



Discordance in diagnoses and treatment of psychiatric disorders in children and adolescents with 22q11.2 deletion syndrome

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ABSTRACT

This study examines the rate of utilization of mental health services in children and adolescents with 22q11.2DS relative to their remarkably high rate of psychiatric disorders and behavior problems. Seventy-two children and adolescents with 22q11.2DS were participants; their parents completed the Diagnostic Interview Schedule for Children (DISC) and the Child Behavior Checklist (CBCL). The results indicated that 22q11.2DS children and adolescents have higher rates of psychopathology than the general pediatric population, with ADHD and anxiety disorders being the most common. However, among youth with 22q11.2DS, those with psychopathology are often no more likely to receive either pharmacological or non-pharmacological mental health care than those without a given psychiatric diagnosis. Thus, although psychopathology is fairly common in this sample, many children with 22q11.2DS may not be receiving needed psychiatric care. These results have significant implications for these children and their families, as well as for the health care providers who treat them. In particular, the results may suggest a need for careful screening of psychiatric disorders that are likely to affect this population, as well as making appropriate treatment recommendations to remedy childhood mental health problems. Since these children face an extraordinarily high risk of psychoses in late adolescence/adulthood, treatment of childhood psychopathology could be crucial in mitigating the risk/consequences of major psychiatric illnesses in later life.

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1. Introduction

Perhaps as evidenced by its many names, 22q11.2 Deletion Syndrome (22q11.2DS; also known as velo-cardio-facial syndrome or DiGeorge syndrome) is a complex condition with multiple phenotypic features. As the most common microdeletion syndrome in humans, it affects 1 in 1600–4000 live births (Driscoll et al., 1992; Wilson et al., 1994; Tezenas Du Montcel et al., 1996; Shprintzen, 2000; Kobrynski and Sullivan, 2007). Common manifestations include heart malformations, palatal abnormalities and typical facial features (Shprintzen et al., 1981; Shprintzen, 2000).

1.1. Cognitive impairments in 22q11.2DS

Cognitive impairments are almost universal in individuals with 22q11.2DS, with a mean IQ of 75 (Swillen et al., 1997; Moss et al.,

1999; Woodin et al., 2001). A complex pattern of impairments occurs, with deficits in sustained attention, working memory, executive function, verbal learning, and visual-spatial processing (Bearden et al., 2001; Niklasson et al., 2001; Campbell et al., 2006; Kiley-Brabeck and Sobin, 2006; Lewandowski et al., 2007).

1.2. Psychiatric/behavior problems in 22q11.2DS

In addition to cognitive deficits, children with 22q11.2DS are highly susceptible to psychiatric problems and disorders such as attention-deficit/hyperactivity disorder (ADHD), obsessive compulsive disorder (OCD), depression, generalized anxiety disorder, separation anxiety, and specific and social phobia (Niklasson et al., 2001; Gothelf et al., 2004; Baker and Skuse, 2005; Antshel et al., 2006; Shashi et al., 2010a,b). High rates of overall behavior problems, elevated internalizing symptoms, and poor social skills are also common (Heineman-de Boer et al., 1999; Kiley-Brabeck and Sobin, 2006). Most remarkably, children with 22q11.2DS are 25 times more likely to experience serious mental illness during late adolescence/early adulthood than the general population (reviewed in Shprintzen, 2008), with up to one-third eventually developing schizophrenia spectrum disorders; also evident with increasing age

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are bipolar disorder and major depression (Shprintzen et al., 1992; Pulver et al., 1994; Papolos et al., 1996; Antshel et al., 2007; Gothelf et al., 2008).

The exact cause of the high rate of psychotic disorders is unclear, although the hemizygous deletion undoubtedly plays a role. It is widely believed that the childhood psychiatric problems may be associated with the later risk of psychosis; thus, early treatment of these may have an effect upon the psychosis risk later in life (Gothelf, 2007; Shprintzen, 2008), underscoring the importance of diagnosing and treating childhood psychopathology.

1.3. The present study

Despite the fact that the condition clearly has an impact upon psychological function and behavior, little effort has been made to design and implement interventions for children with 22q11.2DS (Hatton, 2007). The effectiveness of psychosocial interventions in this group is completely unknown, but there is preliminary evidence that particular pharmacological interventions are effective at treating psychiatric problems in 22q11.2DS. Specifically, metyrosine and clozapine for psychosis resistant to other treatments (Carandang and Scholten, 2007; Gladston and Clarke, 2005), methylphenidate for ADHD (Gothelf et al., 2003), and flouxetine for OCD (Gothelf et al., 2004) may be promising treatment options. It is important to note that these efficacy studies are few and have small sample sizes (ranging from 1 to 12 participants). Furthermore, to date, there are no known studies examining differential utilization of existing interventions or whether service utilization may differ by co-morbid conditions. Based on our clinical observations, we hypothesized that, despite relatively high rates of psychiatric disorders/behavior problems in this population, the reported rate of services being provided would indicate underutilization of mental health services (i.e., the difference in utilization rates between the 22q11.2DS cases with and without a co-morbid psychiatric disorder would not be significant).

2. Methods

2.1. Participants

Participants included 72 children and adolescents with a 22q11.2 deletion, confirmed by fluorescence in situ hybridization or microarray analyses, recruited from genetics clinics at two medical centers located in southeastern United States. The institutional review boards of both medical centers approved the study. Control subjects ($n=58$) consisted of typically developing healthy children, matched to the 22q11.2DS group by age (within 9 months) and gender. The control subjects were recruited from the local public schools and pediatric practices in the community. For controls, exclusion criteria included having a neurodevelopmental disorder or a genetic disorder; however, control children with ADHD were permitted to enroll in the study. It is to be noted that the control group in this study was utilized only to compare treatment rates for ADHD between that group and the 22q11.2DS children who had ADHD, since the incidence of this disorder was similar in both groups. Since the focus of the study is on children with 22q11.2DS, no other comparisons between the control and 22q11.2DS groups were made.

The 22q11.2DS study participants ranged in age from 6 to 16 years, with an average age at study enrollment of 10.49 years ($SD=2.6$). The sample was 54.8% male and largely white (84.7%), with African-Americans (6.9%), Hispanics (6.9%), and Native Americans (1.3%) also being represented in the sample. The Hollingshead Index placed the sample within the middle socioeconomic stratum (SES) ($M=32.29$, $SD=13.69$).

2.2. Procedures

For this study we employed two major data collection tools along with a semi-structured interview. In every instance, the child's primary caregiver was the informant for each of the measures employed. Of the 72 study participants, four had a primary caregiver that was not a parent (i.e., an aunt or grandmother), but in each case, the primary caregiver had raised the child and was thus a reliable informant. To obtain psychiatric diagnoses we utilized the Computerized Diagnostic Interview Schedule for Children (C-DISC), a comprehensive, structured interview that covers 36 mental health disorders for children and adolescents using DSM-IV criteria (NIMH-CDISC, 2004). The C-DISC is the most widely used and studied mental health interview that has been tested in both clinical and community populations. From the C-DISC, we extracted the specific diagnoses, the number of diagnoses, and whether a child received any diagnosis. The C-DISC was administered by trained research personnel (graduate students in psychology or a doctoral level clinician).

In addition, we employed the Child Behavior Checklist (CBCL), a highly reliable (test-retest reliability: $r=0.95$; inter-interviewer reliability: $r=0.93$), widely used parent-rating scale for child social-behavioral problems (Achenbach, 1991; Achenbach and Ruffle, 2000). The CBCL generated standardized scores for Internalizing, Externalizing, and Total Behavior Problems, with subgroups being designated based on summary scores falling at or below the tenth percentile (t -score ≥ 62).

Finally, to obtain data on the type of pharmacological and non-pharmacological treatments the children were receiving, we employed a semi-structured interview that was developed by the researchers. The interview, conducted by the trained researchers mentioned above, provided parent-reported information related to medication use and any type of mental health intervention (e.g., counseling, behavioral therapy). For this study we focused on the response to these questions: (1) has your child ever been diagnosed with an emotional, behavioral, or mental health disorder? (2) Seen a doctor or therapist for emotional, behavioral, or mental health issues? (3) Ever been prescribed medications for these problems?

Although some participants were receiving both types of interventions, these categories were mutually exclusive for this study in an effort to determine if specific diagnoses were aligning with specific types of treatment. Operationally, medication use was defined as a child taking any psychotropic medication for a psychiatric or behavioral problem, whereas the mental health intervention was operationally defined as individual or group counseling, parent-training, behavioral therapy, or participation in family therapy.

2.3. Data analyses

We employed chi-square statistics and Fisher's exact test to examine differences in the rates of mental health services and pharmacology utilization in the 22q11.2DS sample with and without a specific diagnosis. This would allow us to determine whether 22q11.2DS children with co-morbid psychiatric diagnoses were using mental health services at a rate lower than would be expected. All statistical analyses were performed with the use of SPSS version 18.0.

3. Results

3.1. Rates of psychiatric diagnoses in 22q11.2DS

The rates of psychiatric disorders in the sample ranged from 1% (Panic Disorder, Agoraphobia, and Post-Traumatic Stress Disorder)

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