Abstract

An area of research with autism spectrum disorders (ASD), which has received a considerable amount of attention recently is early diagnosis. This phenomenon is due largely to encouraging results from intensive intervention programs for children at very young ages. While five types of ASD exist, efforts in this area have focused almost exclusively on autism. To date, the primary methods of identification have been evidence-based assessment scales using established criteria for differential diagnosis and cognitive/developmental descriptive studies, which attempt to tease out behavior patterns of infants who later evince ASD from their normally developing counterparts. A third focus, which is in early development, involves genetic studies aimed at establishing biological links. However, at present such procedures are not viable for diagnosis. Opinions are rendered on the earliest age at which children can be reliably diagnosed at present, and a review of practical considerations is provided. Future challenges and directions in ASD identification and diagnosis are discussed.

Keywords: Early identification; Diagnosis; Autism spectrum disorders

1. Introduction

In the last decade, one of the major developments in the study of autism spectrum disorders (ASD) has been the recognition that young children can benefit from psychological interventions primarily involving operant conditioning (i.e., Applied Behavior Analysis; Cannella, O’Reilly, & Lancioni, 2006; Ellis, Ala’i-Rosales, Glenn, & Rosales-Ruiz, 2006; Green et al., 2006; Matson & Minshawi, 2006a; Symes, Remington, Brown, & Hastings, 2006). Studies initially involved
single case designs with a few discrete target behaviors such as stereotypies, self-help skills, initiating responses, and eye contact. Over time these procedures have evolved into control outcome studies, often of 20–40 h in duration per week over a period of months (Eikeseth, Smith, Jahr, & Eldevik, 2002; Lovaa, 1987; Matson, Matson, & Rivet, in press; Matson, Sevin, Fridley, & Love, 1990; Matson, Taras, Sevin, Love, & Fridley, 1990; Smith, Groen, & Wynn, 2000; Whittingham, Sofronoff, & Sheffield, 2006). Perhaps the most controversial claim from these studies on comprehensive programs, of which the UCLA-Young Autism Project is arguably the most visible, is the notion that these children’s disorders have been cured. This claim is in contrast to others who have argued that this disorder is chronic and persistent (Howlin, Mawhood, & Rutter, 2000). However, regardless of cure or no cure, little debate lingers regarding the potential for marked gains in socialization, communication, and intellectual test scores (Baird et al., 2001). Many specific core symptoms of ASD are yet to be addressed in these outcome studies (Matson, 2007a), despite the fact that generally agreed-upon core symptoms of communication, social skills, and behaviors problems (e.g., stereotypies, self-injury) have been identified (Holden & Gitlesen, 2006; Schreck & Williams, 2006; Selfe, 2002). Therefore, while a great deal still needs to be learned about how effective these early intervention programs are on specific core symptoms of ASD, preliminary results are encouraging.

As a result of these findings on early intervention, particular impetus, and urgency has been given to the notion that the earlier the child can be identified and treated, the better the long-term outcome. Initial efforts following up these children a few months or years after the initial training provide some but not uniform support for this hypothesis (Matson & Minshawi, 2006a). Other reasons that early identification may be desirable include enabling genetic counseling to parents regarding having more children, to discuss parental concerns and give them information, and to provide psychological support (Williams & Brayne, 2006).

This thinking however is also reflected in nosology. The commonly accepted notion is that onset occurs prior to 30 months of age – a position that has been held for some time (Lund & Jensen, 1989). Irregularity in the attainment of developmental milestones, comorbid psychopathology, and the presence of challenging behaviors can all complicate the diagnostic picture however (Matson & Minshawi, 2007; Matson & Nebel-Schwalm, 2007). Thus, while on the surface of the issue, such goals appear practical, in application they can be quite complicated and hard to attain.

2. How early is too early?

Fifty percent of parents report features of autism in their children by 2 years of age and 93 percent indicate recognition of symptoms by age 3. Similarly, Asperger’s syndrome, while being recognized later than autism, is also noted early by parents, although diagnosis does not typically occur until 11 years of age (Howlin & Asgharian, 1999). About half of these children with autism, conversely, are diagnosed by 5 or 6 years of age in the United Kingdom, while in the United States the average is 3–4 years of age for diagnosis (Filipek et al., 1999; Howlin & Moore, 1997). Baird et al. (2001) note a trend toward a mean diagnosis of 2.7 years. They describe this development as encouraging. We are more cautious, and would note that early diagnosis is a good development only if the diagnoses are reliable, have good predictive validity, and prove useful in assisting in better care and prognosis. At this juncture all of these points require further study and validation, although initial efforts are promising.

There are two issues that need to be overcome for early diagnosis based on behavioral phenomena versus genetic markers. The latter approach is some years off, and even if proven
successful, degree of impairment is likely to be determined by observations of behavior. The first
issue in evaluating observable behavior is the method of data collection. These procedures
primarily entail parental recollections and reports on childhood developmental milestones and
behaviors and/or videotapes taken before the child was diagnosed (Charman, 2000). Along with
this attempt to develop a diagnostic picture is the need to tease out what specific behaviors are
diagnostic. A second issue involves developmental course. Do symptoms appear in a linear
trajectory or do developmental spurts and behavioral regression serve as primary makers for ASD
children? We will address each of these issues next.

3. Infant behaviors that may be diagnostic of ASD

Given that autism and other ASD are routinely diagnosed at 3–4 years of age, moving the
bar to earlier ages may require considerable investigation. Osterling and Dawson (1994), for
example, stated that developmental patterns of ASD children in the first 18 months of life have
largely been a mystery. Initial efforts to describe variations in the behavior of children with
ASD compared to normally developing children were based on retrospective parent report
once a diagnosis had already been made. Some of these events had occurred as much as 4
years in the past (Dahlgren & Gillberg, 1989; Ornitz, Guthrie, & Farley, 1977). Thus, while
these data might help researchers focus on particular groups of behaviors, they are suspect in
terms of accuracy for a host of reasons as researchers attempt to get more specific regarding
symptoms.

A more recently adopted strategy to determine early symptom patterns has been the use of
home videos of the ASD child as an infant. Using tapes of regularly developing and autistic or
PDD-NOS children at 9–12 months of age, differences have been noted between groups on social
behavior, joint attention, and a few “autistic behaviors” (e.g., stereotypies, failing to orient,
covering ears). Additionally, sensory motor behaviors such as poor visual orientation/attention,
prompted/delayed response to name, excessive mouthing of objects, aversion to social touch,
unusual physical posturing, and visual staring and fixation on objects have been mentioned as
potentially being diagnostic (Baranek, 1999; Osterling & Dawson, 1994; Teitelbaum,
a very nice overview of this literature. One obvious limitation to this strategy is that the
videotapes cannot be standardized. The task, length of the tape, setting, number of verbal
prompts, number of people in the video and many other factors may vary. Furthermore, the
research in this area has been sporadic, and the data has not led to any systematic testing
procedures that would extend the ability to diagnose children with ASD or a subset of children
with ASD any earlier than age 2 (Charman et al., 2003). That age appears to be the most reliable
cut-off at present.

4. Children with regressive autism

An important diagnostic issue for young children that has been frequently acknowledged but
infrequently addressed is normal patterns of development up to 18–30 months followed by
marked regression. Within a short time interval, children may lose most or all of their speech,
regress in socialization, and develop “autistic-like” behaviors such as lack of eye contact,
fixation on objects, and ritualistic behaviors. The observation of this phenomenon is not new.
Ritvo (1978), for example, noted that speaking skills might develop normally until 2 years of age,
followed by rapid regression.
Wing and Potter (2002) make the important distinction between regressive autism and childhood disintegrative disorder. The “regression” in the later case is much more “catastrophic” than the regression in language and socialization as would be observed with regressive autism. However, most self-care and other basic skills are lost as well, making the overall skills set almost unrecognizable compared to the preregression phase. Nonetheless, for autistic children who do evince regression, making a diagnosis of ASD prior to age 2 is severely hampered. At this point, the number of people who exhibit regression and methods of predicting this regression are unknown. However, the general lore is that the regression occurs prior to age 2. At least one group of investigators has, as a result, pushed the age for screening from 18 months to 24 months as a means of enhancing reliability of diagnosis (Robins, Fein, Barton, & Green, 2001a). This change would seem to be at least a tacit acknowledgement that reliable and valid diagnosis prior to age 2 may be beyond the reach of investigation at this point.

Population studies are difficult with ASD. Some research on children who attended clinics and two epidemiology, population-based studies has been reported, providing limited samples of children that include regressive autism. Rates vary from 16 to 50 percent of diagnosed children with ASD who fall into the regression group (DeMyer, 1979; Fombonne & Chakrabarti, 2001; Kurita, 1985; Lingam et al., 2003; Lord, 1995; Tuchman & Rapin, 1997). The only study to date that specifically addresses the regression itself in a quantitative nature, was by Ozonoff, Williams, and Landa (2005). They obtained data on 60 autistic children by retrospective questioning of parents. Children were all 9 years of age or younger at the time of assessment. Regression was typically reported as occurring at 14–20 months. About half of those who regressed evinced symptoms of autism before regression. Thus, earlier identification than 2 years may be possible in the future. Nonetheless, more fine-grained analyses to establish various subtypes of early onset and the most relevant diagnostic indicators are needed before such an approach can be justified.

5. Follow-up of early diagnosis

Perhaps the strongest methodology developed to date confirming early diagnosis, is to follow the young children to see if there are consistent diagnosis labels for specific groups of children over time (Stone et al., 1999). A potential problem, particularly as more children are enrolled in early intervention programs, is whether such interventions markedly affect stability of diagnoses. If they do, this particular method of confirming early diagnosis may be markedly compromised in the future as these programs become more effective and widespread. However, to date, early intervention programs are not in play with enough children to invalidate the follow-up of early diagnosis. Therefore, we will briefly review some of the studies using this methodology.

Based on these data, children as young as 20 months may be reliably diagnosed, at least in the short term. Nonetheless, we do not know if such early diagnoses will hold up over time. Secondly, autism appears to be more reliably identified than PDD-NOS or Asperger’s syndrome. A third issue is that of regression. Given the period when regression occurs, it is highly likely that these persons were more difficult to detect, or were not detected at all. This phenomenon can occur because while children diagnosed with ASD were followed up, researchers have not assessed the rate of undetected children at age 20–24 months who may have developed autism or other ASD later. This question awaits future study but is as important as the false positives that are identified (classified as autistic but not confirmed at a later date).

Several preliminary studies have been conducted in recent years. Lord (1995) followed 16 children diagnosed at age 2 and found that 14 were still classified as autistic a year later.
Similarly, Baron-Cohen et al. (1996) diagnosed 10 children at 18–20 months and found the same diagnosis in all cases at 40 months. Stone et al. (1999) assessed 65 children (25 were diagnosed as autistic and 12 as PDD-NOS) with a mean age of 31 months (all were under age 3). They were followed up 1 year later. ASD was reliably diagnosed across time periods for most children, but it was more difficult to distinguish between PDD-NOS and autism. Eaves and Ho (2004) tested 49, 2-year-olds diagnosed with autism or PDD-NOS. The children were reexamined at age 4 1/2 with 88 percent still receiving a diagnosis of autism or PDD-NOS, and with the autism diagnosis being the more stable of the two. Better prognosis of PDD-NOS versus autism may in part account for these findings based on data presented by Charman et al. (2003).

6. Methods of identification and diagnosis

Some experts have flatly stated that there are no definitive diagnostic tests for autism (Wing & Potter, 2002). Nonetheless, the use of reliable and valid scales is considered indispensable for identification and diagnosis of ASD (Lund & Jensen, 1989; Matson, 2007b; Matson, Nebel-Schwalm, & Matson, 2007). With particularly young children, some researchers have hedged their bets by referring to their measures as methods of screening for at-risk children versus actually serving as a method for differential diagnosis (Baird et al., 2001; Dworkin, 1989). However, even where differential diagnosis is possible, scaling methods prove to be better at differentiating ASD from regularly developing children than they are at distinguishing between specific ASD such as PDD-NOS versus autism (Law, Boyle, Harris, Harkness, & Nye, 2000, 1998). Emphasis on initial screening has shifted to infants from birth to 2 years of age (American Academy of Pediatrics, 1994), and the belief that the etiology of autism in particular and ASD in general, have predominantly genetic causes has fueled speculation that such differential diagnosis may be possible in the future as biological variables are teased out and identified (Rutter, Bailey, Simonoff, & Pickles, 1997; Szatmari, Jones, Zwaigenbaum, & MacLean, 1998). Having said this, the genetic model is speculative at present. Currently, checklists and clinical observation still constitute the primary means of early identification of ASD and in practice are central in diagnosis, particularly for autism compared to other ASD.

Williams and Brayne (2006) categorize assessment into primary and secondary screening. A primary method would be brief and relatively universal and would identify a child at risk versus giving a diagnosis. Secondary screening would be selective, only those at risk would be evaluated, the number and comprehensiveness of the tests would likely be greater, and a diagnosis would be made. These researchers note that while a number of tests have been developed for primary screening, only three met criteria for population screening. A detailed list of diagnostic procedures is provided by these authors and others (Matson, Nebel-Schwalm, et al., 2007). For the purpose of this review, only the three instruments they designate will be reviewed. Other measures are available, but the methodology is more suspect (e.g., retrospective scoring of older children by parents based on how they remember their behavior at 14 months; Swinkles et al., 2006).

These three instruments are the Checklist for Autism in Toddlers (CHAT; Baron-Cohen, Allen, & Gillberg, 1992), the modified CHAT (M-CHAT; Robins et al., 2001a), and the Childhood Asperger Syndrome Test (CAST; Scott, Baron-Cohen, Bolton, & Brayne, 2002). Robins et al. (2001a) note that very young children are difficult to diagnose. Factors they posit for this problem are variation in symptoms across children, the limited peer interactions at young ages thus limiting the diagnosis of social and language deficits, limited suspicion of the disorder due to low incidence (we would add that low incidence means that most parents and professionals
are unfamiliar with relevant symptoms), and the fact that motor milestones are usually not affected. They argue, and we agree, that scaling methods become all the more important under these conditions but may not be consistently reliable and valid until 24 months of age.

6.1. CHAT

This measure was developed as a primary screen for children at 18 months. The CHAT can be completed in 5–10 min. The initial study was in a “high-risk” sample of 50 eighteen-month-olds and 41 eighteen-month-old siblings of autism (Baron-Cohen et al., 1992). The test was useful in identifying those at risk for autism and was a useful predictor of children likely to be diagnosed as autistic 30 months later. In a second study, 235 sixteen-month-old children were screened with the test and a subset of those at risk were followed up 20 and 42 months later. Moderate success in identifying autistic children was noted (Baron-Cohen et al., 1996).

6.2. M-CHAT

The authors of the M-CHAT indicate that it extends the original CHAT in several ways. First, they added 21 items to the original 9. The rationale was to extend the checklist symptoms to a broader range of ASD children and to compensate for eliminating the home health visitor’s observation section of the CHAT. They also normed the scale on a US population and conducted screening at 24 months versus 18 months. These are a substantial number of differences and make the scales quite different even though the titles of the measures are similar. They screened 1,293 children through 88 pediatric offices. A somewhat troubling aspect of the study was that the content of the scale was modified halfway through the study; eight items were dropped and one was added. The authors found that the M-CHAT was a reliable and valid means of identifying autistic children, and that the 23-item M-CHAT was more accurate than the nine initial CHAT items. Although some controversy exists among the authors of the two measures (Charman et al., 2001; Robins, Fein, Barton, & Green, 2001b), all researchers from the two teams do agree that the screening process has value but that much more research is needed.

6.3. CAST

Two mainstream school-based samples have shown good specificity and sensitivity with this 31-item measure of Aspergers Syndrome (Scott et al., 2002; Williams et al., 2005). However, it is noted that many false positives arose, thus it may not be a suitable screening instrument (Williams & Brayne, 2006). Asperger’s syndrome as a separate and distinct condition from autism has come into question. Furthermore, the symptoms tend to appear later in life than autism, per se. Thus, difficulty in screening, even where the children were a bit older than 2 (3–9 years of age, as was the case here), should not be particularly surprising. Thus, which of these three measures or some other measure comes to the forefront for early identification is open to question. The big need is a programmatic line of investigation on one scale to systematically address relevant psychometric questions. This task has not been done and appears to be some time off.

7. Conclusions

An important aspect of research in this area is to proceed with caution and compile ample research evidence before drawing conclusions. For example, Bristol-Power and Spinella (1999)
note that “dramatic advances have been made in behavioral diagnostic criteria, lowering the potential age of diagnosis from 5 years to 18 months” (p. 436). While the authors clearly use the word “potential”, the meaning appears to be that reliable and valid diagnosis at 18 months is possible. However, more recently Williams and Brayne (2006) state that based on the existing evidence, screening for autism spectrum disorders is not recommended. We emphasize that there is likely no magic time cut-off for early identification. Children advance in spurts developmentally, which can vary from child to child. More severe symptoms will likely allow for earlier detection than for children with a fewer number or milder symptoms of a particular ASD. Additionally, it appears that autism can, as a general rule, be detected earlier than PDD-NOS or Asperger’s syndrome, although even these dichotomies are far from being totally clear or independent from each other at this time. Along these same lines, at least up to 3 or 4 years of age, the reliability and specificity of diagnosis increases with respect to type and severity of ASD and severity of given symptoms. Given that the previous assumption is accurate, clinically it may make more sense to wait until a highly accurate diagnosis is possible (at an older age, around 24 months at the earliest at present but perhaps as late as 4 years)? More systematic research on defining the three most prevalent ASD is needed along with how symptoms change over time and are interwoven with the establishment of cut scores and cut ages for diagnosis. Additionally, the issue of symptom regression at approximately 1 1/2 to 2 years needs to be studied more carefully. Several important questions need to be considered in this regard. Are there specific symptoms or profiles that predict regression? How many ASD children evince regression and when? Do the quantitative and qualitative aspects of regression vary by type of ASD or the presence of comorbid conditions such as intellectual disability, psychopathologies, or behavior problems? Are children who regress of a different genotype than those who do not? All these issues, and certainly others, need to be addressed before researchers and clinicians can seriously consider definitive answers about when and how children can be identified with ASD at the earliest point in time.

The prevailing wisdom appears to be that early identification is good, and the earlier the better (Swinkles et al., 2006). However, the arguments put forth, while compelling, are largely unsubstantiated by data. Researchers need to establish if early identification, for example, results in better family support. We doubt that is possible unless systems have been built and are readily available to provide this service. Additionally, what is family support and what are the best empirical methods to provide it? This area alone needs considerable experimental development. The same could be argued regarding the “timely planning of educational objectives”. What constitutes timely, who determines it, how does it interface with the individual diagnostic picture of a particular child, what are the goals of these objectives and how are they measured? Unfortunately, this series of practical issues is pervasive with respects to most arguments regarding early identification. As previously noted, early intervention treatments, particularly those using applied behavior analysis, constitute about the only viable argument supported by data at this time. And, even with this area, many questions remain. For example, how much treatment per week is enough and are some skills (“pivotal responses”) really more critical than others and if so how are these skills selected? Additionally, how critical are the pivotal responses, for whom, and how much effort would be needed to improve these responses relative to others in the child’s repertoire? All these questions and many more need earnest consideration and systematic study and evaluation.

References


