





MAKING SENSE OF A COMPLICATED CONDITION

By Mark Harris

Autism spectrum disorder (ASD) is a lifelong neurodevelopmental disorder estimated to affect more than 3.5 million Americans. As a brain-based condition, ASD, or autism as it is commonly described, can impair an individual's ability to communicate and socially interact with others. These behavioral and communication challenges in individuals can range from mild to severe.¹

Perhaps the first thing to know about autism is that, as a brain-based disorder, its signs and symptoms begin early in life. For example, parents may notice their child does not point at objects to show interest or look at objects when another person points at them. Often, children with autism have trouble relating to others. At a young age, they might avoid eye contact, have trouble sharing joint attention with others, or even appear unaware that other people are talking to them. Children who are very young can appear to have delayed or even nonexistent language skill development.²

Many different behaviors and ways of interacting with others and the environment are associated with autism. Some individuals

may engage in repetitive behaviors, such as rocking back and forth.² They may repeat words or phrases they hear, crave routine, and find it difficult to adapt to changes in their environment.³ For some, it can be difficult to express themselves adequately. Wanting to be alone or not wanting to be touched are also symptomatic of an individual on the autism spectrum. In some cases, individuals with autism show no interest at all in other people. Yet, while ASD can often manifest itself as a significant disability in some people, many others possess normal or even above-average IQs and display exceptional academic or artistic abilities.⁴

Defining terms, refining diagnosis

The first description of autism as a medical condition was put forward in 1943 by Leo Kanner, MD, a psychiatrist affiliated with the child psychiatry clinic at Johns Hopkins Hospital in Baltimore, Maryland. Based on observation of a small cohort of young children with developmental disorders, Dr. Kanner described a condition he called *early infantile autism*.⁵

Screening tools for autism

The Centers for Disease Control and Prevention does not endorse any specific tools for autism screening purposes, but it does list a sampling of developmental screening tools for general development and autism spectrum disorder (ASD) to help health care professionals¹³:

- **Ages and Stages Questionnaires (ASQ).** This is a general developmental screening tool that includes a parent-completed questionnaire and a series of 19 age-specific questionnaires that screen for communication, gross-motor, fine-motor, problem-solving, and personal adaptive skills. The ASQ results in a pass-fail score for domains.
- **Communication and Symbolic Behavior Scales (CSBS).** The CSBS is a standardized tool for screening communication and symbolic abilities up to the 24-month level; the Infant-Toddler Checklist, part of the CSBS, is a one-page, parent-completed screening tool.
- **Parents' Evaluation of Developmental Status (PEDS).** This general developmental screening tool employs a parent-interview form; screens for developmental and behavioral problems needing further evaluation; and includes a single response form used for all ages. It may be useful as a surveillance tool.
- **Modified Checklist for Autism in Toddlers (M-CHAT).** This parent-completed questionnaire is designed to identify children at risk for autism in the general population.
- **Screening Tool for Autism in Toddlers and Young Children (STAT).** This is an interactive screening tool designed for children when developmental concerns are suspected. It consists of 12 activities assessing play, communication, and imitation skills and takes 20 minutes to administer.

The condition was marked by such characteristics as a lack of interest in other people, a strong desire for aloneness and persistent sameness in the environment, and other unique behavior patterns.^{4,6} Around the same time, Hans Asperger, MD, an Austrian physician, described a milder form of autism that came to be known as *Asperger's syndrome*.⁵

Before the 1940s, what in some people might have been diagnosed as autism was likely to be identified as a form of schizophrenia. The early understanding of autism also commonly included the psychiatric view that the condition was primarily caused by bad parenting or what was sometimes referred to as the refrigerator mother theory (a term referring to cold or distant parent-child relationships). In the 1960s the well-known psychiatrist Bruno Bettelheim helped popularize this now widely rejected explanation for the causes of autism.⁵

Today, autism is understood to be essentially a physical disorder with a strong genetic component, says Gail J. Richard, PhD, CCC-SLP, president of the American Speech-Language-Hearing Association (ASHA). Accordingly, the developmental disabilities associated with the disorder not only can qualitatively affect a child's education and ability to learn and communicate, but are also associated with restrictive and repetitive patterns of behavior and activity.

"I like to use an operational definition of autism to help people understand its main components," says Dr. Richard, a speech pathologist and director of the Autism Center at Eastern Illinois University (EIU) in Charleston. "First, it's important to know that autism spectrum disorder is an actual physical disorder of the brain, the nature of which is a biochemical neurological disorder. By this we mean that as we are exposed to things in our environment, that

stimulation triggers chemicals that then run our neurology or run the way our brain interacts with other areas of the brain. For individuals within the autism spectrum, their brain actually develops in a different way. We see a different biochemical reaction. Areas of the brain that need to communicate better don't have good synaptic connections."

In humans, explains Dr. Richard, synapses in the brain allow neurons to send and receive signals. During childhood and adolescence, the developing brain learns to limit or prune these synaptic connections to avoid an overload of stimuli. Unfortunately, as Dr. Richard notes, this pruning process is disrupted in individuals with autism.

Much of the current understanding of autism goes back to landmark research in the late 1970s, when studies of identical and fraternal twins helped researchers identify a strong genetic component to autism. In 1980 another landmark occurred when *infantile autism* was for the first time included in the American Psychiatric Association's (APA) *Diagnostic and Statistical Manual of Mental Disorders, Third Edition (DSM-III)*. More recently, the publication of the *DSM-5* in 2013 marked another significant revision to how autism is defined. In this latest edition, *Asperger's syndrome* and *pervasive developmental disorder not otherwise specified* (PDD-NOS) are no longer categorized as distinct diagnostic categories. Instead, the three conditions are now classified under the single diagnostic umbrella of autism spectrum disorder (or *ASD* for quick reference).⁶ In turn, the *DSM-5* now defines the range and severity of autism's characteristics under this single criterion.⁷

Specialty circumstances

Admittedly, diagnosis of ASD, with its varied and complex symptomology, can pose a challenge for clinicians. Indeed, there are no blood or brain imaging tests for the disorder. Nor currently is genetic testing a part of the standard evaluation and assessment. For the most part, diagnosis relies on clinical observation, developmental history, and the expertise of skilled clinicians.

“There’s a saying that if you’ve met one child with autism, you’ve met one child with autism,” says Judith S. Miller, PhD, MS, a psychologist and clinical training director for the Center for Autism Research at Children’s Hospital of Philadelphia. “Some children with autism are very low functioning and may not speak very much or interact with people at all. Then there are other people with autism who could be intellectually gifted, be very talkative and socially interested, but socially awkward. We’re not necessarily looking for one kind of clinical presentation. There’s a really wide range of presentations that make the autism diagnosis tricky.”

For this reason, autism specialty centers, using experienced interdisciplinary teams, often play an essential role in both diagnosis and treatment for patients on the autism spectrum. These teams may include such specialists as psychologists, psychiatrists, speech-language pathologists, developmental pediatricians, and other professionals. “The specialty centers are helpful because those are folks who have usually seen several hundred people with autism across different ages and different levels of functioning,” says Dr. Miller. “They’re especially helpful if there’s anything subtle or sort of complex about the child’s particular situation.”

Typically, establishing a diagnosis involves two or more steps. “Often, families go to their pediatrician or primary care provider with their first concerns that something might not be quite right in their child’s development,” says Jennifer Gerdt, PhD, attending psychologist at Seattle Children’s Hospital and assistant professor of psychiatry and behavioral sciences at the University of Washington. “Or maybe the pediatrician during a well-child visit has done a screening and recognized some concerns. If the pediatrician or the parents have expressed concerns, and they agree that it’s worth investigating, usually then a referral is placed to an autism diagnostic center or to a developmental pediatrician, psychiatrist, or psychologist that have more training and specialty in understanding and diagnosing autism.”

Diagnostic tools for autism

Many tools exist to assess autism spectrum disorder in young children, but no individual tool should form the foundation of a diagnosis. The following are certain examples of some common diagnostic tools¹³:

- **Autism Diagnostic Interview–Revised (ADI-R).** A clinical diagnostic instrument for assessing autism in children and adults, this instrument focuses on behavior in three main areas: reciprocal social interaction; communication and language; and restricted and repetitive, stereotyped interests and behaviors. The ADI-R is for children and adults with mental ages of 18 months and above.
- **Autism Diagnostic Observation Schedule–Generic (ADOS-G).** This semi-structured, standardized assessment of social interaction, communication, play, and imaginative use of materials is used for individuals suspected of having autism spectrum disorder. The observational schedule consists of four 30-minute modules, each designed to be administered to different individuals according to their level of expressive language.
- **Childhood Autism Rating Scale (CARS).** This brief assessment is suitable for use with any child over 2 years of age. The CARS includes items drawn from five prominent systems for diagnosing autism; each item covers a particular characteristic, ability, or behavior.
- **Gilliam Autism Rating Scale–2nd Edition (GARS-2).** This scale assists teachers, parents, and clinicians in identifying and diagnosing autism in individuals ages 3 through 22. It also helps estimate the severity of the child’s disorder.

Accordingly, pediatricians and other primary care providers do not typically diagnose autism on their own. As Dr. Gerdt explains, “This is mostly because either pediatricians are not that comfortable making the diagnosis themselves, because they haven’t enough training in this specialty, or because in some states, such as Washington, a diagnosis from a primary care provider is actually not accepted for insurance purposes.”

The Autism Center at Seattle Children’s Hospital serves an average of 4,000 to 5,000 patients a year, reports Dr. Gerdt, with about half of visits being diagnostic in nature. In addition to diagnostic services, including early intervention and risk screening for children under 16 months, autism specialty centers typically provide a wide range of treatment care and medical management services for patients on the autism spectrum. These can include group and individual therapy, medication management, biobehavioral programs, referral to other medical specialties, and other resources.

Watch and learn

How do clinicians establish a diagnosis of ASD? Among the variety of screening and diagnostic tests to help confirm a diagnosis, one key tool is the Autism Diagnostic Observation Schedule (ADOS), widely considered as a diagnostic gold standard in the comprehensive evaluation. The ADOS represents a “systematic observation tool” that allows qualified clinicians the opportunity to observe a person’s social and communication behaviors in a structured context, according to the Center for Autism Research. There are currently four ADOS modules in use, applicable to the age and level of language of the individual being evaluated. These range from nonverbal children as young as 12 months through verbally fluent adults.⁸

As the Center for Autism Research describes on its website, “specific activities and materials are used to create structured and unstructured social occasions or ‘presses.’ The examiner rates both the spontaneous and prompted social relating,

The meaning of screening

Screening and diagnosing are not one and the same. If a child has a positive screen for autism spectrum disorder (ASD), it doesn't necessarily mean the child will be diagnosed on the spectrum. Furthermore, if a child screens normally, but parents continue to worry about ASD, they should not be shy about it. Screening tests are just that—screening—and do not identify all children with ASD. The rate of success for the Modified Checklist for Autism in Toddlers (M-CHAT), for example, is not 100 percent, so it is used in combination with health and family history to identify children at risk. Parents' opinions are irreplaceable and of the utmost importance.¹⁴

communication, and behaviors that occur during the ADOS.”⁸ There is general agreement among autism experts that behavioral observation using ADOS and other screening and diagnostic tests (e.g., the Modified Checklist for Autism in Toddlers [M-CHAT]⁹), combined with a detailed developmental history, is at the heart of the assessment process.

“Because autism is a developmental disability, we’re looking for signs and symptoms really early in life, at age 3 and under,” explains Dr. Gerdt. “We’ll ask parents or caregivers about what their child was like in those early years. We also look at how the child is doing currently, including medically, and in school and out in the community. Basically, we’re trying to get a sense of how the child is doing in multiple aspects of his or her life, and then pooling that information together from our behavioral observation, developmental history, and sometimes other kinds of IQ tests or cognitive tests. All of this goes together in helping us come to a diagnostic decision,” she says.

Sense of timing

Interestingly, research shows that most referrals for ASD are initiated by parents, adds Dr. Richard—a fact that touches upon concerns about whether the gap between the first signs of autism and the eventual medical diagnosis can be shortened. “It’s actually the parents who usually first notice something different is going on in their child’s development and bring it to the pediatrician’s attention,” says Dr. Richard. “They might notice the child’s not cooing, not facially interacting, not playing games like peekaboo or patty-cake or sharing joint attention with adults.”

Notably, a delayed diagnosis can translate into missed opportunities for treatment intervention during a critical developmental stage for young children. “The research shows that the greatest impact of this disorder occurs before three years of age, as the brain is forming all these connections, but the median age of diagnosis is 5,” reports Dr. Richard. “Thus, we’re missing this diagnosis in a lot of children. In my view, a screening for autism spectrum disorder needs to be part of all general wellness checks, certainly at 18 and 24 months of age. There needs to be screening that pediatricians and family physicians just do as part of the general wellness exam.”

The American Academy of Pediatrics (AAP) agrees, recommending universal screening at 18 and 30 months, and when parents or providers express concerns about a child’s development.⁹ However, not all experts endorse this practice. In early 2016, the U.S. Preventive Services Task Force (USPSTF) decided against endorsing universal screening recommendations, citing insufficient evidence for either benefits or harms from routine screening when there were no existing concerns about autism in individuals.¹⁰

Certainly, discussions within the autism field over screening recommendations will continue. The nonprofit advocacy group Autism Speaks and other organizations have expressed their disagreement with the USPSTF decision, citing both the importance

of early intervention and how symptoms of autism can now be detected at earlier ages than in the past.¹¹

Of course, some delay in the autism diagnosis in young children might be inherent to the nature of early childhood development. “So many things change when children are very little that it can take a while for everyone to agree that the signs of autism are there and significant enough that they need to act,” says Dr. Miller. “Parents might see something that’s a little bit off, but they’re not worried about it yet. But later when they look back they’ll tell you, ‘Yes, that first sign was when he was 6 months old or 12 or 18 months old.’ They recognize later that something was a sign of autism, but at the time it was not always that clear. So, there’s typically some back and forth where the parents are talking to the physician, where you’re both kind of watching and waiting, getting hearing tested, getting speech evaluated, and then hopefully it’s not very long before the decision is made. But it’s not a simple answer like a hearing problem.”

“Get low, go slow”

How prevalent is ASD in the general population? The Centers for Disease Control and Prevention (CDC) estimates that 1 in 68 children in the United States have the disorder. Notably, autism is about 4.5 times more common in boys than girls.³ While the CDC estimate is the official figure, some research indicates the percentage may be even higher. A recent National Health Interview Survey found 1 in 45 children ages 3 through 17 have the autism diagnosis.¹²

The rising percentage of children with autism is certainly a matter of concern. In fact, the diagnosis rate has more than doubled since 2000, when the prevalence of autism was determined to be about 1 in 150 children. However, experts are cautious about how to interpret this upswing. Many believe the increase is influenced by multiple factors, such as greater public awareness of autism, evolving diagnostic criteria, and improved assessments by health

care providers.¹ But there may also be biological and environmental factors at work. Children born prematurely are believed to be at increased risk for autism, which is worth noting because survival rates for premature infants have improved over the years. Evidence also indicates that having older parents (i.e., mother over 35 or father over 40) is linked to increased autism risks.⁷

Whatever the interplay of factors, with an increasing number of children diagnosed on the autism spectrum, pediatricians, family physicians, and other providers must be equipped to offer sensitive, mindful care for these patients. Indeed, for a young child with autism, a negative or upsetting medical visit can set the stage for difficult future visits, or even a lifetime of stressful or avoided medical visits.

At the Center for Autism Research, Dr. Miller has been involved in efforts to help physicians and staff better accommodate the unique health care needs of patients on the autism spectrum and their families. The goal is to make medical appointments as comfortable as possible for patients. Toward this objective, Dr. Miller suggests medical practices employ a practice philosophy she describes as “get low, go slow.” The central idea is to recognize that a child on the autism spectrum might require extra consideration to ensure the appointment goes smoothly. For providers and staff, “get low, go slow,” translates into such advice as remembering to speak directly to the child (not just the parents) and getting on the child’s level when doing so (even if there is little eye contact in return). It means using clear, simple language or breaking down instructions into parts. It could mean using visual prompts such as pictures or other images to help explain or illustrate what will happen in the examination. It can also mean knowing when to back off if any part of the examination causes distress.

“Sometimes children with autism just need more time before they will be comfortable enough to let somebody do an exam,” says Dr. Miller. “In our clinic when we do

evaluations, the physician usually gets about an hour with the patient to take the whole history, and they save the physicals for the end of that hour. By then the child is usually comfortable, and it’s very easy to do the physical. In that hour, they’re talking to the parents as well, while the child is getting comfortable with the room. It’s rare that somebody has a hard time then. But primary care doesn’t have that kind of time. So, figuring out how to either try to make that time, at least in the beginning, or trying to make some sort of thoughtful workaround can be helpful.”

All in all, preparation and communication with parents or caregivers are essential to a successful visit, concludes Dr. Miller. For example, office schedulers might use a simple set of screening questions so that staff know before a medical appointment whether a child has any special behavioral needs or diagnosis that need to be considered. When a child is identified as needing additional support, clinic staff might meet briefly before the visit to discuss the best approach for the appointment. This might include such considerations as what time of day would be least disruptive to a child’s routine. Parents usually have a good idea of what will work best for their child, says Dr. Miller.

Stress-free service

The “get low, go slow” philosophy also embraces a team-wide approach to care. “The medical assistant is often the first one to come out to the waiting room, and it’s potentially that initial interaction that can sort of frighten the patient,” observes Dr. Miller. “Of course, the medical assistant is just doing what they always do; they don’t know the patient has autism, so they’re just going at their regular pace. But if we could alert the medical assistant, that could set a whole different tone for the rest of the visit.”

Indeed, working successfully with patients on the autism spectrum involves cultivating awareness that such patients might be sensitive to stimuli or experiences

Age-appropriate

Here are some examples that may help a parent identify the early signs of autism spectrum disorder¹⁵:

At 12 months

- Children with typical development will turn their head when they hear their name.
- Children with ASD might not turn to look, even after their name is repeated several times, but will respond to other sounds.

At 18 months

- Children with delayed speech skills will point, gesture, or use facial expressions to make up for their lack of talking.
- Children with ASD might make no attempt to compensate for delayed speech or might limit speech to parroting what is heard on TV or what was just heard.

At 24 months

- Children with typical development bring a picture to show their parent(s) and share their joy from it with them.
- Children with ASD might bring their parents a bottle of bubbles to open but will not look at their parents’ faces or share in the pleasure of playing together.

in the medical environment that do not affect other patients. “Sometimes when the phlebotomist draws blood, for example, it’s not the needle that is frightening, it’s the fact that somebody is holding their arm in an unusual way,” explains Dr. Miller. “Or it’s the sight of the blood pressure cuff or the scale. It doesn’t look like something they’ve seen before. Patients with autism will at times have a strong reaction to something that most children are not bothered by. That can throw the medical assistant off because you just don’t see that every day.”

Links to autism resources

American Academy of Pediatrics (AAP): <https://www.aap.org/en-us/about-the-aap/Committees-Councils-Sections/Council-on-Children-with-Disabilities/Pages/Autism.aspx>

American Speech-Language-Hearing Association (ASHA): <http://www.asha.org>

Autism Society: <http://www.autism-society.org>

Autism Speaks: <https://www.autismspeaks.org>

Autism Center, Seattle Children's Hospital: <http://www.seattlechildrens.org/clinics-programs/autism-center/>

Center for Autism Research (CAR), Children's Hospital of Philadelphia: <http://www.chop.edu/health-resources/center-autism-research-car>

Eastern Illinois University (EIU) Autism Center: <https://www.eiu.edu/autismcenter/>

When providing care for those on the autism spectrum, minimizing any potential stressors in the visit or examination is an office-wide challenge that should include not just clinical staff but also registration or front-desk personnel and practice managers. “There can be stress in the waiting room, stress at the first part of the visit, and stress if the patient with autism needs more time, and your office manager is wondering what’s happening,” says Dr. Miller. “So, how do we build a reasonable workflow? Unfortunately, I think we’ve focused most of our attention on the physicians and sometimes the nurses. But actually, the physicians might need to know the least. They are not usually the ones who are trying to take blood pressure or get the vitals. If you think about it, it’s really the nurses and the medical assistants who are doing more of the hands-on work with the patient.”

“You have to realize with autism and other mental or behavioral health issues that you’re often dealing with a very vulnerable group of people,” says Connie Lieseke, CMA (AAMA), MLT(ASCP)PBT, a health clinic coordinator for not-for-profit Kitsap Mental Health Services in Bremerton, Washington. “For those who have autism, in particular, they’re often challenged by that inability to fully engage. As a medical assistant, when you’re talking to a patient with autism,

sometimes it’s hard to know whether they understand. You have to learn to be very patient. You have to be very clear on the explanations for what it is that you’re doing because some patients will not always pick up on the subtleties of what you’re saying. If you speak too fast or are not very clear on what you’re doing, it can be startling for some patients, especially if you’re in a situation where you have to do a blood draw or other procedure.”

As a provider of in- and outpatient care, Kitsap Mental Health Services has seven medical assistants, says Lieseke, all supporting outpatient teams that work with children and youth with a variety of mental and behavioral health diagnoses. She says support staff need to acquire some perspective on the unique challenges involved in working with patients with autism and others with behavioral health diagnoses, especially for those with more severe symptomology. “No matter what you do, it might be difficult to always feel like you’ve really made a connection with an individual,” she says. “So, you just have to try to be as gentle as possible and make sure that you are very good with your processes. You want to make sure you are explaining as clearly as you can and that you’re being kind, while at the same time explaining to the parents in the room what you are doing.”

Developing story

As a complex, heterogeneous disorder, autism necessarily demands research with a multidisciplinary approach. From clinical psychology to developmental pediatrics, developmental and cognitive neuroscience, and genetics, the medical understanding of ASD has drawn upon insights from a range of medical specialties. Despite the many challenges, clinical understanding of autism has made considerable progress since the disorder was first described decades ago.

Certainly, much remains to be learned, particularly regarding the genetic components of autism. But with increasing knowledge comes significant improvements in available treatment options. “When I was training about 20 years ago, it was pretty widely known or accepted that about half of all children with autism would not speak fluently,” observes Dr. Miller. “Now, maybe 10 percent don’t speak fluently. Today, there are a lot of children who get very intensive behavioral intervention around skill development and do really well. And it changes their outcome.”

Dr. Gerdtz shares this optimism about the field’s accomplishments and what it portends for the future of care. “There have been a number of relatively large-scale studies of children who had different types of behavioral intervention early in life, and [the studies] tracked them over time. We’ve seen really dramatic progress in terms of skills that they learned and maintained even after the intervention was complete, even as they went on to get older and go to school. We know that some of the interventions that we’re using are having a lasting impact.”

The future is likely to bring new breakthroughs, especially in the understanding of autism’s complex genetic mysteries. “We used to think cancer was one disease,” concludes Dr. Miller. “But now we understand it’s more like 200 diseases, and they all have their own cause, their own prognosis, and their own course. Autism is probably going to be like that. We are identifying more and more genetic contributions. Hopefully, we’ll be able to eventually map a person’s genetic profile



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to their course of treatment. We will then be able to better predict whether an individual is going to respond to behavioral intervention or respond to a medication. We will be able to better personalize the intervention approach.”

For patients on the autism spectrum and their families, all this suggests hope for the future. ♦

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