Early Expression of Autism Spectrum Disorders

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Overview

• Onset and diagnosis

• Syndrome expression

• Face processing
Autism: Clinical Features

• **Classic triad of symptoms:**
  – Impaired social interaction
  – Impaired communication
  – Presence of restricted interests and activities

• **Onset prior to 36 months**

• **Diagnosis often delayed**

• **Marked heterogeneity**

• **Developmental disorder**

Leo Kanner
Age of Onset: Parental Perception

- **Average age at first concern:**
  - 15 months (SD=6.5)

- **Primary concerns:**
  - Social difficulties
  - Speech delays

- **Why such a variability?**

*Chawarska, Paul, Klin et al., 2007, JADD.*
Patterns of Onset

- **Early onset (1st year):**
  - “Inborn autistic disturbances of affective contact” (Kanner, 1943)

- **Later onset (2nd year):**
  - Regression (15-27% of cases) (Eisenberg & Kanner, 1955; Dawson et al., 2006; Landa et al., 2007)
  - Plateau (Ozonoff et al., 2008; Hansen et al., 2008)
Methodological Approaches to Studying Emergence of Autism

- **Retrospective studies of affected children**
  - Parental report
  - Video diaries analysis

- **Prospective studies of infant siblings at risk for ASD**
  - Recurrence risk for autism: 5-10%
  - High risk for other developmental problems:
    - Language or cognitive delays
    - Broader Autism Phenotype, BAP
Autism in the First Year
Potential Areas of Dysfunction in the 1\textsuperscript{st} Year

1. Typical Development
   - 0 to 3 months:
     • Sensitivity to and preference for face-like stimuli and speech-like sound
   - 3 to 6 months:
     • Emergence of dyadic social interactions
   - 6 to 9 months:
     • Development of face processing skills (identity, affect, gender)
     • Response to name
     • Anticipatory social games
   - 9 to 12 months:
     • Social monitoring and imitation
     • Social referencing
     • Joint attention

2. Many skills affected in toddlers with ASD emerge typically in the first year
Autism at 6 months?

- Reports of typical presentation at 6 months in children with autism
  - Eye contact and affective responses to mother (Young et al., 2009)
  - Social reciprocity and attention (Bryson et al., 2006; Zwaigenbaum et al., 2005)
  - Verbal and nonverbal skills (Landa et al., 2007)

- Emerging consensus that overt symptoms of ASD begin to emerge between 6 and 12 months

- Possible factors responsible for these findings:
  - Natural course of autism
  - Limited sensitivity of existing behavioral methods
Emerging Symptoms of Autism at 12 months

- **Limited response to name**
  - High specificity for ASD (89%)
  - Low sensitivity (50%)

- **Limited eye contact and use of communicative gestures:** pointing, showing

- **Delays in language:** limited range and frequency of vocalizations

- **Atypical behaviors:** Spinning and intense visual examination of objects
ASD in Toddlers
Abnormalities in Social Interaction

- Limited interest in people
- Limited of social reciprocity:
  - Social smile
  - Shared enjoyment
  - Pleasure derived from interactions
- Unusual eye contact
- Limited affective range
- Limited joint attention skills
- Poor observational/imitative learning

Communication

- Low frequency of communication
- Paucity of conventional and descriptive gestures (nonverbal communication)
- Limited goals of communication (instrumental versus declarative)
- Stereotypical/idosyncratic use of language (e.g., echolalia, scripting)
- Use of other’s body to communicate (hand-over-hand gestures)
Abnormalities in Play and Imagination Development

- **Exploratory**: present but often atypical
- **Functional**: may be spared but atypical
- **Pretend**:  
  - Absent  
  - Present but atypical, non-generative
Restricted Interests and Repetitive Behaviors

- **Seeking/avoiding specific visual stimuli** (lights, motion, touch)
- **Seeking sensory input** (jumping, rocking, spinning)
- **Interest in details of objects** (e.g., wheels, dials)
- **Hand and finger mannerisms**
Exceptional Abilities

- **John Longdon Down: Idiot Savant (1887)**

- **Savant skills in autism**
  - More frequent in males
  - Prevalence 1-10% in autism, less prevalent in other disorders

- **Etiology: unknown**

- **Examples of savant skills:**
  - Exceptional memory
  - Computational skills
  - Artistic abilities
  - Musical skills

  Stephen Wiltshire
Unusual Abilities in Toddlers

- **Exceptional skills**
  - Interest in shapes, letters, numbers
  - Early recognition of signs ("reading")
  - Good expressive vocabulary
  - Great memory for movies, books

- **Limited functionality such skills**
  - Isolated
  - Repetitive & restrictive (e.g., labeling, scripting)
  - Potentially transient
Diagnostic Assessment
Diagnostic Assessment Battery for Toddlers

- Developmental tests: e.g., Mullen Scales of Early Development
- Communication: Communication and Symbolic Behaviors Scale
- Adaptive skills: Vineland Adaptive Behaviors Schedules-II
- Medical and developmental history interview
- Genetic and neurological testing
Stability of Early Clinical Diagnosis

• **Short term stability (2nd year to 4 years)** *(Chawarska et al., 2007; 2009)*
  - Very good for ASD diagnosis (80-90%)
  - Changes expected **within** spectrum due to shifts in number of symptoms and intensity

• **Long term stability (2 to 4 to 9 years)** *(Lord et al., 2006)*
  - High stability of ASD diagnosis (90%)
  - Shift from PDD-NOS to Autism Dx: ~20%
  - Shift from Autism to PDD-NOS: ~10%
Stability of Clinical Diagnosis

<table>
<thead>
<tr>
<th>Provisional Diagnosis &lt; 2 yrs</th>
<th>Autism</th>
<th>PDD-NOS</th>
<th>Non-ASD</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autism</td>
<td>32 (74%)</td>
<td>11 (26%)</td>
<td>0 (0%)</td>
<td>43</td>
</tr>
<tr>
<td>PDD-NOS</td>
<td>3 (16%)</td>
<td>15 (83%)</td>
<td>0 (0%)</td>
<td>18</td>
</tr>
<tr>
<td>Non-ASD</td>
<td>1 (4%)</td>
<td>2 (7%)</td>
<td>25 (89%)</td>
<td>28</td>
</tr>
<tr>
<td>Total</td>
<td>36</td>
<td>28</td>
<td>25</td>
<td>89</td>
</tr>
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</table>

Chawarska, Klin, Paul, Macari, & Volkmar, 2009, JCPP
Case Study

Female high-risk infant followed from 6 to 36 months
Case Study: Verbal and Nonverbal Scores

- Nonverbal Skills
- Verbal skills

Mental Age (months)

Chronological Age (months)

Yale Child Study Center
Case Study: ADOS Total Score

- Social Affect
- Repetitive Behaviors

ADOS score vs. Chronological Age:

- 15m: 0
- 18m: 2
- 24m: 4
- 36m: 7
Vineland Adaptive Skills: Communication

- Expressive
- Receptive
- Written

Mental Age (months)

- 12m
- 18m
- 24m
- 36m
Video not available
Eye-Tracking Studies of Face Processing

- **Scanning as active process of seeking task-relevant information**

- **Visual scanning is affected by:**
  - Perceptual factors (e.g., high contrast, motion, etc.)
  - Semantic factors (e.g., familiarity with a specific face, nature of the task)
  - Individual characteristics (e.g., age, disorders)

*From: A.R. Yarbus, 1965*
Face Scanning in Babies
Face Scanning and Recognition

Visual Paired Comparison Paradigm

Familiarization

Test of recognition
Effect of Context on Face Scanning in Infancy
Atypical Face Processing in Toddlers with ASD
Face Processing in Autism

- Deficits in face processing in older children and adults
  - Face recognition
  - Atypical face processing strategies (features over configurations)
  - Atypical brain activation pattern in response to faces

- Developmental end-points versus developmental process
How toddlers with ASD scan and recognize faces?
Face Scanning and Recognition: Toddlers

ASD N=32, TD N=46

Age 1: 26 months (SD=6.5)
Age 2: 47 months (SD=5.5)

Fixed-trial Visual Paired Comparison paradigm

Sequence of six trials using unfamiliar faces

Measure of recognition: Novel/Novel+Familiar

<table>
<thead>
<tr>
<th>Phase</th>
<th>Duration</th>
<th>Display</th>
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<tbody>
<tr>
<td>centering</td>
<td>1 s</td>
<td><img src="image" alt="Centering" /></td>
</tr>
<tr>
<td>Familiarization</td>
<td>(10 s)*</td>
<td><img src="image" alt="Familiarization" /></td>
</tr>
<tr>
<td>blank</td>
<td>5 s</td>
<td><img src="image" alt="Blank" /></td>
</tr>
<tr>
<td>centering</td>
<td>1 s</td>
<td><img src="image" alt="Centering" /></td>
</tr>
<tr>
<td>Recognition</td>
<td>5 s</td>
<td><img src="image" alt="Recognition" /></td>
</tr>
</tbody>
</table>

Chawarska & Shic, 2009
Regions of Interest

Example of the face stimulus and Regions of Interest (ROI) demarcation

- **Eyes**
- **Mouth**
- **Nose**

**Inner**

- **Outer** (cheeks/forehead, neck/body, hair)
- **Non-Face** (screen, background)

<table>
<thead>
<tr>
<th>Region</th>
<th>Color</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eyes</td>
<td>Pink</td>
</tr>
<tr>
<td>Mouth</td>
<td>Yellow</td>
</tr>
<tr>
<td>Hair</td>
<td>Red</td>
</tr>
<tr>
<td>Cheeks/Forehead</td>
<td>Blue</td>
</tr>
<tr>
<td>Nose</td>
<td>Green</td>
</tr>
<tr>
<td>Neck/Body</td>
<td>Brown</td>
</tr>
<tr>
<td>Background</td>
<td>Gray</td>
</tr>
<tr>
<td>Screen</td>
<td>Black</td>
</tr>
</tbody>
</table>
Face Scanning and Recognition

- **Recognition:**
  - **TD:**
    - Novel > Familiar, $p < .01$
    - NP = .56 (.07), $p < .001$
  - **ASD:**
    - Novel = Familiar
    - NP = .51 (.11), $p < .23$

- **Scanning vs. Recognition:**
  More exclusive focus on Eyes associated with less effective encoding, $r = -.42, p < .01$
Face versus Object Recognition

Nineteen toddlers with ASD
(age: M=41 m, SD=14)

Novelty Preference: Objects:
M = .56 (.10), t(18) = 2.5, p < .02

Novelty Preference: Faces
M = .52 (.11), t(16) = .67, p ns.

Bradshaw, Shic, Chawarska, submitted
Are there autism-specific deficits in the attentional aspects of face processing in toddlers with ASD?
Privileged Attentional Status of Faces

Faces are detected faster than objects
Faces hold attention more strongly than objects

- Increased cost of disengagement of attention away from a face: increase in reaction time
- Noted in response to neutral and affective faces
- Possibly modulated by presence of direct eye contact
Attentional Bias for Faces in ASD

Overlap attention cueing paradigm

ASD (N=42), DD (N=31), TD (N=46)

CA-matched groups: 29 months (15 to 60 months)

DD and ASD matched on verbal and nonverbal mental age

Dependent variables:

*Saccadic Reaction Time (SRT)*

Procedure

**NONSOCIAL CONDITION**

**SOCIAL CONDITION**
Saccadic Reaction Time

SRT NonSocial: ASD = DD = TD

SRT Social (p<.008): ASD < DD = TD

DirectGaze = IndirectGaze
Not a Captive Audience...

- Faces do not elicit attentional bias in toddlers with ASD
- This effect is specific to ASD
- Are toddlers with ASD simply faster at processing faces?
Faster to Disengage: What Does it Mean?

- **Speed of processing?**
  - Poor recognition despite attending to faces (Chawarska & Shic, 2009, Chawarska & Volkmar, 2007)

- **Depth of processing?**
  - Perceptual features (e.g., face-no face, male-female)
  - Semantic features (e.g., mom-not mom, nice-mean)

- **Faces in ASD are processed on a more superficial level as indexed by diminished disengagement cost and poor recognition skills**
Face Processing Abnormalities in Toddlers with ASD

- Limited attentional bias for faces
- Deficit in face recognition
- Restricted scanning of key features linked to less effective encoding
- Face scanning pattern might become more abnormal with age
- Next frontier:
  - Primary or secondary impairments?
  - Are other aspects of face processing affected as well?
Early Expression of ASD

- Symptoms of social dysfunction are apparent in a majority of cases by 24 months

- Early Autism Spectrum diagnosis is relatively stable

- High variability in the rate of progress reflected in changes of diagnostic classification within spectrum, IQ, and verbal ability

- Next frontier:
  - ASD in the 1st year
  - Parsing heterogeneity of syndrome expression
Thank you