



A Clinician's Approach to Autism

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Autism Spectrum Disorder

- A psychiatric disorder defined in the “Diagnostic and statistical manual of mental disorder (DSM-5) created as a guide by the American Psychiatric Association”
- A Developmental Disorder whose symptoms usually appear in the first 2 years of life.

- ASD is a spectrum of symptoms that vary in severity in affected individuals but characterized by:
 - Problems with communication with other people. Seen in typical interactions with peers and family.
 - Repetitive behaviors and restrictive interest.
- These symptoms inhibit the child's function at school, home, and work.

Incidence

- 1 in 59 children have been identified with autism according to estimates from CDC Autism and Developmental Disabilities Monitoring Network
- Autism is reported to occur in all racial, ethnic, socioeconomic groups
- 4x more common in boys than girls
- 1 in 6 children in the U.S. had a mild to moderate developmental disability

Symptoms in Social Interaction and Communication Include:

- Inconsistent Eye Contact
- Problems in acknowledging name or getting the child's attention
- Not sharing enjoyment with others
- Problems with engagement and developing a conversation
- Restricted interests and allowing others to share their interests in a conversation
- Mask like facial expressions
- Expressionless or sing song voice
- Inability to understand another persons point of view or inferences

Repetitive Behaviors

- Echolalia
- Interest in favorite objects or subjects (details or numbers)
- Overly fascinated in moving objects or parts
- Easily upset by changes in routines
- More sensitive to sensory input like mechanical noises, clothes, or touch

Diagnosis

General Developmental Screening

- American Academy of Pediatrics
 - Suggests Developmental Screening at 9,18,24, and 30 months of age
 - Referral to specialists experienced in the diagnosis of Autism
 1. Developmental Pediatrician
 2. Pediatric Neurologist
 3. Child Psychologist
 4. Child Psychologist Ph.d.
 5. Neuropsychologist
- Observation, History, and Testing are all used to make a diagnosis in ASD

Risk Factors for Autism

- Sibling with ASD
- Genetic Conditions
- Down Syndrome
- Fragile X
- Tuberous Sclerosis
- Rett Syndrome
- Very Low Birth Weight



Children with ASD may also have problems with:

- Selective Diet
- Sleep
- Irritability



Tests

- Your physician may want:
 - Blood Tests
 - Genetic Testing
 - Possibly Imaging
 - Electroencephalograph
 - Psychoeducational Testing at School
 - Physical and Neurological Examination
 - Cognitive Evaluation
 - Language Skills
 - Age Level
 - Social
 - Fine Motor
 - Gross Motor



Common Tests Used in the Diagnosis of Autism:

- Autism Diagnostic Observation Schedule
- Behavior Assessment System for Children
- Gilliam Autism Rating Scale
- Weschsler Preschool and Primary Scale of Intelligence
- Childhood Autism Rating Scale

Structural Variation of Chromosomes in ASD

- Autism does not have a specific chromosome defect. There are many well known associations recognized in 10% of cases. Most common is:
 - Fragile X
 - Rett Syndrome
 - Tuberous Sclerosis
- Some detectable chromosome abnormality is found in 7.4% of cases

- Most frequent anomaly is the maternally derived duplication of chromosome
 - 15q11 – q13 in 1-3% of cases
 - 22q13, 8,9 the SHANK3 mutation
 - NLGN3 and NLGN4 genes on X chromosome
 - 2p16.4, 11, 12 and as many as probably over 150 mutations

Therapy

- Behavioral Therapy
- Speech
- Occupational
- Physical





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