



Health Sheet

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Post Graduate Training

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Board Certification

Pediatrics; Neurodevelopmental
Disabilities

Clinical Interests

Autism Spectrum Disorders
and early diagnosis of Autism
Spectrum Disorders

DOCTOR'S NOTES

To refer a patient to the Center for
Children with Special Needs (CCSN):

Main Campus, Boston

Little Kids Clinic: Children younger than
3 years: 617-636-8009; All other children
3+ years: 617-636-7242

Pediatric Specialty Center/North Chelmsford

Little Kids Clinic: Children younger than
3 years: 978-937-6362; All other children
3+ years: 978-937-6362

AUTISM Q&A

with Roula N. Choueiri, MD neurodevelopmental pediatrician

ACCORDING TO THE NATIONAL INSTITUTES OF HEALTH, three to six children out of every 1,000 will have an autism spectrum disorder (ASD), and males are four times more likely to be affected than females. Scientists aren't certain what causes ASD, but it is likely that both genetics and environmental factors play a role.

Roula N. Choueiri, MD, Neurodevelopmental Pediatrician in the Division of Developmental-Behavioral Pediatrics at Floating Hospital for Children at Tufts Medical Center, answers some basic questions about this condition.

What is autism?

Autism was first described in 1943, by Leo Kanner, an Austrian psychiatrist. He described children with aloofness, indifference to others, who reacted differently to their environment and had speech delays. Autism: derives from the Greek autos: self.

As we know it today: Autism is part of the Pervasive Developmental Disorders (PDD) which include deficits in three core areas: social skills, language and communication, and repetitive behaviors and/or restricted interests. Onset is seen in the first three years of life.

Under the umbrella of PDD are:

- ▶ Autistic Disorder
- ▶ Asperger Syndrome
- ▶ Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS)
- ▶ Childhood Disintegrative Disorder
- ▶ Rett Syndrome

Autism, Asperger Syndrome and PDD-NOS are grouped into the Autism Spectrum Disorders (ASD). Sometimes autism and ASD are used interchangeably.

Does Autism have grades of severity?

Yes — The "spectrum" in ASD means that there is a wide range of intensity, symptoms and behaviors. Children with Autism Spectrum Disorders may be non-verbal with significant social delays, as in the case of many with "classic" autism, or Autistic Disorder. Or, they can present with delays in their social skills and play, but with normal to superior cognitive skills and normal language development, such as seen in children with Asperger or high-functioning PDD-NOS. All ASDs have delays in the three core areas of: social interaction, communication and repetitive behaviors/restricted interests.

How early can autism be diagnosed?

Autism is usually diagnosed by the age of three years, although it can be recognized and diagnosed by the age of 18 months. Children with Asperger Syndrome and some children with high-functioning PDD-NOS might be diagnosed later, typically by the time they enter kindergarten or first grade.

Previous studies have shown that parents start being concerned between the ages of 12–24 months because of delays in language, play, difficulties with eye contact, but also with repetitive behaviors such as lining up toys or spinning objects or themselves. Other concerns that parents may have at this age: uncontrollable temper tantrums, extreme resistance to change, over- or under-sensitivity to sights and sounds and/or a regression in language and/or play skills.

Can a child be screened for autism?

The American Academy of Pediatrics (AAP) is recommending regular screening for Autism Spectrum Disorders at ages 18 months and 24 months if there are no other risk factors or concerns. This would be part of a well-child visit. However, screening is recommended at any age if there are risk factors and concerns. The AAP has a well detailed algorithm that can be accessed at: <http://www.medicalhomeinfo.org/health/Autism%20downloads/AutismAlarm.pdf>. Several screening tools are validated for ages older than 18 months and there is active research to identify screening tools for children younger than 18 months.

If the child fails the screening test, then he/she needs a referral for early intervention services and at the same time, a referral to a subspecialist for a final diagnosis. Sometimes the pediatrician can give the diagnosis. The earlier the child starts receiving the appropriate intensive services, the better the prognosis.

What causes autism?

We still don't know a lot about the causes of ASDs. We are able to find a cause up to 10 percent of the time. We know that there is a strong genetic basis from family studies. Parents who have a child with an ASD have a 2–8 percent chance of having a second child who is also affected. This genetic basis seems very complex and research is extremely active in this area. ASDs are frequently associated with: Fragile X, tuberous sclerosis, congenital rubella syndrome, and untreated Phenylketonuria (PKU). There are several reported associations with different chromosomal anomalies.

What treatments are available currently?

There are several treatment options currently available but they can be grouped into four categories, with the behavioral and educational interventions being the most proven, researched and the one that addresses treatment of the core features of autism spectrum disorders.

Treatments include:

- ▶ Applied Behavior Analysis (ABA): remains the foundation of most comprehensive programs; it is based on positive reinforcers and breaking down directions and tasks to simple tasks which leads to learning through repetition.
- ▶ Floortime: which is less structured but emphasizes the role of parents and family members; the child leads and the therapist supports the child's social development and play.
- ▶ Treatment and Education of Autistic and Communication-related Handicapped children (TEAACH): This program uses visuals and structure for learning.
- ▶ Relationship Development Intervention (RDI): is a parent-based treatment that focuses on social and emotional development.

Other clinical therapies that are essential in therapeutic programs are speech therapy, occupational therapy, and social skills group teaching.

Alternative treatments such as Chelation, megavitamin doses supplementation, and dietary changes such as the gluten/casein-free diet, are not proven and some can have side-effects. There is an ongoing study looking at environmental factors and heavy metals. The last category is the use of medications to help with associated disorders such as attention, hyperactivity, sleep issues, anxiety, depression, seizures or treating aggressive behaviors.

What advice can one give to parents who have a child with an ASD?

Parenting a child with an ASD can be emotionally and physically exhausting. Keeping up with new research, theories and treatments can be extremely stressful. Parents should be sure to:

- ▶ Take care of yourselves: rest and schedule times for yourselves.
- ▶ Talk to other parents and families and learn what worked and what did not work for them; join support groups, online list serves.
- ▶ Go to seminars and conferences organized by academic centers, scientific and knowledgeable groups to learn more about ASDs and the scientific basis of treatments.

Your child is unique in his/her differences. Learn to appreciate him/her, and be ready to be surprised by his/her progress and don't forget to still enjoy the ride! □

RESOURCES

Recommended web sites for more information: www.firstsigns.org
www.cdc.gov/ncbddd/autism/
www.aap.org/healthtopics/autism.cfm