W01-03 - NEUROPSYCHOLOGICAL PHENOTYPING OF GENETIC SYNDROMES

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Introduction: Over the past years several novel microdeletion syndromes have been reported that may be associated with a specific pattern of psychological dysfunctions. Two major examples are the Kleefstra syndrome (KS) and the 17q21.31 microdeletion syndrome (17qDS). KS was originally described as the 9q subtelomeric deletion syndrome associated with marked mental retardation, specific dysmorphisms, particular sleep disturbances, and progressive deterioration and apathy, suggestive for a 'neurodegenerative phenotype'. Haploinsufficiency of the EHMT1 gene is proven to be the causative factor. 17qDS comprises moderate mental retardation, dysmorphisms and various congenital anomalies. It has been suggested that the behavioural phenotype includes remarkably friendly manners. The syndrome is thought to be caused by MAPT haploinsufficiency.
Objectives: Neuropsychological phenotyping of KS and 17qDS.
Aims: Assessment of neuropsychological functions of patients with either KS or 17qDS.
Methods: Five patients with KS and three patients with 17qDS were extensively investigated by means of broad band neurocognitive instruments as well as specific rating scales.
Results: The behavioural phenotype of KS was characterized by behavioural and motor deficiencies that become apparent at adolescence and increase over time. Motor symptoms could be attributed to a progressive apathy syndrome. In 17qDS the neuropsychological phenotype comprised, as compared to a matched control group, less social fear and more approaching behaviour.
Conclusions: In both syndromes, neuropsychological examination demonstrates specific patterns of (cognitive) development and social interaction. These findings further clarify the genotype-phenotype relation and are of importance for the establishment of an individualized behavioural and clinical management program.

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