Understanding the Diagnostic Evaluation for Autism Spectrum Disorder

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April 26, 2017
Disclosure

- We have no relevant financial relationships with the manufacturers of any commercial products and/or commercial services discussed.
- We do not intend to discuss unapproved/investigative use of a commercial product/device.
Common Questions from Families:

• Who can diagnose ASD?
• What is required for accurate diagnosis?
• How does a “good” or “bad” day affect diagnosis?
• What’s the difference between a medical versus school diagnosis and how can each impact access to services?
• Is Neuropsych testing required for diagnosis?
• Should a child take their ADHD medication for an evaluation?
Outline Part 1

Definition and diagnostic criteria
Recognizing ASD
Challenges in ASD diagnosis
Medical evaluation of ASD
Autism Spectrum Disorder

• Biologically based neurodevelopmental disorder
• Onset in early childhood
• More common in boys (4:1)
• Primarily genetic etiology
• Earlier diagnosis and treatment improves outcome
DSM-5 Diagnostic Criteria for ASD

Social Communication and Social Interaction

- Social emotional reciprocity
- Non-verbal communication
- Developing, maintaining and understanding relationships

Restricted and Repetitive Behavior

- Stereotyped movement or speech
- Insistence on sameness, routines, rituals
- Restricted, fixated interests
- Atypical sensory reactivity

Symptoms must be present from early childhood AND significantly impact functioning APA 2013
DSM-5 Criteria for ASD

➤ Severity Rating – rated for social and behavioral
  ▪ Level 1 – Needing support
  ▪ Level 2 – Needing substantial support
  ▪ Level 3 – Needing very substantial support

➤ Specifiers
  ▪ Cognitive function
  ▪ Language function
  ▪ Associated medical, genetic or environmental factor
  ▪ Other neurodevelopmental, mental, or behavioral disorder
  ▪ Catatonia
Early Red Flags for Autism

- Poor eye contact
- Not responding to name
- Less showing and sharing
- Early language delay
- No gesturing by 12 months (pointing, waving)
- Delayed or unusual play skills
- Tantrums with change or transition
- Loss of language or social skills

Can detect as early as 8-12 months
Lack of Response to Name

CLICK HERE FOR VIDEO
Decreased Reciprocal Interaction

CLICK HERE FOR VIDEO
High Functioning Children With ASD

Missed on early screening
Normal early language and gestures

Pre-School:
- Limited pretend play
- Avoidance of peers
- Odd interests (Thomas, Blues Clues)
- Self-directed, resist transitions, hyperactivity, anxiety

School Age:
- Interest in peers, lack skills
- Poor conversation, difficulty with non-verbal communication
- Trouble understanding emotions, relationships
- Non-compliance, inattention, anxiety

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Boston Children's Hospital Autism Spectrum Center

HARVARD MEDICAL SCHOOL TEACHING HOSPITAL
How are children with ASD identified?

- Family concerns
- Screening by pediatrician
- Daycare, Early Intervention or School concerns
Who can diagnose ASD?

• Physicians
  – Pediatricians
    • Developmental-behavioral pediatricians
    • Neurodevelopmental specialists
  – Neurologists
  – Psychiatrists
• Nurse practitioners
• Psychologists
What other professionals may be involved in the evaluation?

- Speech language pathologists
- Occupational therapists
- Physical therapists
- Educational specialists
- Other medical specialists
What is required for accurate diagnosis?
Evidence base and specialty guidelines support *multidisciplinary, developmentally informed assessment*

- National Research Council 2001
- American Academy of Pediatrics 2007
- American Academy of Child and Adolescent Psychiatry 2014

Diagnosis of ASD is Complex

- Based on *patterns of behavior*
- Spectrum means many presentations possible
- No one symptom is diagnostic or predictive
- Symptoms can change with age and developmental level
- Many conditions share some ASD symptoms
- Differential diagnoses can also occur with ASD
ASD often occurs with other symptoms and conditions

- Sleep disorders
- Toileting problems
- Restrictive feeding patterns
- Obesity
- Anxiety
- Depression
- ADHD symptoms
- Disruptive behaviors (tantrums, aggression, self-injury)
- Seizures
Differential Diagnosis – Consider Level of Social Function and Repetitive Behaviors Compared to Other Skills

- Language delay
- Global delay or Intellectual disability
- ADHD
- Anxiety
- Social communication disorder
Diagnostic accuracy is improved with

• Consideration of cognitive & language function
• Age over 2 years
• Assessment by clinicians who have expertise in ASD diagnosis
• Information from multiple sources
Challenges in Very Young Children

• Children under two years of age may not show enough symptoms under restrictive repetitive patterns of behavior to meet criteria under DSM-5
• Typically developing young children can have some repetitive behaviors

Medical Versus School Evaluation

• Medical evaluation provides diagnosis and is family focused
  – Medical workup
  – Explanation of diagnosis
  – Referral to community services

• Medical diagnosis required to access autism therapies through medical insurance
  – ABA providers currently request letter from physician documenting diagnosis and sometimes require specific diagnostic tools (e.g. ADOS or CARS)

➢ Regulations vary by state
Medical Versus School Evaluation

• School evaluation determines educational needs
  – Required to access IEP services
  – Most school evaluations do not diagnose ASD
  – Diagnosis does not equal eligibility for IEP
• School must consider but not required to accept medical evaluation
• School evaluation may not address family or community needs

➢ Regulations vary by state
Multi-departmental collaborative center providing diagnosis, treatment, continuity of care and support services

**Mission:** To provide excellent, timely, culturally competent, multi-disciplinary care for all children with ASD and their families.

- Contact our intake team: 617-355-7493
  [autismcenter@childrens.harvard.edu](mailto:autismcenter@childrens.harvard.edu)
- Information and resources on our website
  [http://www.bostonchildrens.org/autismspectrumcenter](http://www.bostonchildrens.org/autismspectrumcenter)
Contact our intake team: 617-355-7493
autismcenter@childrens.harvard.edu

Complete intake forms

Triage team review and assignment to next available and most appropriate visit
• Standardized guideline for diagnosis
• Medical model for all diagnostic evaluations
  – MD/NP consultation or Team assessment

➢ Ongoing care, not just diagnosis!
Importance of Medical Model

• Etiology
• Comorbid conditions
• Medical evaluation and testing
• Family centered care
• Ongoing medical care
Autism Spectrum Center (ASC) Assessment Guideline

Introduction

The Autism Spectrum Center (ASC) is a multidisciplinary effort at Boston Children's Hospital integrating the expertise of Neurology, Developmental Medicine, Psychiatry, Genetics and Center for Communication Enhancement.

A key goal for the ASC is to standardize care for patients with ASD at Boston Children’s Hospital:

- We have developed clinical practice guidelines for the assessment and management of ASD.
- This NetLearning module reviews the key information in the Assessment guideline and addresses standardized practice for diagnosis of ASD.
- Completion of the NetLearning module is required to determine eligibility for a clinician to provide diagnostic services for ASD at Boston Children’s Hospital.

ASC Executive Committee

Sarah Spence, MD PhD - Co-Chair
Carolyn Bridgemohan MD - Co-Chair
Eugene D’Angelo PhD
David Miller MD PhD
Howard Shane PhD

Boston Children's Hospital Autism Spectrum Center

Harvard Medical School Teaching Hospital
Accurate diagnosis requires history and direct observation of child

- History of general development, medical history, review of medical red flags such as regression, seizures or spells
- Review ASD symptoms with developmental focus
- Direct observation of communication, play and behavior
- Data considered in conjunction with DSM 5 criteria

Clinical Practice Guideline for Assessment of ASD

Boston Children’s Hospital Autism Spectrum Center

HARVARD MEDICAL SCHOOL TEACHING HOSPITAL
Consider in context of child’s typical behavior: Was this a “good” or “bad” day?
Accurate diagnosis requires multidisciplinary input

- Complete, review results of, or request standardized assessment of
  - Cognitive, language and adaptive skills
  - Social communication skills
    (e.g. CARS, STAT, ADOS, play assessment)
Medical Evaluation Is Individualized Based on the Child’s Presentation

All patients
• Physical examination
• Hearing test
• Genetic testing
  – Microarray and Fragile X

Selected patients
• EEG
• Brain MRI
• Other Genetic Tests
• Metabolic Tests
Recognizing Syndromic ASD Has Clinical Significance

- Testing positive in 10-20%
- Dysmorphic features linked to
  - Genetic abnormalities in ASD
  - Severity (lower cognitive function and seizures)
    (Miles 2000 & 2008; Shaeffer 2008; Miller 2010)
  - Diagnosis of specific etiologic condition may influence management – example PTEN mutation
Case Example:
Why is medical evaluation important?

12 year old patient with ASD and anxiety

- Previous microarray and Fragile X normal
  - On physical exam noted to have very large head
  - PTEN genetic testing sent and positive
  - Patient referred to GI and Endocrine for additional testing (colonoscopy and thyroid scan) and ongoing follow up

➢ Diagnosis of specific condition influences management
When To Get Psychological Evaluation?

• Transition times
  • Age 3 – transition to school services
  • Kindergarten/Elementary school
  • Middle school
  • High school
  • Post graduate

• Change in symptoms/behavior
  • New symptoms
  • Regression
  • Improvement

• For guidance on programming or treatment
Outline Part 2

• Psychological Evaluations
• Neuropsychological Evaluations
• Autism Spectrum Disorder Screeners
• Psychological Evaluation in Autism Spectrum Center at BCH
• Case Presentations
Evaluations

• Children are often referred for evaluations
• These have a variety of names, such as:
  – Psychological evaluation
  – Developmental evaluation
  – Diagnostic evaluation
  – Psychodiagnostic evaluation
  – Neuropsychological evaluation
  – Comprehensive evaluation
• All psychologists and neuropsychologists have differing areas of expertise
Evaluations (Cont’d)

• Important to determine what the psychologist or neuropsychologist specializes in
  – Call their office and ask:
    • What does this psychologist specialize in?
    • What is their area of expertise?
    • With what age groups do they most often work?
    • What type of assessment would they conduct with my child?
    • Does this person have expertise in diagnosing autism spectrum disorders?
Evaluations (Cont’d)

• Also important to determine which assessment tools they administer, such as:

<table>
<thead>
<tr>
<th>Domains of Assessment</th>
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<tbody>
<tr>
<td>Cognitive</td>
<td>Adaptive</td>
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<tr>
<td>Language</td>
<td>Social-Communication</td>
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<tr>
<td>Executive Functioning</td>
<td>Social-Emotional</td>
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<tr>
<td>Memory and Learning</td>
<td>Fine Motor skills</td>
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<tr>
<td>Academics</td>
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</table>
Evaluations (Cont’d)

• It is important to determine a child’s cognitive ability, language functioning and adaptive skills so that the psychologist can provide appropriate referrals

• The goal is to use their strengths to better support their challenges
Neuropsychological Evaluations

• Neuropsychologists often have expertise in head injury, or epilepsy, or learning style, or cancer, etc.
• Neuropsychological evaluations are more likely to include some of the below:
  – Cognitive and learning profile, language, fine motor, memory and learning, auditory processing, visual spatial, social-emotional, academics, attention, executive functioning
• While some neuropsychologists may have expertise in diagnosing autism spectrum disorders, this is often not the case
**Autism Spectrum Screeners**

- Determine whether a child requires further evaluation to rule out Autism Spectrum Disorder

<table>
<thead>
<tr>
<th>Screeners</th>
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<tbody>
<tr>
<td>MCHAT- R- Autism spectrum Screener- 16-30 months</td>
<td>Social Communication Questionnaire (SCQ)- Autism Spectrum Screener (over age 4)</td>
</tr>
<tr>
<td>Social Responsiveness Scale-2 (4-18 yo)</td>
<td>Autism Spectrum Rating Scale (2-18 yo)</td>
</tr>
<tr>
<td>Ages and Stages Questionnaire-developmental (birth-6 yo)</td>
<td>Parent Evaluation of Developmental Status-developmental (0-8 yo)</td>
</tr>
<tr>
<td>Communication and Symbolic Behavioral Scales- research based (9-24 months)</td>
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Psychological Evaluation in Autism Spectrum Center at BCH

• Psychologist or assessment team will meet with the family and take a detailed developmental and behavioral history based on the DSM-5 criterion

• We will review previous assessments and evaluations of your child
  – School based, Early Intervention, Private evaluations, Speech, Occupational Therapy, Physical Therapy
Psychological Evaluation in Autism Spectrum Center at BCH

• We will conduct a play evaluation/observation of your child to look at their social communication skills
• Psychologist does not conduct a physical evaluation
• We offer a variety of assessments, based on how old your child is and the questions you would like answered for your child
Psychological Evaluation in Autism Spectrum Center at BCH

- Depending on the age of the child, we may work with the child independently, or with the parent present
- The assessment may include: cognitive, ADOS-2, adaptive, behavioral, social-emotional, executive functioning
- We will collect ancillary data, such as teacher/therapist observations, Early Intervention Staff, Daycare staff, rating scales, and reports
Psychological Evaluation in Autism Spectrum Center at BCH

• High levels of concern that require action (e.g. referral to another department or agency and/or prioritization of appointment)
  – Hearing loss
  – Seizures
  – Developmental regression
  – Psychiatric crisis (e.g. suicidality, extreme aggression or self injury)
  – Safety concerns
Measures

• **Cognitive:** Thinking and learning style, verbal, nonverbal, visual-spatial skills

• **Social-Emotional and Behavioral:** Determine whether there are areas of concern as compared to same-aged peers, such as internalizing concerns or acting out behaviors

• **Adaptive:** Understand child’s adaptive abilities as compared to same-aged peers, such as receptive and expressive language, written skills, daily living skills, social interaction skills, fine motor, gross motor skills, atypical behaviors
Autism Diagnostic Observation Schedule—Second Edition (ADOS-2)

- Semi-structured
- Play-based standardized assessment
- Requires significant training
- Clinician will observe child’s social strengths and challenges, provide social milieu, provide a variety of materials, examine child’s social interaction and communication, observe play skills and any unexpected or atypical behaviors
- Takes about 60 minutes
- Sometimes the parent is present, especially for younger children
The ADOS-2 provides estimates of current levels of autism spectrum symptomatology, but cannot be used independently in the diagnosis of an autism spectrum disorder.

Information must be collected from a variety of measures (cognitive, language, behavioral, and adaptive measures) and multiple sources.

Some examples:
- Autism Diagnostic Interview (ADI-R) or similar developmental and behavioral interview
- Social Communication Questionnaire (SCQ)
- Cognitive and Adaptive Assessments
- Teacher interviews
Measures (Cont’d)

• **Language**: Determine strengths and challenges with receptive, expressive, and social pragmatic language

• **Pragmatic/Social language**:
  
  – The ways people produce and comprehend meanings through language  
  (Bishop, 1997)
  
  – Language that we use socially
Examples of Difficulties with Social Pragmatics

- Inappropriate initiation
- Eye contact
- Approaching strangers
- Talking repetitively about topics
- Focusing on themes
- Using formal language
- Difficulty describing certain events or a sequence of events
- Correcting others
- Literal language
- Misinterpreting the language/nonverbal cues of others
- Missing social cues of others
- Not using gestures/facial expressions to add meaning (Bishop and Baird 2001)
Measures (Cont’d)

• Executive Functioning:
  – Controlling impulses
  – Problem-solving
  – Flexibility
  – Regulating emotions
  – Initiating activities
  – Actively holding information via working memory
  – Planning and organization
  – Monitoring and checking behavior
Comorbidities

• Co-occurring conditions
• As part of the evaluation we also look for comorbidities
• Child may have additional diagnoses along with an autism spectrum disorder, such as
  – ADHD, anxiety, global developmental delay, intellectual disability, speech and language delay, etc.
Medication

• Should my child receive his or her ADHD medication the day of the evaluation?
  – Important to discuss this with your individual provider
  – Share your concerns
  – Depends on the diagnostic question
  – We want your child to do their best
  – Can bring medication to the appointment
Recommendations and Referrals

• After the evaluation or medical visit, the family will receive a diagnostic formulation and treatment recommendations of evidence based, ASD-specific educational interventions that are tailored to the patient's cognitive, language and behavioral skills.

• This may include follow-up evaluation, recommendations for the IEP, recommendations for Early Intervention, further speech and language evaluation, therapy, etc.
Support for the Family

• Allow family to process the information and receive support around this new diagnosis, such as psychologist, social work, therapy, support group

• Sometimes talking with someone who really understands autism spectrum disorders can help the family process this information and receive help with the next steps moving forward
Support for the Family

- The goal of the evaluation is to help you understand the diagnosis, determine the severity of some of your child’s behaviors, understand their language functioning, and determine how much support they will need.
- We would like to set a benchmark for where your child is currently, in order to use this information to monitor their response to intervention over time.
- This evaluation can help the family, teachers, and therapists better understand the child, so they can act from an informed position.
Recommendations and Referrals (Cont’d)

- All families are offered genetic counseling and genetic testing, but coverage for genetic testing varies by insurance plan
- Families are offered written materials, email messaging service, and resource support
- We provide referral information as needed, such as Speech and Language testing, Psychiatry, Occupational Therapy, Physical Therapy, Sleep referral, Nutrition, Feeding therapy, Pain and Incontinence Clinic, Gastrointestinal follow-up
Further Evaluation

• Determine if child requires neuropsychological or learning disability assessment:
  – Child is receiving supports in school for learning disability but not making progress
  – Has specific concerns with executive functioning, attention, language, learning and memory, etc.
Case Presentation

• 14 year old boy
• Previous neuropsychological evaluation
• Differing reasons for evaluation: e.g., focus
• Evaluation included assessment of cognition, language, visual-spatial, memory and learning, executive functioning, attention, fine motor, and social-emotional
• Did not examine question of autism spectrum disorder
Case Presentation

• 8 year old girl
• Previous neuropsychological evaluation
• Differing reasons for evaluation: e.g., Attention, executive functioning, and auditory processing
• Evaluation included assessment of cognition, academics, attention, executive functioning, memory and learning, language, auditory processing, visual spatial, and social-emotional
• Reported that she did not meet for autism spectrum disorder diagnosis, but did not conduct the ADOS-2 or in depth developmental and behavioral history specific to the question of autism spectrum disorder
Outline Part 3

• Family supports and resources
• Questions
ASC Staff

• Child Life Specialist
  – Patient preparation and support
  – Staff consultation and training

• Social Worker
  – Guardianship and transition support
  – Care coordination

• Patient Family Educator/Resource Specialist
  – E-news and social media
  – Patient resource materials

617-355-7493
autismcenter@childrens.harvard.edu
Family Resources:

- Parent Information Packet provided at time of diagnosis
- Email News Service – educational emails for the first year after diagnosis; quarterly blasts for continued updates on timely health information, research and educational opportunities to all families
  - Available in English and Spanish
- Trivox – secure web-based system to gather information from parents and teachers prior to visit
  - Available in English and Spanish
- Forum Series – evening talks for families and staff; archived on website, over 4600 downloads to date
Parent Reported Outcome Measure (ASD-PROM ©)

TriVox Health

Behavioral Functioning

To what extent does Riley do each of the following?

Has compulsions or rituals (e.g., needs things to be in a certain order, certain place, or certain color)

Avoids or is upset by visiting new places or meeting new people

Becomes easily upset by changes in routine, new activities, or surprises

Has difficulties with transition

Needs you to change your behavior to avoid becoming upset (e.g., needs you to avoid eating certain foods, needs you to avoid driving a certain way, needs you to say things a certain way)

Communication and Social Skills (higher is better)

M (Mother)

<table>
<thead>
<tr>
<th>Date</th>
<th>Expressive Language</th>
<th>Receptive Language</th>
<th>Nonverbal Communication</th>
<th>Social Skills</th>
<th>Play</th>
<th>Adaptive Skills</th>
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<td>Nov 15, 2016</td>
<td>2.5/5</td>
<td>2.6/5</td>
<td>3.0/5</td>
<td>2.8/5</td>
<td>1.7/5</td>
<td>2.4/5</td>
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Behavioral Functioning (lower is better)

M (Mother)

<table>
<thead>
<tr>
<th>Date</th>
<th>Restricted/Repetitive Behaviors</th>
<th>Sensory Preferences</th>
<th>Atypical Language</th>
<th>Psychiatric Consequences and Maladaptive Behavior</th>
<th>Maladaptive Toiletting</th>
<th>Feeding Difficulties</th>
<th>Seizure Screening</th>
<th>Sleep</th>
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<tr>
<td>Nov 15, 2016</td>
<td>2.4/5</td>
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Overall Improvement (higher is better)

M (Mother)

<table>
<thead>
<tr>
<th>Date</th>
<th>Language and Communication</th>
<th>Social Skills</th>
<th>Behavior</th>
<th>Toiletting</th>
<th>Dressing</th>
<th>Eating</th>
<th>Sleeping</th>
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</thead>
<tbody>
<tr>
<td>Nov 15, 2016</td>
<td>A little worse</td>
<td>A little worse</td>
<td>Much worse</td>
<td>A little worse</td>
<td>A little worse</td>
<td>Much worse</td>
<td>A little worse</td>
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Autism Friendly Hospital Initiative!
Challenges for children with ASD in the clinical setting

• Change in routine
• Sensory overload (auditory, olfactory, tactile)
• Uncertainty causes anxiety
• May be ill or in pain
• Need to interact with multiple strangers
• Too much language
BCH Staff Education Program

• Providing ASD training to targeted departments
• Monitoring impact via
  – Staff reported knowledge and comfort
  – Patient reported reduced barriers to care
“Precautions B” Behavior Support Plan

- Customized coping/care plan
- Promote safe and successful hospital visits
- Flagged via banner in medical record
Visual Schedules

My Daily Schedule

- get off bus
- backpack in cubby
- gym
- bathroom
- table work
- circle time
- snack time
- chores
- bathroom
- recess
- physical therapy
- centers
- speech
- bathroom
- lunchtime
- occupational therapy
- goodbye circle
- get on bus
Developmental Medicine Center Visit

Boy Version

My Hospital Story

Boston Children's Hospital

August 2016
My doctor is Dr. Bridgemohan.
I will take an elevator to get to the Developmental Medicine Center.
Then I will walk on the colorful floor.
Then I might sit at the table with the doctor and play with puzzles, games, and look at pictures.
Sometimes I will sit in a chair at the table with no toys and talk to the doctor.
The doctor will use a special tool to look at my ears while I sit very still.
Finally, my appointment at the Developmental Medicine Center is all done. Time to say goodbye.
In Summary….

• ASD is a complex neurodevelopmental disorder with a strong genetic basis
• Accurate diagnosis must include
   – consideration of a child’s cognitive and language function
   – thorough history with information from multiple sources
   – direct observation of a child’s communication, play and behavior
• The ASC uses a family-centered, evidence-based medical model of assessment and care
Questions?

Thank you!
# Diagnostic Services in Neurology & Developmental Medicine

## Boston Main Campus

- Peabody
- Waltham
- Weymouth
- Lexington