

Given the potential benefits of early diagnosis, is it possible to reliably diagnose autism before age 2? The answer is yes. A number of researchers have now studied the early development of children with autism—a not so easy task given that the average age of diagnosis in the United States is 3 to 4 years of age.

Early development of children with autism has been studied by:

- * screening large numbers of children for autism-specific behaviors during well-baby checks at 18 months of age;
- * asking parents of children with autism to remember back to whether their child showed certain behaviors at certain developmental ages;
- * and having parents bring in home videos of children diagnosed with autism when these children had their first or second birthday parties (which parents often videotape).

From these studies, five distinct areas of development are flagged for consideration. All parents should consider these "Big Five" if they suspect that their child may have autism.

1) Does the baby respond to his or her name when called by the caregiver? Within the first few months of life, babies respond to their own name by orienting toward the person who called them. Typical babies are very responsive to the voices of familiar people, and often respond with smiles and looks.

In contrast, infants later diagnosed with autism often fail to respond to their own name. That is, when called by name, they tend to turn and look at the person only about 20% of the time as found in the videotaped one year-old birthday parties of children with autism. They also are often selectively responsive to sounds. They may ignore some sounds and respond to others that are of the same loudness. Thus, they may fail to respond to their parent calling their name, but immediately respond to the television being turned on. It is not unusual for parents to suspect their child has a hearing loss.

2) Does the young child engage in "joint attention"? Near the end of the first year of life, most infants begin to join with their caregivers in looking at the same object or event. To aid in this process of "joint attention", typical infants begin to shift their gaze from toys to people, follow other's points, monitor the gaze of others, point to objects or events to share interest, and show toys to others. These behaviors have a distinct sharing quality to them. For example, the young infant may point to an airplane flying over head, and look to the parent, as if to say, "do you see that!"

In contrast, young children with autism have particular difficulties in jointly attending with others. They rarely follow another's points, do not often shift their gaze back and forth from objects to people, and do not seem to share "being with" the caregiver as they watch and talk about objects, people, or events. They also tend not to "show" a toy to the parent.

3) Does the child imitate others? Typical infants are mimics. Very young infants can imitate facial movements (e.g., sticking out their tongue). As early as 8-10 months, mothers and infants say the same sounds one after another, or clap or make other movements. Indeed, imitation is a major part of such common infant games as pat-a-cake and So Big ("How big is baby? Soooo big!" as infant raises hands to sky).

Young children with autism, however, less often imitate others. They show less imitation of body and facial movements (waving, making faces, playing infant games), and less imitation with objects.

4) Does the child respond emotionally to others? Typical infants are socially responsive to others. They smile when others smile at them, and they initiate smiles and laughs when playing with toys and others. When typical infants observe another child crying, they may cry themselves, or look concerned. Somewhat older infants may crawl near the person, pat, or in other ways offer comfort. These latter behaviors are suggestive of empathy and are commonly observed among children in the second year of life.

In contrast, children with autism may seem unaware of the emotions of others. They may not take notice of the social smiles of others, and thus may not look and smile in response to other's smiles. They also may ignore the distress of others. Several researchers have now shown that when an adult feigns pain and distress after hitting herself with a toy, or banging her knee, young children with autism are less likely to look at the adult, or show facial concern.

5) Does the baby engage in pretend play? Someone once noted that "play is the work of children." Young children love to pretend-to be a mother, father, or baby, to be a firefighter or police officer. Although children start to play with toys around six months or so, play does not take on a pretend quality until the end of the first year. Their first actions may involve pretending to feed themselves, their mother or a doll, brush the doll's hair, or wipe the doll's nose. Nearer their second birthday, children engage in truly imaginative play as dolls may take on human qualities of talking or engaging in household routines. Children may pretend that a sponge is a piece of food, a block is a hat, or a plastic bowl is a swimming pool that contains water.

In contrast, the play of children with autism may be lacking in several ways. The young child may not be interested in objects at all, paying more attention to the movement of his hands, or a piece of string. If interested in toys, only certain ones may catch his interest, and these may be used in a repetitive way that is not consistent with how most children would play with the toy. They may be more interested in turning a toy car upside down and spinning the wheels than pushing the car back and forth. Overall, pretend qualities are nearly absent in the play of children with autism under 2 years of age.

It is important to note that in each of the 5 areas we have flagged, we are most concerned with behaviors that are absent or occur at very low rates. The absence of certain behaviors may be more difficult to pinpoint than the presence of atypical behaviors. But concerns in any of the above areas should prompt a parent to investigate screening their child for autism. Several screening measures are now available, and information from the screener will help to determine if the parent should pursue further evaluations. If the parent is convinced their child has autism, then they should seek an evaluation with an expert in autism. Most likely, this evaluation will involve an interview with the parents to obtain a complete developmental history of the child, and direct observations of the child in different situations.

The development of tools for earlier diagnosis of autism is moving quickly.

By Beth Azar
Monitor staff

To the consternation of many parents, children with autism rarely receive a diagnosis before age 3 or 4. But behavioral researchers believe they are homing in on specific behaviors that should drop the age of first diagnosis down to as young as 18 months.

Early diagnosis has become increasingly critical as treatments for the potentially devastating developmental disorder advance and research begins to show that the earlier the disorder is diagnosed, the better the prognosis.

Developmental researchers increasingly find that there are critical periods of child development after which certain systems—such as language, vision and motor skills—become less malleable.

And the same is likely to be true for social behaviors and intellectual abilities—skills often affected in children with autism, says University of Washington psychologist Geraldine Dawson, PhD. Therefore, if researchers can learn how to diagnose autism in children at birth or several months after, they may be able to design interventions that derail the disorder before it develops, some researchers speculate.

The development of tools for earlier diagnosis of autism is moving quickly, say researchers. This summer, the National Institutes of Health (NIH) hosted a conference on autism diagnosis to discuss the state of the science and to begin to develop practice guidelines for diagnosing autism. And, as geneticists begin to locate potential 'autism genes' (see article on page 13), behavioral researchers are fast developing behavior-based diagnostic tools that will work at increasingly younger ages.

A tricky diagnosis

Autism is difficult to diagnose in very young children because several of the disorder's main symptoms—such as the child's relationship with peers—involve behaviors that don't fully develop in children until later in childhood, says Fred Volkmar, MD, of the Yale Child Study Center.

According to the Diagnostic and Statistical Manual of Mental Disorders-IV (DSM-IV), the essential features of autistic disorder include trouble interacting with others and a tendency to have narrowly focused and odd interests, as with the boy who was fascinated by watches to the exclusion of anything else. Most researchers admit that these traits are the hallmarks of autism, but they also say that they don't help much when trying to diagnose autism in children younger than age 2 or 3.

Diagnosis of autism is also difficult because the best early indicators involve the absence of consistent social and communication behaviors rather than the presence of an abnormality. 'There could be lots of reasons you're not seeing a behavior,' says autism researcher Wendy Stone, PhD, of Vanderbilt University School of Medicine. 'You could not be seeing it because it's not developing, or it could be that the child is not showing it to you at that time.'

University of Chicago researcher Catherine Lord, PhD, has tried to address this issue by designing an observational diagnostic tool—the Autism Diagnostic Observation Schedule-Generic—which creates situations that attempt to elicit certain behaviors in children such as pointing or asking for something. Autistic children, researchers find, often fail to draw others' attention to objects by pointing and tend not to ask for things in the same way as other children, says Lord. Her diagnostic technique also

includes a parent interview, called the Autism Diagnostic Interview–Revised, which asks parents about how the child acts in typical situations.

Lord’s longitudinal work suggests that her tools can diagnose children as young as 18 months, she says. The diagnosis is somewhat inconclusive when children are this young, but becomes more stable at age 3 and again at age 5, she says.

Research by Lord and others finds that social and communication behaviors, such as imitation and use of gestures, are most indicative of autism in very young children, says Stone, who reviewed the research on early behavioral indicators of autism.

Based on her own research, Stone is developing a screening tool that she hopes will allow clinicians to check for autism in children age 2 and possibly younger.

In a study that followed a group of children from age 2 to 4, she identified three specific skill areas, all in the realm of social and communication skills, which appear to indicate autism:

- Play. Children with autism fail to engage in reciprocal play, where there is a back-and-forth between two people (as when rolling a ball) and functional play, where children play with toys such as dolls or cars in the way intended.
- Motor imitation. Children with autism copy others’ motor movements far less than other children.
- Joint attention. Unlike most children, children with autism don’t often use pointing or other techniques to direct another person’s attention.

A pattern of behaviors

What is particularly striking about children with autism is how different they are from each other, say researchers such as Stone and Grace Baranek, PhD, at the University of North Carolina, Chapel Hill.

In an attempt to delineate behaviors that distinguish children with autism from other children, Baranek examined early home videos taken when the children were 9 months to 12 months old—well before they were diagnosed with autism. When she compared the videos with home videos of children with other developmental disabilities and children with no disabilities, she found that the children with autism didn’t all have the same symptoms but instead showed a pattern of behaviors as a group.

Overall, the autistic children showed subtle problems in sensory attention and arousal. In particular, they oriented less to visual information in their environments; they put objects in their mouths more often; they needed more cues before they would look when someone called their names; and they pulled away from social touch slightly more than either of the other groups of children.

But 'you can’t tell these kids apart by glancing at their home movies—it takes a detailed analysis of their subtle behaviors,' adds Baranek. In fact, at the study’s end, the students who coded the specific behaviors from the videos were unable to reliably determine whether a child had autism, another developmental disability or no disability.

Even trained clinicians have a hard time seeing behavioral problems in very young children later diagnosed with autism, says Dawson of the University of Washington. She’s conducted three studies

using home videos, and in each she asked experienced pediatricians to look at the tapes and make a qualitative judgment about which children had autism. When the children were 1 year old or older, the pediatricians did reasonably well. But when the children were 8 months to 10 months old, the pediatricians were unable to tell the difference.

In fact, even trained video coders had a hard time finding differences in behaviors of the youngest children. In Dawson's first home video study, she examined videos of babies' first birthday parties and, similar to what others find, identified four behaviors that distinguished the children with autism from the children with no developmental disability: failure to make eye contact with others, to point, to show objects to others and to orient to one's name being called.

When she examined videos of children 8 months to 10 months old, the inability to make eye contact with others and to respond to one's name continued to distinguish the children. But joint attention behaviors, such as the inability to point and to show, did not.

This isn't surprising, says Dawson, since such behaviors are not typical of children much younger than 1, but it limits the number of criteria clinicians will be able to use for diagnosis at such young ages, she adds.

A Catch-22?

If researchers develop behavioral tools for screening and diagnosing children with autism at age 2 and younger—and most researchers believe they're close—they will face a seeming paradox. How can they prove their early diagnosis was right if the children they diagnose at 18 months enter into interventions that help treat their dysfunction?

'It's a huge problem,' says Stone. 'You don't know if you were wrong at age 2 or whether the intervention worked so well that your diagnosis is no longer valid.'

That's why most researchers hope they will find biological or genetic markers for autism that could accurately diagnose autism at birth and would bolster the behavioral measures. Some teams are well on the way toward finding one or several autism genes, and others are using neuroimaging to search for specific anatomical or functional differences in the brains of people with autism as well as for biochemical markers. However, having something that is solid enough for diagnosing the disorder is still a long way off, agreed researchers at the NIH 'Working conference on the state of the science in autism: screening and diagnosis' held in June.

That conference, sponsored by the NIH Autism Coordinating Committee, which represents several NIH institutes, and the Office of Behavioral and Social Sciences Research, brought together researchers, clinicians and representatives of parents' groups to review the research on autism diagnosis and draft an outline for autism screening and diagnosis practice guidelines.

Although NIH is not in the business of setting diagnostic criteria or practice guidelines, it tries to help professional organizations do so when appropriate, said National Institute of Child Health and Human Development Director Duane Alexander, MD, as he welcomed people to the conference.

And it's time to do so for autism because it is so often mis-diagnosed or mistaken for other developmental disorders. Also, although the DSM-IV provides diagnostic criteria, it doesn't provide clinicians with guidelines on how to perform initial screenings or on which tools to use to measure behavior.

Several professional organizations, including the American Academy of Neurology, the American Academy of Pediatrics, the Society for Research in Child Development and the American Academy of Child and Adolescent Psychiatry, fully backed the initial conference. And many others, including APA, were invited to participate. The same professional groups are invited to meet this winter to review a draft of the proposed guidelines and negotiate a final product they can all agree on and endorse, says Pauline Filipek, MD, co-chair with Marie Bristol-Power, PhD, of the first meeting, and chair of the next meeting.

'It is very important to get everyone on board and on the same wavelength,' says Filipek, a child neurologist at the University of California, Irvine. That will be the only way to ensure accurate and early diagnosis of autism.

Early Diagnosis of Autism

Psychologist Philip Teitelbaum, a Graduate Research Professor at the University of Florida, is researching early signs of autism in infants with the ultimate goal of enabling children to begin treatment for the disorder early enough to curtail and possibly even reverse its effects.

According to Professor Teitelbaum, autistic children learn to sit up, turn over and crawl in noticeably different manner than normal children. He believes that this difference in movement is caused by the same wiring problem in the central nervous system that later causes the social/verbal symptoms commonly associated with the condition. Since Teitelbaum's research indicates it may be possible to detect these movement problems at three to six months of age, autistic babies may soon be able to receive early treatment therapies during crucial brain development (zero to two years of age).

Several years ago, while studying a gait disorder commonly found in patients suffering from Parkinson's disease, Dr. Teitelbaum attended a talk that changed the focus of his research when a colleague, Dr. Ralph Maurer, delivered a paper on similarities between the way Parkinsonian adults and autistic children walk. Teitelbaum's curiosity was piqued, and after he and his wife, Osnat, a movement analysis expert, examined videos of autistic children, they felt sure that a movement disorder was indeed a key component of the condition.

Next, the Teitelbaums obtained from parents 17 videos of infants who were later diagnosed as autistic. The Teitelbaums taped key parts of these videos--the children attempting to master developmental milestones like turning over, walking and crawling--on a optical disc recorder, which made careful, blur-free, frame-by-frame movement analysis possible. Using footage of 15 "normal" babies as a control, they examined and documented the children's movements using Eshkol-Wachman Movement Notation (EWMN). EWMN is a general analysis system in which spherical coordinates are applied independently to each segment of the body. By distinguishing between which segments are actively moving versus those that are being carried passively along, a deeper understanding of abnormal movement is possible.

The results of their analysis were astounding. Every single autistic child demonstrated at least one movement disturbance by six months of age. The group published an article in the November 10, 1998 issue of the Proceedings of the National Academy of Sciences detailing their findings. Osnat, who studied EWMN with the Noa Eshkol in Tel Aviv for 20 years, points out that this is the first time

EWMN has been used in a medical diagnostic context. "It's a pioneering use of the technology," she says, "and may eventually prove useful in the diagnosis of other developmental disorders."

Using segments of video, Teitelbaum can demonstrate that among other things, autistic babies have difficulty supporting themselves to crawl. In an ineffectual attempt to move forward, they may rest weight on their elbows and forearms, dig their toes in and lift their rumps. Or, struggling to pull themselves along, these babies might leave one arm underneath the torso while attempting to crawl with the free arm or may crawl atypically, with one leg stepping while the other leg moves normally. Similar movement problems are seen earlier in turning over and later in walking.

Moebius mouth (flat lower lip and arched, oval shaped upper lip) is also present in a number of the children the Teitelbaums observed on video. According to Dr. Teitelbaum, not all autistic children have this feature, but when a child does have it, it's a possible indicator of autism. And since the condition is noticeable in autistic infants as young as one month old, moebius mouth may prove to be one of the very earliest signs of the disorder.

Professor Teitelbaum is pioneering a more extensive study into early diagnosis and needs additional early home videos of children (0-18 months) later diagnosed with autism. For more information, please call (352) 392-0615, e-mail Jennifer Nye nye@psych.ufl.edu or write Dr. Teitelbaum at Campus Box 112250, UF, Gainesville, FL 32611.