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Autism symptoms in toddlers with Down syndrome: a descriptive study

Susan Hepburn[*], **Amy Philofsky**[*], **Deborah J. Fidler**[†], and **Sally Rogers**[‡]

[*]Department of Psychiatry, University of Colorado Health Science Center, Denver, CO, USA

[†]Human Development & Family Studies, Colorado State University, Denver, CO, USA

[‡]M.I.N.D. Institute, Department of Psychiatry, University of California, Davis, CA, USA

Abstract

Background—Research suggests that children with Down syndrome may be at increased risk of having an autism spectrum disorder; however, previous studies have not utilized comprehensive, state-of-the-art diagnostic tools to address the question of comorbid autism and Down syndrome.

Method—Comprehensive evaluations for autism were conducted in 20 2-year-old children with Down syndrome. The Autism Diagnostic Observation Schedule-Generic (ADOS-G) and the Autism Diagnostic Interview–Revised (ADI-R) were administered by experienced clinicians, who then determined if the child presented with significant symptoms of autism. Eighteen children participated in a follow-up evaluation at age 4 years.

Results—Three children (15%) met criteria for an autism spectrum disorder at both time points, and two (10%) for autistic disorder. Case descriptions are provided. Several children presented with difficulties in communication and play, but did not show problems in core social relatedness. The ADOS-G, if used alone, over-identifies autism in toddlers with Down syndrome.

Conclusions—Clinical judgement is critical in diagnosing autism in Down syndrome. Young children may show some signs of autism (particularly in the area of communication) and not have the disorder. Children who have comorbid Down syndrome and autism are particularly challenged, relative to their peers without autism.

Keywords

autism; comorbidity; Down syndrome; early development

Introduction

Individuals with Down syndrome have frequently been described as having charming personalities, often showing relative strengths in social functioning (Gibbs & Thorpe 1983; Rodgers 1987; Wishart & Johnston 1990). Many studies also report that individuals with Down syndrome show lower levels of psychopathology than individuals with other forms of

Correspondence: Any correspondence should be directed to Susan Hepburn, Department of Psychiatry, University of Colorado Health Sciences Center, 4200 E. 9th Ave., Box C-268-30, Denver, CO 80220, USA (susan.hepburn@uchsc.edu).

mental retardation (Dykens & Kasari 1997; Myers & Pueschel 1991; Stores *et al.* 1998), including a protection against specific disorders like autism (Gath & Gumley 1986; Howlin *et al.* 1995). However, other researchers have suggested that individuals with Down syndrome may actually be at increased risk for autism spectrum disorders (Collacott *et al.* 1992; Kent *et al.* 1999; Lund 1988). These studies suggest that the stereotype of the 'affectionate and outgoing' (Gibbs & Thorpe 1983) child may not hold true for all individuals with Down syndrome.

Studies to date report that somewhere between 2% (Collacott *et al.* 1992), 5% (Ghaziuddin *et al.* 1992), 7% (Kent *et al.* 1999) and 10% (Paly & Hurley 2002) of individuals with Down syndrome meet the criteria for autism or autism spectrum disorders. Symptoms of autism that are noted in studies of persons with Down syndrome include: social isolation, poor use of eye gaze, restricted/idiosyncratic interests, pre-occupation with parts of objects, hand-flapping, body rocking, compulsivity and lack of awareness of surroundings (Ghazuiddin 1997; Kent *et al.* 1999). These descriptions focus less on impairments of core social reciprocity and more on the third triad of symptoms (i.e. restricted interests), which have been shown not to differentiate between children with mental retardation and children with autism (Lord *et al.* 1994). Given that autism is currently understood as a deficit of core social relating, this overlap of symptoms may not be clinically significant. In addition, the methods of assessing autism symptoms in persons with Down syndrome have been limited to the use of a screening tool (Kent *et al.* 1999) and qualitative case study approaches (Bregman & Volkman 1988; Ghazuiddin 1997; Wakabayashi 1979). The impact of developmental level on symptoms has also not been adequately explored (Howlin *et al.* 1995). In order to understand how symptoms of autism manifest in some children with Down syndrome, it is important to include a group of well-characterized children with Down syndrome and to use state-of-the-art diagnostic tools to detect autism symptoms. Considering that autism symptoms arise prior to the age of 3 years in classic autism, it is also essential to evaluate the children with Down syndrome prior to their third birthday. Examining relations between autism symptoms and other child characteristics (e.g. developmental level, temperament) will be important for understanding more about who is at risk for a comorbid diagnosis, which has implications for practice.

The research questions posed in this study are as follows: (1) How many young children with Down syndrome exceed criteria for an autism spectrum disorder on the Autism Diagnostic Observation Schedule-Generic (ADOS-G) and/or the Autism Diagnostic Interview-Revised (ADI-R)? (2) Are results of the ADOS-G and ADI-R consistent with expert clinical judgement? (3) Are these symptoms stable across time – in this study, from age 2 to 5 years? And (4) Are there any child characteristics that could serve as risk factors for a comorbid autism diagnosis? In addition to examining data on a group level, we will also present case descriptions of the children who met criteria for autistic disorder as well as Down syndrome.

Methods

Participants

This study is part of a larger longitudinal study of the developing phenotypes of autism and other neurodevelopmental conditions. Participants were 20 2- to 3-year-old children with Down syndrome (mean age of 34 months) (see Table 1). All had a genetic diagnosis of trisomy 21. Participants were recruited from the Front Range/Denver Metropolitan Area parent support organizations for families of children with Down syndrome (Mile High Down Syndrome Association). The study was also advertised in clinic waiting rooms of The Children's Hospital of Denver and the JFK Child Development Center. Eighteen of these participants were seen for a follow-up assessment at age 4–5 years. Of the two that were unable to return, one family dropped out because of the time demands of the study, and one family moved out of state and there were no funds to travel to visit her.

Measures

Child and family history

Demographics questionnaire—Parents were asked about information regarding their age, socioeconomic status, education level and ethnicity.

Medical history—Complete medical records were sought for each participant through parents and primary care physicians.

Child temperament—The Toddler Temperament Scale (TTS) (Fullard, McDevitt & Carey 1995) was administered in a questionnaire format to the parents prior to their child's participation in the developmental or diagnostic portions of the study. Scores are obtained on nine dimensions, such that higher scores indicate more extreme temperamental features. Temperament data were correlated with autism symptom severity during exploration of possible child characteristics that may mark a risk for autism.

Developmental functioning

Mullen Scales of Early Learning—The Mullen Scales of Early Learning (MSEL; Mullen 1995) is a standardized developmental test for children aged 3–60 months, consisting of five subscales: gross motor, fine motor, visual reception, expressive language and receptive language. The MSEL allows for separate standard verbal and nonverbal summary scores to be constructed and demonstrates strong concurrent validity with other well-known developmental tests of motor, language and cognitive development (Mullen 1995). The MSEL was administered to all subjects at both time points according to standard instructions by raters with advanced degrees, trained in assessing young children with autism and other developmental disorders. Reinforcers for all subjects in all groups were used at times to reward cooperation and attention. Developmental quotients (DQ) were calculated (ma/ca), in order to eliminate floor effects on standard scores.

Vineland Scales of Adaptive Behavior, Interview Edition—The Vineland Adaptive Behavior Scales (Sparrow *et al.* 1984) is a standardized parent interview designed to assess

adaptive behaviour across four domains: socialization, communication, daily living and motor skills. The Vineland provides norm-referenced information based on the performance of representative national standardization samples of 4800 typically and atypically developing children. Mothers were interviewed at both time points, with a subsample completing the interview at time 2 (n = 12).

Autism symptoms

Autism Diagnostic Interview–Revised—The Autism Diagnostic Interview–Revised (ADI-R; Lord *et al.* 1994) is a structured, standardized parent interview developed to assess the presence and severity of symptoms of autism in early childhood across all three main symptom areas involved in autism: social relatedness, communication, and repetitive, restrictive behaviours. The ADI-R has been carefully psychometrically validated across a wide range of ages and severity levels in autism. The interview consists of over 100 questions. Considered the 'gold standard' in assessment in autism, this instrument yields an algorithm score and cutoffs for a diagnosis of autism. An algorithm has been established that differentiates autism from other developmental disorders at high levels of sensitivity and specificity (over 0.90 for both) for subjects with mental ages (MA) of 18 months and older. Inter-observer reliability of 80% or better was maintained across the duration of the project and systematically evaluated for more than 50% of subjects. The ADI-R was administered to the child's mother at both time points.

Autism Diagnostic Observation Schedule–Generic—The Autism Diagnostic Observation Schedule–Generic (ADOS-G; Lord *et al.* 1999) is a semi-structured standardized assessment tool that uses developmentally appropriate social and toy-based interactions in a 30- to 45-min interaction to elicit symptoms of autism in four areas: social interaction, communication, play and repetitive behaviours. The ADOS-G consists of four different modules, each directed at a particular level of language ability. In the present study, all subjects received module 1, for preverbal children or those just beginning to speak. Reliability was maintained at 85% and checked for 50% of participants across the period of data gathering. The ADOS-G was administered to all subjects at both time points in the study as part of the diagnostic qualification process.

Clinical judgement—For all participants, an experienced clinical psychologist with expertise in autism observed and interacted with the child and reviewed the child's records, including a review of all data collected within a 10-h research battery. This battery includes measures of social–emotional functioning, imitation, motor planning, play, language, cognition, nonverbal communication, executive function and sensory reactivity. Based upon this comprehensive assessment, the psychologist made a clinical diagnosis using the DSM-IV-TR criteria (APA, 2000), suggesting that the child either (1) meets full criteria for a clinical diagnosis of autism and demonstrates impairment in social, communication, and restricted activities (i.e. 'autism'); (2) meets partial criteria for autism and demonstrates impairment in social interaction and *either* communication *or* restricted activities (i.e. 'Pervasive Developmental Disorder - not otherwise specified' (PDD-NOS)); or (3) does not meet criteria for autism, but does have a developmental delay (i.e. not autism).

Procedures

The entire study was carried out under IRB approval. Consent forms were reviewed with each family and all questions were answered before consent was obtained and before any measures were gathered. The MSEL, ADOS-G and ADI-R were administered in a laboratory visit in a standardized fashion. All examiners were masters- or doctoral-level clinicians with several years of clinical experience working with young children with developmental disabilities. Mothers were interviewed for the Vineland, generally during a home visit. Approximately 2 years after participating in the initial evaluation, the family was contacted and asked to return for a follow-up visit. Eighteen children returned for a follow-up assessment, which was comprised of the MSEL, ADOS-G and ADI-R (among other measures).

Results

How many children meet criteria for an autism spectrum disorder on the ADOS-G or ADI-R?

See Table 2 for a presentation of ADOS-G and ADI-R scores by initial diagnostic classification.

ADOS-G scores—As previously described, the ADOS-G provides cutoff scores on the classification algorithm for: (a) autism and (b) autism spectrum, on two domains: (1) communication, and (2) reciprocal social interaction. The cutoff scores for autism are higher (more restrictive) than those for autism spectrum, and hence a ‘spectrum’ classification suggests a milder set of symptoms. To be classified as having autism on the ADOS-G, a child must exceed the autism cutoff on both social and communication domains. To be classified as having an autism spectrum disorder, a child must exceed spectrum cutoffs on both domains. It is important to note that restricted activities and impairments in play are not included in the diagnostic classification.

Of the 20 toddlers with Down syndrome in this study, one child (A.B.) exceeded ‘autism cutoff’ on both social and communication. Two children (E.D. and L.K.) exceeded ‘autism spectrum cutoff’ on both social and communication. Three children (15%) exceeded criteria for spectrum for the ADOS-G communication domain, but not the social domain; and one child (5%) exceeded criteria for spectrum on ADOS-G social domain, but not on the communication domain.

ADI-R scores—In contrast with the ADOS-G, none of the toddlers with Down syndrome met criteria for autism on the ADI-R. Furthermore, none of the toddlers with Down syndrome met the ADI-R cutoff for the ADI-R social scale. However, three of the toddlers (A.B., E.D. and L.K.) (15%) with Down syndrome met the ADI-R cutoff on the communication scale – all of whom were classified as having autism or autism spectrum on the ADOS-G (see above). In addition, two more children met the cutoff on the ADI-R repetitive/stereotyped behaviour scale.

These results suggest that the ADOS-G and the communication subscale of the ADI-R were very consistent in identifying the children with Down syndrome who met criteria for autism

or spectrum disorder. However, children who met ADOS-G criteria for autism or spectrum disorder were not identified via the social domain of the ADI-R. Their impaired communication development appears to be the area primarily responsible for their elevated scores on these scales.

How consistent are ADOS-G and ADI-R scores with overall clinical judgement?

The psychologists on the team reviewed all data from the participant's developmental history, standardized testing and laboratory measures, and determined that two children (A.B. and E.D.) presented with comorbid autism and Down syndrome. One of these children exceeded the cutoffs for autism on both the ADOS-G and ADI-R (A.B.), and the other exceeded cutoffs for spectrum on the ADOS-G and communication on the ADI-R (E.D.). These two children will be described in more detail below.

The third child (L.K.) who was classified as having autism spectrum by the ADOS-G was not classified as having a clinical diagnosis of autism. Her developmental delay was significant (i.e. she functioned at approximately 10 months developmentally when she was 2 years chronologically) and she had very poor motor skills and difficulty initiating movements. Her difficulties in joint attention, imitation, play and coordination of nonverbal behaviours were attributed by the clinicians to her overall cognitive and motor impairments. Notably, she demonstrated a reciprocal social smile, directed her facial expressions towards adults, and shared affect during play, providing evidence of dyadic social interaction.

None of the nine children who met partial criteria on either the ADOS-G or ADI-R were clinically diagnosed as having autism or PDD-NOS, primarily because they did not present with core deficits of social relating. Developmentally, seven of these nine children were estimated to be functioning below 18 months, which may help to explain the immaturity of their social and communicative behaviours. While all these children demonstrated limited play, repetitive motor behaviours (particularly hand-flapping), and were nonverbal, none of them demonstrated a core difficulty in social relatedness. Emotional contagion and sharing of affect with an adult during play was noted in all nine of these children, as were attempts to imitate and to follow an adult's bid for joint attention. Seven of these children were not observed to initiate joint attention during the ADOS-G, and eight did not demonstrate any shows or gives; however, these difficulties in regulating triadic social interactions may be mediated by overall developmental level.

Stability across time: from age 2 to 5 years

Eighteen children returned for a follow-up evaluation approximately 2 years later (see Table 2). The two children who were not observed at follow-up were dropped for the following reasons: one family withdrew due to time and scheduling constraints, and one child moved out of state and funds prohibited our travel to visit her family. Three other families expressed concern about the time commitment for the follow-up visit; however, they allowed the clinical psychologist to travel to the child's school to conduct the evaluation. None of the three children evaluated at the school presented with significant symptoms of autism at follow-up.

The same three children who met criteria on the ADOS-G and ADI-R at age 2 (E.D., A.B. and L.K.), also met criteria at age 4 years; however, symptom presentation was more dramatic for two of the children (E.D. and A.B.; see 'Case descriptions' below). L.K. continued to present with significant developmental delay (mental age of <12 months) and was observed to use her eye gaze effectively to modulate social interactions, share affect and attention with adults, and demonstrate some nonverbal reciprocity within sensorimotor play. Her social development, consisting of responsive dyadic exchanges, appeared to be congruent with her cognitive maturity. Therefore, she once again did not receive a clinical diagnosis of autism.

The remaining 15 children did not meet ADOS-G or ADI-R criteria for autism, nor were they clinically diagnosed as having autism. Five (30%) continue to demonstrate qualitative impairments in communication and exceeded the spectrum cutoff on the communication domain of the ADOS-G, but none exceeded the cutoff on the ADI-R. None of these children met criteria for autism on the social domain.

Examination of the distribution of scores between groups suggests that social and communicative symptoms of autism are bimodally distributed at both time points – such that either many symptoms occur together or few actually occur. Many of the children who demonstrated a few symptoms but were clearly not autistic demonstrated poor requesting and impaired play skills. Social responsivity, initiations, joint attention, imitation and reciprocity were not impaired in these children.

Finally, autism symptoms did not emerge after the age of 3 years for any of the participants. However, for those children already presenting with early symptoms, the severity of their social and communicative impairments seemed to worsen with age, as reflected in higher (more severe) scores on both the ADOS-G and the ADI-R.

Are there any child characteristics that could serve as risk factors for a comorbid autism diagnosis?

One hypothesis is that children with Down syndrome who are lower functioning may present with more significant social and communicative impairments than those with higher functioning. The present sample is too small to evaluate this conclusively; however, for the three children who met criteria for an autism spectrum disorder, we observed very little variability in their developmental skills. All three children, at the chronological ages of 52–58 months, obtained mental ages of 27–28 months on the MSEL. All three fell within the lower 20% of the IQ distribution of this sample of children with Down syndrome. Further studies, with larger sample sizes, will be necessary to elucidate this hypothesis.

We hypothesized that child temperament could be related to parent report of symptoms associated with autism spectrum disorders; however, correlations between ADI-R total scores and parent report of child temperament on the TTS (Fullard *et al*. 1995) were not statistically significant and were as follows: ADI-R x activity level: $r = -0.21$, ADI-R x rhythmicity: $r = -0.38$; ADI-R x approach/withdrawal: $r = -0.33$; ADI-R x adaptability: $r = -0.12$; ADI-R x emotional intensity: $r = -0.23$; ADI-R x negative mood: $r = -0.05$; ADI-R x persistence: $r = -0.24$; ADI-R x distractibility: $r = -0.21$; ADI-R x threshold of

responsiveness: $r = -0.26$. Further examination of some of these dimensions of child temperament should be examined in studies with more statistical power. Perhaps a child's tendency to withdraw from novelty (approach/withdrawal), or to be dysregulated in daily behaviours (rhythmicity) could be markers of children with Down syndrome at risk for autism.

Case descriptions: a closer look at two toddlers with Down syndrome and autism spectrum disorders

A.B—A male child, age 26 months at the initial evaluation and 50 months at follow-up, A.B. met autism criteria on the ADOS-G, ADI-R and clinical judgement at both evaluations. Medically, he has a history of heart problems that have required monitoring, but no surgery. His motor milestones were delayed (sitting at 11 months, walking at 20 months) and he was reported to begin using words at 18 months and then lost their spontaneous use for a period of 8 months. He is currently functionally nonverbal and communicates by reaching, manipulating another's hand, and vocalizing without coordinated eye gaze. His overall developmental quotient is estimated to be 45 (mental age/chronological age x 100) and his nonverbal problem-solving skills are a relative strength on his MSEL profile.

A.B. presents as a lethargic, irritable child who is easily frustrated and cries in the context of challenges. During the ADOS-G at the time of initial evaluation, he rarely directed affect or vocalizations to others, did not use any gestures, did not integrate verbal and nonverbal behaviours to communicate intention, did not initiate or respond to joint attention, and did not initiate any showing or giving behaviours. His functional play was limited to cause-and-effect actions. At follow-up, he continued to show impairments in social and communicative behaviour (e.g. no gesture use, no integration of nonverbal and verbal, poor social orienting).

His mother's report on the ADI-R was consistent with the observations on the ADOS-G. She also reported that he shows limited attention to her voice, does not seem to comprehend language, and does not come for comfort or lift his arms to be held. She also reported that he has difficulty initiating appropriate play activities and does not seem to show interest in other children. She expressed particular concern over his exhibition of self-injurious behaviours, which emerged around the age of $3\frac{1}{2}$ years, and his difficulty 'being happy'. Overall, A.B.'s mother reported that she worries constantly over her son's behaviour and future and has been very frustrated that their attempts to provide social and language intervention have not been fruitful. An examination of his intervention plan reveals that primarily naturalistic strategies have been employed and A.B. is fully included in an integrated preschool with a full-time aide. He has had little direct instruction in social or communicative behaviours. Because he is not officially identified as a child with autism in the public schools, he has not had access to specialized instruction for children with autism.

E.D—E.D. was a 33-month-old male when he was initially tested and met autism spectrum criteria on both the ADOS-G and ADI-R. Clinically, he met criteria for an autism diagnosis. He was evaluated again at 52 months and presented with a worsening of autism symptoms, now scoring within the full autism range on all instruments. E.D. was born 6 weeks

premature. Little is known about his early history, as he was adopted in his second year of life, although there is some speculation that he experienced some neglect during infancy. He was delayed in achieving his motor milestones, sitting at 22 months and walking at 32 months.

Initially, E.D. presented as a happy, relatively easygoing child who was content to twirl objects for long periods of time. He obtained a developmental quotient of 45 on standardized testing. During the ADOS-G at the initial evaluation, his social responsiveness was inconsistent. He responded to his name and oriented to others when they vocalized or had a toy of interest to him. He was observed to direct some of his vocalizations to others, and sometimes directed his facial expressions and shared his enjoyment with others. He did not initiate joint attention. He was not able to imitate others and participated passively in adult-directed activities, such as the birthday party. He showed no gestures and did not integrate his nonverbal behaviours to communicate intention.

In the follow-up assessment, E.D. presented as a more irritable child who was easily frustrated and avoidant of activities. During the ADOS-G, E.D. demonstrated more impaired social responsiveness, as evidenced by a lack of social orienting (did not turn when his name was called, did not direct facial expressions or vocalizations to others). He actively resisted adult-directed activities and refused to participate in both the imitation or birthday party activities. He communicated primarily by manipulating the adult's hand and did not coordinate his vocalizations or eye gaze with his communications. In fact, he rarely vocalized.

His mother also reported a worsening of symptoms over time, stating that he seems 'more remote than ever', and is 'increasingly sinking into his own little world'. She described that he spends the majority of his time engaging in repetitive activities, such as twirling and rocking. He has become increasingly aggressive towards teachers and other children and sometimes tries to bite himself when frustrated. She expressed concern over his early intervention programme, as he only receives 1 h of speech consultation per week and is educated in an inclusive setting without specific supports or structure. He has access to a teaching assistant, but E.D.'s mother fears that she is not well trained and tends to allow E.D. to escape demands instead of acting out. The family is now seeking services specifically designed for children with autism.

Discussion

The results of this descriptive study suggest that some young children with Down syndrome, 15% in this small sample of 20 2- to 3-year-old children, meet criteria for an autism spectrum disorder. Two of three of the children who met criteria on the ADOS-G and ADI-R for an autism spectrum disorder met criteria for autism 2 years later, at age 4–5 years. None of the children who presented as non-autistic at the initial evaluation acquired significant symptoms over time. Of the three children presenting with significant autism symptoms at age 2–3 years, one female child demonstrated marked improvement in social symptoms over time; however, her communication skills remained significantly impaired. The two boys who presented with the most autistic symptoms at age 2 years were still

nonverbal at age $4\frac{1}{2}$ years and had made few functional gains, despite access to early intervention services.

Autism symptoms appear to distribute bimodally within Down syndrome, such that a few children have a lot of symptoms and many children demonstrate just a few. Symptoms in the communication and play domains are evident at both time points in children with Down syndrome who do not present as autistic.

Although we observed limited variability in the developmental functioning of children who met criteria for autism spectrum disorders in this sample, we do not have sufficient power to test for the effects of developmental functioning on autism symptoms.

A larger study is necessary to examine whether comorbid Down syndrome and autism constitutes ‘classic’ autism, or whether an alternative explanation accounts for the social and communication difficulties we recognize as autism. Children who present as autistic may be lower functioning across domains; or perhaps the severity of specific deficits associated with Down syndrome – such as motor planning or memory skills – impacts upon the development of social reciprocity and communicative intent. Working with the children in this study has prompted us to examine the possible role of temperament and mood issues on social style in children with Down syndrome.

If autism truly occurs at a higher rate in Down syndrome than in other developmental disabilities and if that increased rate is not a function of overall level of impairment or severity of impairment in specific core skills (such as memory), then there may be implications for a shared genetic aetiology of autism and Down syndrome. A recent study has suggested that chromosome 21 may be involved in the epigenesis of some cases of autism (Molloy, Keddache & Martin 2005). Research examining a large community-based sample of children with Down syndrome is needed to address these questions.

Although the present study includes a very small sample of children with Down syndrome, we have attempted to conduct a more thorough diagnostic evaluation than has been conducted in previous studies. This is one of a few studies examining social and communicative functioning in children younger than 3 years, which is the age of onset of classic autism. However, given the small sample, the results of this study should be interpreted with caution. Future studies, drawing from large, population-based samples of children with Down syndrome would be very informative for the field.

While many children (9/20) demonstrated some behaviours that are consistent with autism during the toddler period (such as impairments in initiating joint attention through an isolated point or gesture, repetitive behaviours and limited play), these impairments occurred in the context of some critical social and affective strengths that were appropriate to the child’s developmental level (e.g. sharing affect, following joint attention, directing vocalizations). In fact, the developmental level of the child may impact upon the presentation of social and communicative behaviours. The three children who demonstrated the most severe autism symptoms all had developmental quotients of less than 55 and developmental ages of less than 18 months at the initial evaluation, as did seven of the nine children who demonstrated communicative symptoms. Interestingly, approximately 30% of

the other children with Down syndrome in the sample also presented with developmental quotient of less than 50, but did not present with significant symptoms of autism.

Early identification of children with comorbid Down syndrome and autism may be very important so that appropriate social and communicative interventions can be designed and implemented for these dually challenged children. Given the lack of progress evidenced by the children who met criteria for comorbid autism and Down syndrome at both time points, one wonders about the implications for early intervention for these children. There is no evidence in the literature about response to treatment or educational strategies, which would be an important contribution to the field.

In our clinical experience, family members of persons with Down syndrome and autism face additional challenges in coping with their child's disability. The literature available to parents often stresses the social strengths of children with Down syndrome and teachers and therapists may expect these strengths to be pervasive in the population. Inconsistent or poor social relating in a child expected to be 'charming' and 'outgoing' may create some frustration for all concerned and may lead to a poor fit between the child and the expectations in the environment.

Given that this study is limited by the small sample, it will still be important to consider that perhaps 10–20% of children with Down syndrome present with significant social and communicative difficulties at young ages and that these children and their families may require qualitatively different interventions than other children with the same genetic condition.

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References

American Psychological Association (APA). Diagnostic and Statistical Manual of Mental Disorders. 4. APA; Arlington, VA: 2000. Text Revision (DSM-IV-TR)

Bregman JD, Volkman FR. Autistic social dysfunction and Down syndrome. Journal of the American Academy of Child and Adolescent Psychiatry. 1988; 27:440–441. [PubMed: 2972692]

Collacott RA, Cooper SA, McGrother A. Differential rates of psychiatric disorders in adults with Down's syndrome compared to other mentally handicapped adults. British Journal of Psychiatry. 1992; 161:671–674. [PubMed: 1422617]

Dykens EM, Kasari C. Maladaptive behavior in children with Prader-Willi syndrome, Down syndrome, and non-specific mental retardation. American Journal on Mental Retardation. 1997; 98:580–587. [PubMed: 8192903]

Fullard, W.; McDevitt, S.; Carey, W. Toddler Temperament Scale (TTS). Behavioral - Developmental Initiatives; Scottsdale, AZ: 1995.

Gath A, Gumley D. Behaviour problems in retarded children with special reference to Down's syndrome. British Journal of Psychiatry. 1986; 149:156–161. [PubMed: 2946349]

Ghazuiddin M. Autism in Down's syndrome: family history correlates. Journal of Intellectual Disability Research. 1997; 41:87–91. [PubMed: 9089464]

Ghaziuddin M, Tsai L, Ghaziuddin N. Autism in Down's syndrome: presentation and diagnosis. Journal of Intellectual Disability Research. 1992; 36:449–456. [PubMed: 1458002]

Gibbs MV, Thorpe JG. Personality stereotype of noninstitutionalized Down Syndrome children. American Journal of Mental Deficiency. 1983; 87:601–605. [PubMed: 6223525]

Howlin P, Wing L, Gould J. The recognition of autism in children with Down syndrome: implications for intervention and some speculations about pathology. Developmental Medicine & Child Neurology. 1995; 37:406–413. [PubMed: 7768340]

Kent L, Evans J, Paul M, Sharp M. Comorbidity of autistic spectrum disorders in children with Down syndrome. Developmental Medicine and Child Neurology. 1999; 41:153–158. [PubMed: 10210247]

Lord, C.; Rutter, M.; DiLavore, P.; Risi, S. Autism Diagnostic Observation Schedule – WPS Edition. Western Psychological Services; Los Angeles, CA: 1999.

Lord C, Rutter M, LeCouteur A. Autism Diagnostic Interview Revised: a revised version of a diagnostic interview for caregivers of individuals with possible pervasive developmental disorders. Journal of Autism and Developmental Disorders. 1994; 24:659–685. [PubMed: 7814313]

Lund J. Psychiatric aspects of Down's syndrome. Acta Psychiatrica Scandinavia. 1988; 78:369–374.

Molloy CA, Keddache M, Martin LJ. Evidence for linkage on 21q and 7q in a subset of autism characterized by developmental regression. Molecular Psychiatry. 2005; 10(8):741–746. [PubMed: 15940295]

Mullen, EM. Mullen Scales of Early Learning. American Guidance; Circle Pines, MN: 1995. AGS Edition

Myers BA, Pueschel SM. Psychiatric disorders in persons with Down syndrome. Journal of Nervous and Mental Disease. 1991; 179:609–613. [PubMed: 1833506]

Paly RJ, Hurley AD. Down syndrome and autistic disorder. Mental Health Aspects of Developmental Disabilities. 2002; 5:64–65.

Rodgers C. Maternal support for the Down's syndrome stereotype: the effect of direct experience of the condition. Journal of Mental Deficiency Research. 1987; 31:217–278.

Sparrow, SS.; Balla, DA.; Cicchetti, D. Vineland Adaptive Behavior Scales. American Guidance; Circle Pines, MN: 1984.

Stores R, Stores G, Fellows B, Buckley S. Daytime behaviour problems and maternal stress in children with Down's syndrome, their siblings, and non-intellectually disabled and other intellectually disabled peers. Journal of Intellectual Disability Research. 1998; 42:228–237. [PubMed: 9678407]

Wakabayashi S. A case of infantile autism associated with Down's syndrome. Jouranl of Autism and Developmental Disorders. 1979; 19:31–36.

Wishart JG, Johnston FH. The effects of experience on attribution of a stereotyped personality to children with Down's syndrome. Journal of Mental Deficiency Research. 1990; 34:409–420. [PubMed: 2148352]

Table I

Demographic and developmental information by initial diagnostic classification

	Time 1 (2–3 years)		Time 2 (4–5 years)	
	Not autism spectrum (n = 17)	**Autism spectrum (n=3)**	**Not autism spectrum (n = 15)**	**Autism spectrum (n=3)**
Chronological age				
Mean (SD)	34.41 (6.87)	28.00 (4.34)	55.67 (6.96)	57.72 (3.21)
Range	24–45	25–33	47–71	52–58
Mental age				
Mean (SD)	22.73 (5.12)	14.42 (0.29)	32.35 (5.28)	27.75 (0.25)
Range	14–33	14.25–14.75	26–42	27–28
Developmental quotient (ma/ca)				
Mean (SD)	65 (0.09)	0.52 (0.08)	0.58 (0.06)	0.50 (0.03)
Range	0.54–0.86	0.43–0.59	0.48–0.73	0.48–0.53
Vineland Adaptive Behavior Scales				
Mean (SD)	65.31 (6.4)	61.33 (6.43)	58.87 (7.9)	51 (4.58)
Range	58–81	54–66	50–80	47–56
Gender				
Male:female	12:5	2:1	11:4	2:1
Socioeconomic status (Hollingshead scores)				
Mean (SD)	55.73 (6.81)[1]	64.75 (1.77)[2]	58.88 (5.35)[2]	65.25 (6.06)[3]
Range	40–66	64–66	49–66	64–66

[1] *n*= 12;

[2] *n* = 10;

[3] *n* = 2.

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Table 2

ADOS-G and ADI-R scores by diagnostic group at both timepoints

	Time 1 (age 2–3) (n = 20)		Time 2 (age 4–5) (n = 18)		
	Not autism spectrum (n = 17)	**Autism spectrum (n=3)**	**Not autism spectrum, module I (n = 11)**	**Not autism spectrum, module II (n = 4)**	**Autism spectrum, module I (n=3)**
ADOS-G					
Social					
Mean (SD)	2.77 (1.52)	7.0 (2.0)	(1.18)	2.25 (1.63)	7.67 (2.51)
Range	0–6	5–9	0–3	0–8	5–10
Communication					
Mean (SD)	1.18 (1.19)	4.0 (2.0)	1.54 (1.29)	2.00 (1.69)	4.00 (2.0)
Range	0–4	2–6	0–4	0–4	2–6
Communication + social					
Mean (SD)	3.94 (2.44)	11.00 (3.46)	2.54 (2.01)	4.25 (5.31)	11.67 (4.51)
Range	0–10	9–15	0–6	0–12	7–16
Play					
Mean (SD)	1.18 (0.88)	2.0 (0.00)	1.55 (0.82)	0.67 (0.58)	3.33 (1.15)
Range	0–3	2–2	0–3	0–1	2–4
Restricted activities					
Mean (SD)	1.29 (1.16)	(1.73)	1.27 (1.19)	0.75 (0.98)	1.33 (2.31)
Range	0–3	0–3	0–4	0–2	0–4
ADI-R					
Social					
Mean (SD)	2.44 (1.67)	4.67 (2.52)	2.27 (1.23)	(2.45)	13.33 (7.27)
Range	1–6	2–7	0–4	0–5	5–18
Communication					
Mean (SD)	3.44 (2.7)	7.33 (4.73)	3.73 (1.19)	6.0 (2.16)	11.0 (3.0)
Range	0–10	2–11	2–6	4–9	8–14
Restricted activities					
Mean (SD)	1.31 (1.1)	(1.0)	1.36 (1.57)	1.25 (0.96)	4.00 (1.7)
Range	0–3	0–2	0–4	0–2	2–5
Total ADI-R score					

	Time 1 (age 2–3) (n = 20)		Time 2 (age 4–5) (n = 18)		
	Not autism spectrum (n = 17)	**Autism spectrum (n=3)**	**Not autism spectrum, module I (n = 11)**	**Not autism spectrum, module II (n = 4)**	**Autism spectrum, module I (n=3)**
Mean (SD)	7.06 (4.12)	13.00 (5.0)	7.36 (2.50)	9.25 (2.5)	28.33 (.96)
Range	1–19	8–18	3–12	6–12	18–37