Methods of Early Screening, Detection, and Diagnosis for Autism Spectrum Disorders

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Comprehensive Developmental Approach to Diagnostic Evaluations
1. Screening & Detection of Risk
2. Developmental/Cognitive Assessment
3. Diagnostic Assessment (by history & direct assessment)
4. Speech, Language, & Communication Assessment
5. Assessment of Adaptive Behavior
6. Assessment of Behavioral/Emotional Regulation
7. Assessment of Motor Development
8. Medical Assessment
9. Integrated Transdisciplinary Diagnostic Formulation
10. Recommendations for Treatment & Intervention
11. Follow-up and Re-Evaluation

Risk for Autism

IN THE GENERAL POPULATION:
• ASD: 1 in 68 (More prevalent than all childhood cancers combined)
• Median Age of Diagnosis: 4-5 years
  • Much later for disadvantaged populations
• When ASD can be reliably diagnosed: 18-24 months when diagnosed by experienced clinicians

IN SIBLINGS OF CHILDREN WITH ASD:
• ASD: 1 in 5 (~20% risk)
• Broader Autism Phenotype (“shadow symptoms”): 1 in 5
• Non-ASD developmental delays: 1 in 10

Approximately 50% of subsequent siblings are at risk for developmental vulnerabilities, including autism

Risk Factors for Autism in Infancy

• No babbling or cooing by 12 months
• No single words by 16 months
• No spontaneous two-word phrases by 24 months (not including echolalia/scripting)
• No use of social gestures by 12 months (point, wave, grasp)
• No response to name
• Limited imitation skills
• Regression or plateau of skills

Typical Development (“Low Risk”) – 9 Months
High Risk Infant – Younger Sibling

Who is on the Front Line for Early Detection?

- Parents/Caregivers
- Pediatricians/Primary Care Physicians
- Daycare Providers/Preschools
- Babies Can’t Wait
- Specialty Clinics (e.g., CHOA, Marcus, Private Practice Clinicians)

[cdc.gov/ncbddd/actearly]

How often should we screen for ASD?

American Academy of Pediatrics
Developmental Surveillance:
www.aap.org

Autism-specific screening is recommended for all children at the 18 and 24 month well-child visits.

Is this enough???

Screening Measures for ASD

Best Practices in Diagnosing ASD under the age of 2 years

1. Diagnostic process led by an experienced clinician knowledgeable of infants/toddlers & ASD
2. Standardized observation & assessment of:
   * Social, communication, & play skills
   * Cognitive, language, & adaptive skills
   * Repetitive behaviors, unusual interests, etc.
3. Parent report & developmental history of:
   * Social, communication, & play skills
   * Repetitive behaviors, unusual interests, etc.
   * Sleep, feeding, sensorimotor issues
4. Clinical judgment of experienced clinicians consulting w/ & interpreting results from a multidisciplinary team

Assessing Developmental/Cognitive Skills

Need for Establishing a Baseline of Functioning

- General IQ Tests:
  * Wechsler Scales (WPPSI; WISC; WAIS)
  * Differential Ability Scales (DAS)
  * Stanford-Binet
- Nonverbal IQ Tests:
  * Leiter International Performance Scale
  * Test of Nonverbal Intelligence
- Developmental Assessments:
  * Mullen Scales of Early Learning
  * Bayley Scales of Infant Development

Caution about using the terms “IQ” or “cognitive ability” in very young children (i.e., < age 6)
Assessing Developmental Abilities

16 Gestures by 16 Months

Wetherby, 2014; First Words Project, Autism Navigator; Florida State University

9 Months:
  • Give; Shake Head
10 Months:
  • Reach; Raise Arms
11 Months:
  • Show; Wave
12 Months:
  • Open Hand;Point; Tap
13 Months:
  • Clap; Blow a Kiss
14 Months:
  • Index Finger Point; Shhh Gesture
15 Months:
  • Head Nod; Thumbs Up; Hand Up
16 Months:
  • Symbolic Gestures (e.g., High Five; Shrug for "I Don’t Know")

Assessing Learning Readiness Skills

1. Sitting
   • Establishing a “ready response”
2. Attending to Adult Instruction
   • Tolerating adult-led activities
3. Establishing Eye Contact
   • Monitoring another person
4. Imitation
   • Modeling the actions & speech of others

Imitation and Modeling

THESE ARE THE BUILDING BLOCKS OF LEARNING!

Assessing Diagnostic Profile

The Autism Diagnostic Observation Schedule (ADOS-2; Lord et al.; 2012)

Western Psychological Services (WPS)

www.wpspublish.com
The Autism Diagnostic Observation Schedule

5 Modules based on age and language level
• Toddler Module: Between 12 and 30 months < phrase speech
• Module 1: 31 months + with no phrase speech
• Module 2: 31 months + with phrase speech
• Module 3: Verbally fluent children & young adolescents
• Module 4: Verbally fluent older adolescents & adults

Items Coded on 4-point severity scale
• 0 = symptom not present
• 3 = symptom severe/atypical

Diagnostic Algorithm for Modules 1–4:
• Autism
• Autism Spectrum
• Non Autism Spectrum

Assessing Adaptive Behavior

• Cognitive ability is generally defined an individual’s repertoire of skills that are either innate or acquired.
  —Skills that an individual is capable of performing
• Adaptive behavior is generally defined as performance of skills that are necessary for personal and social sufficiency
  —Skills an individual does perform, independently, in daily activities

Adaptive impairment = when an individual is capable of performing a skill but does not do so, independently

Measures of Adaptive Functioning

Vineland Adaptive Behavior Scales
(Sparrow, Balla, & Cicchetti, 1984; Sparrow, Cicchetti, & Balla, 2005)

Domains of Functioning (birth – 90 years)
• Communication: Receptive; Expressive; Written
• Daily Living: Personal; Domestic; Community
• Socialization: Interpersonal; Play/Leisure; Coping
• Motor: Fine; Gross Motor

Maladaptive Social Interaction

Challenges Associated with Early Detection
Zwaigenbaum, Bryson, Lord et al., Pediatrics, 2009

1. Diagnostic differentiation early on
  • Global/severe developmental delays vs. ASD
  • Specific language impairments vs. ASD
  • Visual/hearing impairments vs. ASD
  • Siblings who present w/ ASD-like symptoms
2. Subtle symptom-expression early on
  • Higher functioning children
  • Absence of language delays
  • Plateau/regression occurring after 18-months
3. Stability of diagnosis
  • Uncertain trajectories for those diagnosed under the age of 2
    (at age 2, research shows generally reliable & stable diagnoses)
**Diagnostic Stability**

**Short term stability (From 2 to 4 years)**
- 80-90% for ASD diagnosis around age 2
- Changes expected within spectrum due to shifts in number of symptoms and intensity

Chawarska, Klin, Paul et al., JCPP 2009

**Long term stability (From 2 to 9 years)**
- 90% stability of ASD diagnosis for autism vs. non-spectrum

Lord, Risi, DiLavore et al., JCPP 2006

**Stability for ASD diagnosis in infant siblings:**
- ????
- High-risk infants can have variable diagnostic trajectories, with some evidencing symptoms before 18m, others not until after 24m, & others with broader phenotype

Chawarska, Shic, Mazori, et al., 2014

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**Do Early Risk Factors for ASD even Resemble ASD Symptomatology???**

**Delays/Deviance in Motor and Postural Development**
- Delays in sitting in standing
- Impaired ability to change postures

Nickel, Thatcher, Keller et al., 2013

**Reduced Grasping and Object Manipulation**
- 6-month-old high-risk infants exhibit reduced grasping activity and object manipulation during play

Libertus, Shepherd, Ross, & Landa, 2014

**Sensory-Motor Impairments**
- NICU infants later diagnosed with ASD exhibit asymmetric visual tracking, tone deficits, and declining motor performance in the first year of life

Karmel, Gardner, Meade et al., 2014

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**Parental Concerns and Perceptions**

Chawarska, Paul, Klin, et al., JADD, 2006

- For toddlers diagnosed by age 3, the average age of first concerns is 14 months
- Typical parent concerns involve the following:
  - Absence of expected milestones
  - Delays in or slowing of developmental skills
  - Regression or loss of previously acquired skills
- In a clinically-referred sample, parents who had an older child with ASD did not have an earlier age of concern
- Parents with the latest concerns tended to be older and have a history of infertility treatments

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**Where Parents Seek Information about ASD**

Rhoades, Scorpa, & Soley (BMC Pediatrics, 2007)

- Media: 71-73%
- Conferences/Workshops: 42%
- Other Parents: 42%
- Health-care Professionals: 15-20%
What the General Public is Learning from the Media...

If We Believed that Correlation = Causation

The real cause of increasing autism prevalence?

Benefits of Social Media...

Risk Perception & Risk Communication

Risk Perception

• Clinician vs. parent perception: what is the knowledge about risk signs for ASD?
• Are perceptions influenced by comparisons (e.g., profile of older sibling)?
• Are perceptions influenced by “diagnosis” vs. “risk”?
• Do perceptions alter actions (e.g. whether or not to treat)?

Risk Communication

• How much/little do we know about risk signs?
• “At risk” vs. “diagnosis”
• What are the benefits/consequences to making erroneous judgments?
• False positives
• False negatives
• What do we know about treatment efficacy for children this young?

Recommendations for Risk Communication

Working Group in Autism Risk Communication & Ethics, 2012

1. Require risk communication as part of CE/CME training
2. Develop “tool kits” for risk communication distribution to providers
3. “Meet families where they are”
4. Train professionals on how to communicate risk in the face of scientific uncertainty
5. Address risk factors as they become public knowledge (e.g., Nature paper)
6. Establish centralized resource of current risk factors vetted by stakeholders
7. Address the need for families to understand risk by increasing access to evidence-based resources

Take Home Messages

• We can reliably diagnose ASD by age 2 in clinically referred samples – but diagnostic validity & stability is more questionable for younger siblings in multiplex families
• Both technology-based and clinically-based measures allow us to improve upon detecting risk for ASD in the first year of life
• Clinicians need to be well informed about risk factors for ASD in order to best inform parents/caregivers about next steps
• Clinicians need to be sensitive to how risk is communicated to parents (esp. to parents who already have a child with ASD)
• Children need to receive multiple evaluations across development to assess and re-assess developmental and diagnostic profiles
• Early and intensive intervention is associated with optimal outcome
Thank you!

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