22q11 Deletion Syndrome
Neuropsychological and Neuropsychiatric Correlates
A Clinical Study of 100 Cases

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av
Lena Niklasson
Leg. psykolog

Fakultetsopponent:
Professor Ann Swillen
Centre for Human Genetics, University Hospital Gasthuisberg
Leuven, Belgien

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I. Niklasson, L., Rasmussen, P., Óskarsdóttir, S., Gillberg, C. (2002). ”Chromosome 22q11 deletion syndrome (CATCH 22): neuropsychiatric and neuropsychological aspects”. *Developmental Medicine and Child Neurology*, 44:44-50.

II. Niklasson L., Rasmussen P., Óskarsdóttir S., Gillberg C. (2007). “Autism, AD/HD, learning disability, and behaviour problems in 100 individuals with 22q11 deletion syndrome”*(submitted).*

III. Niklasson, L., Rasmussen P., Óskarsdóttir, S., Gillberg, C. (2005). ”Attention deficits in children with 22q11 Deletion Syndrome”. *Developmental Medicine and Child Neurology*, 47:803-807.

IV. Niklasson L. & Gillberg C. (2007). “The neuropsychology of 22q11 deletion syndrome. A clinical study of 100 individuals”*(submitted).*

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GÖTEBORG UNIVERSITY

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Lena Niklasson
Institute of Neuroscience and Physiology, Child and Adolescent Psychiatry, Göteborg University, Sweden

Abstract

Objectives: Examine the prevalence and type of Autism Spectrum Disorder (ASD) and Attention-Deficit/Hyperactivity-Disorder (AD/HD), Learning Disability (LD), behavioural profile, intellectual ability/profile, and executive function in 22q11 Deletion Syndrome (22q11DS) and to study the impact of AD/HD and ASD on these functions.

Methods: One hundred individuals (58 female, 42 male; 1-35 years of age) with 22q11DS, confirmed by FISH analysis, were included. They were the first 100 referred 22q11DS cases, of whom 92 came from a multidisciplinary team as part of routine 22q11DS assessments, and 8 were referred directly to a Child Neuropsychiatry Clinic for learning and/or behaviour problems. Neuropsychological evaluation made use of a test battery designed to provide information concerning developmental/intellectual level and profile, visuomotor development, executive functions (planning ability and attention), and mentalisation skills. Neuropsychiatric assessments included structured and semistructured interviews with parent(s), an evaluation of the individual including psychiatric assessment, physical examination, and age-appropriate neurological examination. Parents completed the Autism Spectrum Screening Questionnaire, the Conners Brief Parent Rating Scale, the Child Behavior Checklist, and the Five To Fifteen (FTF) questionnaire. Comprehensive diagnoses of ASD and AD/HD were made by a psychiatrist according to the DSM-IV taking the results of the various examinations (interview, medical examinations, observation, and the FTF questionnaire) into account.

Results: The prevalence of ASD and/or AD/HD with or without LD was 44%, of whom 21% had AD/HD "only", 14% ASD "only", and 9% a combination of these two diagnoses. In addition, 23% had LD "only", meaning that there were 33% without any of these diagnoses. Autistic disorder was found to be quite rare (5%). Other psychiatric diagnoses were found mainly among the adults. Altogether 51% met criteria for LD, and the mean IQ was 71 with a normal distribution around this mean. Higher IQ for females compared to males and a negative trend for IQ with increasing age were found. An overrepresentation of girls was found only in the group without ASD/AD/HD/LD. In the school age group and in the adult group significantly higher verbal IQ than performance IQ was found. In contrast, in the youngest group the lowest result was found in the "Hearing and Speech" subscale (Griffiths´ Mental scale) reflecting a delay in expressive language in the early years. The strength within the verbal area was mainly due to good Vocabulary. Deficits in performance ability were found. The intellectual and the visuomotor impairments were related to 22q11DS per se while the presence of ASD/AD/HD had a negative impact on planning ability in children. The ability to sustain attention was found to be critically impaired in school age children with 22q11DS. According to results of the questionnaires a variety of behaviour problems were reported. A characteristic combination of initiating difficulties and a "lack of mental energy" was observed in the majority.

Discussion and conclusions: The vast majority of all with 22q11DS had behaviour and/or learning problems and more than 40% met criteria for either ASD, AD/HD or both (even though typical autistic disorder was rather uncommon). Half the group had LD. The majority of the group with IQ in the normal to low normal range had learning difficulties. Many individuals with 22q11DS had social interaction difficulties that, in the presence of relatively good word skills, appeared to be related to initiation problems and language use deficits. Given the high rate and variety of problems found, a neuropsychiatric assessment, including neuropsychological testing, should be performed in all cases of 22q11DS. Such assessment will provide essential information about strengths and difficulties, crucial for providing optimal support for individuals with 22q11DS.

Key words: 22q11 Deletion Syndrome, Autism Spectrum Disorder, Attention-Deficit/Hyperactivity Disorder, Learning Disability, neuropsychology

Correspondence: lena.niklasson@vgregion.se

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