Autism is uncommon in 22q: the how and why of wrong diagnoses

Kathleen Angkustsiri, Khyati Brahmbhatt, Lesley Deprey, Beth Goodlin-Jones, Tony J. Simon

UC Davis MIND Institute, Sacramento, CA

Disclosures

• Co-investigator on clinical trials in autism and fragile X syndrome
  – Novartis
  – Roche
  – Seaside Pharmaceuticals
  – Forest Laboratories
22q and Autism Spectrum Disorders

- Autism Spectrum Disorders are behavioral diagnoses based on impairments in 3 domains
  - Social Interaction
  - Communication
  - Repetitive/Restricted Interests

- Children with 22q11.2DS have social and communication impairments, sometimes along with repetitive behaviors, but is it really autism?
  - What features are shared with ASD, and what features distinguish between the two?

Superficial similarities

- Many (20-50%) children *screen positive* for ASD symptoms or meet criteria based on the ADI-R questionnaire, which is only *one part* of the gold-standard assessment for ASD *diagnosis* (Antshel et al., 2007; Kates et al., 2007; Vorstman et al. 2006)

- To our knowledge, there are no published studies using the gold-standard for ASD diagnosis, which includes BOTH:
  - Autism Diagnostic Interview-Revised (ADI-R)
  - Autism Diagnostic Observation Schedule (ADOS)
Objective

- To determine the prevalence of ASD diagnoses in children with 22q11.2DS using best-practice standards:
  - Social Communication Questionnaire (SCQ) Questionnaire based on the ADI-R
  - Autism Diagnostic Observation Schedule Interactive assessment directly administered by trained personnel

Demographics

- 29 children with 22q11.2DS
  - 16 boys; 13 girls
  - Mean FSIQ: 74.6 ± 14.3
- ages 7-14, mean age: 10.7 ± 2 years
  - SCQ:
    - >15 “at-risk” for ASD
  - ADOS:
    - module 2: 1, module 3: 22, module 4: 5
ASD is uncommon in 22q based on best practice assessment

• Using only one assessment, 7-18% demonstrated concerns for ASD, but no child met strict diagnostic criteria for ASD using both SCQ and ADI-R

<table>
<thead>
<tr>
<th></th>
<th>positive</th>
<th>total</th>
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<tbody>
<tr>
<td>SCQ</td>
<td>2 (7%)</td>
<td>2 (7%)</td>
</tr>
<tr>
<td>ADOS</td>
<td>4 (15%) ASD</td>
<td>1 (3%) Autism</td>
</tr>
<tr>
<td>SCQ+ADOS</td>
<td>none</td>
<td>0</td>
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22q: Different patterns on ADOS

<table>
<thead>
<tr>
<th></th>
<th>Strengths</th>
<th>Weaknesses</th>
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<tbody>
<tr>
<td>Type A (+ADOS)</td>
<td>Social: Reciprocal Social Interaction</td>
<td>Communication: Conversation, Gestures, Insight</td>
</tr>
<tr>
<td>Type B (-ADOS)</td>
<td>Social: Reciprocal Social Interaction</td>
<td>Communication: Reporting</td>
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Implications

• Small sample, needs replication
• Appropriate treatments and interventions
  – Co-morbid conditions?
    • Anxiety in 4 of 5 children with +ADOS
  – Different causes leading to overlapping behaviors
    • Poor eye contact: anxiety?
    • Social interaction: poor emotional recognition?
      – Cognitive and linguistic difficulties
    • Repetitive and restrictive interests: cognitive control difficulties, response to stress?

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