

Beyond Behavior—

Autism Spectrum Disorders: Beyond Behavior

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The autism spectrum disorders (ASD) are behaviorally defined disorders characterized by impaired social interaction, delayed and disordered language and isolated areas of interest. Symptoms can vary over time and with functional level but may include poor eye contact, insistence on sameness, atypical cognitive development, repetitive and stereotypic behaviors, deficits in joint attention and a normal physical appearance.

Although the cause of ASD remains unknown, there is strong evidence that genetics plays a significant role. Although once considered rare, current prevalence rates suggest that one in every 166 children may be affected with ASD (Fombonne, 2003).

Since the initial description of infantile autism by Dr. Leo Kanner in 1943, much of the clinical research related to the ASDs has centered on investigations of cognition, behavior, social skills and language, with relatively little attention to the possible significance of associated medical conditions. Physical examination of ASD children can be challenging and often is limited by poor patient cooperation and difficult office behavior, as well as the fact that many of these children are nonverbal and therefore unable to describe or localize discomfort. In addition, some of these children may present with symptoms that are atypical and whose causes are not easily recognized. Research suggests that children with ASD are less likely than those with mental retardation or other special needs to obtain specialty medical care.

The fact that a child has autism does not rule out the possibility that he may have one or more other illnesses or disorders, similar to those that affect typically developing children.

Failure to diagnose and treat these disorders may compromise the child's ability to function in a classroom and to take advantage of therapeutic services; negatively impact quality of life for the child and his family; and, in some cases, even lead to hospitalization and perhaps death. In addition, identifying associated medical disorders may amplify the phenotypic description (observable traits or characteristics of a living being) of subsets of ASD children, and defining these subsets may have genetic implications.

Space does not allow a detailed description of the multiplicity of medical conditions that may affect the child with autism. Therefore, only some of the more common disorders will be highlighted here. These include seizures, sleep disturbances and gastrointestinal disorders, with brief comments regarding metabolic, urologic and hormonal dysfunctions. This is not an all-inclusive list, and the primary care physician and specialist working with ASD children must remain alert to a wide range of medical possibilities at any one time.

Seizure Disorders. Seizure disorders are said to affect approximately one third of people with ASD at some time during their lives, with peak risk periods in early childhood and during adolescence (Volkmar and Nelson, 1990). No one seizure type has been reported to be specifically associated with autism, and most electroencephalographic (EEG) and seizure patterns have been observed in ASD.

As with typically developing children, if seizures are suspected, appropriate diagnostic procedures, including the performance of an EEG, should be implemented, the type(s) of seizures identified and treatment started. In some cases,

the onset of seizures may signal the need for more extensive evaluations and specialty referrals to rule out underlying metabolic disorders, syndromes, degenerative disorders, head trauma or mass lesions.

Atypical behavioral patterns and body movements often can be observed in children with autism, complicating the ability to accurately diagnose a potential seizure disorder. But not all body movements or mannerisms observed in ASD children are seizure-related. Some may be related to other medical conditions, such as gastroesophageal reflux disease (GERD) or other gastrointestinal disturbances (Buie, 2005). Thus, carefully analyzing the behaviors of concern is critical to targeting the most appropriate and, therefore, effective treatment. Since the child does not usually exhibit the concerning behaviors during the office visit, ask the family or the school to obtain a videotape of these events for diagnostic purposes.

Sleep Disorders. Sleep disorders are said to occur in approximately 30 percent of typically developing children and appear to be more common in early childhood (Ferber, 1996). Sleep disturbances include delayed sleep onset, frequent nighttime awakenings, sleeping too much, nightmares or night terrors. Among children with autism, parents most frequently report difficulty getting to sleep, frequent nighttime awakenings and/or early morning arousals followed by the child remaining awake for the day.

While the causes of sleep disturbances may be related to central nervous system dysregulation of arousal and/or abnormal REM sleep patterns, physicians and families should consider the potential contribution of enlarged tonsils and adenoids, or gastroesophageal reflux. Urinary tract infections associated with nocturnal enuresis (bed-wetting) also could contribute to nighttime awakenings. Because there is growing evidence that disordered sleep can negatively impact daytime behavior and learning, it is important to determine the cause of the sleep disturbance and treat the underlying condition.

Gastrointestinal Disorders. Parents frequently describe gastrointestinal disorders—usually diarrhea, chronic constipation, food intolerances, gas, bloating and abdominal pain/discomfort—as issues for children with ASD. However, the percentage of children with autism suffering from GI disorders is not known; nor is it known whether these disorders are more common in ASD than in typically developing children. Disorders such as celiac disease, gastroesophageal reflux, colitis, esophagitis, gastritis, food allergies and motility dysfunction have all been reported in those with ASD.

Although typical GI symptoms often are apparent in some children with ASD, others may present with episodes of aggression and/or self-injurious behavior (SIB) without evidence of GI symptoms. These behaviors are observed most frequently in lower-functioning children who are nonverbal and who have no other means of expressing their discomfort or pain. Well-designed research is needed to define the prevalence of GI disturbances in ASD, the types of disorders most often found, and the signs and symptoms with which these children present. It is important to consider gastrointestinal dysfunction in ASD, especially in those children who are nonverbal or hypoverbal, and who have developed behavioral outbursts without obvious cause.

Metabolic Disorders. Metabolic disorders only recently have become a potentially important area of investigation in ASD.

Several reports have suggested, for example, an association between ASD and mitochondrial disorders (Oliveira et al., 2005; Miles et al., 2005). Possible clinical "red flags" that may suggest such a diagnosis include low muscle tone, easy fatigability and poor physical endurance, and repeated regressions. If there is a suspicion of an underlying metabolic disorder, a referral to a medical geneticist should be considered, since some of these disorders are treatable.

Hormonal Imbalance. Hormonal imbalance has been found in some children with autism, most often during preadolescence and adolescence. Precocious puberty has been reported in both ASD boys and girls. Behavioral disruptions that seem to have a relationship to the onset of the menstrual period should suggest the possibility of disordered estrogen/progesterone levels and a referral to an endocrinologist could be beneficial.

Other Concerns. Other health care concerns include recurrent ear infections, hearing impairment, urinary tract infections,

spastic bladder leading to new onset of bedwetting at any age, attention deficit hyperactivity disorder, disordered sensory processing and almost any other illness commonly seen in typically developing children.

Regardless of the challenging behaviors with which many ASD children present, the physician must remain mindful of the fact that these children may have any number of common childhood illnesses and disorders, but their presentation may be atypical and thus may create a diagnostic dilemma, especially in very young children and in those who are nonverbal. However, many of these medical conditions are treatable, and effective diagnosis and intervention can substantially improve the child's daytime behavior, his attention and ability to learn, and his overall quality of life as well as that of his family. Quality health care should be considered a high priority for all children with autism. Their futures may depend on it.

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