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Autism Spectrum Disorders: Identification and Diagnosis

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Autism spectrum disorders (ASD) are a complex group of related disabilities marked by impairments in communication and socialization, a limited range of interests, and the presence of repetitive behaviors. Although often not diagnosed until preschool age, ASDs develop early in life and are generally lifelong, with implications for education, social development, and community adjustment.

Autism became a federal special education disability category in 1991. Since that time, an enormous amount of information has become available for parents and teachers and has resulted in a broader definition of autism, often referred to as autism spectrum disorder. Today there are better tools to identify the disorder, and schools are often the primary source of referral for evaluation. Parents and educators can acquire the knowledge and skills they need to support children at home and at school. As more research yields new information, parents and teachers must stay informed and work together as a team to help children achieve positive learning outcomes.

ASD is diagnosed based on behaviors, not medical tests. In order to accurately diagnose ASD, the child should have a comprehensive evaluation by professionals with expertise in identifying differences in language, behavioral, social, and cognitive skill development. An accurate and early diagnosis of ASD will enable parents to gain an understanding of their child and his or her specific needs while the child is very young. Specialists can reliably diagnose children at age 2.

BASIC FACTS ABOUT AUTISM SPECTRUM DISORDERS

Autism affects as many as 1 out of 500 children. When the other two autism spectrum disorders are included, Asperger syndrome and pervasive developmental disorder not otherwise specified (PDD-NOS), that number rises to as many as 1 out of 150 children. Although some individuals with ASD may make significant improvements over time, relative weaknesses in social and communication skills remain.

Causes of ASD

The cause or causes of ASD are unknown. Most experts hypothesize that autism is the result of many factors that impact brain development before or shortly after birth, possibly in varying combinations at different levels. These include genetic, environmental, and other factors. ASD is not the result of parenting style.

Characteristics of ASD

Socialization, communication, and restricted patterns of behaviors and interests are the core areas of impairment in ASD. Children can meet different combinations of the diagnostic criteria. Children with autism have problems in all three core areas and lack the capacity to understand social behavior. Children with Asperger syndrome do not meet the full criteria for autism, do not have a history of early language delay, and have average to above-average intelligence, but with significant problems in socialization and restricted patterns of interests. These children may not be identified until they enter elementary school or later. Children with PDD-NOS have problems in socialization and one or both of the two other areas (communication or restricted patterns of behaviors and interests), but not enough symptoms to meet criteria for autism.

In addition to the meeting the diagnostic criteria for ASD, many children may also have intellectual impairment and/or other disorders such as fragile X syndrome or tuberous sclerosis. Similarly, children with ASD may have features of other disorders, such as hyperactivity associated with ADHD, anxiety, or behaviors that may be associated with obsessive-compulsive disorders.

General characteristics of children with ASD often observed by parents and teachers include the following:

Cognitive. Uneven development of cognitive skills, with relative strengths in processing visual/nonverbal information and rote learning, and relative weakness in processing verbal or abstract information.

Social skills. Difficulty understanding social rules such as taking turns and sharing, perceiving and understanding the emotions of others, taking the perspective of others, initiating and maintaining interactions, responding to the social overtures made by others, and having appropriate conversations with others.

Communication. Difficulty responding to verbal information presented at a fast pace, understanding multiple-step commands, and expressing wants and needs; inconsistent understanding of verbal information; a need for verbal information to be repeated, particularly new information.

Organization/self-direction. Difficulty screening out distractions, completing activities independently, initiating work activities, organizing free time, stopping one activity and moving on to the next, being flexible, shifting attention to a new focus, sharing another person's focus of attention, and doing more than one thing at a time.

Red Flags: Early Signs of ASD

Although the early signs of autism may be difficult to detect before age 2, the American Academy of Pediatrics has recommended that pediatricians screen for ASD at ages 18 and 24 months as part of routine health visits. Red flags that, when observed, should result in immediate referral to the early intervention system (for children birth to 3 years) or the school system (3 years and older) for further evaluation (Filipek et al., 1999) include:

- By 12 months, not babbling or gesturing (pointing, waving)
- By 16 months, not using single words meaningfully
- By 24 months, not using two-word spontaneous phrases (not just repeating)
- Any loss of language or social skills at any age

EVALUATION AND DIAGNOSIS

Early and accurate diagnosis of ASD is critical to long-term positive outcomes. Educators are often aware of potential drawbacks to labeling children, such as reduced expectations for learning or a lack of correspondence between a label and effective interventions. For children

with ASD, however, the presumed stigma of labeling is outweighed by the clear benefits of a diagnosis.

The label of autism serves many purposes. It helps professionals and families communicate, allows children to access specialized intervention approaches, provides a basis from which treatment and prevention research can occur, leads to appropriate intervention and program planning, and provides a framework for gathering information on outcome, causes, and associated problems. A label allows teachers and parents to become informed because it gives them the basis to gather information, read, join support groups, advocate, and become organized in their efforts to obtain resources and improve outcomes. Thus, when it is suspected that a very young child shows possible red flags associated with ASD, it is necessary to refer the child immediately to specialists for an evaluation.

The Diagnostic Evaluation

When concerns are raised about the development of a young child, including the presence of the above red flags or failing an autism screener, the child should be referred for evaluation and early intervention services, preferably before age 3.

The evaluation. A formal diagnostic evaluation should be performed by a trained experienced professional or multidisciplinary team of professionals who utilize a standardized assessment instrument (such as the Autism Diagnostic Observation Schedule [ADOS]) as part of a comprehensive assessment. Detailed developmental history and information about autism symptomatology from the parent are also needed. The use of a standardized parent questionnaire (such as the Social Communication Questionnaire [SCQ]) is also recommended.

Diagnostic and educational classifications. Note that there may be differences between a medical diagnosis of ASD and an educational classification of ASD. Schools must provide services for all children with disabilities, including ASD, beginning in early childhood (age 3 or earlier, as defined by state regulations). Two autism spectrum disorders, Asperger syndrome and PDD-NOS, are not recognized education disability categories. Some states allow these students to receive services using the autism eligibility category or a different category such as speech-language impaired or other health impaired. Young children (usually under age 9, depending on state regulations) might also be served under a noncategorical early childhood disability umbrella. Because of the significant problems in social interaction development common to all autism spectrum disorders, it is recommended that children with

any ASD be eligible for special education services under the category of autism.

The symptoms associated with an ASD diagnosis in very young children can be subtle and sometimes difficult to distinguish from developmental delays. Longitudinal research studies have also demonstrated that the symptoms observed in very young children change over time, but for children diagnosed with autism, the label is usually stable. It is therefore recommended that children under age 3 who are given a diagnosis of PDD-NOS, or deemed to be at risk for a later diagnosis of autism, be reevaluated within 1 to 2 years of initial diagnostic evaluation.

Reporting results to parents. The results of the diagnostic evaluation should be carefully discussed with the parents, with a focus on helping them to understand the meaning of the diagnostic terms, the differences between a medical diagnosis and an educational classification, the unique strengths and weaknesses of their child, and their role in helping their child succeed.

NEXT STEPS: INTERVENTION PROGRAMS

A primary reason for a diagnosis is obtaining specialized interventions. Effective intervention programs address the core characteristics of ASD described earlier (National Research Council, 2001). The handout "Autism Spectrum Disorders: Intervention Options for Parents and Educators" (Ruble & Akshoomoff, 2010) provides an overview of interventions and next steps following a diagnosis. Other helpful resources can be found below.

REFERENCES

Filipek, P. A., Accardo, P. J., Baranek, G. T., Cook Jr., E. H., Dawson, G., Gordon, B., et al. (1999). The screening and diagnosis of autistic spectrum disorders. *Journal of Autism & Developmental Disorders*, 29, 439-484.

National Research Council. (2001). *Educating children with autism*. Washington, DC: National Academy Press. Retrieved July 28, 2009, from <http://www.nap.edu/books/0309072697/html/R1.html>

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RECOMMENDED RESOURCES

Print

Hall, L. J. (2008). *Autism spectrum disorders: From theory to practice*. Upper Saddle River, NJ: Merrill.

Ozonoff, S., Sawson, G., & McPartland, J. (2002). *A parent's guide to Asperger syndrome and high-functioning autism*. New York: Guilford Press.

Online

Autism Resources: <http://www.autism-resources.com>

Autism Society of America: <http://www.autism-society.org>

Autism Speaks, *Diagnosis/early intervention*: http://www.autismspeaks.org/community/resources/resources_diagnosis.php

Center for Disease Control and Prevention, *Autism spectrum disorders*: <http://www.cdc.gov/ncbddd/autism/facts.html>

First Signs: <http://www.firstsigns.org>

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