

•> CASE 46

A 2½-year-old boy is brought to a pediatrician by his parents for his regular yearly examination. He is the couple's only child. The parents relate a normal medical history with a single episode of otitis media. They recently placed their son in day care for 2 half-days a week. However, he has not adjusted well, crying and having tantrums during the first hour of school. Then he usually quiets down, but he does not interact with the rest of the children. The teacher cannot seem to make him follow directions and notes that he does not look at her when she is near him and attempting to interact with him.

On further discussion with the parents, the pediatrician finds that the patient has only a limited vocabulary of perhaps 10 words. He does not use these words in any greater length than two words in a row and often uses them inappropriately. He did not speak his first clear word until 6 to 9 months previously. The patient does not interact well with other children but does not seem upset by them. His favorite toys are often used inappropriately—he performs single, repetitive movements with them for what seems like hours on end. The pediatrician picks the child up to help him onto the examination table and notices that he seems quite stiff, pushing himself away from the examiner with his hands. Although his hearing and eyesight appear to be intact, the child does not respond to requests by the pediatrician and does not make eye contact. All other gross neurologic and physical features are within normal limits.

- **What is the most likely diagnosis?**
- **What is the most likely prognosis for this condition?**

ANSWERS TO CASE 46: Autistic Disorder

Summary: A 272-year-old child is brought to a pediatrician for his yearly physical examination. His history reveals various behavioral issues, including poor interaction with peers and family, delayed language development, repetitive movements, and difficulty accepting change. The child stiffens physically when touched, does not respond to the examiner, and does not make eye contact.

- **Most likely diagnosis:** Autistic disorder.
- ^ **Prognosis:** The child will likely experience a number of developmental delays, but with intensive treatment at home and at school, he could achieve near-normal or normal development. Language development is the most important indicator of future developmental potential in autistic children.

Analysis

Objectives

1. Recognize autistic disorder based on symptoms.
2. Understand the unique symptomatic aspects of autistic disorder.
3. Understand the importance of early treatment.

Considerations

The patient's presentation and history are typical of a child with autistic disorder. The symptoms of autism often go unrecognized until the child is placed in an environment with other children of a similar age. This lack of recognition of the problem is especially likely to occur in a family with no other children, where it is not possible to compare developmental milestones. The patient displays typical symptoms: difficulty with social reciprocity, poor peer interaction, poor language development, and repetitive and odd play. His language development is poor for his age. A normal 2½-year-old child should have a much larger vocabulary—hundreds of words—and should easily be able to use several words in a sentence. His comprehension of words should be even better than his ability to use them. The presence of autism does not necessarily indicate mental retardation, but a large percentage of autistic individuals have this disorder. The parents have come for help relatively early in the course of the illness. Intensive behavioral and educational interventions will be necessary to help accelerate the child's development.

Definitions

Asperger disorder: A disorder of unknown etiology in which affected individuals display social impairment and restricted interests and behavior (stereotyped behavior) but have normal language and cognitive skills.

Mental retardation: A classification of cognitive functioning involving both a low intelligence quotient (IQ) and an impairment in adaptive functioning

Rett disorder: A childhood developmental disorder of unknown etiology in which the patient develops progressive encephalopathy, loss of speech capacity, gait problems, stereotyped movements, microcephaly, and poor social interaction skills. The child must have shown normal development in early infancy, and only females are affected.

Social reciprocity: The ability to read and to exhibit verbal and nonverbal behaviors as a result of interacting with others.

Stereotyped behaviors: Purposeless, repetitive movements and behaviors such as spinning toys, toe walking, or hand flapping.

Clinical Approach

Boys are more often affected by autistic disorder than girls by a three- to five-fold increased prevalence. Typically, the disorder is noticed by the parents of an affected child before 3 years of age and is characterized by developmental delay, aloofness, and stereotyped behavior. The etiology of autistic disorder is unknown, but a genetic etiology is likely. Family studies show a markedly increased incidence in monozygotic twins and a low risk in dizygotic twins. Elevated serum serotonin levels can be a clue to the neurochemical abnormality. Approximately 40% of children with autistic disorder are mentally retarded; however, some demonstrate unusual or extremely precocious abilities, so-called islets of precocity. One such talent is the ability to perform extraordinary mathematical calculations although the child is cognitively impaired in other ways.

Diagnostic Criteria

Patients demonstrate a qualitative impairment in social interaction skills, manifested by symptoms such as **a marked impairment in nonverbal behaviors, a failure to develop appropriate peer relationships, or a lack of social reciprocity**. There are also qualitative impairments in their ability to communicate, manifested by a delay in learning or a failure to learn spoken language. Patients exhibit **repetitive and stereotyped patterns of behavior**, including inflexible adherence to rules or stereotyped motor mannerisms. They can also be persistently preoccupied with parts of objects.

Differential Diagnosis

Autistic disorder must be differentiated from other pervasive developmental disorders, including Asperger disorder, childhood disintegrative disorder, Rett disorder, and pervasive development disorder not otherwise specified (NOS). This can usually be done on the basis of age of onset (autistic disorder and Rett disorder have an age of onset prior to 36 months), whether or not there was a period of normal development (present in childhood disintegrative and Rett disorders), and whether or not skills were attained and subsequently lost (also typical of childhood disintegrative and Rett disorders). Autistic children often function within the mentally retarded range, however, unlike autistic children. mentally retarded children generally do not exhibit restricted activities and interests or impairments in communication and social skills. Although a child with schizophrenia can exhibit poor social functioning and affective withdrawal, the onset of childhood schizophrenia usually occurs later, there is a family history of schizophrenia, and the child is less impaired in the area of intellectual functioning. Children with obsessive-compulsive disorder (OCD) can display stereotypical behavior or perform rituals, but they have a more normal course of development otherwise. They also do not exhibit an impairment in social interaction or communication.

Treatment

Autism, perhaps more than any other child psychiatric disorder, requires a well-rounded, multisystemic treatment approach: **family education, behavior shaping, speech therapy, occupational therapy, and educational planning.** Care should be taken to coordinate these activities across school and home settings. Parental support and training are essential to a successful outcome. Applied behavioral analysis can be helpful in autistic patients, especially those with limited verbal skills. This treatment involves an intensive behavioral program that works best if started early in the course of the illness. The goals of this treatment are to teach the child a variety of **basic skills**, such as attending to adults, language use, and how to interact with their peers, all of which can increase the child's ability to be more successful in educational as well as social settings.

No specific medications are used in treating the core symptoms of autism. although some recent studies using low dose Risperdal (risperidone) show some promise. Other psychiatric disorders such as attention-deficit/hyperactivity disorder. OCD, behavior disorders, and psychotic disorders can be present in children with autism. These conditions should be targeted and treated if the symptoms meet the diagnostic criteria for that particular illness. Proper recognition and treatment of comorbid psychiatric disorders can have a significant impact on the overall outcome for children with autism.

Comprehension Questions

- [46.1] Which of the following factors is most likely associated with a more positive prognosis for autistic disorder?
- A. Physical development/performance IQ
 - B. Family socioeconomic status
 - C. Language development/verbal IQ
 - D. Presence of a seizure disorder
 - E. Sibling order
- [46.2] What percentage of children with autistic disorder are mentally retarded?
- A. 100%
 - B. 50% to 75%**
 - C. 25% to 50%**
 - D. 1% to 5%
- [46.3] Stereotyped behaviors in autistic individuals are best illustrated by which of the following?
- A. Interacting preferentially with other children of a specific race
 - B. Preference for similar schedules of play everyday
 - C. Focus of play on only one aspect of a toy
 - D. Difficulty playing with a toy creatively beyond its most obvious use
 - E. Spinning or repetitively using a toy in a specific manner for extended periods of time
- [46.4] Autism can be distinguished best from Rett disorder by which of the following?
- A. Age of onset
 - B. Early language development
 - C. Presence of a period of normal development
 - D. Evidence of mental retardation
 - E. Sex of the child

Answers

- (46.1] C. One of the best predictors of the ability to improve the development of autistic children is the extent to which they have, or have begun to develop, language skills. This ability is often measured in verbal IQ testing.
- [46.2] B. Because 50% to 75% of autistic individuals are mentally retarded, it is important to have an autistic child thoroughly evaluated for mental retardation so that appropriate educational programs can be developed as quickly as possible.

- [46.3] E. Spinning or repetitively using a toy in a specific manner for extended periods of time is characteristic of stereotyped play. Autistic individuals often play with toys or objects in odd or eccentric ways. A typical example of stereotyped behavior in playing with toys is spinning or moving an object or toy in the same way for long periods of time.
- [46.4] C. Both syndromes show evidence of presence prior to the age of 36 months; however, in Rett disorder, the child has an early phase of normal development then gradually loses those skills toward further decline.

CLINICAL PEARLS

Language development is the best predictor of future outcome in autistic disorder.

Mental retardation is often but not always associated with autism.

In cases where early autism is suspected, a full medical workup should always be scheduled to rule out hearing or vision difficulties that can result in poor language development.

REFERENCES

- Ebert M. Loosen P. Nurcombe B. eds. Current diagnosis and treatment in psychiatry. New York: McGraw-Hill. 2000:546-551.
- Kaplan H. Sadock B. Synopsis of psychiatry. 9th ed. Baltimore: Lippincott Williams & Wilkins. 2003:1642-1646.