

P-700 - SOCIAL COGNITION AND THE BEHAVIOURAL PHENOTYPE OF 17Q21.31 MICRODELETION SYNDROME

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Introduction: Recently, the 17q21.31 microdeletion syndrome was described with characteristic features including developmental delay, moderate intellectual disability, facial dysmorphisms, and anomalies of the brain and multiple organ systems. With respect to behaviour, scarce data from clinical observations have suggested a remarkably amiable, friendly disposition, to some extent comparable to that observed in Williams syndrome.

Objectives: Delineation of the behavioural phenotype in 17q21.31 microdeletion syndrome.

Aim: Investigating the various aspects of neurocognitive functioning in 17q21.31 microdeletion syndrome and social cognition in particular.

Methods: Neuropsychological assessment was performed in three out of the four known Dutch patients with a proven 17q21.31 microdeletion syndrome as well as in three control subjects with intellectual disability. Apart from intelligence, attention, memory and executive functioning, social-emotional functioning was extensively assessed.

Results: The general cognitive profile of all patients and controls was in accordance with their lowered intellectual capacities, albeit that in the patients a relatively strong memory for social-contextual information was established. Basic emotion perception was intact in both patients and controls; but patients showed less social fear and more approaching behaviour. Interestingly, alexithymic traits, being marked difficulties in the recognition and expression of emotions, were more prevalent in control subjects.

Conclusion: Hypersocial behaviour with a high level of frustration tolerance was demonstrated with a hypothetico-deductive assessment strategy in the three patients with 17q21.31 microdeletion syndrome, and may be part of its core phenotype.