FETAL ALCOHOL SPECTRUM DISORDERS (FASD) IN AUSTRALIA:

PROVISIONAL FINDINGS OF NATIONAL CASE SURVEILLANCE

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What do we know?

FASD epidemiology in Australia is poorly known

APSU study of Fetal Alcohol Syndrome (FAS) / Partial FAS (PFAS) 2001-2004 (Elliott et al)
- FAS Incidence per annum 0.6 per 100,000 children aged <15 years
- Considered a significant underestimate

Lililwan Project, Fitzroy Valley (Fitzpatrick et al)
- FASD prevalence ~20% - population based, active case ascertainment study
- High risk community

FASD is not a rare disease
- US and Canadian data suggests prevalence 1-2% of the population (May 2014 2-5%)
What this study adds

Aim was to conduct the first national case surveillance study to identify cases across the entire FASD spectrum.
Methods

Prospective active national case-finding using monthly reporting by paediatricians to APSU

December 2014 – May 2015 (ongoing)

Children under 15

Using Australian FASD diagnostic criteria (2013)
FASD diagnostic criteria

4 key aspects
Prenatal alcohol exposure
- Confirmed exposure

CNS impairment (neurodevelopment)
- Structural – microcephaly (HC <3rd %ile) and/or
- Functional – 3 domains significantly impaired <3rd %ile
  (e.g. Cognition, Communication, ADHD, Adaptive Behaviour)

Facial features
- Palpebral fissure length (PFL) - <3rd %ile
- Smooth philtrum – Rank 4 or 5 on UW Lip-Philtrum guides
- Thin upper lip - Rank 4 or 5 on UW Lip-Philtrum guides

Growth deficit
- Height or weight <10th %ile
Diagnostic categories

FASD

FAS
- PAE +/-
- CNS impairment
- 3 Facial features
- Growth deficit

PFAS
- PAE
- CNS impairment
- 2 Facial features

ND-AE
- PAE
- CNS impairment

FAS = Fetal Alcohol Syndrome
PFAS = Partial Fetal Alcohol Syndrome
ND-AE = Neurodevelopmental Disorder – Alcohol Exposed
PAE = prenatal alcohol exposure
Diagnostic facial features

- Small eyes (distance from A to B)
- Smooth philtrum
- Thin upper lip
Facial features of FASD

Not associated with prenatal alcohol exposure, below diagnostic threshold for FASD

Frontal view ¾ view
Notifications and confirmed reports

- Total notifications: n=86
- Case reports: n=58
  - Duplicates: n=9
  - Criteria not met: n=8
- Cases confirmed: n=41
### Results - confirmed cases

<table>
<thead>
<tr>
<th>Condition</th>
<th>Cases</th>
<th>Percentage</th>
<th>Median Age</th>
</tr>
</thead>
<tbody>
<tr>
<td>FAS</td>
<td>12</td>
<td>29%</td>
<td>5.0y</td>
</tr>
<tr>
<td>PFAS</td>
<td>20</td>
<td>49%</td>
<td>5.6y</td>
</tr>
<tr>
<td>ND-AE</td>
<td>9</td>
<td>22%</td>
<td>9.5y</td>
</tr>
</tbody>
</table>

>50% from the 3 specialised FASD clinics
## Demographics

<table>
<thead>
<tr>
<th>Category</th>
<th>FASD 2014-16</th>
<th>FAS 2001-04</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at diagnosis (median)</td>
<td>6.3y</td>
<td>3y</td>
</tr>
<tr>
<td>Male:female ratio</td>
<td>2:1</td>
<td>1:1</td>
</tr>
<tr>
<td>Child protection services (current/past)</td>
<td>73.2%</td>
<td>67.4%</td>
</tr>
<tr>
<td>Out of home care (foster/adoptive)</td>
<td>61.0%</td>
<td>38.0%</td>
</tr>
<tr>
<td>Biological parents’ care</td>
<td>22.0%</td>
<td>40.2%</td>
</tr>
<tr>
<td>Grandparents’ care</td>
<td>12.2%</td>
<td>20.7%</td>
</tr>
<tr>
<td>Sibling with FASD</td>
<td>19.5%</td>
<td>51.0%</td>
</tr>
<tr>
<td>Indigenous</td>
<td>41.5%</td>
<td>65.0%</td>
</tr>
</tbody>
</table>
Who first suspected FASD?

- Paed
- Other
- Parent/caregiver
- Teacher
Who made diagnosis?

- Paed + multi D
- Paed + using reports
- Paed solo
Prenatal alcohol exposure

Standardised approach to asking about alcohol allows for assessment of exposure risk

- 12 cases (29%) a standardised tool was used (i.e. AUDIT-C)

High risk exposure

- Nearly 50% >7 std drinks per week
- Nearly 40% 5 or more std drinks on a single occasion
- Risk unknown in ~40%
Central nervous system - structural

48.8% had microcephaly
(47.9% FAS 2001-04 study)
## CNS Impairment - functional

<table>
<thead>
<tr>
<th>Domain</th>
<th>Impairment (%)</th>
<th>Standardised testing (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>ADHD</td>
<td>48.8</td>
<td>29.3</td>
</tr>
<tr>
<td>Cognition</td>
<td>46.3</td>
<td>61.0</td>
</tr>
<tr>
<td>Communication</td>
<td>41.5</td>
<td>51.2</td>
</tr>
<tr>
<td>Adaptive Function</td>
<td>36.6</td>
<td>31.7</td>
</tr>
<tr>
<td>Exec Function</td>
<td>12.2</td>
<td>24.4</td>
</tr>
<tr>
<td>Memory</td>
<td>9.8</td>
<td>14.6</td>
</tr>
<tr>
<td>Global dev delay &lt;5yo</td>
<td>39</td>
<td></td>
</tr>
</tbody>
</table>
Facial features

80.5% had a *smooth philtrum*
65.9% had a *thin upper lip*
61.0% had *short palpebral fissure length*
Facial features - assessment

<table>
<thead>
<tr>
<th>Assessment method (can be multiple)</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Direct PFL measurement ruler</td>
<td>56.1</td>
</tr>
<tr>
<td>Lip-philtrum guide</td>
<td>46.3</td>
</tr>
<tr>
<td>Facial photo analysis</td>
<td>56.1</td>
</tr>
<tr>
<td>Visual/gestalt</td>
<td>78</td>
</tr>
</tbody>
</table>
Genetics

70.7% (n=29) had chromosomal microarray testing

19 normal

4 variants - *not clinically significant*
Other drug exposures (all cases)

<table>
<thead>
<tr>
<th>Drug</th>
<th>FASD 2014-16 (%)</th>
<th>FAS 2001-04 (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cigarettes</td>
<td>43.9</td>
<td>67.4</td>
</tr>
<tr>
<td>Marijuana</td>
<td>31.7</td>
<td>25.0</td>
</tr>
<tr>
<td>Amphetamines</td>
<td>17.1</td>
<td>4.4</td>
</tr>
<tr>
<td>Heroin</td>
<td>12.2</td>
<td>4.3</td>
</tr>
<tr>
<td>Cocaine</td>
<td>4.9</td>
<td>3.3</td>
</tr>
<tr>
<td>Phenytoin/Valproate</td>
<td>4.8</td>
<td></td>
</tr>
<tr>
<td>Methodone, diazepam</td>
<td>2.4</td>
<td>6.6</td>
</tr>
<tr>
<td>Opioids (?oral/IVDU)</td>
<td>2.4</td>
<td></td>
</tr>
</tbody>
</table>
What are the data telling us?

Paediatricians are diagnosing across the FASD spectrum
  But underdiagnosing FASD without physical features
  Lack of awareness that FASD spectrum includes children without physical features
What are the data telling us?

Paediatricians are not using:

- Standardised tools to assess *prenatal alcohol exposure*
- Direct measurement or photographic analysis to determine palpebral fissure length
  
  *The most objective diagnostic measure of facial feature, consistent across racial groups*

- Standardised or validated psychometric assessment tools to determine severity of CNS functional impairment
Strengths

- Detailed demographic and diagnostic data for each case
- Provides critical information re diagnostic patterns in Australia
- Increases awareness of FASD among paediatricians
- Allows comparison with previous FAS study
- Will enable incidence estimation
Limitations

Many case reports pending (from existing notifications)
Under-reporting of cases of ND-AE
Limited use of standardised assessment methods may affect classification
  Prenatal alcohol exposure
  CNS/neurodevelopmental impairment
  Facial features
Take home messages

Need for clinician education about diagnostic criteria

*National diagnostic tool* will standardise approach and allow for international comparison

Importance of *asking about alcohol use in pregnancy*, including before pregnancy awareness, particularly in children presenting with neurodevelopmental disorders

Informs planning diagnostic, support and disability services

Need for high-index of suspicion children in high-risk groups (e.g. out of home care)

*Children and adolescents with FASD may be hidden in all of our practices*

Current diagnosis may be ADHD or ASD......*keep thinking and reporting FASD*
QUESTIONS

Thank you