W01-04 - PSYCHOPATHOLOGY IN RARE GENETIC DISORDERS

V.M.A.Verhoeven[1,2], J.I.M.Egger[1,3,4], H.G.Brunner[5]

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

Introduction: Deinstitutionalization of psychiatric patients and people with intellectual disabilities has markedly increased the number of referrals of patients with behavioural disturbances and/or psychiatric to outpatient psychiatrist, frequently because of 'treatment resistant' psychiatric syndromes. Given the rapidly growing knowledge of clinical and molecular genetics, co-occurrence of psychological and behavioural dysfunctions with a chromosomal anomaly in an individual patient greatly increases the possibilities for an etiological explanation of the clinical picture, which in turn may serve as guidance for a specific treatment.
Objectives: To elucidate the phenotype-genotype relation in patients with unexplained behavioural problems.
Aims: To advocate the clinical awareness of so called behavioural phenotype.
Methods: Detailed presentation of some cases (diagnosis and treatment) that are referred to the outpatient department for neuropsychiatry and who are published by the research group 'Psychopathology and Genetics'.
Results: Female (25yrs;IQ:50), reason of referral: psychotic disorder. D/ anxieties due to overestimation; de novo duplication 13(q14.1q21.3). Female (30yrs;IQ:< 60) referred for psychotic and autistic symptoms. D/ cognitive alexithymia; de novo r21. Female (17yrs;IQ:81), referred for difficulties in school performance. D/ panic disorder; distal 22q11 microdeletion. Male (57yrs;IQ:128), reason of referral: treatment resistant depression. D/ no psychopathology; Robertsonian translocation 13;14. Female (57yrs;IQ< 60), referred presenile dementia. D/ mucopolysaccharidosis SanfilippoB. Male (21yrs;IQ:52). Referred for mood instability. History: XXY. D/ atypical bipolar disorder; PWS + XXY.
Conclusions: In patients referred for recurrent challenging behaviours and/or psychiatric symptoms the search for a genetic etiology is mandatory in order to avoid erratic treatment advises.

https://doi.org/10.1016/S0924-9338(12)75671-7 Published online by Cambridge University Press