

Autism Spectrum Disorders – Beyond Behavior

Margaret L. Bauman, MD

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 Kanner in 1943, much of the clinical research related to the ASDs has largely 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 is often limited by poor patient cooperation and difficult office behavior, the fact that many of these children are non-verbal and therefore unable to describe or localize discomfort, and the possibility that at least some of these children may present with symptoms which may be 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 fact 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, take best advantage of therapeutic services provided, negatively impact quality of life for the child and his family and in some cases, could lead to hospitalization and perhaps death. In addition, identification of associated medical disorders may amplify the phenotypic description of subsets of ASD children, and delineating these subsets may have genetic implications.

Space does not allow a detailed description of the multiplicity of possible medical conditions that may affect the child with autism. Therefore, only some of the more common disorders will be highlighted herein. These will include seizures, sleep disturbances and gastrointestinal disorders, with brief comments regarding metabolic, urologic and hormonal dysfunctions. This is not an all inclusive list, and it goes without saying that 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 are said to affect approximately one third of individuals 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 begun. In some cases, the onset of seizures may signal the need for more extensive evaluations and specialty referrals to rule out the possibility of underlying metabolic disorders, syndromes, degenerative disorders, head trauma or mass lesions previously unsuspected.

Atypical behavioral patterns and body movements can often be observed in children with autism, complicating the ability to accurately diagnose a potential seizure disorder. It is worth stating at this point that all body movements and/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, it is critical to carefully analyze the behaviors of concern in order to target the most appropriate and therefore effective treatment. Since the child does not usually exhibit the concerning behaviors during the office visit, it can often be informative to ask the family and/or the school to obtain a videotape of these events for diagnostic purposes.

Sleep disorders are said to occur in approximately 30% 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 and remaining up for the day. While the causes of sleep disturbances may be related to central nervous system dysregulation of arousal and normal 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 could also contribute to nighttime awakenings. Because there is growing evidence that disordered sleep can negatively impact on daytime behavior and learning, it is important to determine the etiology of the sleep disturbance and treat the underlying causative condition.

Gastrointestinal Disorders are frequently described by parents, usually diarrhea, chronic constipation, food intolerances, gas, bloating, and abdominal pain/discomfort. However, the percentage of autistic children suffering from GI disorders is unknown 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 ASD. Although typical GI symptoms are often apparent in some ASD children, 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 non-verbal and who has 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, most especially in those children who are non-verbal or hypoverbal and who have developed behavioral outbursts without obvious cause.

Metabolic Disorders have only recently become a potentially important area of investigation in ASD. Several reports have suggested an association between ASD and Mitochondrial Disorders for example (Miles et al.,). 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 has been found in some autistic children, most often in 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 health care concerns include recurrent ear infections, hearing impairment, urinary tract infections, spastic bladder leading to new onset enuresis at any age, attention deficit hyperactivity disorder, disordered sensory processing, and/or almost any other illness commonly seen in typically developing children.

Regardless of the challenging behaviors with which many ASD children may present, the physician must remain mindful of the fact that these children may have any number of common childhood illnesses and disorders but that their presentation may be atypical and may thus create a diagnostic dilemma, most especially in very young children and in those who are non-verbal. 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 children with autism. Their future may depend on it.

Bibliography:

Buie, TM. Gastrointestinal issues encountered in autism. In: Bauman, ML and Kemper, TL, eds. The Neurobiology of Autism. Johns Hopkins University Press: Baltimore. Pp. 103-117, 2005.

Ferber, R. Childhood sleep disorders. Neurologic Clinics. 14:493-511, 1996.

Fombonne, E. The prevalence of autism. J. Am Med Assoc. 289:87-89, 2003

Kanner, L. Autistic disturbances of affective contact. Nervous Child. 2: 217-250, 1943.

Volkmar, FR and Nelson, DS. Seizure disorders in autism. J. Amer Acad Child Adolesc Psychiatr. 29:127-129, 1990.