Adrift in the Social Universe

Prof. Louise Gallagher
First descriptions
Simple Facts:

- Affects 1/150 children
- M>F
- High rates of intellectual disability
- High rates of epilepsy (~30%)
- 50% of children are non-verbal
- High economic, familial and personal costs
- 90-95% - causes are undetermined
Autism is distinguished by a pattern of symptoms rather than one single symptom. The main characteristics are impairments in social interaction, impairments in communication, restricted interests and repetitive behavior.
What is the Autistic Spectrum?

- Language impairment
- Cognitive impairment
- Social impairment
- Other behaviours

Autistic Disorder, Asperger's Disorder, Childhood Disintegrative Disorder, Rett's Disorder, Pervasive Developmental Disorder - Not Otherwise Specified
Early signs of autism

No big smiles or other warm, joyful expressions by six months or thereafter

No back-and-forth sharing of sounds, smiles or other facial expressions by nine months
No babbling by 12 months
No words by 16 months
No meaningful, two-word phrases (not including imitating or repeating) by 24 months

No back-and-forth gestures such as pointing, showing, reaching or waving by 12 months

Any loss of speech, babbling or social skills at any age
Best practice autism assessment

- Lifetime and family history
- Review of medical and educational records
- Behavioural observation
- Physical examination
- Behavioural assessment - standardised instruments, e.g. ADI, ADOS, DISCO
- Cognitive/adaptive assessment
- Review of DSM or ICD diagnostic criteria
- SLT
- OT
Comorbidities

- Influence of comorbidities - presentation, severity

Table 2 CBCL syndrome profiles in ASD and control groups

<table>
<thead>
<tr>
<th>CBCL/6–18 syndromes profile</th>
<th>ASD group</th>
<th>Control group</th>
<th>P-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anxious/depressed</td>
<td>63.33</td>
<td>64.54</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Withdrawn/depressed</td>
<td>63.99</td>
<td>54.27</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Somatic complaints</td>
<td>58.48</td>
<td>56.64</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Social problems</td>
<td>65.46</td>
<td>55.09</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Thought problems</td>
<td>57.39</td>
<td>55.54</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Attention problems</td>
<td>77.10</td>
<td>70.07</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Rule breaking behaviour</td>
<td>56.01</td>
<td>59.76</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Aggressive behaviour</td>
<td>56.82</td>
<td>62.51</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Internalising problems</td>
<td>63.46</td>
<td>59.34</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Externalising problems</td>
<td>54.51</td>
<td>61.46</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Total</td>
<td>64.82</td>
<td>62.31</td>
<td>&gt;0.05</td>
</tr>
</tbody>
</table>

CBCL, Child Behavior Checklist; ASD, autistic spectrum disorders.

Table 3 CBCL DSM-orientated scales

<table>
<thead>
<tr>
<th>DSM-orientated scales</th>
<th>ASD group</th>
<th>Control group</th>
<th>P-value</th>
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</thead>
<tbody>
<tr>
<td>Affective problems</td>
<td>60.43</td>
<td>61.57</td>
<td>&gt;0.05</td>
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<tr>
<td>Anxiety problems</td>
<td>66.61</td>
<td>61.33</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Somatic problems</td>
<td>57.01</td>
<td>53.22</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Attention deficit/attention problems</td>
<td>70.1</td>
<td>65</td>
<td>&lt;0.05</td>
</tr>
<tr>
<td>Oppositional defiant problems</td>
<td>55.07</td>
<td>58.19</td>
<td>&gt;0.05</td>
</tr>
<tr>
<td>Conduct problems</td>
<td>57.1</td>
<td>62.1</td>
<td>&gt;0.05</td>
</tr>
</tbody>
</table>

CBCL, Child Behavior Checklist; ASD, autistic spectrum disorders.

Skokauskas and Gallagher, 2011
Challenging behaviours

• Common, 10-15%
• Self-injury, aggression, destructive behaviour
• High risk of injury to self and carers
• Impact on academic and social engagement
• Poorer outcomes – residential placement
Diagnosis <12m highly unreliable, unstable <30m

Diagnostic complexities

Developmental nature of diagnosis
Differentiation from ID, mental health disorders
Poor diagnostic instruments

Multiple care givers
Poor history from early years
Comorbidity - e.g. ADHD, attachment disorders
Personality trait vs disorder

- Social communication deficits common in population
- 40% of cases in CAMHS - social communication deficits
- When is a diagnosis important?
- When and what type of autism specific treatments are important?
Managing autism

Behavioural interventions

Social Skills Training

Following Directions at School

Educational approaches

Medication for comorbidity
What causes this profound neurodevelopmental condition......?

- 90% of the time....We don’t know

- Known genetic causes 5-10%
- Medical causes 1-2%
- Unknown 90%
Autism Genetics
Abnormal connectivity

STRUCTURAL BRAIN ANATOMICAL DATA

STRUCTURAL BRAIN ABNORMALITIES

ACCELERATED EARLY BRAIN GROWTH

WHITE MATTER ENLARGEMENT

REDUCED INTEGRITY OF WHITE MATTER

MINICOLUMNAR ABNORMALITIES

COULD ALL COMPROMISE CONNECTIONS BETWEEN BRAIN REGIONS IN AUTISM

Slide thanks to Dr. Jane McGrath
Environmental Factors

- In utero exposures - Rubella, sodium valproate, thalidomide
- ?↑ Paternal age
- Epidemiological studies - season of birth phenomena, pesticides

<table>
<thead>
<tr>
<th>Ethnic descriptor</th>
<th>N</th>
<th>%</th>
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<tbody>
<tr>
<td>Irish</td>
<td>104</td>
<td>30.4%</td>
</tr>
<tr>
<td>African</td>
<td>54</td>
<td>15.79%</td>
</tr>
<tr>
<td>Asian</td>
<td>15</td>
<td>4.39%</td>
</tr>
<tr>
<td>American/Canadian</td>
<td>3</td>
<td>0.87%</td>
</tr>
<tr>
<td>South/ Central America</td>
<td>3</td>
<td>0.87%</td>
</tr>
<tr>
<td>Europe</td>
<td>13</td>
<td>3.80%</td>
</tr>
<tr>
<td>EU Accession states 2004/2007</td>
<td>13</td>
<td>3.80%</td>
</tr>
<tr>
<td>Unknown</td>
<td>137</td>
<td>40.0%</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>342</td>
<td>100%</td>
</tr>
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</table>

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Group A No. (%)</th>
<th>Group B No. (%)</th>
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</thead>
<tbody>
<tr>
<td>Autism</td>
<td>14 (35%)</td>
<td>13 (59%)</td>
</tr>
<tr>
<td>Autism Features</td>
<td>5 (12.5%)</td>
<td>0</td>
</tr>
<tr>
<td>PDD-NOS</td>
<td>9 (22.5%)</td>
<td>5 (22.7%)</td>
</tr>
<tr>
<td>ASD</td>
<td>7 (17.5%)</td>
<td>4 (18%)</td>
</tr>
<tr>
<td>Asperger’s</td>
<td>5 (12.5%)</td>
<td>0</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>40 (12.5%)</td>
<td>22</td>
</tr>
</tbody>
</table>
What are the important research questions?

• Is there a better way to diagnosis autism - e.g. biomarkers?
• Is there a better way to identify people at risk?
• Can you prevent autism by intervening early?
• What are the best ways of intervening in autism?
Summary

- Profound neurodevelopmental disorder
- Many traits of autism seen widely in general population
- High personal and familial costs
- Need better diagnosis and treatment
- Research beginning to yield some clues about causes that will help towards better diagnosis and treatments
Disorder vs Diversity?

Famous People with Autism