

Learning Objectives

- Emphasize that the diagnosis of ASD is often delayed to show the importance of screening and diagnosis
- Highlight the key features of the history and physical exam when assessing for ASD
- · Review the key diagnostic tools for ASD
- Identify other evaluations important in the assessment of ASD
- Evaluate treatment options for ASD, including behavioral and educational interventions, pharmacologic interventions, and complementary and alternative therapies

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Delayed Diagnosis Multicenter surveillance study 85% caregivers expressed concern by 36 nonths of age 61% undergone a comprehensive evaluation by 48 months Median age diagnosis 52 months

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Delayed Diagnosis

Challenges

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- Time constraints of office visit
- Vague nature of social developmental milestones
- Variability of signs and symptoms of ASD

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Delayed Diagnosis: Factors

- Less severe symptoms
- Female sex

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- Coexisting conditions
- Lack of continuity of care
- Hearing impairment
- Attribution of regression of skills to stressors

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Delayed Diagnosis: Factors

- Language barriers
- Underrepresented ethnic group
- Living in rural area
- Lower socioeconomic status

Formal Evaluation

- Multidisciplinary team
- Meet diagnostic criteria?
- Level of function, neurodevelopmental strengths and weaknesses
- Different condition?

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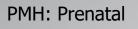
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Associated conditions?

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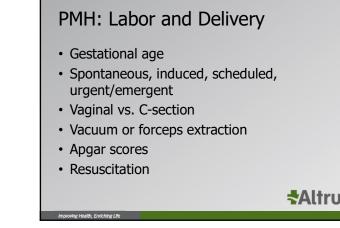
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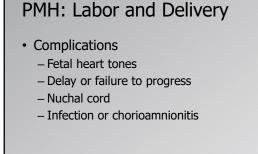
- Maternal and paternal age
- Gravida para, miscarriages
- Ultrasounds
- Fetal movements
- Maternal history during pregnancy
 - Medical and surgical history
 - Weight gain

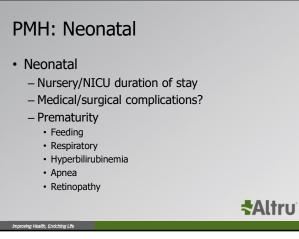
- Prescription and recreational drug use

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PMH: Development

- Autistic behaviors and traits
- Speech and language development
- Gross and fine motor development
- Previous evaluations
- Previous and current interventions

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Physical Exam

- · Vitals, esp. weight
- General appearance and observations
- Head: shape, fontanelles, OFC, cephalic index
- EENT
- Neck: Palpate, auscultate carotids

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Neurologic Exam Mental status Awakeness, alertness, NAD? Orientation Mood and affect Speech and Language Rate, volume, articulation

- Vocabulary and sentence formation
- Receptive and nonverbal language

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Past Medical History

- Acute or chronic illnesses
- Major hospitalizations or surgeries
- Psychiatric history
- Medications and supplements
- Allergies

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 Family history: 1st degree relatives, multiple individuals, consanguinity

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Physical Exam

- Heart and Lungs
- Abdomen: Organomegaly
- Skin: Neurocutaneous findings
 Wood lamp
- Spine: curvature, bony prominences or masses, sacral dimple or hair tuft

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Neurologic exam Cranial nerves Motor exam Muscle tone Reflexes

- Sensory exam
- Coordination and gait

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Autism Diagnostic Observation Schedule-2nd edition (ADOS-2)

- Administered by specialist, requires intensive training
- More sensitive than other diagnostic tools (94%), similar specificity (80-88%)
- Assesses severity
 - In conjunction with an adaptive scale such as Vineland Adaptive Behavior Scale

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Differential Diagnosis

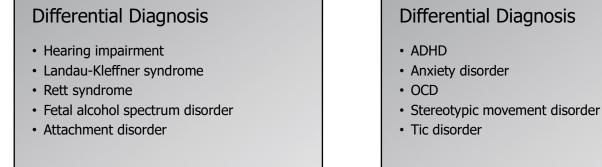
- Developmental delay/intellectual disability
- Intellectual giftedness
- Social (pragmatic) communication disorder
- Language disorder
- Language-based learning disorder
- Nonverbal learning disorder

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Evaluations: Labs

- Lead
- Metabolic
 - CBC, CMP, TSH/free T4, CK, lactate, ammonia, pyruvate, amino acids, acylcarnitine panel, urine organic acids
- Genetic

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- First tier: Fragile X testing and chromosomal microarray
- Second tier: MECP2, PTEN, WES
- Neuropsych

 Ceruloplasmin, ANA, autoimmune encephalopathy panel
- Lumbar puncture and CSF analysis



Evaluations: Referrals Audiology Neuropsychology/ Psychology/Psychiatry Opthalmology/ Developmental and Optometry behavioral pediatrics • PT, OT, speech Other pediatric Educational subspecialties evaluations Pediatric neurology subspecialties Altru

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Decrease nonfunctional/negative behaviors Decrease nonfunctional/negative behaviors Permote academic and occupational performance Neurodiversity vs. disorder Person-first and "treatment" verbage

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Treatment Behavioral and educational interventions Pharmacologic interventions Complementary and alternative therapies

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Behavioral and Educational Interventions

- Close monitoring and modification as the child's needs change
- Curriculum that emphasizes attention, imitation, communication, play, social interaction, regulation, and self-advocacy
- Highly supportive teaching environment
- Predictability and structure
- Transition planning

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Behavioral and Educational Interventions

- Minimum of 25 hours and 12 months per year
- High staff-to-student ratio (at least 1:2)
- Individualized
- Family involvement
- Functional analysis of behavioral concerns
- Ongoing program evaluation and adjustment

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Applied Behavioral Analysis (ABA)

- Behavioral analysis
 - Positive and negative behaviors
 - External stimuli

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- Impact on learning and ADLs
- Applies that information to:
 - Increase language and communication skills
 - Improve attention, focus, social skills, memory, and academics
 - Decrease problem behaviors

Applied Behavioral Analysis (ABA)

- · Flexible and individualized
- · Positive reinforcement, reward-based
- Antecedent, Behavior, Consequence (ABC)
 - Antecedent: what occurs prior to the behavior
 - Behavior: response to the antecedent
 - Consequence: what comes after the behavior
 - All 3 can be targeted to reinforce positive behaviors and improve negative behaviors

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Applied Behavioral Analysis (ABA)

- Ongoing monitoring, assessment, and planning
- Considered an evidence-based best practice treatment by the US Surgeon General and by the American Psychological Association.
- Covered by Medicaid and many private insurances
- Limited resource

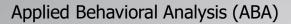
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Other Interventions

- Specific behavioral interventions
- · Communication, speech, and language
 - Speech therapy
 - Social skills
- Occupational therapy
- Sensory integration therapy
- Transition programs

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- · Flexible and individualized
- Positive reinforcement, reward-based
- Antecedent, Behavior, Consequence (ABC)
 - Antecedent: what occurs prior to the behavior
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Developmental and Educational Interventions

- Especially effective in conjunction with medical-based therapies
- Early intervention

 In-home therapies and other services
 School based recourses
- School based resources
 - School-based therapies
 - Academic support

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– IEP's and 504 plans

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Pharmacologic Interventions

- May be a useful **adjunct** to behavioral and educational interventions
- Children w/ ASD can be more sensitive to medications
- Can be difficult to identify the target symptom
 - Behavioral analysis can help
- Can be challenging to monitor side effects

Behavioral Concerns

- Irritability, aggression, self-injury
- Risperidone (FDA-approved)
 - Weight gain, increased appetite, fatigue, drowsiness, dizziness, drooling, tremor, constipation
- Aripiprazole (FDA-approved)
- Olanzapine

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Repetitive Behaviors and Rigidity

- SSRIs: Fluoxetine and Sertraline – Increased SI
- Clomipramine: serotonin-selective TCA
 More established with OCD
- Risperidone
- Valproate
- Bispirone

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Depression and anxiety

- SSRIs and SNRIs
- Mood lability
 - Risperidone, SSRIs, mood stabilizers (lithium)

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Behavioral Concerns

- Other atypical antipsychotics

 Clozepine, quetiapine, ziprasidone
- Haloperidol

 Less effective than risperidone,
 - extrapyramidal symptoms
- Alpha-2 agonists (esp. clonidine)
- N-acetylcysteine

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 ASMs, mood stabilizers, SSRIs, stimulants, beta blockers

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Hyperactivity, Impulsivity, and Inattention

- Stimulants
 - Methylphenidate: response lower, side effects more frequent
- Alpha agonists (guanfacine, clonidine)

 Hypotension, rebound HTN, HA, nocturnal enuresis
- Atomoxetine
- Risperidone

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Social Deficits

- Investigative
- Oxytocin may improve social interaction and function
- D-cycloserine, tetrahydrobiopterin, cognition enhancers (galantamine, memantamine, rivastigmine)

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Associated Conditions

Seizures

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- GI problems
- Sleep disturbance
 - Melatonin
 - Iron supplementation as necessary
 - Meds used to treat other symptoms
- Parasomnias: Clonazepam and TCAs

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Complementary and Alternative Therapies Biologic hypotheses: GI, food sensitivities

- and allergies, inflammation/autoimmune, metabolic, heavy metal toxicity, nutritional imbalances
- Not without side effects
- May compete with validated treatments for time, effort, and financial resources
- Healthy lifestyle encouraged

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No Benefit

- · Secretin: GI hormone inhibits intestinal motility and release of gastric acid, stimulates secretion of pancreatic fluid and bicarbonate
- Facilitated communication: fascilitator physically guides hand of nonverbal child in using output device for communication

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Potential Benefit, Potential Risk · Gluten-free casein-free diet: only if evidence of celiac disease or true gluten sensitivity - Evidence is limited and weak - Strict adherence difficult - Potential nutritional deficiencies Altru 46

Unknown Benefit, Potential Risk

- IVIG: only if indicated for something else
- · Chelation: Not recommended
- Hyperbaric oxygen: Costly, Efficacy unknown
- Antimicrobial agents: Not recommended
- Vitamins B6, A, and D; Magnesium
- · Homeopathic remedies: unknown ingredients
- VNS: epilepsy, unproven benefit with ASD
- Stem cell transplant: expensive

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Potential Benefit, Low Risk

- Music therapy
- Melatonin
- Oxytocin
- Hippotherapy and other animal therapies
- · Sulforaphane: antioxidant derived from broccoli sprout exctracts More studies necessary
- Transcranial magnetic stimulation

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Potential Benefit, Low Risk

• Yoga

- Body work and energy therapies
- Biofeedback/neurofeedback
- Hypnotherapy
- Low dose vitamins
 - Vitamin C

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- Vitamin B12
- Folinic acid and folic acid

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Unknown Benefit, Low Risk

- Amino acids (dimethyglycine)
- Digestive enzymes
- Mindfulness therapy
- Acupuncture
- Craniosacral manipulation
- Chiropractic

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Resource

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Unknown Benefit, Low Risk

- Auditory integration training
- Omega-3 fatty acids
- Probiotics
- Cannabinoids
- Methylcobalamin and N-acetylcysteine
- Zinc

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Herbal products

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