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Autism

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Case Study

Bradley is a 2½ year old boy who presents to a primary care provider's (PCP) office for his 30 month check-up. Upon administration of the screening tool "Modified Checklist for Autism in Toddlers" (M-CHAT), Bradley scored High Risk for Autism. While discussing with

mom, she reveals that he has a vocabulary of about three words, poor eye contact and doesn't point to things. She has to anticipate his needs because he cannot tell her when he's hungry or thirsty. She states he has some "flailing of his hands and arms" throughout the day and "babbling to himself." During the interview, the primary care doctor notices

that Bradley is playing in the corner alone and is lining up the toy cars on the exam table. Bradley doesn't seem to notice the doctor's presence in the room. He does not turn when the examiner or mother calls his name. His primary care doctor refers him to a developmental pediatrician. What is Bradley's diagnosis and treatment plan?

Case Discussion

Autism Spectrum Disorders (ASD's) are not rare and many PCPs care for several children affected by autism, but not all pediatricians screen on a regular basis. It is very important that PCPs recognize the early signs of ASD as early identification and intervention lead to a more positive outcome. Early identification also allows for etiologic investigation and counseling regarding recurrence risk.

The American Academy of Pediatrics (AAP) recommends all children should be assessed with a standardized developmental tool at specific intervals (9, 18, 24 and 30 month visits) regardless of a concern or risk identified. The following "red flags" warrant evaluation: no babbling, pointing or other gesture by 12 months, no single words by 16 months, no 2-word spontaneous phrases by 2 years, and loss of language or social skills at any age (regression). (Plauche Johnson)

ASD's are neurodevelopmental conditions without an exact etiology, but have strong genetic foundations with environmental

factors also playing a role (to a lesser extent). Some ASD diagnoses are related to other medical conditions such as Fragile X Syndrome, tuberous sclerosis, fetal alcohol syndrome and Rett syndrome.

The increased rate of ASD diagnosis may be attributed to better parental and public awareness, professionals increasing proficiency in recognizing and diagnosing, and changing DSM-V diagnostic criteria to a broader range of symptoms. Although there are not diagnostic imaging studies to confirm ASD, neuropathologic studies have found structural abnormalities in the brain. (Plauche Johnson)

Symptoms of ASD consist in three areas of concern, including social and communication deficits, and restricted interested/ repetitive behaviors. Even though PCP's gather data during brief observations of around 10-20 minutes, these observations are key to early detection and referrals. (Gabrielsen)

Clinical signs of ASD present in varying degrees. Early social deficits include delayed and/ or absent joint attention (JA) or a lack of shared enjoyment in

an activity (where a child looks from person to object and back). Other social deficits may include ignoring parents attempts for attention, content in being alone, decreased eye contact, and decreased attempts to get others attention. The child may not follow a point, respond to their name being called or using facial expressions. The child may not use pointing and/or reaching to show or meet their needs. They may have trouble making or keeping friends, and have trouble understanding another person's feelings or perspective. They may have limited or no speech to communicate, or may use scripted speech called "echolalia." They may demonstrate repetitive behaviors such as hand flapping, rocking or other atypical body movements, and some may be self-injurious. They may demonstrate unusual attachments to inanimate objects or have a fixed, restricted and/or exaggerated interest on one topic. This would be more than typical for their age and to the exclusion of everything else. (Plauche Johnson)

Based on the DSM-V criteria, Bradley would meet the diagnostic measures for ASD.

Treatment: After diagnosis is confirmed by a trained professional (in this case, a Developmental Pediatrician), treatment consists of first providing the family with information about ASD. Treatment plans will be highly individualized to the patient. No two children with ASD will be alike and they will have different challenges in management and diagnosis. They should have an audiology exam and speech evaluation. Treatments may involve the use of genetic laboratory blood tests or imaging. The professional should also rule out any comorbid conditions, such as ADHD, seizures or cognitive delay. Benefits to having a known diagnosis include genetic and diagnosis specific, anticipation of a possible later associated comorbidity, prevention of secondary disorders, access to public support systems/resources, and psychological benefits. (Plauche Johnson)

Pharmacologic treatments have not been proven to correct the core deficits of ASD. Although medications are not a primary treatment, they may be used to treat associated behaviors or psychiatric comorbidities that may interfere in the child's life (seizures, gastrointestinal issues, sleep issues). Specific, specialized and structured educational programs may be used as necessary, as well as social skills instruction/classes. Occupational or sensory integration therapy may be used with some children. Treatment over the course of the lifetime is required as ASD's are not "curable" and require long term management. Chronic management goals are to maximize the child's functional independence and quality of life, minimize core ASD symptoms and maladaptive behaviors, facilitate development/learning, socialization, and to support families. (Myers)

Bradley and his family are thankful for all the services and support from their community and medical professionals. He has been diagnosed and referred for appropriate early intervention programs. He has all the resources he needs at this time and will continue to be monitored for any additional services he may need throughout his life.

References

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Featured Specialist



**Alissa Flach, MSN,
CPNP-PC**

- Has worked at Children's for over five years collectively starting as a Nurse Extern, then PCA, then Registered Nurse on 3 West, now Nurse Practitioner in the Developmental Pediatrics Clinic/ Autism Clinic after a short time as a Primary Care Provider at a private practice.
- Received BSN in 2009, then MSN in 2012, both from Wright State University with a concentration in pediatric primary care.
- Current focus is in the First Steps screening clinic for Autism Spectrum Disorders and also managing behavioral and psychological disorders, as well as medication management for disorders such as ADHD.

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