BERKOWITZ'S PEDIATRICS

INSTRUCTOR'S GUIDE

Carol D. Berkowitz, MD, FAAP

Distinguished Professor of Clinical Pediatrics,
David Geffen School of Medicine at
University of California Los Angeles
Executive Vice Chair,
Department of Pediatrics,
Harbor-UCLA Medical Center
Torrance, CA

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Contents

Part 4: Adolescent Health

Part 5: Acute and Emergent Problems

Part 6: Head, Neck, and Respiratory System

Part 7: Hematologic Disorders

Part 8: Cardiovascular System

Part 9: Genitourinary Disorders

Part 10: Orthopedic Disorders

Part 11: Gastrointestinal Disorders

Part 12: Neuropsychiatric Disorders

Part 13: Dermatologic Disorders

Part 14: Social Determinants of Health

Part 15: Chronic Diseases of Childhood and Adolescence

CHAPTER 132

Autism Spectrum Disorder

CASE STUDY

The mother of 18-month-old twin boys is concerned because 1 twin is not talking as much as his twin sibling. Both twins are quite active. The mother feels that even though the child is quiet, he is very smart. He likes to figure out how things work. He seems very sensitive to sounds and covers his ears around loud noises. He loves music and even knows which CD his favorite song is on. He will interact with his sibling but does not seem interested in other children.

During the office visit, both boys are quite active. It is difficult to perform an adequate examination because the twin with limited language is crying the entire time. He does not seem to seek out his mother for comfort. Although both children have stranger anxiety, the twin about whom the mother is concerned seems to have extreme stranger anxiety. He appears well otherwise.

Questions

1. What is autism spectrum disorder?

Autism spectrum disorder (ASD) is characterized by impairments in social communication as well as restrictive, repetitive, and stereotypic behaviors or interests. According to the *Diagnostic and Statistical Manual of Mental Disorders*, 5th Edition (*DSM-5*), a person with ASD must display persistent communication, interaction, and behavioral challenges across multiple contexts.

This new term, ASD, includes the previous terminology of autistic disorder, Asperger syndrome, and pervasive developmental disorder–not otherwise specified; the term ASD no longer includes Rett syndrome. Although criteria differ somewhat, all these disorders had in common an impairment in social communication and repetitive or unusual interests of varying degrees. These disorders require similar management, and assessing the level of impairment is somewhat subjective. Therefore, a single term—ASD—best incorporates all those individuals who are significantly affected by its symptomatology.

2. How does autism spectrum disorder differ from language delay?

Language delay is isolated delay in the acquisition and expression of language. Autism spectrum disorder, however, is truly a spectrum of social communication deficits, often including developmental delays in multiple areas. Some affected individuals, because of an incredible ability to recognize patterns, can read as early as 2 years of age, even though they can neither speak functionally nor comprehend what they read.

Inconsistent symptoms are the hallmark of this disorder. Some parents or guardians of children with ASD describe a phenomenon whereby the children are developing normally until 12 to 15 months of age and then suddenly lose skills or stop progressing. This finding is particularly concerning.

3. How does the physician evaluate a child for autism spectrum disorder?

No single diagnostic test, blood or otherwise, can confirm the diagnosis of ASD. Diagnosis is based on history, interaction with the child, and meeting *DSM-5* criteria.

Regular developmental surveillance and screening should be part of every well-child evaluation, especially between ages 9 and 30 months. The Modified Checklist for Autism in Toddlers, Revised, with Follow-Up (M-CHAT-R/F) is an excellent autism-specific screening tool with moderate sensitivity and high specificity for use at the 18- and 24-month visits to identify individuals at high risk for ASD.

Family history is also important, because ASD is presumed to have a genetic contribution and it may be helpful in identifying other etiologies.

Understanding family structure is helpful in determining whether abuse, neglect, or maternal depression play a role in the child's delay. It is important to remember, however, that ASD is not caused by poor parenting.

4. Where can a physician refer a patient with autism spectrum disorder?

Waitlists to see specialists and a limit on the number of specialists in this field makes it imperative for the primary care physician to be able to make the diagnosis of significant autism. Because early intervention can have such a vital effect on patient outcomes, the American Academy of Pediatrics endorses this approach.

Autism spectrum disorder is a neurologic condition that can improve with intensive multimodality interventions. Behaviors such as impulsive aggression, repetition, resistance to change, and obsession are frequently targeted by systematic interventions. The best studied therapy is known as applied behavior analysis, in which a child's behavior is scrutinized by a trained behaviorist.

5. What types of treatment are available?

The mainstay for treatment of ASD remains behavioral. Interventions include speech and language services, occupational therapy, social skills training, and special education in the school setting.

Although medications do not seem to help the core symptoms of ASD, almost 2 in 3 children with ASD receive medications for behaviors that, despite intensive behavioral intervention, continue to obstruct progress or become dangerous.

A variety of alternative treatments have been suggested, but none of these methods is considered traditional or the standard of care because minimal empiric data exist to support their use.

6. Should a child suspected of having autism spectrum disorder receive further immunizations?

The child with ASD should undergo routine health maintenance, including all recommended immunizations. No evidence exists linking ASD with immunizations.

CASE RESOLUTION

The child's parent completed an M-CHAT-R/F, and the child scored a 4 (ie, intermediate risk). A follow-up interview confirmed that the risk for ASD was significant, and the child was evaluated by a developmental-behavioral pediatrician and the local governmental agency, where he underwent a comprehensive assessment by a multidisciplinary team. The diagnosis of autism was confirmed, and his brother was noted to have a language delay. Both children were placed in an early intervention program. The primary patient was placed in a 1:1 structured teaching environment for 4 months. After exhibiting significant improvement, he was moved to a therapeutic preschool setting that emphasized generalization of his newly acquired skills, speech therapy, occupational therapy, and social skills. His brother received speech therapy 2 times per week. Both are due to start a regular kindergarten class in the fall, with ongoing speech and social support. The primary patient has been placed on a stimulant medication to control hyperactivity and problems with attention.