Screening and Assessment of ASD in the primary care setting

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Disclosures

If you do not have disclosures, state:

“Neither I nor my spouse/partner has a relevant financial relationship with a commercial interest to disclose.”

I am developing a Level 2 screening Autism Test but have no royalties from it
Prevalence and Early Signs

- Autism Spectrum Disorder (ASD) is defined as delayed social communication skills and restricted, repetitive behaviors (RRB).
- Prevalence 1 in 68 [CDC 2014]
- Symptoms visible as early as 12 months in some
- Variation linked to Social Economic Status and culture
Average age of diagnosis

Most still diagnosed after 4 years of age

Almost five times more common among boys than girls...but this may be changing

Special populations

White children are more likely to be identified as having ASD than are black or Hispanic children.
RED FLAGS for ASD

• No babbling by 12 months
• No gesturing by 12 months (pointing, waving bye bye)
• No single words by 16 months
• No 2-word spontaneous by 24 months
• LOSS of ANY LANGUAGE or SOCIAL skills at ANY age

Screening programs

Several programs to increase awareness:

- CDC campaign: Learn the signs Act Early
  http://www.cdc.gov/ncbddd/autism/screening.htm
- www.MAActearly.org

AAP (November 2013):

- Universal Screening for ASD at 18m and 24 m if no concerns before- Autism Clinician Kit

Massachusetts:

- Access to intensive services: final diagnosis ASD needed (January 2009)
- Mandatory developmental and autism screening (January 2008)
Recommendations in Toddlers

1. General observation and milestones
2. Ongoing developmental screening and surveillance
3. Autism screen at 18 and 24 mo
4. Refer to Early Intervention and specialist for formal diagnostic evaluation
5. Case management

• **History** is primary. Ask about **REGRESSION** and lack of progress *(all ages)*

• Open Ended questions

• **OBSERVATION** of Behavior: Does the child...
  – Make eye contact with you?
  – Engage in back and fro babbling
  – Have extremes of temperament
  – Answer to his/her name
  – Look across to see what you point at? “Look” while pointing
  – Play pretend play
  – Point with his index when asked to?
  – Interact nicely with his/her mother/parent?
Current Screening Methods for ASD

- **Level 1 - Universal Screen**

- Autism screen recommended by AAP at 18 and 24 mos.

- Example: MCHAT
  - Low Positive Predictive Value (PPV) for ASD (0.54)
  - High PPV for Developmental Delay (0.98)

➤ **Over-referral for ASD evaluations**
  ➔ **Longer wait delaying those who really need diagnosis**
Current Screening Methods for ASD

- Level 2 – Disorder-specific screener
  - Differentiates between ASD and other delays
    ➔ Referrals for ASD evaluations more appropriate
  - Interactive Level 2 ASD screeners preferred
    ➔ Triggers atypical behaviors
Two-Level ASD Screening

- **Risk ASD+++**
- **Level 2**
- **Risk for Developmental Delays/ASD**
- **Level 1**
- **Well Child Visits or high risk General Pediatrics**
# Selected ASD Level 1 and Level 2 Screening tools

*Choueiri & Zimmerman “Novel Assessments and Treatments of ASD”; in print*

<table>
<thead>
<tr>
<th>Screening Test</th>
<th>Age</th>
<th>Format</th>
<th>Time to complete mn</th>
<th>Sensitivity/Specificity</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Level 1</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>MCHAT-R</td>
<td>16-36 m</td>
<td>Questionnaire</td>
<td>5-10</td>
<td>0.92/0.83 PPV: 98% DD; 54% ASD</td>
</tr>
<tr>
<td>CAST</td>
<td>4-11 y</td>
<td>Questionnaire</td>
<td>10</td>
<td>0.88/0.97</td>
</tr>
<tr>
<td><strong>Level 2</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>SCQ</td>
<td>4y &amp; up</td>
<td>Questionnaires</td>
<td>10 each</td>
<td>0.85/0.96</td>
</tr>
<tr>
<td>CARS-2</td>
<td>2y &amp; up</td>
<td>Observation</td>
<td>15-20</td>
<td>Agreement with DSM 5: 84%</td>
</tr>
<tr>
<td>SRS-2</td>
<td>3y &amp; up</td>
<td>Questionnaires</td>
<td>15-20</td>
<td>Agreement ADI-R: 0.75-0.91</td>
</tr>
<tr>
<td>GARS-3</td>
<td>3-22 y</td>
<td>Questionnaires</td>
<td>10</td>
<td>GARS-2: 0.65/0.81 GARS-3: 0.97/0.97 (ProED)</td>
</tr>
</tbody>
</table>

*MCHAT-R: modified Checklist for Autism in Toddlers revised; CAST: Childhood Autism Screening test; SCQ: Social Communication Questionnaire; CARS-2: Childhood Autism Rating Scale; SRS: Social Responsiveness Scale; GARS: Gilliam Autism Rating Scale; ADI-R: Autism Diagnostic Interview*
## ASD Level 2 Interactive Screening tests & other useful tools

<table>
<thead>
<tr>
<th>Screening test</th>
<th>Age</th>
<th>Format</th>
<th>Time to complete in mn</th>
<th>Sensitivity/Specificity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Level 2 Interactive</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>RITA-T</td>
<td>18-36m</td>
<td>Interactive Training needed</td>
<td>10</td>
<td>1/0.84</td>
</tr>
<tr>
<td>STAT</td>
<td>24-36m</td>
<td>Interactive Training needed</td>
<td>20</td>
<td>0.92/0.85</td>
</tr>
</tbody>
</table>

**Other useful assessment tools**

<table>
<thead>
<tr>
<th>Test</th>
<th>Age</th>
<th>Format</th>
<th>Time to complete in mn</th>
<th>Measure of ADHD, ODD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Vineland Adaptive Behavior Scales</td>
<td>Birth &amp; up Survey Rating</td>
<td>25-60</td>
<td>Adaptive behavior, &amp; communication, social</td>
<td></td>
</tr>
<tr>
<td>Vanderbilt</td>
<td>School age Questionnaires</td>
<td>5-10</td>
<td>Measure of ADHD, ODD</td>
<td></td>
</tr>
</tbody>
</table>

*RITA-T: Rapid Interactive Screening Test in Toddlers; STAT: Screening Tool for Autism in Toddlers and Young Children*
Level 1: MCHAT-R

- 20 Questions simplified, examples added for each one
- All answers matter (no critical items anymore)
- Scoring system simplified
- Can download from www.mchatscreen.com
- Already translated in several languages
- Improved PPV though still high number of false positives
  - PPV for ASD: 54%
  - PPV for developmental concerns/delays: 89-98%
MCHAT-R scoring

- **LOW-RISK: Total Score is 0-2**
  - If child is younger than 24 months, screen again after 2y
  - No further action required unless surveillance indicates risk for ASD.

- **MEDIUM-RISK: Total Score is 3-7**
  - Administer the Follow-Up (second stage of M-CHAT-R/F)

- **HIGH-RISK: Total Score is 8-20**
  - It is acceptable to bypass the Follow-Up
  - Refer immediately for diagnostic evaluation and eligibility evaluation for early intervention.
Level 1: The CSBS-ITC
Communication & Symbolic Behavior Scales: Infant Toddler Checklist

- For those <18 months
- Between 6 to 24 months
- Evaluates gestures, eye contact, facial expressions, vocalizations
- Scorable by anyone but requires clinical interpretation
Considering Culture in Autism Screening

Massachusetts Act Early

www.MAActEarly.org
www.cdc.gov/actearly
1-800-CDC-INFO
Level 2: The Rapid Interactive screening test of Autism in Toddlers (RITA-T)
Description of the RITA-T

- 9 interactive presses
- Assesses developmental constructs delayed in early ASD such as:
  - *Joint Attention (JA)*
  - *Reaction to Emotions*
  - *Awareness of Human Agency*

- Each item coded from 0 to 2, 3 or 4 (typical to atypical)
- Total score generated; maximum: **30**
- The **lower** the score, the **more typical** the reaction

- Administration and scoring time: **5-10 minutes**
<table>
<thead>
<tr>
<th>RITA-T Total score</th>
<th>Sensitivity</th>
<th>Specificity</th>
<th>PPV</th>
<th>NPV</th>
</tr>
</thead>
<tbody>
<tr>
<td>8</td>
<td>1</td>
<td>0.05</td>
<td>0.56</td>
<td>1</td>
</tr>
<tr>
<td>9</td>
<td>1</td>
<td>0.11</td>
<td>0.58</td>
<td>1</td>
</tr>
<tr>
<td>10</td>
<td>1</td>
<td>0.16</td>
<td>0.59</td>
<td>1</td>
</tr>
<tr>
<td>11</td>
<td>1</td>
<td>0.26</td>
<td>0.62</td>
<td>1</td>
</tr>
<tr>
<td>12</td>
<td>1</td>
<td>0.37</td>
<td>0.66</td>
<td>1</td>
</tr>
<tr>
<td>13</td>
<td>1</td>
<td>0.53</td>
<td>0.72</td>
<td>1</td>
</tr>
<tr>
<td>14</td>
<td>1</td>
<td><strong>0.84</strong></td>
<td><strong>0.88</strong></td>
<td>1</td>
</tr>
<tr>
<td>15</td>
<td>0.96</td>
<td>0.84</td>
<td>0.88</td>
<td>0.94</td>
</tr>
<tr>
<td>16</td>
<td>0.83</td>
<td>0.89</td>
<td>0.90</td>
<td>0.81</td>
</tr>
<tr>
<td>17</td>
<td>0.74</td>
<td>0.95</td>
<td>0.94</td>
<td>0.75</td>
</tr>
<tr>
<td>18</td>
<td>0.65</td>
<td>1</td>
<td>1</td>
<td>0.70</td>
</tr>
<tr>
<td>19</td>
<td>0.61</td>
<td>1</td>
<td>1</td>
<td>0.68</td>
</tr>
<tr>
<td>21</td>
<td>0.48</td>
<td>1</td>
<td>1</td>
<td>0.61</td>
</tr>
<tr>
<td>22</td>
<td>0.35</td>
<td>1</td>
<td>1</td>
<td>0.56</td>
</tr>
<tr>
<td>23</td>
<td>0.30</td>
<td>1</td>
<td>1</td>
<td>0.54</td>
</tr>
<tr>
<td>24</td>
<td>0.13</td>
<td>1</td>
<td>1</td>
<td>0.49</td>
</tr>
<tr>
<td>25</td>
<td>0.09</td>
<td>1</td>
<td>1</td>
<td>0.48</td>
</tr>
<tr>
<td>26</td>
<td>0.04</td>
<td>1</td>
<td>1</td>
<td>0.46</td>
</tr>
<tr>
<td>27</td>
<td>0</td>
<td>1</td>
<td>1</td>
<td>0.45</td>
</tr>
</tbody>
</table>

Sensitivity & Specificity for all cut-off scores
Other Level 2 Interactive ASD Screener Toddlers

The Screening Test for Autism in Toddlers (STAT)
20 minutes to administer
Complex administration and scoring

At cut-off score of 2:
- Good sensitivity & specificity for Autism at 24-36 mos
- Misses milder forms of ASD (e.g. PDD-NOS)
- Weak sensitivity & specificity for ASD < 24 mos
DSM-5: Diagnostic Criteria of ASD

**DSM-IV**

Multiple ASD categories, e.g., Asperger’s, PDD-NOS

**Three core symptom domains:**

I. Qualitative abnormalities in Reciprocal Social Interaction

II. Qualitative abnormalities in Communication

III. Restricted, Repetitive and Stereotyped Patterns of Behavior (RRB)

AND Onset at/before 36 months of age

**DSM-5**

Single category with levels of severity, i.e., ASD

**Two core symptom domains:**

I. Qualitative abnormalities in Social Communication; Marked by deficits in social-emotional reciprocity; Deficits in nonverbal communicative behaviors, & deficits in developing relationships.

II. RRBs, inclusive of repetitive speech, hyper/hypo-reactivity to sensory input

AND symptoms limit & impair everyday functioning
I-DSM-5 ASD Criteria for Social Communication (all must be met)

• Deficits in socio-emotional reciprocity

• Deficits in nonverbal communication behaviors used for social interaction

• Deficits in developing and maintaining relationships appropriate to developmental level
II. DSM-5 Criteria for Restricted/Repetitive Interests and Behaviors (RRB)  
(At least 2 of 4 met)

A. Stereotyped or repetitive speech, motor movements or use of objects

B. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior or excessive resistance to change

C. Highly restricted, fixated interests that are abnormal in intensity or focus

D. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment
“Specifiers” and “Moderators”

- Examples: ASD “associated with known medical or genetic condition” (e.g., Fragile X, VCFS, TS)
  - Verbal Abilities
  - Cognitive Abilities
  - ADHD
  - Severity
## Severity Levels

<table>
<thead>
<tr>
<th>Severity Level</th>
<th>Social Communication</th>
<th>RRBs</th>
</tr>
</thead>
<tbody>
<tr>
<td>I (mild)</td>
<td>Inclusion support with peers; child shows age level speech</td>
<td>Cues &amp; reminders for transitions to manage reluctance, organization and planning</td>
</tr>
<tr>
<td>II (moderate)</td>
<td>Inclusion support/partial separate class depending on variability in behaviors; inability to engage with peers; immature and diminished talk, and talk topics limited to interests</td>
<td>Step plans for transitions to manage inflexibility; distress around change, visible to casual observer</td>
</tr>
<tr>
<td>III (severe)</td>
<td>Separate class due to limited &amp; minimal initiations, responses, little intelligible speech &amp; shows responses limited to self needs</td>
<td>Need to reduce demands due to limited coping, level of RRBs interfere with function, &amp; frequent distress reactions with change</td>
</tr>
</tbody>
</table>
DSM-5 New Diagnostic Category

- **Social Communication Disorder (SCD)**

  ALL criteria for category I met, but no or <1 criteria met in category II

  1) Is an impairment disorder of pragmatics
  2) Diagnosed based on difficulty in the social uses of verbal and nonverbal communication
  3) Affects the functional development of social relationships and discourse comprehension
  4) Cannot be explained by low abilities in the domains of word structure and grammar or general cognitive ability
  5) Still unclear what services provided by schools
Screening & Assessment recommendations in primary care

• Discussion of Algorithm by age

• Factors to consider:
  – Age
  – Medical, Developmental History
  – History of Regression
  – Level 1 screen or concerns for ASD
  – Level 2 ASD screening
  – Awareness of limitations of screening tools
  – Observation of child: language, non verbal communication, play, conversation skills
  – Physical and neurological exam
  – Can use:
    • DSM-5 criteria: older children better sensitivity than younger children
    • Vineland
    • Previous testing
Flow Chart: assessments and screening

Choueiri R & Zimmerman A “Novel Assessments and Treatments of ASD”; in print

<table>
<thead>
<tr>
<th>Child with Concerns for ASD or with positive ASD level 1 screening test</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>HISTORY:</strong> Developmental History and Trajectory; Regression; Detailed Medical History</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Toddler: 1-3 y</th>
<th>Preschooler: 3-5 y</th>
<th>School Age: 5-18 y</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Observation &amp; Surveillance</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Play, communication, language, non-verbal</td>
<td>Play, communication, language, non-verbal</td>
<td>Communication, language, non-verbal</td>
</tr>
</tbody>
</table>

| **Testing & Exam** |
|---|---|---|
| Level 2 Interactive Review EI, Developmental Consider Vineland; DSM 5; ADOS Physical and Neurological Exam (PNE) | Level 2 screening test Review Developmental, cognitive, ST, EI, school testing; teacher's concerns; rating scales; IEP Consider Vineland; DSM5; ADOS; PNE | Level 2 Screening test Review Developmental, cognitive, ST, school testing; teacher's concerns; rating scales; IEP Consider Vineland; DSM5; ADOS; PNE |
Flow Chart: assessments and screening

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<table>
<thead>
<tr>
<th>Toddler 1-3y</th>
<th>Preschooler: 3-5y</th>
<th>School age: 5-18 y</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Other assessments and referrals</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hearing test</td>
<td>Hearing test</td>
<td>Hearing test</td>
</tr>
<tr>
<td>MRI/EEG if indicated</td>
<td>MRI/EEG if indicated</td>
<td>MRI/EEG if indicated</td>
</tr>
<tr>
<td>Genetic testing</td>
<td>Genetic testing</td>
<td>Genetic testing</td>
</tr>
<tr>
<td>Early Intervention</td>
<td>School Evaluation</td>
<td>School &amp; academic evaluation</td>
</tr>
<tr>
<td>Specialized services: ABA, ST, OT, PT as indicated</td>
<td>Specialized services: ABA, ST, OT, PT, Social skills, counseling</td>
<td>Specialized services: ABA, ST, OT, PT, Social skills</td>
</tr>
<tr>
<td>-Other medical referrals as needed</td>
<td>-Other medical referrals as needed</td>
<td>Psychiatry for co-morbidities &amp; psychopharmacology;</td>
</tr>
<tr>
<td></td>
<td></td>
<td>-Other medical referrals as needed</td>
</tr>
</tbody>
</table>
Conclusions

• The assessment of ASD is complex and remains clinical

• In primary care, it is important to provide developmental monitoring at all ages, surveillance, and to be familiar with screening tools and their limitations, with DSM-5 criteria and limitations, and with other useful assessment tools