P-701 - EXECUTIVE DYSFUNCTIONS AS PART OF THE BEHAVIOURAL PHENOTYPE OF AARSKOG-SCOTT SYNDROME

J.I.M.Egger[1,2,3], W.M.A.Verhoeven[1,4], G.Janssen[1,3], L.Van Aken[1], A.J.M.Hoogeboom[5]

[1]Centre of Excellence for Neuropsychiatry, Vincent van Gogh Institute for Psychiatry, Venray, [2]Donders Centre for Brain, Cognition and Behaviour, [3]Behavioural Science Institute, Radboud University Nijmegen, Nijmegen, [4]Department of Psychiatry, [5] Department of Clinical Genetics, Erasmus University Medical Centre, Rotterdam, The Netherlands

Introduction: Aarskog syndrome (AAS) also called Aarskog-Scott syndrome faciodigitogenital syndrome or faciogenital dysplasia is a genetically heterogeneous developmental disorder, first described in 1970 by the Norwegian pediatrician Dagfin Aarskog and further delineated by Scott in 1971. It is a predominantly X-linked disorder, phenotypically characterized by short stature, craniofacial dysmorphisms, brachydactyly and urogenital abnormalities. The level of intelligence shows a great variability and no specific behavioural phenotype has been described so far. In about 20 percent of Aarskog families, a mutation in the *FGD1* gene located in Xp11.21 can be identified.
Objectives: The delineation of the potential behavioural phenotype of AAS.
Aims: Neuropsychological and neuropsychiatric investigation of four males from one kindred.
Methods: Four affected affected males from the fourth generation of a previously published large Dutch family (Van de Vooren et al., 1983) are assessed in detail by means of an extensive neuropsychological battery and semi-structured psychiatric examination. In addition, mutation analysis was performed.
Results: A novel *FGD1* missense mutation (R402W) at position 1204 (1204C>T) was demonstrated. In the patients, the level of intelligence varied between normal and severely disabled. Their behavioural profile showed, among others, elements of attention deficit hyperactivity disorder, primarily reflected by impaired executive attentional processes that may be sensitive to systematic training.
Conclusions: In AAS, dysfunctional executive cognitive processes can be considered as part of the behavioural phenotype of the syndrome. Cognitive training and structuring of daily life may therefore reduce the intensity of disinhibited behaviours that are reported in AAS patients.

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