AUTISM IN INFANTS AND TODDLERS

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Angel was referred for language delay. At 24 months, he had not yet begun to speak. He had no way to communicate his wants other than crying. His parents also felt that he didn’t seem to understand well. Parents found it difficult to get Angel’s attention. He seldom looked when called. His parents wondered if he could hear. Angel also had limited interest in other children and little interest in toys. He continued to put a lot of objects in his mouth and to seek out the wheels on his toys to explore.

Eric, age 21 months, was very confusing to his parents. He spent hours each day running back and forth in their small apartment. He appeared quite bright, able to turn on the VCR from a young age and open any child safety latch. He could recognize the alphabet and even a few words. He had an excellent memory and was able to recite extensive dialogue from his favorite videos. However, he still did not say “mommy” and only rarely used words to make his wants known. He tended to walk on his tiptoes. His parents agreed that he was very bright. His mother was concerned that he spent too much time “in his own world” and sought evaluation.

Luis was developing well for his first year of life. His mother described feeling confident that Luis would speak early since he was such a vocal and imitative infant. When Luis was 14 months of age, a young sibling was born and the family moved to a larger apartment. In retrospect his parents felt that Luis changed at that time. He stopped waving bye-bye and playing patty-cake. He was often irritable and difficult to soothe. Now at 18 months they find that he’s “in his own world” with little interest in the adults and children around him.

Angel, Eric, and Luis, although quite different, have several things in common. They are not developing language or communication abilities as expected for age. They are not as social as is typical. They display some unusual interests, and may lack interest in the toys that interest their peers. They share the typical findings of toddlers with autistic spectrum disorders (ASDs). Probably, though, the most common finding in young children diagnosed with ASDs is that their parents are generally quite confused. They may see their child as progressing well, even advanced in some regards and behind in others. The child may use language at times if he chooses to, but not as reliably as other children. The child is often selectively responsive, and may not respond when called by name, but may come running when a favorite television commercial jingle comes on the air.

Although toddlers with ASDs are rarely frankly manneristic\(^1\) (e.g. displaying spinning, hand movements, atypical gaze patterns) they do tend to engage in repetitive motor activities such as running back and forth, and playing with parts of objects such as the wheels of toy cars or the door of a toy doll house.

**Diagnosis**

The DSM-IV, the Diagnostic and Statistical Manual of Mental Disorders, defines the pervasive developmental disorders, also known as the autistic spectrum disorders, as a group of conditions characterized by three core deficits: impairment in social interaction, impairment in communication, and the presence of repetitive and stereotyped patterns of behavior, interests, and activities. The diagnosis of autistic disorder (or autism) is given when an individual meets at least six of a set of formal diagnostic criteria across all three core domains. This condition generally applies to the most severely affected individuals. When qualitative impairments in each of the three areas of deficit are present without formally meeting the diagnostic criteria for autistic disorder, a diagnosis of pervasive developmental disorder – not otherwise specified (PDD.NOS) may be given.

In the youngest children coming for evaluation of language and social/behavioral concerns, it is common to utilize the general category of autistic spectrum disorder for diagnostic purposes since some of the clinical findings such as mannerisms may unfold as the child matures. This developmental progression can make it difficult to distinguish between autistic disorder and PDD.NOS in young children. In this instance, a child who does not originally meet formal criteria may go on to meet formal criteria for autistic disorder at a later date. The use of the diagnostic category of autistic spectrum disorder (ASD) in the toddler population takes into account the complexities of making a diagnosis in a developmental moving target.

The average age of diagnosis for children with autism is reported to be over 4 years of age. Yet, 50% of parents of a child with autism report that they suspected a problem before their child was 1 year of age and most parents initially express concern to their pediatrician by the time their child is 18 months of age\(^2\). With the advent of early intervention and the findings that young children benefit greatly from intervention, especially when it is begun before 30 months\(^3\), there has been a thrust to identify autism in younger and younger children.
From a clinical perspective, the question then becomes: how early can autism be diagnosed? The data support the idea that signs of autism are present from a very young age in the majority of children. A look at videotapes of first birthday celebrations in children subsequently diagnosed with autism showed that these 1-year-olds could be distinguished from their unaffected peers in four ways: lack of eye contact, failure to orient to name, lack of pointing behaviors to indicate wants, and lack of showing behaviors.

The emphasis next turns to the reliability of the diagnosis between clinicians for this age group and the stability of the diagnosis over time in young children. Reliability refers to the likelihood that a child would be given the same diagnosis by another qualified evaluator. The reliability of the diagnosis has been found to be quite good in children by the age of 2 years when evaluated by experienced clinicians with experience with the toddler population. The diagnosis of autism, the condition found in the most severely affected children, was more reliable than differentiation between the various autistic spectrum disorders. Similarly, the diagnosis of autistic disorder was found to be quite stable whereas the milder autistic spectrum disorder diagnosis of PDD.NOS was found to be a less stable diagnosis at the age of 2 years.

**Multidisciplinary Evaluation**

Since the diagnosis of autistic spectrum disorder (ASD) has profound implications for a child’s future, evaluation of the young child with language and social delays should always include a multidisciplinary assessment. A neurodevelopmental examination includes a physical examination which may highlight certain medical conditions known to be among the possible causes of autistic spectrum disorder, such as tuberous sclerosis and Fragile X. An audiologic assessment looks for the presence of a hearing impairment which may affect the child’s development of language and responsibility. The psychological assessment looks at the child’s intelligence, as cognitive impairment is commonly associated with autistic spectrum disorders. The speech and language evaluation can document delays as well as atypical features in the language development. These assessments taken together allow the diagnosis to be made based on several separate observations of the child by clinicians with differing but relevant clinical perspectives.

The Infant and Toddler Team of the Children’s Evaluation and Rehabilitation Center (CERC) sees children birth through 3 years of age. Roughly 20% of the 300 children evaluated each year are given a diagnosis along the autistic spectrum. The diagnosis is made with a multidisciplinary approach involving a developmental pediatrician, psychologist, speech and language pathologist, audiologist and special educator. The diagnosis is a clinical one utilizing standardized instruments to aid in the diagnostic process: the Checklist for Autism in Toddlers (CHAT), Childhood Autism Rating Scale (CARS), and DSM-IV criteria. Later, the team’s diagnostic impressions are shared with the family in a parent conference with one of the team members. In addition to sharing diagnostic impressions, the team makes a set of recommendations to the family. These recommendations often involve a referral to the Early Intervention program to make arrangements for a therapeutic preschool program, intensive speech and language therapy and occupational therapy, parent support and parent/child dyadic work for behavior management. Some children with autistic spectrum disorders are also recommended to receive intensive home-based services with an approach such as applied behavioral analysis. Children may be offered interim speech and language or parent-child dyadic therapy to address behavior management at CERC while they await more intensive services at home or school.

**Developmental Follow-up**

Follow-up of children diagnosed early is critical to the care we provide, and offers many benefits to children and families. In this age group, especially, it is important to reassess the child every few months for diagnostic purposes. There are children who do very well with intervention and may no longer warrant the diagnosis of an autistic spectrum disorder by school age. It is unclear whether this population was “cured” by intervention or whether the original diagnosis made in the toddler was an unreliable one. Other children may require an increase or change in their program in order to progress optimally. The follow-up visit presents an opportunity to advocate for the child’s needs and provide feedback to the parent regarding the child’s progress and current therapeutic needs. At follow-up appointments, other treatment modalities can also be put into place, such as making orthotics for the shoes to treat the common complaint of toe-walking in children with ASDs. Since children with ASDs often eat a highly restricted diet, due to their dislike of certain textures, they may be referred to a nutritionist to help them achieve a more typical diet. The use of medications for treatment of target symptoms can be discussed with the family over time. Parents may also be guided in how to approach the deluge of alternative treatment modalities offered through the media. Approximately 20% of children with ASDs have a history of language and/or social loss as seen in the description of Luis. It is recommended that such children be evaluated by a child neurologist with an assessment that includes a 24 to 48 hour EEG to look for the presence of subclinical seizure activity in the language portions of the brain. Such abnormalities may be treated with anticonvulsant therapy, with subsequent improvement in language and social functioning.

**Conclusion**

Making the diagnosis of autistic spectrum disorders in toddlers represents a challenging but worthwhile clinical activity. There are data suggesting that a reliable and valid diagnosis can be made in infants and toddlers. There are data to suggest an improved outcome for children diagnosed early who receive an appropriate, intensive therapeutic program. Here at CERC we continuously work to hone our diagnostic skills and expertise in this dynamic diagnostic and treatment area.
References


