



ELSEVIER

Contents lists available at ScienceDirect

Research in Developmental Disabilities



Autism, ADHD, mental retardation and behavior problems in 100 individuals with 22q11 deletion syndrome

Lena Niklasson^{a,b,*}, Peder Rasmussen^{a,b}, Sólveig Óskarsdóttir^{c,d}, Christopher Gillberg^{a,b}

^a Child Neuropsychiatry Clinic, The Queen Silvia Children's Hospital, Göteborg, Sweden

^b Institute of Neuroscience and Physiology, Child and Adolescent Psychiatry, The Sahlgrenska Academy, University of Gothenburg, Göteborg, Sweden

^c Sólveig Óskarsdóttir, the Queen Silvia Children's Hospital, Göteborg, Sweden

^d Department of Pediatrics, Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, Göteborg, Sweden

ARTICLE INFO

Article history:

Received 19 September 2008

Received in revised form 23 October 2008

Accepted 23 October 2008

Keywords:

22q11 deletion syndrome

Autism

ADHD

Mental retardation

ABSTRACT

This study assessed the prevalence and type of associated neuropsychiatric problems in children and adults with 22q11 deletion syndrome. One-hundred consecutively referred individuals with 22q11 deletion syndrome were given in-depth neuropsychiatric assessments and questionnaire screens.

Autism spectrum disorders (ASDs) and/or attention deficit/hyperactivity disorder (ADHD) were diagnosed in 44 cases. ASD was diagnosed in 23 cases of whom only 5 had autistic disorder. ADHD was diagnosed in 30 individuals. In nine of these cases with ASD or ADHD there was a combination of these diagnoses. Mental retardation (MR) with or without ASD/ADHD was diagnosed in 51 individuals. ASD, ADHD, and/or MR were present in 67 cases. Females had higher IQ than males.

The results of this study showed that the vast majority of all individuals with 22q11 deletion syndrome have behavior and/or learning problems and more than 40% meet criteria for either ASD, ADHD or both. Neuropsychiatric and neuropsychological evaluations are indicated as parts of the routine clinical assessment of individuals with 22q11 deletion syndrome.

© 2008 Elsevier Ltd. All rights reserved.

* Corresponding author at: Child Neuropsychiatry Clinic, The Queen Silvia Children's Hospital, Kungsgatan 12, SE-411 19 Göteborg, Sweden. Tel.: +46 313425950; fax: +46 31848932.

E-mail address: lena.niklasson@vgregion.se (L. Niklasson).

1. Introduction

The 22q11 deletion syndrome (22q11DS) is one of the most common genetic deletion syndromes, with a rate of approximately 1 in 4000 live births (Oskarsdottir, Vujic, & Fasth, 2004). The genotype and physical phenotype have been described in many publications (McDonald-McGinn et al., 1999; Oskarsdottir, Persson, Eriksson, & Fasth, 2005b). The inheritance pattern is autosomal dominant, but 85–90% of all cases appear to be caused by de novo deletions (Digilio et al., 2003). The diagnosis can be confirmed by fluorescent in situ hybridization (FISH) technique (Driscoll et al., 1992). The phenotypic spectrum includes abnormalities with variable severity. Common features include cleft palate/velopharyngeal insufficiency, cardiac anomalies, immune deficiency due to hypoplasia of the thymus, and hypoparathyroidism. The syndrome is sometimes referred to as velocardiofacial syndrome or DiGeorge syndrome.

Relatively mild cognitive deficits have been documented in a large proportion of cases. Mean full-scale IQ (FSIQ) is usually in the low borderline range (De Smedt et al., 2007; Moss et al., 1999; Niklasson, Rasmussen, Oskarsdottir, & Gillberg, 2001; Swillen et al., 1997) with a bell-shaped distribution of cases around this mean (Niklasson, Rasmussen, Oskarsdottir, & Gillberg, 2002). Mild mental retardation (MR) is very common, while severe MR is rare.

Attention deficits are common in individuals with 22q11DS with attention deficit/hyperactivity disorder (ADHD) rates of about 40% in several studies (Antshel et al., 2006; Gothelf et al., 2004; Papolos et al., 1996). Anshel et al. found that 43% (36/84 individuals) had ADHD and that in this group the inattentive subtype (25/36 with ADHD) was the most common. In an earlier report (Papolos et al., 1996) ($n = 25$) 36% met DSM-IV criteria for ADHD (about half of whom had the combined subtype and half inattentive subtype). In a previous study from our group ($n = 32$, all included in the present study) we found DSM-IV ADHD (usually inattentive subtype) in 44% (Niklasson et al., 2001). On the basis of these rather smaller studies, ADHD appears to be present in about 40% of all young people with 22q11DS.

Other studies have reported withdrawn behaviors and shyness, difficulties initiating and maintaining social interaction, and limited facial expressions (Gerdes et al., 1999; Niklasson et al., 2002; Swillen et al., 1999). These are traits commonly encountered in individuals with autism spectrum disorders (ASDs). In one study the prevalence of ASD in 22q11DS ($n = 32$) was found to be 31%, but only one person had autistic disorder (Niklasson et al., 2001). In another study, Fine et al. (2005) reported that 11/98 children with 22q11DS assessed with ADI-R (autism diagnostic interview-revised, Lord, Rutter, & Le Couteur, 1994) had autistic disorder. In a recent study of 60 child patients, high rates of ASD (50%) and psychotic symptoms (27%) were found (Vorstman et al., 2006). The authors concluded that autism/psychotic disorders should be considered to be main elements in the behavioral phenotype of children with 22q11DS. In another recent study the phenotype in two groups with 22q11DS, with or without ASD, was studied (Antshel et al., 2007). In the whole group ($n = 41$; age range 6.5 years–15.8 years) 17 individuals (41%) met criteria, based upon ADI-R, for ASD (of whom 8 children (20% of the whole group) met criteria for autistic disorder). In this subgroup with ASD, 94% had a co-occurring psychiatric disorder compared with 60% in the group with 22q11DS “only”. In a further study the behavioral phenotype of autism in samples with either 22q11DS or idiopathic autism were studied (Kates et al., 2007). The diagnoses were based upon the ADI-R. Both groups exhibited less make believe play and more rituals, motor stereotypes and repetitive use of objects compared to children with 22q11DS “only”. However other core autism behaviors, as lack of sharing attention, deficits in gestural communication and initiating conversation, and presence of circumscribed interests, appear to be phenotypic 22q11DS behaviors, characterizing children with 22q11DS regardless of an autism diagnosis. There is, thus, some consensus that ASD are common in children with 22q11DS, but it is unclear as to whether autistic disorder is a common concomitant of the syndrome or not.

The 22q11DS disorder is also associated with non-ADHD/non-ASD psychiatric disorders. In a group of 25 adults, 16 (64%) met DSM-IV criteria for bipolar spectrum disorders (Papolos et al., 1996). In another study ($n = 15$) 30% with 22q11DS had psychotic disorder, of whom 80% met DSM-IV criteria for schizophrenia. In addition, 12% of these adults had depression without psychotic features (Murphy, Jones, & Owen, 1999).

متن کامل مقاله

دریافت فوری ←

ISIArticles

مرجع مقالات تخصصی ایران

- ✓ امکان دانلود نسخه تمام متن مقالات انگلیسی
- ✓ امکان دانلود نسخه ترجمه شده مقالات
- ✓ پذیرش سفارش ترجمه تخصصی
- ✓ امکان جستجو در آرشیو جامعی از صدها موضوع و هزاران مقاله
- ✓ امکان دانلود رایگان ۲ صفحه اول هر مقاله
- ✓ امکان پرداخت اینترنتی با کلیه کارت های عضو شتاب
- ✓ دانلود فوری مقاله پس از پرداخت آنلاین
- ✓ پشتیبانی کامل خرید با بهره مندی از سیستم هوشمند رهگیری سفارشات