1 in 40:
Diagnosis and Treatment of Autism Spectrum Disorder

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Disclosure

- I have no commercial interests or conflicts

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Objectives

- Learn about the early signs and symptoms of Autism Spectrum Disorder
- Learn about screening of ASD and the referral process
- Learn the diagnostic criteria for ASD
- Learn about current treatment options for ASD

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Autism Spectrum Disorder

- DSM-5 Criteria
  - Persistent deficits in social communication and interaction
  - Restricted, repetitive patterns of behavior, interests or activities
  - Symptoms must be present in early development

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Autism Spectrum Disorder

"Autistic disturbances of affective contact"

- Described by Dr. Leo Kanner in 1943
- Profound lack of reciprocal social engagement
- Disturbances in communication
- Unusual response to environment
- Insistence on sameness

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Prevalence

- National Health Interview Survey 2016
  - Approximately 1 in 40
  - Increased male to female ratio
    - 1:26 for boys and 1:93 for girls
    - 3 to 4 times more common in boys
- Sibling risk of 3-10%
  - American College of Medical Genetics and Genomics
  - 7% risk if affected child is female
  - 4% risk if affected child is male
  - 30% risk if 2 or more siblings previously diagnosed

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Prevalence

- Increasing incidence
  - Changes in definition
  - Increased public and professional awareness
  - Early detection and screening
  - Availability of specialized developmental services
  - Change in special education laws
  - Recognition that ASD is a spectrum

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Pathogenesis

- Genetic factors that alter brain development
  - Altered neural connectivity due to abnormal neuronal differentiation in prenatal development
  - Accelerated head growth with increased overall brain size
  - Changes in brain chemical concentrations, neural networks and cortical structure and organization
- Interactions between multiple genes or gene combinations
  - Unequal sex distribution, increased prevalence in siblings, and high concordance with monozygotic twins

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Pathogenesis

- Most individuals do not have an identifiable etiology
- Approximately 10-20% have a genetic syndrome, mutation, or gene copy variants
  - Tuberous Sclerosis Complex
    - 40% patients have ASD
    - Only 0.4-4% of patients with ASD have Tuberous Sclerosis
  - Fragile X
    - 30% patients have ASD
    - Fragile X is only found in 0.5% patient's with ASD

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Early Signs and Symptoms

Lack of communication skills before two years of age

- No babbling by 9 months
- No pointing or gestures by 12 months
- No orientation to name by 12 months
- No single words by 16 months
- No spontaneous meaningful two-word phrases by 24 months

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Early Signs and Symptoms

Deficits of social skills or plateau of development

- Do not smile responsively
- No desire to be picked up
- Indiscriminate affection
- Lack of interest in other people
- Poor eye contact or gaze aversion
- No pretend or symbolic play by 18 months

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Early Signs and Symptoms

Autistic Regression

- 20-35% children have regression or plateau of language and interactive skills
- Occurs after typical development
- Usually between 15-24 months
- Gradual or sudden
- Loss of meaningful spoken language

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Surveillance and Screening

- Routine developmental screening
  - 9, 18, and 24 or 30 months
  - Ages and Stages Questionnaire
  - Additional screening if there are concerns about development or communication
- Autism Specific screening
  - MCHAT-R at 18 and 24 months

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Surveillance and Screening

- LISTEN TO PARENTS
  - Most parents (85%) had concerns about develop when their child was under 3 years of age but only 60% of children had initial evaluations before 4 years of age
  - Median age of diagnosis 46-67 months
  - Most common concerns are speech delays or frequent tantrums
  - Delay in evaluation and diagnosis due to the variability of signs and severity of symptoms

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Referral

- Children with Michigan Medicaid
  - Community Mental Health Organization
  - Referral process varies by county
- Private Insurance
  - BCBS requires diagnosis from an approved Autism Evaluation Center
  - Currently 18 centers in the state
  - BCBS will not cover services if diagnosis is from a medical provider not associated with an approved center

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Diagnostic Day

- Team approach
  - BCBA (Board Certified Behavior Analyst), Speech-Language Pathologist, Psychologist and Pediatric Physician
- Standardized Tests
  - ADOS: Autism Diagnostic Observation Schedule
  - ADI-R: Autism Diagnostic Interview
  - Various speech and language evaluations
- Family Interview
  - Developmental Milestones
  - Medical, social and family history

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DSM-5 Criteria

A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history:

1. Deficits in social-emotional reciprocity
2. Deficits in nonverbal communicative behaviors used for social interaction
3. Deficits in developing, maintaining, and understanding relationships

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DSM-5 Criteria

B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following:

1. Stereotyped or repetitive motor movements, use of objects or speech
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior
3. Highly restricted, fixated interests that are abnormal in intensity or focus
4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment

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DSM-5 Criteria

C. Symptoms must be present in early developmental period

D. Symptoms cause clinically significant impairment in social, occupations, or other important areas of current functioning

E. These disturbances are not better explained by intellectual disability or global developmental delay.
- Intellectual disability and autism spectrum disorder frequently co-occur
- For comorbid diagnoses of both, social communication should be below that expected for general developmental level

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Criteria A-1: Social-Emotional Reciprocity

- Abnormal social approach
  - "No personal space"
  - Getting too close to a social partner and not recognizing when the partner is uncomfortable
  - Indifference to socially motivated physical contact or affection
- Reduced sharing of interests, emotions or affect
  - Limited interest in social interaction
  - Limited attempts to draw the attention of others to objects by pointing or showing
  - Lack of joint attention

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Criteria A-1: Social-Emotional Reciprocity

- Failure to initiate or respond to social interactions
  - Limited interest in social interactions with other children
  - Content playing passively by themselves
- Failure of normal back-and-forth conversation
  - Difficulty initiating or sustaining conversation
  - One-sided conversations with limited reciprocal exchange
  - Difficulty staying on topic, not taking turns, or becoming overly focused on personal interests
  - Overly literal – difficulty understanding sarcasm, irony or jokes
  - Abnormal prosody – volume, rhythm, intonation

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Criteria A-2: Nonverbal Communication

- Impaired ability to use and interpret nonverbal behavior
- Abnormalities in eye contact and body language
  - Avoid eye contact or gaze too intently
  - Turning away or not engaging
- Limited facial expressions
  - Do not change facial expressions or expressions are overly exaggerated
- Deficits in understanding and use of gestures
  - Awkward or absent gestures – pointing, waving, showing
  - Only uses primitive gestures – leading or pulling a hand

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Criteria A-3: Relationships

- Absence of interest in peers
  - Little or no interest in relationships
  - "Independent" or "Self-sufficient"
- Difficulties in shared imaginative play
  - Impaired pretend play or playing alone
  - Difficulty imitating others
  - Lack of cooperative play

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Criteria A-3: Relationships

- Difficulties making friends
  - Failure to form and maintain developmentally appropriate peer relationships
  - No sharing of mutual interests, activities or emotions
  - Inability to understand the difference between acquaintances and friends
- Difficulties adjusting behavior to suit various social contexts
  - Lack of empathy
  - Misunderstanding the emotional response of others
  - Responding inappropriately to another's distress
  - Socially immature

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Criteria B-1: Stereotypical Behavior

- Motor stereotypes
  - Hand flapping, finger flicking or posturing
  - Rocking, swaying, jumping, spinning, prancing on toes
- Repetitive motions or use of objects
- Idiosyncratic phrases
  - Pronoun reversal
- Echolalia
  - Immediate – usually in response to questions
  - Delayed – scripted verses or memorized dialogue

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Criteria B-2: Routines, Rituals

- Insistence on sameness and inflexible adherence to routines
  - Extreme distress at small changes in routine
  - Difficulties with transitions
- Rigid thinking patterns
  - Do not tolerate deviations from normal or expected rules of conduct
- Nonfunctional rituals or routine
  - Lining up and arranging objects
  - Repetitive questions about a particular topic

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Criteria B-3: Restricted interests

- Excessively circumscribed or perseverative interest
  - Difficulty shifting their attention away
  - Very specific and intense
- Preoccupation with specific objects or parts of objects
- Strong attachment with unusual objects

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Criteria B-4: Abnormal Sensory Response

- Apparent indifference to pain or temperature
- Hypersensitivity to certain types of sound or frequencies
- Adverse response to specific textures
  - Restriction of certain food textures
  - Increased sensitivity to certain kinds of touch – light touch, clothing
- Excessive smelling or touching of objects
  - Compulsive touching of certain textures
  - Sniffing or licking nonfood objects
- Visual fascination with lights or movement
- Peering

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DSM-5 Diagnosis

- Must meet all 3 of Criteria A: Deficits in social communication and interaction
- Must meet 2 out of 4 of Criteria B: Restricted, repetitive behaviors
- Symptoms must be present in early childhood

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Treatment

- Goal is to maximize functioning and improve quality of life
  - Improve social functioning and play skills
  - Improve communication
  - Improve adaptive skills
  - Decrease nonfunctional or negative behaviors
- Individualized to child's age and specific needs

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Treatment

- Treatment settings
  - Early intervention program
  - School-based special education with IEP
  - Private therapies

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Treatment

- Treatment Program Features
  - Early and intensive
  - High staff-to-student ratio with small group instruction
  - Minimum of 25 hours per week and year long
  - Structured and predictable
  - Ongoing evaluation and program adjustment
  - Supportive teaching environment
  - Transition planning
  - Family involvement

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Treatment

- Applied Behavior Analysis
  - Intensive intervention that focuses on behavior modification
    - Reinforce desirable behaviors and decrease undesirable
    - Teach new skills by breaking them down into the simplest elements
    - Repeat reward-based trials
  - Most well-established, evidence-based treatment
    - Improvement in core symptoms and maladaptive behaviors
    - Most significant improvement within the first 12 months of treatment

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Treatment

- Speech and Communication Therapy
  - Promote communication skills
    - Expressive, receptive, and pragmatic language
  - Traditional speech and language therapy along with behavioral strategies
  - Most effective when incorporated into daily routine
  - Augmentative communication for nonverbal patients
    - PECS – Picture Exchange Communication System

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Treatment

- PECS: Picture Exchange Communication System

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Treatment

- Occupational Therapy
  - Focus on deficits in fine motor skills
  - Sensory integration therapy
    - Introduction of intensive sensory inputs
    - Theory is to remediate deficits in neurological processing and integration of sensory information
    - Goal is to allow child to interact with the environment
    - Validity remains controversial

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Treatment

- Social Skills
  - Improve overall social competence and friendship quality
  - Learn to initiate social interactions and maintain joint attention
  - Taught through modeling, peer training, or story-based

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Treatment

- Parental Involvement
  - Training parents in specific behavior management
  - Improvement in disruptive and noncompliant behaviors
  - Help families interact with their child
  - Increase parental satisfaction, empowerment, and mental health

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Treatment

- Medical management
  - Psychotropic medications have not been proven to correct the core deficits of ASD
  - Directed to specific symptoms or coexisting conditions
    - ADHD, ODD, Anxiety, Depression
  - Initiated after educational and behavioral interventions
  - Risperidone and Ariprazole
    - Only two medications specifically approved for treatment of ASD
    - Effective for irritability, tantrums, aggression, self-injurious behavior

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Treatment

- Complementary and Alternative Medicine (CAM)
  - "Group of diverse medical and health care systems, practices, and products that are not presently considered to be part of conventional medicine"
  - Most are inadequately evaluated and cannot be recommended
  - Potential risks
    - Medication interactions, Toxic effects of biological agents
    - Delay in starting validated therapy, Financial cost

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Treatment

- Complementary and Alternative Medicine (CAM)
  - Gluten-free, casein-free diet – possible nutritional deficiencies
  - Hyperbaric oxygen – very costly
  - Music Therapy – inconsistent data
  - Animal Therapy – only observational studies
  - Auditory Integration training – efficacy unproven and expensive
  - Omega-3 – randomized trials have not shown benefits
  - Probiotics – not adequately tested
  - Cannabinoids (CBD) – limited research

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Role of Primary Care Provider

- Providing routine health maintenance and preventative care
- Monitor dietary intake and nutritional deficiencies
- Treating GI symptoms - constipation or diarrhea
  - Behavioral and/or related to dietary intake
- Discussion of sleep problems in 40-80%
  - Difficulty falling asleep, staying asleep or early awakening
- Monitor for seizures, 20-35% have epilepsy

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Role of Primary Care Provider

- Safety guidance
  - Frequent wandering
  - Approximately 50% of children will try to elope at least once
- Family support and guidance
  - Enrolling in therapies
  - Special education laws – IEP, 504 plan
  - Discussion about complementary and alternative therapies

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Questions???

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References

- American Psychiatric Association. (2013) *Diagnostic and Statistical Manual of Mental Disorders, 5th Edition.* American Psychiatric Publishing.
- Augustyn, MD , M., & von Hahn, MD, E. (2018, December) *Autism spectrum disorder: Clinical features.* Retrieved from UpToDate.
- Augustyn, MD, M. (2019, January) *Autism spectrum disorder: Terminology, epidemiology, and pathogenesis.* Retrieved from UpToDate.
- Augustyn, MD, M., & von Hahn, M. E. (2019, January) *Autism spectrum disorder: Evaluation and diagnosis.* Retrieved from UpToDate.

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References

- Robert G. Voigt, MD. (2011) *Developmental and Behavioral Pediatrics.* American Academy of Pediatrics.
- Weissman, MD, L., & Bridgemohan, MD, C. (2018, September) *Autism spectrum disorder in child and adolescents: Overview of management.* Retrieved from UpToDate.
- Weissman, MD, L., & Bridgemohan, MD, C. (2018, August) *Autism spectrum disorder in children and adolescents.* Retrieved from UpToDate.
- Weissman, MD, L., & Bridgemohan, MD, C. (2018, June) *Autism spectrum disorder in children and adolescents: Behavioral and educational interventions.* Retrieved from UpToDate.

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