

Evaluation and Management of Autism Spectrum Disorder

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Disclosures

Relevant Financial Relationship(s)

None

Off Label Usage

None



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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 months of age
 - 61% undergone a comprehensive evaluation by 48 months
 - Median age diagnosis 52 months



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Delayed Diagnosis

- Challenges
 - 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
- 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



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Formal Evaluation

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



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PMH: Prenatal

- 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: Labor and Delivery

- Gestational age
- Spontaneous, induced, scheduled, urgent/emergent
- Vaginal vs. C-section
- Vacuum or forceps extraction
- Apgar scores
- Resuscitation



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PMH: Labor and Delivery

- Complications
 - Fetal heart tones
 - Delay or failure to progress
 - Nuchal cord
 - Infection or chorioamnionitis



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PMH: Neonatal

- Neonatal
 - Nursery/NICU duration of stay
 - Medical/surgical complications?
 - Prematurity
 - Feeding
 - Respiratory
 - Hyperbilirubinemia
 - Apnea
 - Retinopathy



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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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Past Medical History

- Acute or chronic illnesses
- Major hospitalizations or surgeries
- Psychiatric history
- Medications and supplements
- Allergies
- Family history: 1st degree relatives, multiple individuals, consanguinity



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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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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

- 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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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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Differential Diagnosis

- Hearing impairment
- Landau-Kleffner syndrome
- Rett syndrome
- Fetal alcohol spectrum disorder
- Attachment disorder



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Differential Diagnosis

- ADHD
- Anxiety disorder
- OCD
- Stereotypic movement disorder
- Tic 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
 - First tier: Fragile X testing and chromosomal microarray
 - Second tier: MECP2, PTEN, WES
- Neuropsych
 - Ceruloplasmin, ANA, autoimmune encephalopathy panel
- Lumbar puncture and CSF analysis



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Evaluations: Imaging

- MRI brain with and without contrast
 - Low yield in children with ASD and no other neurologic findings
 - Helpful in finding etiology and associated conditions



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Evaluations: Referrals

- Audiology
- Ophthalmology/ Optometry
- PT, OT, speech
- Educational evaluations
- Neuropsychology/ Psychology/Psychiatry
- Developmental and behavioral pediatrics
- Other pediatric subspecialties
- Pediatric neurology subspecialties



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Treatment

- Comprehensive and multidisciplinary
- Individualized
- Goals
 - Improve social function and communication
 - Improve adaptive and independent skills
 - Decrease nonfunctional/negative behaviors
 - Promote 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

- 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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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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Applied Behavioral Analysis (ABA)

- Behavioral analysis
 - Positive and negative behaviors
 - External stimuli
 - 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



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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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Developmental and Educational Interventions

- Especially effective in conjunction with medical-based therapies
- Early intervention
 - In-home therapies and other services
- School based resources
 - School-based therapies
 - Academic support
 - IEP's and 504 plans



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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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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



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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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Behavioral Concerns

- Other atypical antipsychotics
 - Clozapine, quetiapine, ziprasidone
- Haloperidol
 - Less effective than risperidone, extrapyramidal symptoms
- Alpha-2 agonists (esp. clonidine)
- N-acetylcysteine
- ASMs, mood stabilizers, SSRIs, stimulants, beta blockers



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Repetitive Behaviors and Rigidity

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



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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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Depression and anxiety

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



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Social Deficits

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



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Associated Conditions

- Seizures
- 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



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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
- Sulfuraphane: antioxidant derived from broccoli sprout extracts
 - 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
 - Vitamin B12
 - Folic acid and folic acid



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Unknown Benefit, Low Risk

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



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Unknown Benefit, Low Risk

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



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Questions?



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