42. Prediction of course and outcome in psychiatry

Chairs: C. Höschl (CZ), W. Gaebel (D)

S42.01

PRODROMAL SYMPTOMS IN SCHIZOPHRENIA: STATE OF THE ART

W. Gaebel*, M. Jänner, N. Frommann. *Department of Psychiatry, Heinrich-Heine-University Düsseldorf, Germany*

The vulnerability-stress-coping-model is the most influential heuristic concept in understanding the course of schizophrenia, whose prodromal status still offers unsolved conceptual and methodological issues. Improved knowledge about the prodromal phase could provide a better understanding of the developing psychopathology and psychophysiology of schizophrenia and could also be of predictive value in order to attune therapeutic actions to the illness course more precisely. To shed more light on the characteristics of prodromal states, data of a German multicenter study on intermittent vs maintenance neuroleptic long-term treatment in schizophrenia (ANI-study) were reanalysed with respect to prevalence and profile, nature, time course and predictive value of

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