Co-Occurring Autism in Down syndrome: Considerations for Early Development

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Research Patterns

Autism vs. Down Syndrome Publications in the Last 20 Years

- Autism
- DS
What are the **Implications** of this trend?

### Autism
- More Knowledge – risk factors, development, co-occurring diagnoses, phenotype
- Sources of variability
- Importance of early intervention
- Autism-specific interventions (Early Start Denver Model)

### Down syndrome
- Minimal understanding of early development
- Risk factors
- Lack of Down syndrome-specific interventions
- Policy implications
<table>
<thead>
<tr>
<th><strong>Refine</strong></th>
<th>Identification and Behavioral Profile</th>
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<tbody>
<tr>
<td><strong>Characterize</strong></td>
<td>Factors of Risk and Resilience</td>
</tr>
<tr>
<td><strong>Develop</strong></td>
<td>Targeted Treatment Strategies</td>
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</table>
What is Autism??
Autism – Core Features

Social Interaction
- Social Emotional Reciprocity
- Nonverbal Communication
- Relationships

Restricted & Repetitive Behaviors
- Stereotyped Repetitive
- Insistence on Sameness
- Restricted Interests
- Abnormal Sensory
Importance of Early Detection
EARLY SIGNS OF ASD
First Year Signs
### Signs

<table>
<thead>
<tr>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
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</thead>
<tbody>
<tr>
<td><strong>ATTENTION</strong>&lt;br&gt;Attention Disengagement&lt;br&gt;Reduced Attention to Faces or Objects</td>
<td><strong>SENSORY PROCESSING</strong>&lt;br&gt;Hypo or Hyper-sensitivity to sound or touch&lt;br&gt;Excessive Mouthing&lt;br&gt;Fine or Gross Motor Delays or Atypical Movements</td>
<td><strong>SOCIAL FEATURES</strong>&lt;br&gt;Diminished Response to Name&lt;br&gt;Eye Contact potentially reduced</td>
<td><strong>COMMUNICATION</strong>&lt;br&gt;Reduced affection, less interest in others (reaching, vocalizing, imitating)</td>
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Second Year Signs
Signs

1. **ATTENTION**
   - Reduced attention to whole object
   - Decreased complexity of play

2. **SENSORI-MOTOR**
   - Repetitive motor behaviors
   - Continued sensory sensitivities

3. **SOCIAL FEATURES**
   - Reduced interpersonal synchrony
   - Decreased Social Responsivity and Initiation

4. **COMMUNICATION**
   - Reduced gesture use
Third-Fifth Year Signs
**Signs**

1. **ATTENTION**
   - Over-focus on objects
   - Interest in unusual objects
   - Decreased complexity of play

2. **SENSORI-MOTOR**
   - Repetitive motor behaviors
   - Hand-flapping or Rocking

3. **SOCIAL FEATURES**
   - Peer interaction limited, restricted or unusual
   - Reduced interest in others

4. **COMMUNICATION**
   - Expressive Language
   - Reduced or absent
How Signs May Differ in Down Syndrome

**Timing**
- May show similar features but take more time to emerge.
- Continued delay of expected milestones during 3 - 5th year rather than 18 - 24 months.
- Absence of functional play into 3 - 5th year.
- More prolonged lack of communication.

**Quality**
- Rather than absence of skill, is there an absence of skill foundation?
- For example, rather than lack of or minimal expressive communication, is there a lack of *intent to communicate*?

**Coordination**
- A child with DS may use eye contact and vocalization with a delay between these to communicate, rather than at the same time.
- A child with ASD may use only one mode to communicate, or no modes of communication.
Phenotypic Overlap & Distinction

Intellectual Disability

Down Syndrome

Autism Spectrum
Phenotypic Overlap & Distinction

Down Syndrome

Intellectual Disability

RRBs in ID

Communication Delays

Atypical Motor

Impaired Attention

Cognitive Delays

Autism Spectrum
Parent Reported Developmental Trends

![Graphs showing developmental trends across different domains: Communication, Daily Living, Socialization, Motor, and Adaptive Composite. Each graph plots domain standard scores against chronological age (months). The graphs illustrate disparities between DS (Down Syndrome) and TD (Typically Developing) groups.](image-url)
Diagnostic challenges in Down syndrome

**Motor Delays**
- Late Independent Walking
- Poor Motor Planning
- Gesture use, play

**Medical Comorbidities**
- Surgeries
- Effect of Physical symptoms on behavior
- Full Medical history
- Vision & Hearing

**Info Processing & Attention Regulation**
- Impaired engagement:
  - Showing
  - Giving
  - Joint Attention
  - Eye contact
  - coordination

**Speech and Language**
- Low Intelligibility
  - Difficult to catch words & phrases
- Use of ASL
  - Can resemble motor mannerisms
VIDEO EXAMPLES – Diagnostic Overlap
ASD in DS – what do we really know?

- Epidemiological study estimates a ~17% prevalence rate of co-occurring ASD in DS
- Knowledge is limited due to several factors
- Prior work suggests milder profile of symptoms
- Need to consider developmental and phenotypic variation
- Medical conditions
Implications for Families
# ASD in Medical vs. Educational Settings

<table>
<thead>
<tr>
<th></th>
<th>DSM-5/ICD-11</th>
<th>Special Education Eligibility</th>
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<tbody>
<tr>
<td><strong>Diagnosis</strong></td>
<td>Medical</td>
<td>Educational</td>
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<tr>
<td><strong>Classification</strong></td>
<td>Neurodevelopmental Disorder</td>
<td>Educational Classification/Disability</td>
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<tr>
<td><strong>Purpose</strong></td>
<td>Insurance reimbursement, provide information about strengths/challenges, determine severity and level of impairment, inform treatment</td>
<td>Give access to federally funded services within the public education system, determine educational needs, inform educational decisions (e.g., special education programming, classroom placement)</td>
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<tr>
<td><strong>Criterion</strong></td>
<td>Not better explained by another factor, interferes with and/or reduces the quality of social, academic, or occupational functioning</td>
<td>Not better explained by another factor, adverse effects of the disability on the child’s educational performance that requires specialized instruction and/or related services</td>
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Educational Classification Patterns

Distribution of Students Ages 3-21 Served Under IDEA 2020-2021

- Autism
- Developmental Delay
- Intellectual Disability

(National Center for Education Statistics, 2022)
Evaluation Considerations for Dual Diagnosis of Autism in Down syndrome

- **Purpose of evaluation**
- **Provider**
  - Training, credentials
- **Access to services**
  - Educational supports vs. insurance reimbursement
  - State differences
- **Developmental level, co-occurring conditions**
  - Hearing, vision, medical complications
- **Cultural differences**
<table>
<thead>
<tr>
<th>Supports &amp; Services</th>
<th>School Based</th>
<th>Community Based</th>
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<tr>
<td></td>
<td>• IFSP, IEP</td>
<td>• Early intervention services (EI)</td>
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<td></td>
<td>• OT/PT/Speech</td>
<td>• Family training</td>
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<tr>
<td></td>
<td>• Behavior management program</td>
<td>• OT/PT/Speech</td>
</tr>
<tr>
<td></td>
<td>• Learning strategies/study skills support</td>
<td>• Service coordinator/case management services</td>
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<tr>
<td></td>
<td>• Social skills</td>
<td>• Psychological services</td>
</tr>
<tr>
<td></td>
<td>• Adaptive equipment/special communication systems</td>
<td>• Applied Behavior Analysis (ABA)</td>
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<tr>
<td></td>
<td></td>
<td>• Eligibility/requirements</td>
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<tr>
<td></td>
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<td>• Waitlists</td>
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EVIDENCE BASED SUPPORTS

General
North Carolina
Regional
SUPPORTING YOUR CHILD’S DEVELOPMENT VIDEO EXAMPLES
NATURALISTIC ENVIRONMENT AND EMBEDING SKILLS INTO ROUTINE

ESDM
SYNCHRONY
VIDEO EXAMPLE OF RESPONSIVITY
What to do?

• Monitor your child’s development.
• Watch her/his progress with early intervention services.
• Watch for skills to emerge later than expected.
• Keep routine medical check-ins for vision and hearing – difficulties in these areas can mimic features of ASD.

• Don’t put too much weight in unusual behaviors like flapping or looking closely at objects.
• Children with DS may show a developmentally appropriate behavior like flapping longer than neurotypical children.
• Encourage your child to give equal amounts of attention to people and toys – especially if you notice extra attention to one over the other.
Questions?

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