

Speaking of Autism Part II: Diagnosing Autistic Spectrum Disorders

Among parents autistic spectrum disorders (ASD) generate more fear than perhaps any other threat to our children's wellbeing. Last month we reviewed current scientific thinking about how autism develops, looking at what we've learned (some), what we're in the process of learning (a lot), and what early theories have fallen by the wayside (the most popular ones). Today we'll look at the rapidly growing science of diagnosing ASD.

Why diagnose autism early?

Many developmental abnormalities stem from conditions that can be treated. The first reason to get an autism diagnosis is to look for an underlying cause such as seizure disorder, fragile X syndrome or Rett Syndrome. Second, early intervention does affect outcome in ASD, as we'll discuss in detail next month. Third, siblings of autistic children are at higher risk of having ASD, which may factor in parents' future reproductive choices.

How early can you diagnose ASD?

Many children are diagnosed around age two, sometimes even later. But often parents become concerned much earlier, typically around 15 to 18 months of age when most children start talking. Speech delay is the most obvious clue for identifying children with ASD. By age eight to ten months most children recognize their names and respond when called. Children who don't respond to their names may have hearing impairment, ASD, or other developmental delays.

Most children with ASD start talking late. Around a quarter to a third of autistic children begin using some words, then stop, usually between 15 and 24 months of age. When they do speak it may be scripted, including phrases or sentences from TV shows or videos. Another common feature of autistic speech is echolalia, an automatic repetition of words. Toddlers may imitate words and sentences normally, but a speech-language pathologist can usually differentiate between normal copying and echolalia.

According to the Child Neurology Society the following "red flags" should prompt an evaluation for autism:

- No babbling or pointing or other gesture by 12 months
- No single words by 16 months
- No 2-word spontaneous phrases by 24 months
- Loss of language or social skills at any age

What about non-verbal communication?

Some of the most intriguing studies of autism in infancy involve a form of non-verbal communication called Joint Attention (JA). Joint attention is not, as you might think, the act of staring at your knees. JA is the process of taking enjoyment in sharing an event or object with another person. It can best be summed up as, "Hey, Mom, check that out!" Infants display this behavior by looking back and forth between the person and the object of interest. Researchers analyzing home videos have identified deficits in joint attention long before other signs of autism become obvious.

Pointing is another form of non-verbal communication that provides an early red flag for ASD. Around ten to twelve months of age children develop the ability to follow a point. When a parent points and says, "Look," these children instinctively turn their attention to the object or event of interest then back to the parent in order to share an emotional response. Children with ASD, on the other hand, may not look even when parents use a loud voice or physical direction like tapping them on the shoulder. Likewise children with ASD are less likely to point things out to their parents. The idea that another person might share a response to a stimulus is part of what seems to be missing in ASD.

Are motor skills impaired in autism?

Motor skills (crawling, walking, climbing) may be delayed in the most severe form of ASD, pervasive developmental delay. But many autistic children display average or above-average motor skills. They may be especially independent, helping themselves to food or toys other children might ask an adult to get for them. Autistic children will play with toys, but they often seem more interested in the parts of the toy than in the toy as a whole. A child with ASD may spin the wheels on a car rather than race it along the floor. Nurturing play, like feeding a baby doll or hugging a teddy bear, is usually absent. Stereotypies are repetitive self-stimulatory behaviors such as rocking or hand-flapping. These movements are a late sign of autism, usually emerging around age three years or later.

Should my child be screened for autism?

Developmental surveillance is part of what your child's doctor does at each wellness exam. Additionally the American Academy of Pediatrics recommends universal developmental screening using a standardized tool like the Ages and Stages exam at ages 9, 18, and 24 to 36 months. But these tests are not specific for autism, so they also recommend an autism-specific screen like the Modified Checklist for Autism in Toddlers (M-CHAT) at 18 and 24 months. Any child should be screened whenever a parent expresses a concern about development that may suggest ASD.

What if the screening exam comes back positive?

Screening exams are designed only to identify people who are at high risk for certain problems. They are not diagnostic tools. The diagnosis of ASD requires a much more thorough evaluation, often involving a team of child specialists and a battery of developmental tests.

Are there labs or studies all children with ASD should have?

Only hearing testing is universally recommended. Other tests, including genetic studies, EEG, and MRI, should be ordered based on the findings of a thorough history and physical exam. Labs such as heavy metal screening in hair or urine are notoriously unreliable and have no known role in the management of ASD. There is a tremendous amount of ongoing research on autism, and any number of tests may be done as part of a research trial, but no child should be enrolled in a study without a thorough and formal informed consent process. Even labwork comes with risks and benefits, and investigators are obligated to make sure parents understand both.

Next month: Treating autism