The continuing challenge of diagnosing autism spectrum disorder in children with Down syndrome

L. Gray,* P. Ansell,† G. Baird‡ and J. R. Parr*

*Institute of Neuroscience, Newcastle University, Newcastle upon Tyne †Department of Health Sciences, University of York, York, and ‡Guy’s and St Thomas’ NHS Foundation Trust, London, UK

Accepted for publication 17 December 2010

Traditionally, the association between autism spectrum disorder (ASD) and Down syndrome (DS) was considered uncommon, as the stereotype image of an individual with DS was (and continues to be) a friendly, affectionate and outgoing personality (Ghaziuddin et al. 1992). Identifying children with DS who also meet ICD-10/DSM-IV criteria for ASD (DS-ASD) continues to be a challenge for clinicians and parents. Following 20 years of research in which the prevalence of DS-ASD varied from 1% to 12% (Ghaziuddin et al. 1992; Kent et al. 1999; Capone et al. 2005; Starr et al. 2005; Carter et al. 2007; Hepburn et al. 2008; Molloy et al. 2009; Reilly 2009), DiGuiseppi and colleagues (2010) have used screening tools and standardized diagnostic measures in a population-based cohort of 123 children with DS (mean age: 6.1 years) and reported 18% to have ASD and 7% autism. DiGuiseppi’s study is the most robust to date and, while limitations remain and further research is necessary, we now know more than ever about DS-ASD. Taking DiGuiseppi’s prevalence findings into account, we believe DS-ASD remains under-diagnosed. There are many reasons for this, some of which are discussed below, with quotes from some parents and clinicians who were canvassed in preparation for this editorial. Some quotes sum up the challenges for parents and clinician alike. Opinions differ between professionals about whether, and when, an additional neurodevelopmental diagnosis should be given. Parental opinion on the desirability of the diagnosis is similarly variable – some parents find an ASD diagnosis valuable and others do not.

Although there are often many other conditions associated with DS (Roizen & Patterson 2003), some parents report that the ASD behaviours displayed by their child are hardest to cope with, and have the biggest impact: ‘Of [my child’s] three conditions autism is most difficult, then Hirschsprung’s then DS (which is by far the easiest to deal with!)’. Parents reported that professionals have a wide-ranging level of understanding about DS-ASD: ‘It is hard on us all. We cope and put on a good front of coping (as you have to) but to be honest we don’t cope. It is hard to explain to other people and professionals what it is really like…some professionals identify completely…others have little understanding and expect him to act like other DS children’. Some parents faced absolute disbelief from professionals about a possible DS-ASD diagnosis: ‘Everyone I approached thought he either couldn’t, definitely didn’t or shouldn’t have autism’. Other clinicians may be unable to see what benefit an additional diagnosis may bring: ‘[The Consultant said] he didn’t think anything would be gained by having [my child] assessed’. Some parents commented that their child had been diagnosed later than hoped for, delaying an explanation for certain behaviours and impacting on the availability of early intervention and support for ASD-related issues from health, education and social care services, and from their own family. As discussed by Lenhard
and colleagues (2005), there may be relief that differences between their child and other children with DS, and worries about parenting, were finally explained: ‘Although we didn’t want to hear she had autism, it explained the behaviour – maybe it wasn’t us being poor parents!’ Parents also reported that the ASD diagnosis helped them find the required support, although this may ‘require huge personal input’ as inadequate information leads to parents searching themselves: ‘Trying to find information was difficult . . . Finding it too late happens too often . . . Not being given enough information or not being steered to people with the right information happens too often . . . Finding the energy to find information is difficult . . . Acting upon information can be daunting, expensive, tiring or impossible’. Similarly, one clinician commented, ‘A secondary ASD diagnosis is often late. Features of ASD, such as narrow diets and sleep disorders, are often identified first. The family does not have a happy experience and often a less than desirable process of assessment. Then they receive an additional diagnosis, which is hard in itself. But why is the diagnosis of DS-ASD late?

Many quotes from parents suggest a lack of awareness by clinicians of the co-occurrence of ASD and DS. It may be that some practitioners are not referring children and their families for second opinions when there is diagnostic uncertainty. In our experience, some clinicians continue to find separating ASD from behaviours relating to intellectual disability or DS difficult: ‘Although we [local clinicians] pursued a diagnosis no one would give him the actual diagnosis of Autism. They [the parents] were also told by independent source that he was not [autistic]! . . . it is difficult to separate what is true autism from severe learning difficulties. All of the children who we would consider as having ASD also have severe learning difficulties’. Some parents are not comfortable with yet another diagnosis. An ASD diagnosis could be the ‘nth’ additional diagnosis that some parents have to consider. Parents and families may also be worried by the implications for their child – such as differences in the opportunities or educational placements offered – and may think they are protecting their child’s interests by not seeking a DS-ASD diagnosis, or rejecting it if one is suggested. Clinicians report a mixed response from parents when a dual diagnosis is raised, and need to tread a thin line in order to maintain a long-term, constructive, working relationship with a family: ‘I know what you are saying but don’t want you to say it’ [said the mum whose child has ASD]. We agreed that I would make it clear in the report to the local team [who had requested an opinion about ASD diagnosis] that her child had ASD, but would not include the ‘A’ word; we parted on good terms, ‘both happy that we had achieved our goals’. Upsetting a family by alluding to the possibility of an additional diagnosis may mean conflict and a negative experience for both clinician and parent. The parent may express dissatisfaction or complain and search for support elsewhere; these may then lead to the clinician being more hesitant when discussing these or other issues in the future: ‘Most people are positive, but I remember one notable exception of a parent who never wanted to see me again when I raised the possibility [of an additional diagnosis]’. The position of the clinician is a complex one, as the aim is always to work with parents over time, in terms of both diagnosis and decision making, while acting in the best interest of the child to provide support and appropriate intervention. Parents are the experts in terms of what would be beneficial in their own unique set of circumstances – and although from a clinician’s viewpoint a diagnosis would seem to be the correct path to take, the parent may very well be correct as to whether it would help or hinder their child, and affect the family as a whole. All of these difficult issues are part of clinical skill and should be considered and evaluated with parents before the suggestion of further diagnosis is broached. Doing this is sometimes interpreted by parents who are keen to move quicker as clinicians delaying, or being reluctant to offer a diagnosis. This could in turn become a source of some frustration.

We recommend that before this point is reached, and before parents seek support elsewhere, clinicians seek additional support in the form of a local colleague or referral to a regional or national assessment service. We believe a DS-ASD diagnosis is important, as this helps parents and professionals understand behaviours, and plan for appropriate behavioural strategies and intervention/educational provision. In turn, this is likely to have benefits for both the child and family, and professionals working closely with them. We have recently started what we hope will be the definitive study of DS-ASD, answering questions about prevalence and clinical and behavioural differences between DS and DS-ASD. In collaboration with the Children with Down Syndrome Study (http://www.cdss.org.uk), and funded and supported by the Down’s Syndrome Association (http://www.downs-syndrome.org.uk), we are sequentially using ‘screening’ questionnaires, around the child’s 3rd, 4th and 5th birthdays. By using clinical notes and standardized assessments, we will investigate the detected and true prevalence of DS-ASD and, critically, report on the sensitivity and specificity of the screening measures between 3 and 5 years. These data, and identifying behavioural and developmental ‘red flags’, which predict DS-ASD and differentiate it from DS alone, will improve our knowledge about earlier detection of DS-ASD. However, we suspect DS-ASD will remain a difficult clinical diagnosis to make and, we suspect, one about which parental and clinicians’ opinions will on occasion be divided.
References


This document is a scanned copy of a printed document. No warranty is given about the accuracy of the copy. Users should refer to the original published version of the material.