AUTISM SYNDROMES IN THREE BEHAVIOURAL PHENOTYPE CONDITIONS

A clinical psychiatric study of 76 individuals with Möbius sequence, CHARGE syndrome, and oculo-auriculo-vertebral spectrum

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II. Johansson M, Råstam M, Billstedt E, Danielsson S, Strömland K, Miller M, Gillberg C. (2006) Autism spectrum disorders and underlying brain pathology in CHARGE association. Developmental Medicine and Child Neurology 48: 40-50.

III. Johansson M, Billstedt E, Susanna D, Strömland K, Miller M, Granström G, Flodmark O, Råstam M, Gillberg C. (2007) Autism spectrum disorders and underlying brain mechanisms in the oculoauriculovertebral spectrum. Developmental Medicine and Child Neurology 40: 280-288.

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A clinical psychiatric study of 76 individuals with Möbius sequence, CHARGE syndrome, and oculoauriculovertebral spectrum

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Abstract

Objectives: (1) Examine the prevalence of autism syndromes in three different Behavioural Phenotype Conditions (BPCs), (2) examine background and associated factors/conditions, and (3) describe and evaluate diagnostic difficulties in this field of research. **Method**: As part of multidisciplinary surveys of Möbius sequence (Möbius) (n=25), CHARGE syndrome (CHARGE) (n=31) and oculoauriculovertebral spectrum (OAV) (n=20), the occurrence of autism symptoms was assessed utilizing the DSM-III-R and DSM-IV checklists for autistic disorder (AD), the Autism Diagnostic Interview-Revised (ADI-R), the Childhood Autism Rating Scale (CARS) and the Autistic Behavior Checklist (ABC). Mental level was evaluated using standardized IQ tests or the Vineland Adaptive Behavior Scales. Results from previously performed radiological imaging/laboratory tests, and data on pre/perinatal nonoptimal conditions and family factors were scrutinized. The applicability of the autism diagnostic instruments used in individuals with multiple disabilities (such as in these BPCs) was analysed. **Results**: There was a high rate of autism syndromes (Möbius 48%, CHARGE 68%, OAV 42%) across all BPCs. Severe behaviour disturbances with major impact on family life were common in the individuals with autism syndromes, especially in the CHARGE group. Learning disability (LD) was a common finding (Möbius 32%, CHARGE 72%, OAV 25%), possibly reflecting the link between autism syndromes and LD. Visual and/or hearing impairments affected only a few subjects with Möbius, but were very common and associated with autism syndromes in the CHARGE/OAV groups. Cerebral abnormalities were recorded in one fifth of radiologically examined individuals with Möbius, 74% with CHARGE, and 63% with OAV. Autism syndromes, LD and cerebral abnormalities tended to occur together in the same individuals. Cranial nerve dysfunction was present in all Möbius individuals, in 55% of the CHARGE, and in 60% of the OAV group. Pre-, perinatal and/or family factors of possible interest were recorded in several individuals in each BPC. The diagnostics of autism syndromes in these BPCs presented difficulties, notably due to sensory impairments, cranial nerve palsies and LD. The diagnostic difficulties increased with the number and severity of disabilities. **Discussion and conclusions:** This study suggests that autism syndromes always should be considered in subjects with Möbius/CHARGE/OAV. In the CHARGE/OAV groups, cerebral abnormalities occurred frequently in subjects with autism syndromes, indicating that autism symptoms were not only attributable to sensory impairments. The frequent occurrence of cerebral abnormalities in those with autism syndromes, together with the fact that the majority of those with LD in all the three BPCs had an autism syndrome, could be suggestive of a specific link between autism syndromes and Möbius/CHARGE/OAV. The associated overall clinical findings, including the frequent occurrence of cranial nerve dysfunction found in all three BPCs, implicate the early embryonic brain, including the brain stem, as a possible area of core dysfunction. The use of an extensive battery of autism diagnostic instruments is essential in individuals with multiple disabilities. Current autism diagnostic instruments are insufficiently tailored to deaf-blind individuals.

Key words: autism syndromes, behavioural phenotype conditions, Möbius, CHARGE, OAV, ADI-R, CARS, ABC, brain stem

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