

Early Identification of Autism Spectrum Disorder: Present and Future Directions

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Objectives for Today

Participants will be able to:

- Recognize the importance of early identification of children with autism spectrum disorder (ASD).
- Recognize early behaviors that indicate risk of ASD.
- Describe screening processes used to indicate risk of ASD.
- Identify barriers to ASD screening, referral, and diagnosis in NC.
- Identify potential future directions in NC to enhance ASD services.

Where We're Going Today

- Current diagnostic framework for ASD
- Barriers to early identification of at-risk children
- Early behaviors indicating risk
 - What are they?
 - When do they appear?
- Surveillance and screening for ASD in infants and toddlers
- North Carolina barriers to accessing services
- Ideas for what enhancements NC could make
- Available resources
- Discussion



What is Autism Spectrum Disorder and How Does it Differ From Autism?

- Autism once viewed as subtypes (Autistic Disorder, Pervasive Developmental Disorder, Asperger's, High Functioning Autism)
- Researchers and clinicians have found it hard to distinguish between these groups
- Diagnostic and Statistical Manual 5 (2013) now identifies one disorder = Autism Spectrum Disorder
- Children/Individuals viewed on spectrum
- Some advocates prefer term "Autism" or "Autistic" to avoid "disorder"

Diagnostic and Statistical Manual of the American Psychiatric Association – Fifth Edition (DSM-5), 2013

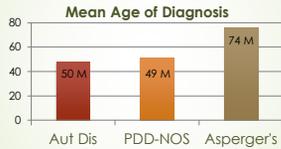
- Two broad domains of diagnostic symptoms
 - Social-communication
 - Restricted and repetitive behaviors and interests (RRBIs, includes sensory features)
- Individuals with ASD vary on multiple dimensions
 - Severity of symptoms – level of support needed associated with each symptom domain
 - Co-morbid diagnoses or associated characteristics – intellectual deficits, ADHD, language delay/disorder, sensory, repetitive behaviors, etc.
- Diagnosis of ASD should be accompanied by specification of levels of support needed in each symptom domain, and other diagnoses & characteristics, etc.

Factors in ASD Identification

- Continued rise in prevalence of ASD diagnosis (1/68 in U.S., 1/42 boys, 1/189 girls; **1/59 in NC**; CDC, 2016)
- Boys outnumber girls 4:1
- No expectation of differences in **prevalence** across racial, ethnic, geographic groups (U.S. vs other countries), **but there are differences in identification**
- High likelihood that range of professionals will see toddlers with ASD pre-diagnosis
- Growing body of research on ASD in children age two years and younger
- Potential for very early identification & intervention

How Early is ASD Typically Diagnosed?

Pre-DSM-5 categories for 8 year olds (CDC, 2016)



Fewer than half of children with ASD identified in their communities by age 5 (Maenner et al., 2013)

Disparities in Prevalence of ASD Diagnosis

- Prevalence in 8 year olds:
 - White, non-Hispanic: 1/65
 - African-American: 1/75
 - Hispanic: 1/99
- Substantial numbers of children (especially those from underrepresented groups) continue to miss the opportunity for early intervention



(CDC, 2016)

Barriers To Early ID

- Relatively "late" identification has made it difficult to follow children from early ages to know course of early development
- Difficulty identifying behaviors that could be markers for signs of **ASD vs other disabilities**
- Need to look for both **absence** of typical behaviors and **presence** of atypical behaviors
- Limited knowledge of developmental course of behaviors that may be common in young children (e.g., repetitive movements, mouthing)

Statement of Need

- Early screening is critical to the well-being of families
 - Prevents secondary consequences of late entry into EI services & social isolation
 - Addresses mission of effective health care for NC families
 - Growing underserved populations nationally
- Need to translate research into viable clinical practice models.
 - Few efforts to screen for ASD in infancy (before 18 months)
 - Primary care providers are increasing ASD screening efforts, but not enough to identify all at-risk children early

Early Social-Communication Symptoms



Social Communication/Interaction Impairments

Infants and Toddlers may show:

- ▶ Less responsiveness to people's overtures
- ▶ Lack of response to name
- ▶ Atypical eye contact (more aversion, less looking at face/eyes & at others)
- ▶ Limited interest in other children



Social Communication/Interaction Impairments (continued)

- Limited interest in reciprocal/social games like peek-a-boo (unless tickle & chase & roughhouse)
- Less likely to draw others into play
- May not want parents to do things with them (e.g. read books)
- Poor or limited imitation of others
- Simple pretend play not emerging



What Do Parents Describe?

- May describe child as affectionate - this does not rule out ASD!
- Often hear child described as "in his own world".
- Limited facial expressions
- Parents may consider child is hearing impaired.
- Differences between "shy" children and ASD.



Social Communication/Interaction Impairments (continued)

- ▶ Delayed speech/language
- ▶ Loss of acquired words
- ▶ Fewer social gestures (wave bye, patty-cake, nodding head) than children with other DD
- ▶ Echolalia/stereotyped speech
- ▶ Unusual rhythm, intonation of speech



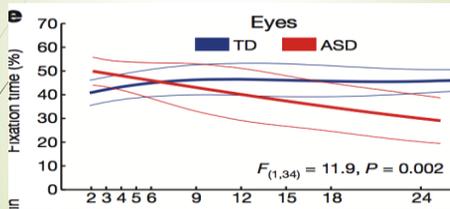
Social Communication/Interaction (continued)

- ▶ **Joint Attention (RJA and IJA)**
- ▶ No showing, giving, or pointing to **share interest** compared to other children with DD **but may give to get things to happen** (Watson, Crais, et al., 2013)
- ▶ Doesn't attract attention to own activities
- ▶ Use of another's hand as a tool



Attention to Eyes Across First two Years

Jones & Klin (2013)



Restricted/Repetitive Behaviors, Interests or Activities

- ▶ Unusual or repetitive play
- ▶ Interest in parts of objects
- ▶ Attachment to unusual objects
- ▶ Repetitive, stereotyped movements
- ▶ Unusual sensory interests
- ▶ Insistence on sameness



Other ASD concerns

- ▶ Over and/or under reaction to sensory stimuli (hyper-responsive, hypo-responsive)
- ▶ **More children with ASD (than children with other DD) have mixed pattern, also more with ASD have hypo-responsive pattern**
- ▶ Irregular sleep-arousal rhythms
- ▶ Picky eaters/gastro issues



[Baranek et al., 2006; Rogers et al., 2003; Watson et al., 2011; Wiggins et al., 2009]

Screening Guidelines (AAP, 2007)

- Surveillance at every visit (developmental updates)
- Four risk factors for surveillance:
 - ✓ Sibling with ASD
 - ✓ Parent concern, inconsistent hearing, unusual responsiveness
 - ✓ Other caregiver concern
 - ✓ Pediatrician concern
- Specific inquiries about social-emotional milestones



Surveillance & Screening: AAP Guidelines Follow-up

- Two or more risk factors → parent education, referral for ASD evaluation & EI services, follow-up visit
- One risk factor, <18 months → evaluate social-communication development
- One risk factor, >18 months → use ASD specific screener
- **ALL children at 18 & 24 months** → use ASD specific screener (but no ASD screener recommended)
- Any positive screen → parent education, referral for ASD evaluation & EI services, follow-up visit

Why Not Just Surveillance?

- Evidence suggests that action on these concerns is often delayed ("wait & see")
- Absence of a concern doesn't mean there is not a problem
- Some parents report concerns, others not sure what "red flag" behaviors are or how to interpret their child's behaviors
- The younger the child, the harder to recognize the red flags without a standardized tool
- Considerable evidence that surveillance alone results in under-referral of young children with DD including ASD

Parent & Physician Recognition of First Concerns

- Parent initial concerns (Chawarska et al., 2007)
 - 14.7 months for Autism
 - 20% at <11 m
 - 36% at 11-18 m (56% of families by 18 months)
 - 44% at >18 m
 - Types of concerns
 - Language & speech 71%
 - Social difficulties 61%
 - Medical problems/motor delays 29%
 - Stereotyped behaviors 17%

Parent & Physician Recognition of Concerns

- Mean time delay of 5.2 months from first concerns to consultation with a professional (Guinchat et al., 2012)
- Time lag of more than 3 years (mean = 39.3 months) before diagnosis (Guinchat et al., 2012)
- Pediatrician made observations of autistic-like behaviors in children later diagnosed (Niehus & Lord, 2006)
 - 10% of children with ASD during 1st year
 - 59% of children with ASD during 2nd year (only 1/2 referred)
- Number of professionals consulted was positively correlated with parents' stress (Moh & Maaliati, 2012)
- Extent parents engaged as collaborative partners in process negatively correlated with stress (Moh & Maaliati, 2012)

North Carolina Needs of Young Children with Autism Survey (HRSA State Implementation Grant, Hooper & Pretzel)

Conducted statewide survey of families with young children with ASD (effort led by Rob Christian, CIDD)

- Children under 9 years of age
- Web & paper surveys
- Recruited through Autism Registry (CIDD) & ASNC parent meetings
- N = 450 families, 80/100 counties represented
- Racial & ethnic breakdown - 78% White, 14% Black, 8% Hispanic/Latino, 4% Asian, 2% American Indian

(Martinez, M., Thomas, K., Williams, C., Christianson R., Crais E., Edmondson R., & Hooper, S, in preparation)

North Carolina Needs of Young Children with Autism Survey (HRSA State Implementation Grant, Hooper & Pretzel)

PRELIMINARY RESULTS:

- Less than 1/3 of children screened by a professional
- 25% families told "It's not autism" by a professional before later ASD diagnosis
- Almost 1/2 saw 3 or more professionals before ASD diagnosis
- Majority of families report difficulty accessing mental health/behavioral health services (Psychologist, Psychiatry, Behavioral Therapist)

(Martinez, M., Thomas, K., Williams, C., Christianson R., Crais E., Edmondson R., & Hooper, S, in preparation)

North Carolina Needs of Young Children with Autism Survey (HRSA State Implementation Grant, Hooper & Pretzel)

- Mean age of first developmental concerns = Typically before 21 months of age
- Parents most likely to notice concerns
- More likely to enter EI services earlier if parents had concerns
- More likely to get diagnosis if previously screened for ASD
- Mean age of initial intervention started = 2 1/2 years
- Mean age of ASD diagnosis = 3 years 2 months
- More data to come

(Martinez, M., Thomas, K., Williams, C., Christianson R., Crais E., Edmondson R., & Hooper, S, in preparation)

Level I Autism Screening Tools



- *Modified Checklist for Autism in Toddlers Revised with Follow-Up (M-CHAT-R/F)* (Robins, Fein, & Barton, 2009)
- Valid for screening between 16 and 30 months of age
- Sensitivity: 85% Specificity: 93%
- 20 Yes/No questions
- First stage, parent completes MCHAT-R
- Second step, professional asks follow-up questions for items failed
- Expect "Yes" for all items ("No" = ASD risk), except 2, 5, and 12 ("Yes" on these indicates ASD risk)
- http://mchatscreen.com/Official_M-CHAT_Website.html

M-CHAT-R/F and Follow-up Questions

Have you ever wondered if your child might be deaf?

Is your child interested in other children?

If you point at something across the room, does (child's name) look at it? If no, ask:

How he/she will respond if you point at something?

PASS Examples:

Looks at object
Points to object
Looks and comments on object
Looks if parent points and says "look!"

FAIL Examples:

Ignores parent
Looks around room randomly
Looks at parent's finger

MCHAT-R/F Scoring Algorithm

- **LOW-RISK: Total Score is 0-2;** if child is younger than 24 months, screen again after second birthday. No further action required unless surveillance indicates risk for ASD.
- **MEDIUM-RISK: Total Score is 3-7;** Administer Follow-Up to get additional information about at-risk responses. If M-CHAT-R/F score remains at 2 or higher, the child has screened positive. Action required: refer child for diagnostic evaluation and eligibility evaluation for early intervention. If score on Follow-Up is 0-1, child has screened negative. No further action required unless surveillance indicates risk for ASD. Child should be rescreened at future well-child visits.
- **HIGH-RISK: Total Score is 8-20;** It is acceptable to bypass the Follow-Up and refer immediately for diagnostic evaluation and eligibility evaluation for early intervention.

MCHAT-R/F Outcomes

- Significant false positives **without** follow-up questions
- Even with follow-up questions, significant number of children who fail M-CHAT-R will not be diagnosed with ASD. **However, they are at risk for other DDs**
- **Recent study: 48% of screen positive children diagnosed with ASD, 95% with ASD or other DD (Robins, et al., 2014)**
- **Therefore, follow-up is warranted for any child who screens positive!!!**
- Use of standardized tools (MCHAT and Infant Toddler Checklist) identified significantly more children with ASD than did either clinical judgment of primary care providers or parent report alone (Miller et al. 2011)

Program for Early Autism Research, Leadership, & Service (PEARLS)

www.med.unc.edu/ahs/pearls

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PEARLS' Early Research Efforts

Retrospective video analysis to identify at-risk behaviors:

- Home videos before diagnosis
- Range of contexts in home setting (outdoor & indoor play, bath time, birthday parties)
- Three groups of participants = Children with typical development; and children later diagnosed with ASD, or developmental disabilities
- Our research has targeted children 9-12 months & 15-18 months
- Interest in identifying early behaviors indicative of ASD
- Symptoms of ASD apparent by 12 months of age in some children, some behaviors recognized in majority of children between 18-24 months

Early Identification of Children At Risk for ASD

- Many children seen by primary care provider at 12 months.
- UNC-CH research team developed parent report tool to identify risk for ASD in 12 month olds - *First Year Inventory* (Baranek, Watson, Crais, & Reznick, 2003; Turner-Brown, et al., 2013).
- With early identification, some children can be diagnosed as young as 2 years of age with good reliability and stability.
- Symptoms emerge differently in different children – some children who do not meet criteria for an ASD diagnosis at 2 years may do so at 3 years.

(Baranek, 1999; Bryson et al., 2007; Landa et al., 2007; Mandell et al., 2009; Ozonoff et al., 2010; Pierce et al., 2011; Wetherby et al., 2008; Zwaigenbaum et al., 2005)

First Year Inventory (FYI, Baranek, Watson, Crais, & Reznick, 2003)



- 12 month parent report
- Social-communication items
- Sensory-regulatory items
- 63 items total
- 46 items; parents check "never," "seldom," "sometimes," or "often"
- 14 multiple choice items
- 1 item on sound production
- 2 open-ended questions regarding concerns

FYI Sample Questions: Checklist & Multiple-choice Formats

- Does your baby turn to look at you when you call your baby's name?
- Does your baby seem overly sensitive to your touch?

When you introduce your baby to a new game (peek-a-boo, so-big, patty-cake, etc.), how does your baby respond?

- Almost always joins in immediately without any help.
- Usually joins in, with a little help.
- Joins in only with a lot of help.
- Doesn't seem very interested in new baby games.

What do you typically have to do to get your baby to look up from playing with a favorite toy?

- Just show him or her a different toy.
- Move, shake or make a noise with the different toy.
- Take the favorite toy away and give your baby the different toy.

Identification of ASD and Other DDs

- Using cut-off for both Social-Communication & Sensory-Regulatory domains yielded best performance of FYI in screening for ASD:



Turner-Brown, Baranek, Reznick, Watson, & Crais. (2013).

- Positive Predictive Value = .31 (31% chance child identified by FYI will have ASD at age 3)
- Sensitivity = .44 (44% of children with ASD by age 3 were identified by FYI)
- 85% of children had DD or ASD**
- In infants/toddlers, symptoms may be more subtle. That's why screening tools are invaluable!

Clinical Use of FYI?

- Not ready for clinical use yet!
- Only normed at 12 months of age
- Too long
- Scoring is complicated
- Working on First Year(s) Inventory (expand age range to 9–16 months)
- FYI-Life (20 questions)
- The FYI identifies children who are at high risk for ASD or other developmental disabilities
- ASD screening can provide opportunities for a broader range of children to receive early intervention (EI) services
- Keep up with our progress on our website: <http://www.med.unc.edu/ahs/pearls>

SIG Focus Group Study with NC Parents of Children with ASD

- Eight focus groups (4 English speaking, 2 Spanish speaking, 2 American Indian) with **60+ caregivers of young children (birth to eight)** with ASD.
- Six regions of North Carolina.
- Caregivers asked to describe facilitators & barriers within three time periods: between first concerns & professional consultation, up through the diagnosis, & through entry into early intervention.
- Caregivers asked how these processes could be improved.

(Crais, McComish, Kertcher, Hooper, Pretzel, et al.; in preparation)

Focus Group Results

Caregivers reported few facilitators:

- Small minority of families noted professionals (physicians, early care provider) or a family member first raised concerns and linked them with professional services.
- Small group reported helpful professionals who guided their path.

(Crais, McComish, Kertcher, Hooper, Pretzel, et al.; in preparation)

Caregivers Highlighted Many Barriers

- Each step of the early identification and intervention process
- Not knowing who/where to go
- Getting conflicting advice
- Disagreements within and outside the family
- Their own uncertainty or "denial" (as described by caregivers)
- Moving through multiple providers to get answers
- Negative experiences with professionals
- Limited resources
- Professional and parental lack of knowledge
- Family and cultural beliefs (autism as stigma)

What Parents Said: Hard to Find the Way

My pediatrician looked at his file and said "He did pass the MCHAT at 2, so what makes you think he has autism?" The doctors involved had just about the worst bedside manners I have EVER dealt with.

And then somebody on a post-it note would write down their person that they called...and it's like, gosh, I live in the USA, and it's like somebody in an alley handed me a phone number to call (and it's like gold!).

You have to constantly fight being isolated...we're not hot play dates...so on top of that you also feel isolated in terms of the resources.

Sometimes the best resources are often other confused mothers out there.

Caregivers Provided Many Suggestions

- Having navigators to guide them through the process
- Clear guidelines/roadmaps of printed materials for decision making
- Better connections with local resources
- Enhanced education for professionals (physicians, teachers, early care providers)
- More parent-to-parent opportunities

Additional NC Trends

- CDSA & TEACCH serve as possible locations for ASD diagnosis (usually team, MD &/or psychologist as key).
- CDSA System encountered state budget cuts & many CDSAs lost positions, especially psychologists.
- TEACCH is enhancing services to provide ASD diagnosis, especially for children birth to three.
- SIG faculty worked with CDSAs to support ASD diagnosis & provide educational opportunities to primary care clinicians (pediatricians & family doctors/nurses).

Resources for Families

- CDC Learn the Signs, Act Early, Milestone checklists (2 months – 5 years), information about sharing their concerns, referrals, understanding evaluation process <http://www.cdc.gov/ncbddd/actearly/index.html>
- Family Support Network <http://fsp.unc.edu/>
- TEACCH www.teacch.com
- Autism Society of NC <http://www.autismsociety-nc.org/>
- Autism Navigator <http://autismnavigator.com/resources-and-tools/#about>
- Early Intervention Program <http://www.ncei.org>
- Children's Developmental Services Agency (CDSA) <http://www.ncei.org/ei/tip/cdsa.html>

Web Resources for Providers

- American Academy of Pediatrics
 - <http://www.aap.org/healthtopics/autism.cfm>
- Autism Speaks ASD glossary & Tool Kits for parents & providers (blood draw, dentist visits) <http://www.autismspeaks.org/>
- National Professional Development Center on Autism Spectrum Disorder (24 evidence-based practices, training modules, online course) (<http://www.fpg.unc.edu/~autismpdc/>)
- Autism Navigator <http://autismnavigator.com/resources-and-tools/#about>
- CDC Autism Case Training, videos, web-based CE <http://www.cdc.gov/ncbddd/actearly/autism/video/index.html>

CDC Learn the Signs/Act Early Autism Case Training



- Individual Modules
 - Identifying
 - Diagnosing
 - Managing
- Online Course Available
- CE credit

CDC Milestones



CDC Milestone Checklist

Your Baby at 2 Months

Child's Name _____ Child's Age _____ Today's Date _____

How your child plays, learns, speaks, and acts offers important clues about your child's development. Developmental milestones are things most children can do by a certain age. Check the milestones your child has reached by the end of 2 months. Take this with you and talk with your child's doctor at every visit about the milestones your child has reached and what to expect next.

What most babies do at this age:

Social/Emotional

- Begins to smile at people
- Can briefly calm himself (may bring hands to mouth and suck on hand)
- Tries to look at parent

Act early by talking to your child's doctor if your child:

- Doesn't respond to loud sounds
- Doesn't watch things as they move
- Doesn't smile at people

Suggestions for Enhancements? Questions and Discussion

