
Early Identification of Autism Spectrum Disorder

The HELP Group Summit
October 14, 2016

Sally Ozonoff, Ph.D.
Professor and Vice Chair
MIND Institute, UC Davis

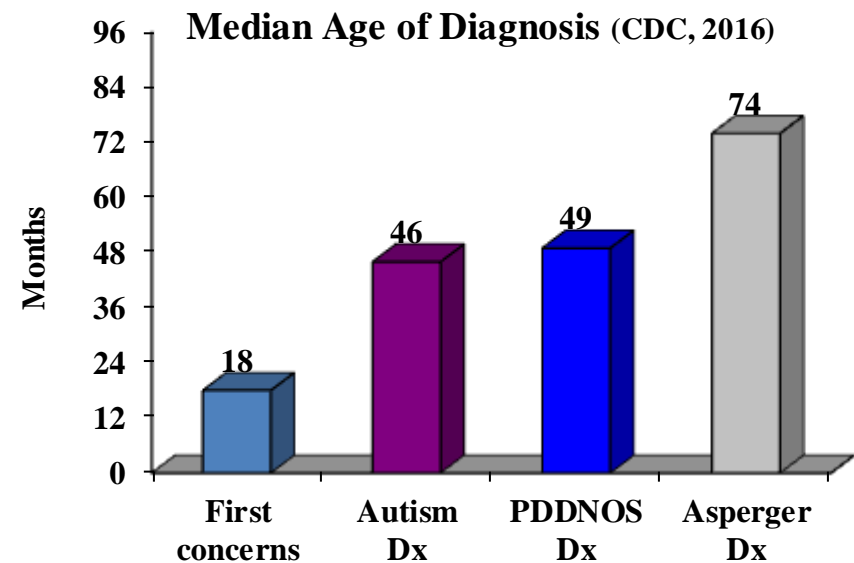
Outline

- Early signs of ASD
 - Stability of early diagnoses
 - Other challenges in siblings
 - Screening for ASD in infants and toddlers
 - Working with families
-

Earlier Identification

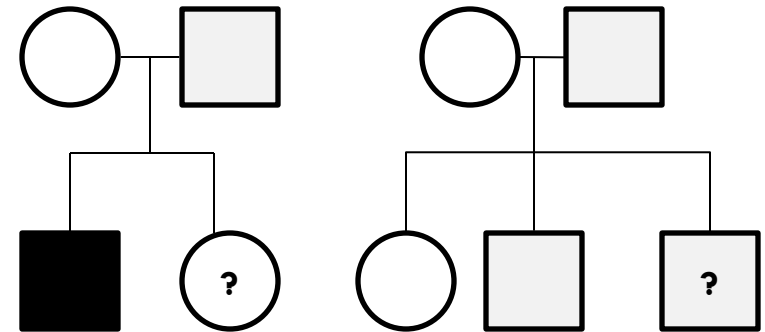
- Median age of diagnosis = 50 months (CDC, 2016)
- Mean age of first parental concern = 18 – 19 months
 - 30–50% report concerns before 1st birthday

**Earlier identification →
Earlier intervention**



Methods to Study Early Symptoms

- Retrospective
 - Parent report
 - Home video analysis
- Prospective
 - High risk samples
 - Longitudinal assessments of development



Infant Sibling Design

UC Davis – UCLA Infant Sibling Study



Phase I 2003 - 2008

N = 350



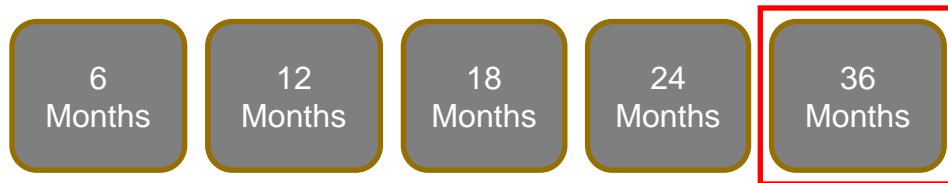
Phase II 2008 - 2013

N = 300



Phase III 2013 - 2017

N = 200



UC Davis – UCLA Infant Sibling Study



■ Goals

1. How early can we identify ASD in babies?
 - What are the earliest reliable markers?
2. How stable is early diagnosis?
3. What other developmental problems occur in infant siblings?

Measures

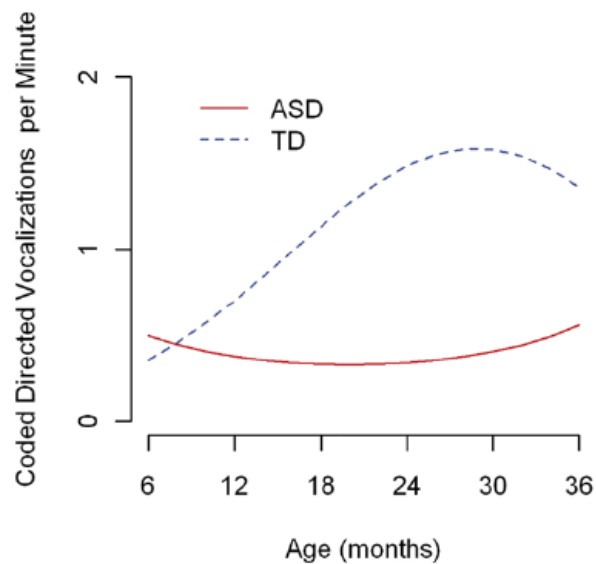
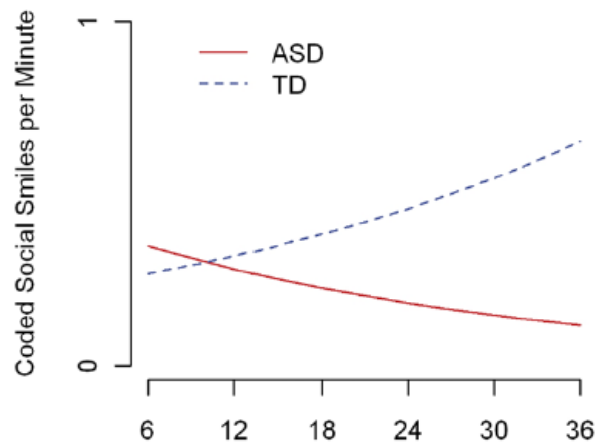
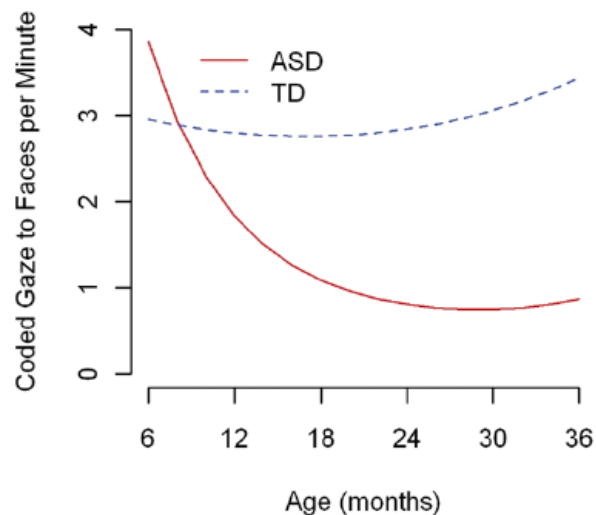


Communication	<u>ESCS</u> , Mullen, MacArthur CDI, Language Use Inventory
Social-Emotional	Peek-a-boo, imitation, empathy, parent-child play, <u>response to name</u>
Face Processing	Eye gaze during still face paradigm
Autism Symptoms	AOSI, ADOS, ADI, SRS
Other	Cognitive, adaptive, motor, head circumference, <u>object use/exploration</u> , form and motion processing, smooth pursuit, audiovisual integration of speech

Group Differences: ASD Outcomes v Low Risk Infants

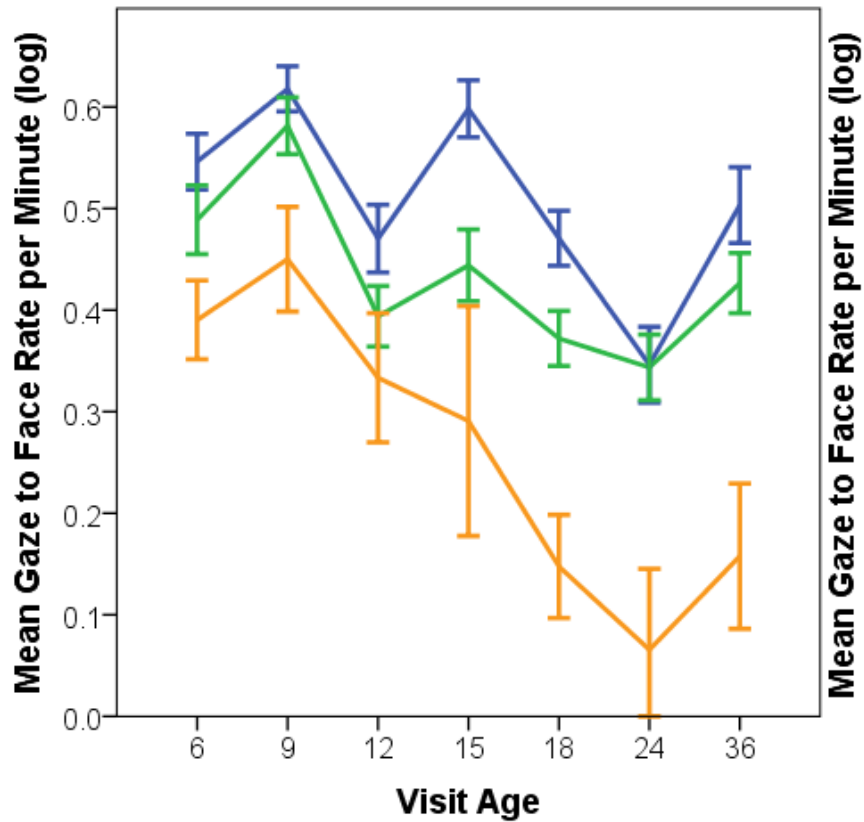
	6 m	12 m	18 m	24 m	36 m
Mullen language		✓	✓	✓	✓
MCDI vocabulary		✓	✓	✓	
Vocalization freq		✓	✓	✓	✓
Empathy		✓	✓	✓	✓
ESCS joint attention		✓			
Response to name		✓	✓	✓	
Face processing					
Imitation		✓	✓	✓	✓
Parent-infant synchrony		✓			
Eye contact frequency		✓	✓	✓	✓
Examiner social ratings		✓	✓	✓	✓
Parent social ratings		✓	✓	✓	✓
Repetitive object use		✓	✓	✓	✓

Declining Trajectories: Behavior with Examiners



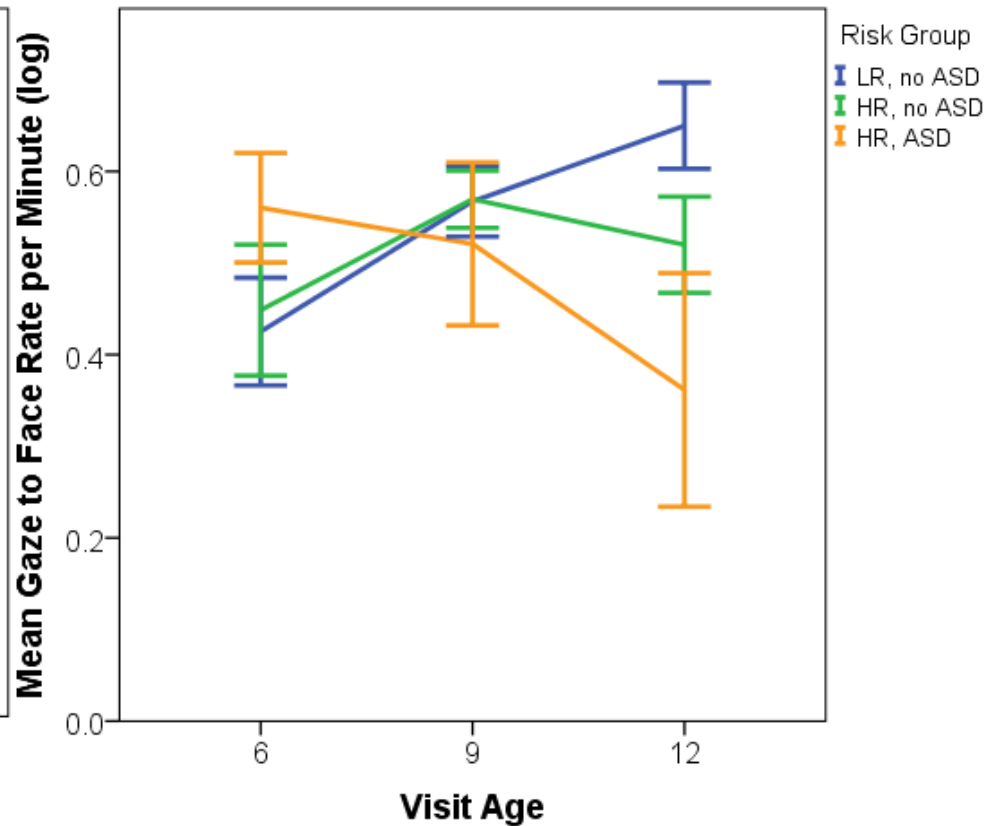
Eye Contact

With Examiner



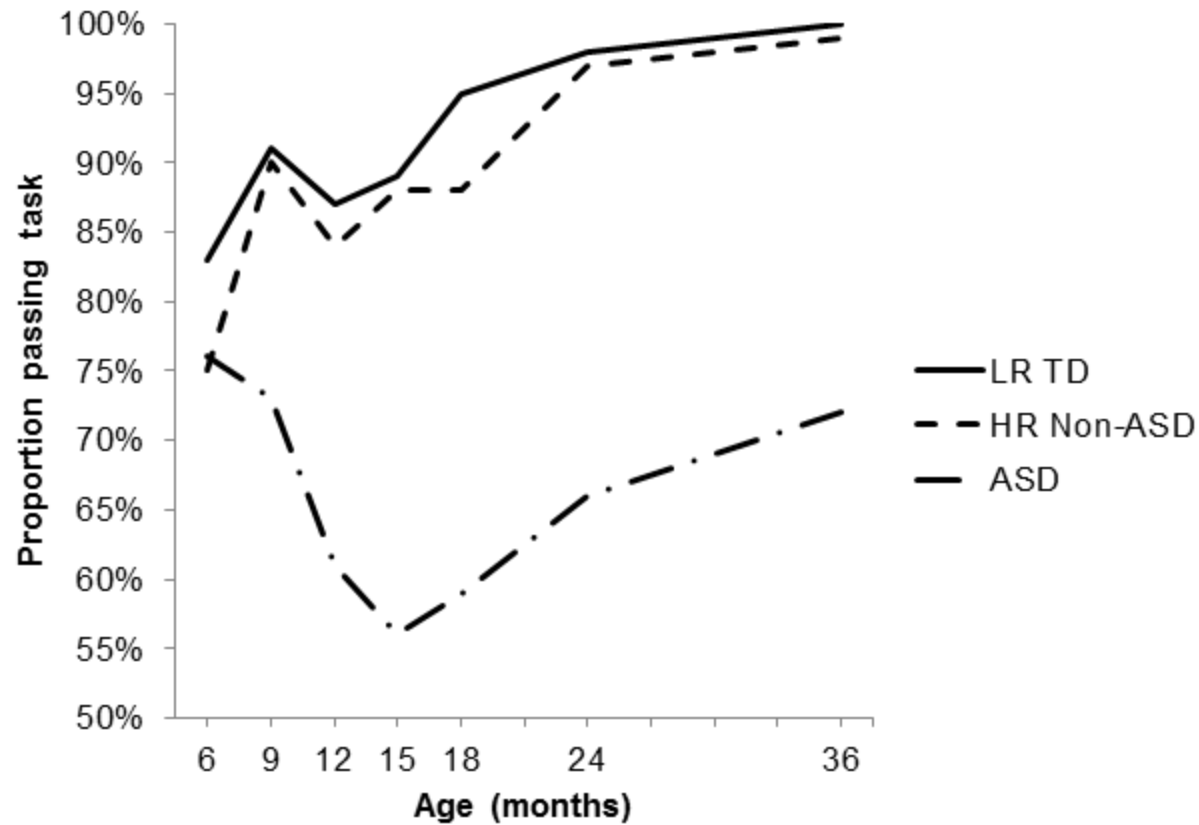
Error Bars: +/- 1 SE

With Mother

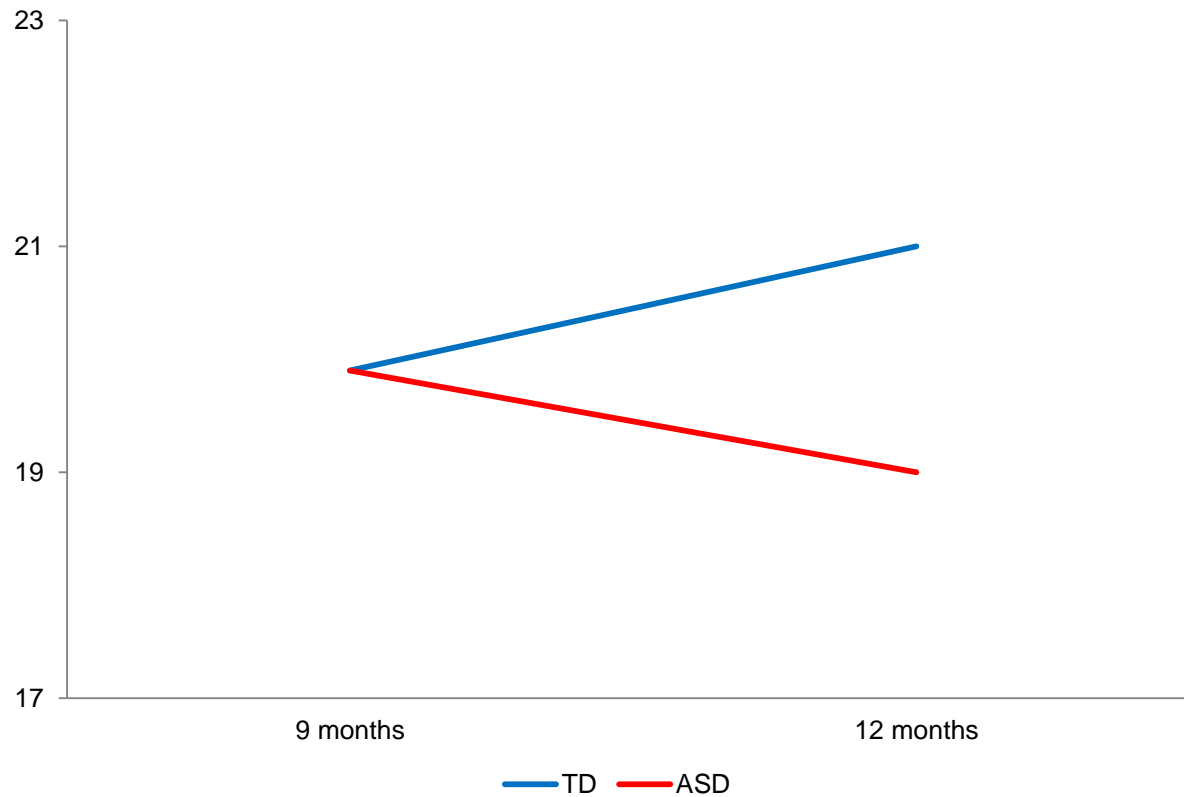


Error Bars: +/- 1 SE

Response to Name

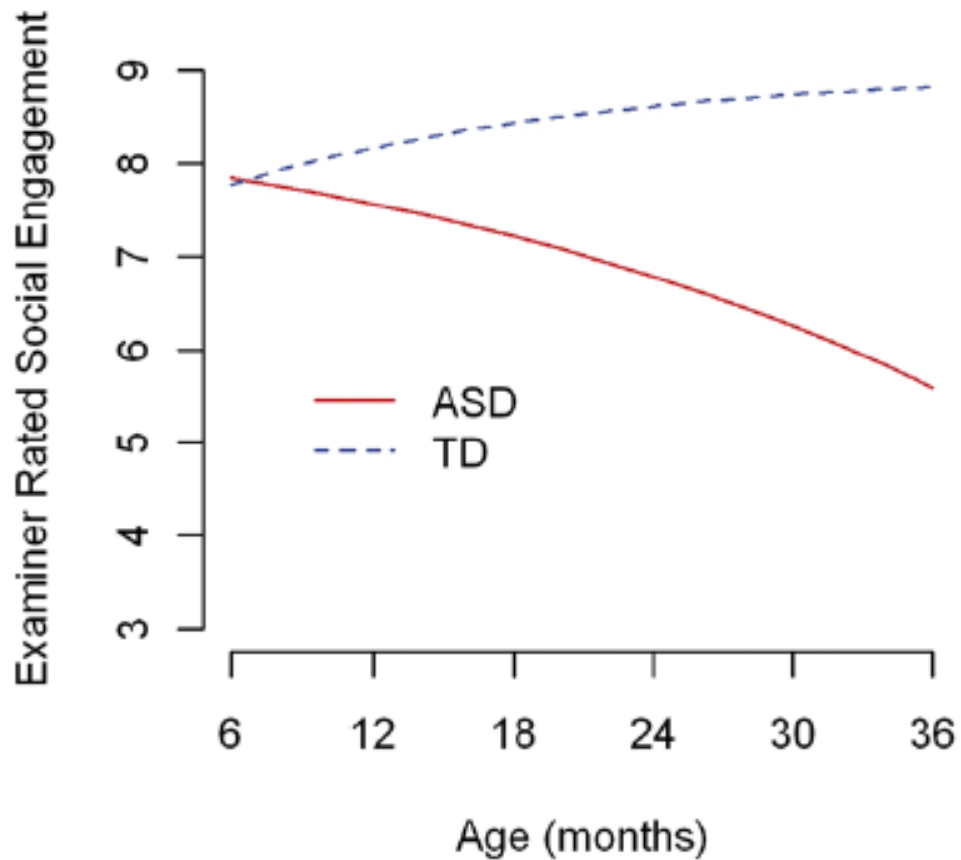


NCAST Feeding Scale



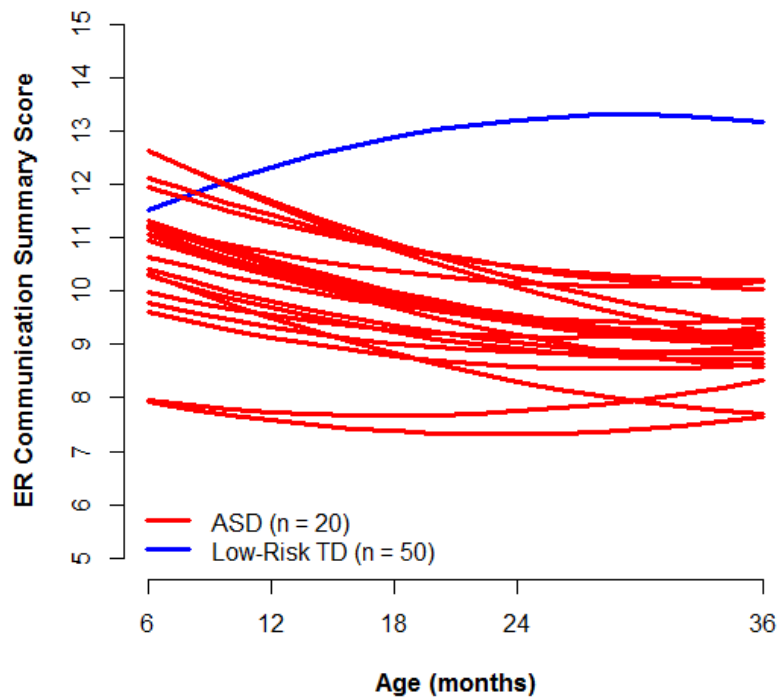
Informant Ratings

Examiner Ratings, 2010

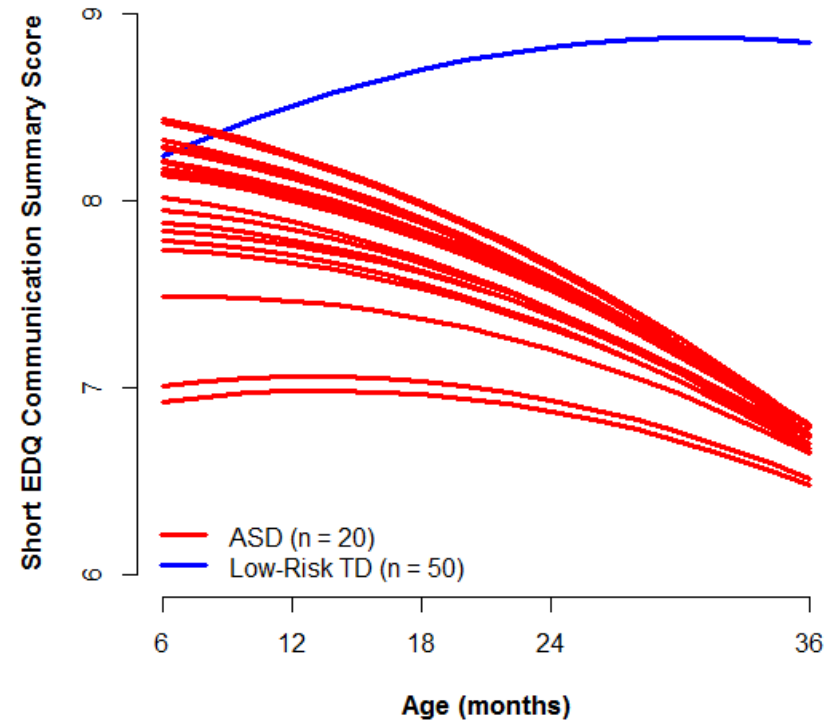


Informant Ratings

Examiner Ratings, 2014

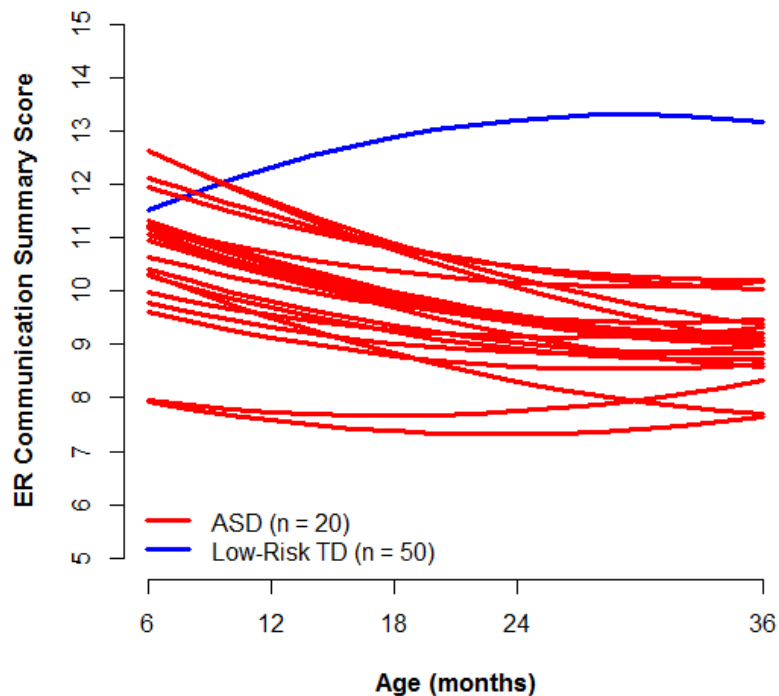


Parent Ratings, 2014



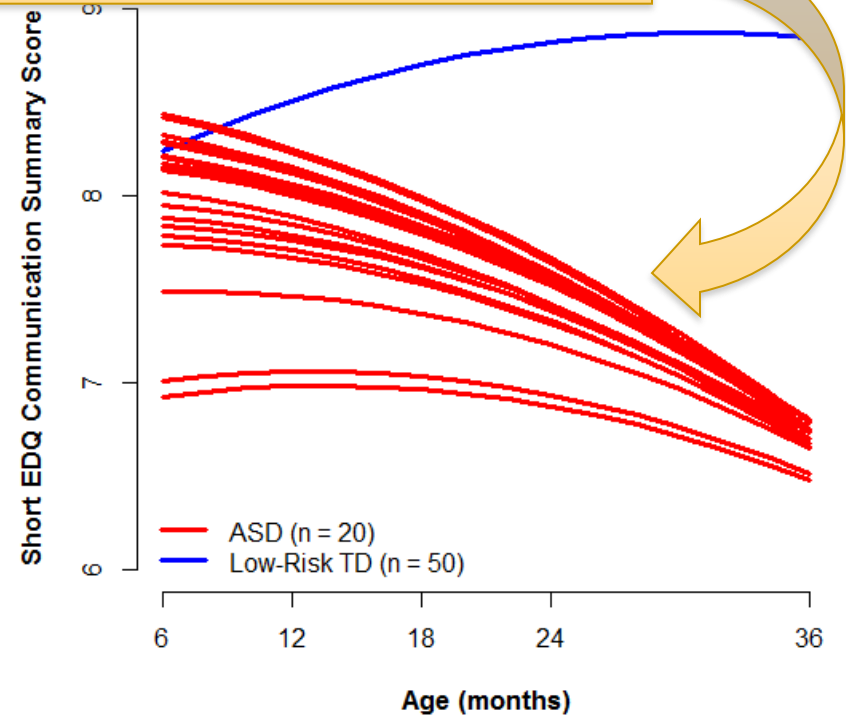
Informant Ratings

Examiner Ratings, 2014



Parent Ratings, 2014

Only 6 of 18 parents endorsed a regression retrospectively



Prospective Tracking of Symptoms

	6m	12m	18m	24m	36m
1	Green	Green	Green	Green	Red
2	Green	Green	Green	Red	Red
3	Green	Green	Yellow	Yellow	Red
4	Green	Green	Yellow	Red	Red
5	Green	Green	Yellow	Red	Red
6	Green	Green	Yellow	Red	Red
7	Green	Green	Yellow	Red	Red
8	Green	Green	Yellow	Red	Red
9	Green	Green	Yellow	Red	Red
10	Green	Green	Yellow	Red	Red
11	Green	Green	Yellow	Red	Red
12	Green	Green	Yellow	Red	Red
13	Green	Green	Yellow	Red	Red
14	Green	Green	Yellow	Red	Red
15	Green	Green	Red	Red	Red
16	Green	Green	Red	Red	Red
17	Green	Green	Red	Red	Red
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28	Green	Yellow	Red	Red	Red
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40	Green	Red	Red	Red	Red
41	Yellow	Yellow	Yellow	Red	Red
42	Yellow	Yellow	Red	Red	Red
43	Yellow	Yellow	Red	Red	Red
44	Yellow	Red	Red	Red	Red
45	White	Yellow	Red	Red	Red
46	White	Yellow	Red	Red	Red
47	White	White	Yellow	Yellow	Red
48	White	White	Yellow	Yellow	Red

M age of diagnosis =
23.3 months

50% diagnosed by 18m
75% diagnosed by 24m

Group Differences: ASD Outcomes v Low Risk Infants

	6 m	12 m	18 m	24 m	36 m
Mullen language Landa et al., 2013		✓	✓	✓	✓
MCDI vocabulary		✓	✓	✓	
Vocalization freq		✓	✓	✓	✓
Empathy		✓	✓	✓	✓
ESCS joint attention		✓			
Response to name Gammer et al., 2015; Zwaigenbaum et al., 2005		✓	✓	✓	
Face processing Jones & Klin, 2013					
Imitation		✓	✓	✓	✓
Parent-infant synchrony Wan et al., 2013		✓			
Eye contact frequency		✓	✓	✓	✓
Examiner social ratings		✓	✓	✓	✓
Parent social ratings		✓	✓	✓	✓
Repetitive object use Wolff et al., 2014		✓	✓	✓	✓

Conclusions

- *ASD emerges over the first 2 years of life*
 - Not present at birth as Kanner suggested
 - Most children developing ASD show declining trajectories
 - Regressive onset is the norm, not the exception
 - Parents identify declining trajectories prospectively, but less so retrospectively
 - ADI-R report of regression is only the tip of the iceberg
 - Promise for development of screening measures
-

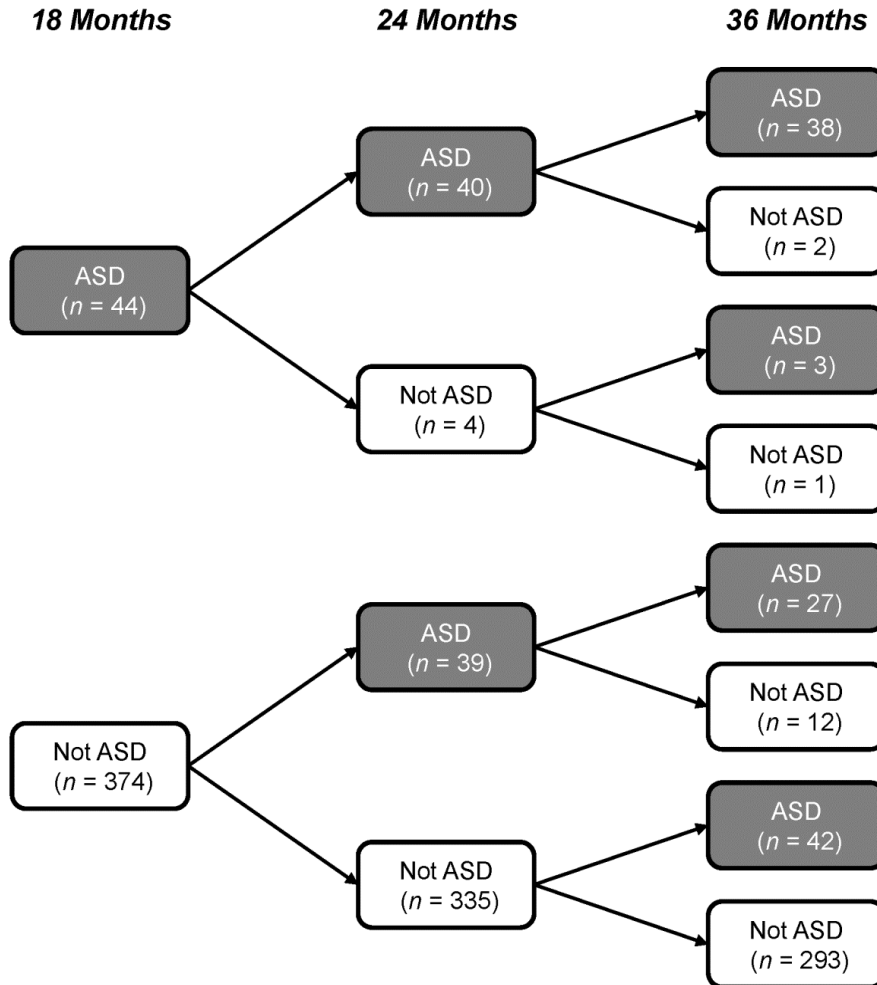
UC Davis – UCLA Infant Sibling Study



■ Goals

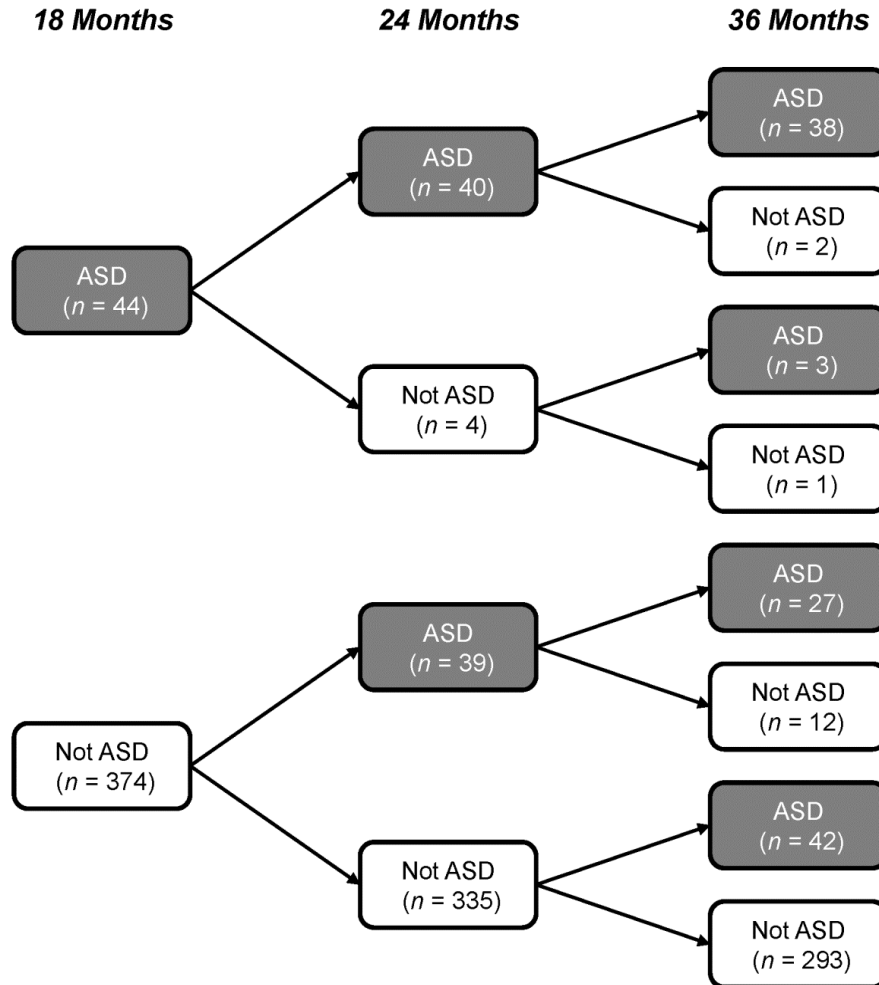
1. How early can we identify ASD in babies?
 - What are the earliest reliable markers?
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3. What other developmental problems occur in infant siblings?

Stability of Early Diagnosis



	Sensitivity	Specificity	PPV/Stability
18 Months	37%	99%	93%
24 Months	59%	95%	82%

Stability of Early Diagnosis



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18 Months	37%	99%	93%
24 Months	59%	95%	82%

Stability of Early Diagnosis



- Diagnosis before 36 months is very stable
 - Over 90% retain the diagnosis
 - If diagnosed early, intervene
 - No need to confirm diagnosis at older ages
- Very few “false positives”
- But... many children with ASD are not identified at 18 or 24 months
 - Symptoms emerge over time

Conclusions and Clinical Implications

Few markers of ASD at 6 months or earlier

Continued search for markers that identify risk before the onset of behavioral signs

Declining developmental trajectories that clinicians and parents can identify

Early diagnosis is very stable – if diagnosed, intervene

Screen early, screen often, screen later

Family history of ASD is a significant risk factor for later-born siblings

Medical Risk-Prevention Model

	Cardiovascular Disease	Autism
Undesired outcome	heart attack, stroke	functional impairment
Screening for risk factors	family hx, hi chol, smoking, diabetes, hypertension	older sibling, failed screening
Prevention/Monitoring	lifestyle changes (e.g., exercise, diet)	parent training
More intensive intervention as needed	medication, stent, bypass surgery	in-home or center-based beh thx

AAP Practice Recommendations (2007)

- Primary care providers should ...
 - Be aware of early signs
 - Perform developmental surveillance and elicit parent concerns at every visit
 - Administer standardized screening measure if indicated
 - Perform formal screens for autism at 18 and 24 months for all children



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Infant Social-Communication Milestones

1 month

**Gaze to
Faces and
Eyes**

1-2 months

**Social
Smile**

3 months

**Cooing,
Turn-Taking
Vocals**

6-9 months

**Consonant-
Vowel
Babbling**

6-9 months

**Responds
to Name**

9-15 months

**Gestures
&
Pointing**

12-15 months

Imitation

Red Flags for Autism in Infancy

Babies should be referred who are not...

- 6 – 9 months
 - Looking at faces
 - Smiling at others
 - Cooing
- 9 – 12 months
 - Responding to name
 - Babbling
 - Playing social games
 - Displaying bright affect
- 12 – 18 months
 - Pointing and showing
 - Using single words
 - Using gestures
 - Imitating
 - Interested in peers



Screening Tools

- Autism-specific screeners
 - Infant-Toddler Checklist (ITC)
 - 6 – 24 months
 - <http://firstwords.fsu.edu/pdf/checklist.pdf>
 - Modified Checklist for Autism in Toddlers (M-CHAT)
 - 16 – 30 months
 - <https://www.m-chat.org>





John Constantino MD
Professor of Psychiatry
Washington University in St. Louis
School of Medicine



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Journal of Child Psychology and Psychiatry **:* (2015), pp **~**

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Rapid video-referenced ratings of reciprocal social behavior in toddlers: a twin study

Natasha Marrus,¹ Anne L. Glowinski,¹ Theodore Jacob,² Ami Klin,^{3,4,5} Warren Jones,^{3,4,5}
Caroline E. Drain,^{1,6} Kieran E. Holzauer,¹ Vaishnavi Hariprasad,¹ Robert T. Fitzgerald,¹
Erika L. Mortenson,¹ Sayli M. Sant,¹ Lyndsey Cole,¹ Satchel A. Siegel,¹ Yi Zhang,¹
Arpana Agrawal,¹ Andrew C. Heath,¹ and John N. Constantino¹

¹Department of Psychiatry, Washington University, St. Louis, MO; ²Department of Psychology, Palo Alto University, Palo Alto, CA; ³Marcus Autism Center, Children's Healthcare of Atlanta, Atlanta, GA; ⁴Division of Autism & Related Disabilities, Department of Pediatrics, Emory University School of Medicine, Atlanta, GA; ⁵Center for Translational Social Neuroscience, Emory University, Atlanta, GA; ⁶Department of Neurology, Washington University, St. Louis, MO, USA

Video Referenced Infant Rating System for Autism (VIRSA)

- Video-based screening instrument for 6, 9, 12, and 18-month-olds
 - Measuring social-communication behavior and repetitive, unusual use of objects
- Examine psychometric properties
 - Test-retest reliability
 - Inter-rater reliability
 - Convergent and discriminant validity
 - Predictive validity

VIRSA

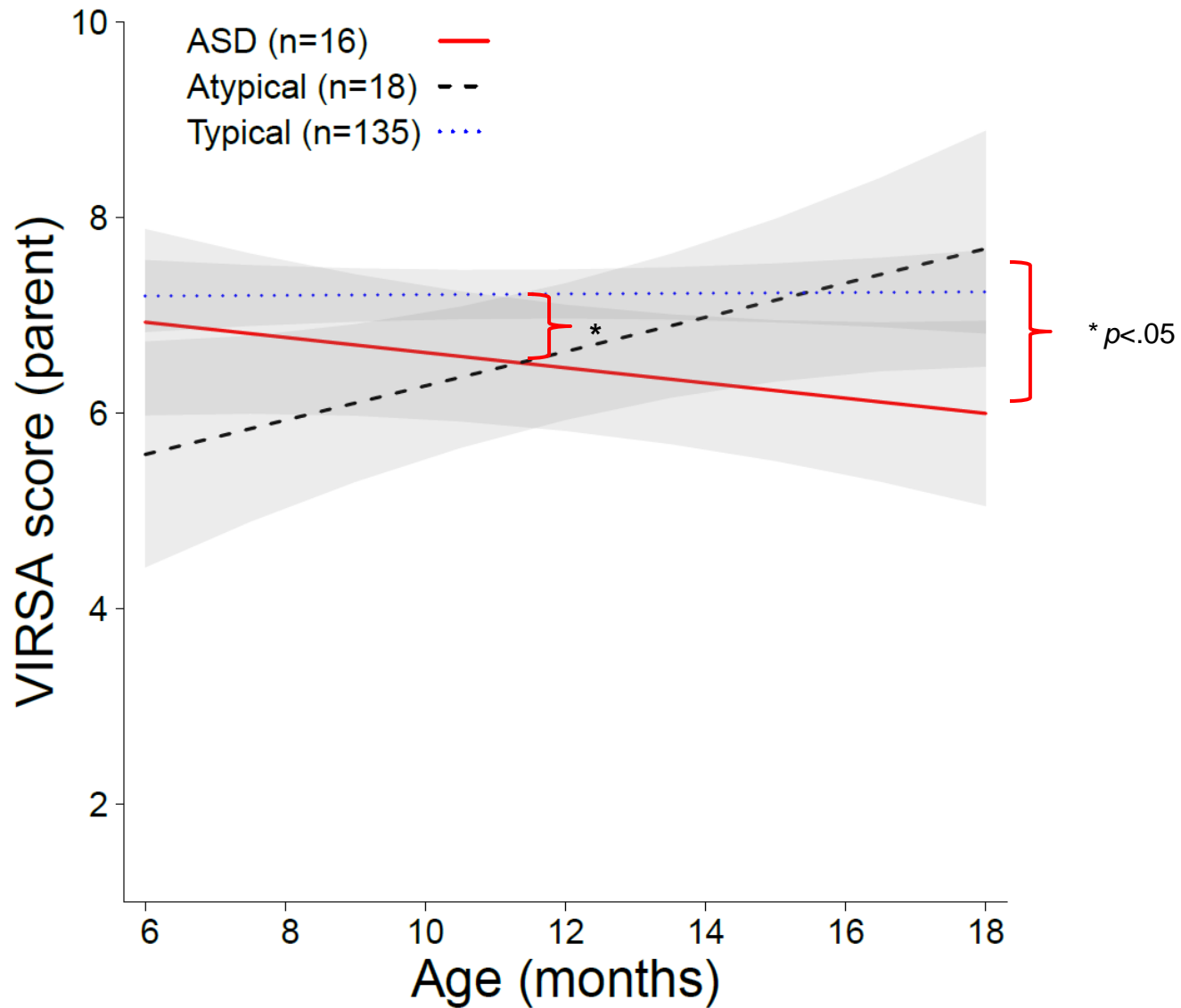
- 20-second video segments
- Paired to contrast
- Babies playing socially with their parents
- Clips display varying degrees of sociability



Completed VIRSA ratings

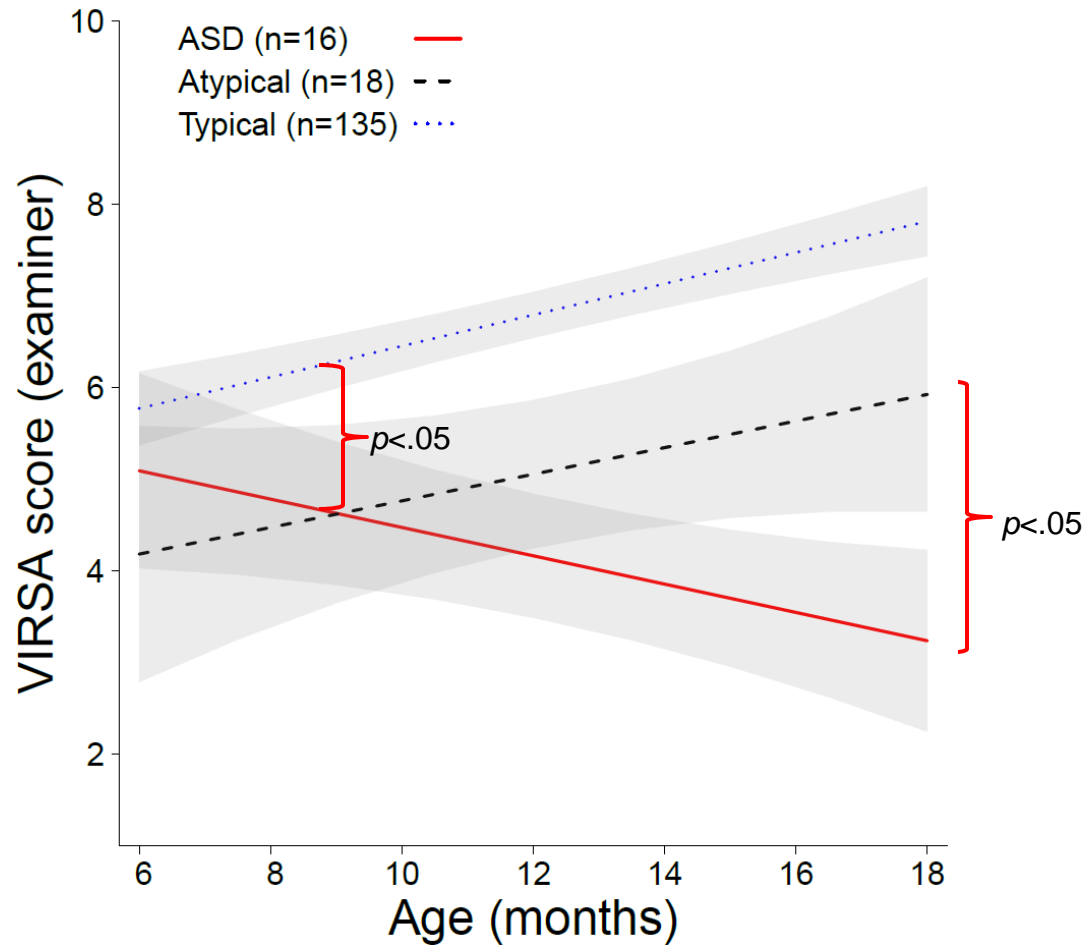
	6 mos	9 mos	12 mos	18 mos
Parent	81	111	136	87
Parent retest	62	85	90	48
Examiner	84	45	144	89

VIRSA Ratings: Parent



Main effect for Group: $X^2 = 6.68, p < .05$
Group x Age interaction: $X^2 = 6.41, p < .05$

VIRSA Ratings: Examiner



Main effect for Group: $X^2 = 55.4, p < .001$
Group x Age interaction: $X^2 = 20.7, p < .001$

VIRSA Screening Efficacy

No differences among groups at 6 months, replicating other measures

Declining trajectories over time, replicating other measures

By 9 months, the screening instrument can identify broad/general delays

By 18 months, it can differentiate which infants have ASD

This low-burden, video-based, informant-report screening measure is promising for earlier identification

Working with Families in the Face of Diagnostic Uncertainty

PROMOTE:

- Transparency and trust
- Monitoring



AVOID:

- Undue alarm
- Negative changes in relationship with child

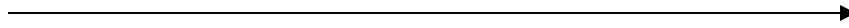
Working with Families in the Face of Diagnostic Uncertainty

What we see

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What parents see

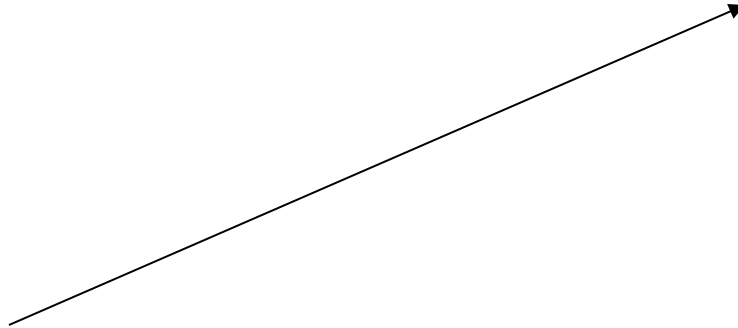
Inconsistent eye contact



Independent



Flat affect



Strong-willed



Low facial tone

How to convey concerns to parents?

Communicating Concerns

- Acknowledge, do not dismiss, parent concerns
- Have open and honest discussions
 - Be willing to express uncertainty
 - Acknowledge range of outcomes, imperfection of prediction of severity
- Discuss repercussions of false negatives and false positives
 - Families will differ in their values
- Provide parenting strategies and practical advice

Resources

- *Does My Child Have Autism? A Parent's Guide to Early Detection and Intervention* by Stone & DiGeronimo (2006)
 - ASDetect.org – a video-based app for early identification of ASD
 - *An Early Start for Your Child with Autism* by Rogers et al (2012)
 - *The Activity Kit for Babies and Toddlers at Risk: How to Use Everyday Routines to Build Social and Communication Skills* by Fein et al (2016)
 - <http://www.autismspeaks.org>
 - First 100 Days kit
 - Learn the Signs campaign
 - Video Glossary
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