Let’s Talk Into the future: Diagnostics

Harmonizing FASD Diagnosis Around the Globe

Elizabeth Elliott, Sydney; Christine Loock, Vancouver
World Health Organization Global Alcohol Summit

SDG (Target 2, 10% rdn)
Global Action Plan NCDs
FASD morbidity
FASD prevalence
ICD 11, beyond?
Take home messages:

- PAE is common, harmful
- Harms unpredictable, variable
  - PAE, genetics, environment
- FASD is an acquired brain injury
disability, lifelong
- Diagnosis makes a difference
clinical benefits, optimal
treatment, financial support, expectations
- No diagnostic test, clinical
  ‘syndrome’, overlap, comorbidity
- No single internationally
  accepted diagnostic criteria
In your (WHO) region, which FASD diagnostic guidelines are followed?

1. Institute of Medicine (Stratton et al, 1996)
4. Australian Guidelines (Bower & Elliott, 2016) (3%)
5. US - Updated Clinical Guidelines (Hoyme et al. 2016)
7. DSM-5 (American Psychiatric Association)
8. Combination of above
9. I don’t know.

https://www.who.int/about/regions/en/
Q 15. In your region which FASD diagnostic guidelines used?

257 (51%) response; 84% Americas, 72% Canada

- IOM: 4%
- UW: 5%
- Canadian: 51%
- Australian: 3%
- US updated: 4%
- German: 1%
- DSM-5: 4%
- Combination: 11%
- Don’t know: 20%
Can we agree globally on a single (or harmonised) diagnostic system?

- Yes 53%
- No 16%
- Uncertain 31%
Diagnosis of FASD: common criteria and data, but differences in how defined or combined

- Prenatal Alcohol Exposure (PAE)
- Neurodevelopmental impairment
  - Severe, multiple domains
- dysmorphic facial features +/-
  - Narrow palpebral fissure
  - Flat philtrum
  - Thin upper lip
- +/- Growth
- +/- other birth defects
- Exclude: genetic, other teratogens
- Document: ACES, PTSD, other comorbidities
Commonwealth* Approach to Diagnosis

Prenatal alcohol exposure (PAE)

- Confirmed absent
- Confirmed
- Unknown

Neurodevelopmental criteria Not Met* (≤3 domains severely impaired)

- < 3 Sentinel Facial Features
  - No FASD diagnosis
  - Follow-up, therapy and support as indicated

Neurodevelopmental criteria Met* (1 or more domains severely impaired)

- 3 Sentinel Facial Features
  - FASD
  - FASD diagnosis

Neurodevelopmental criteria Met* (3 or more domains severely impaired)

- 3 Sentinel Facial Features
  - No FASD diagnosis

Neurodevelopmental criteria Not Met* (≤3 domains severely impaired)

- < 3 Sentinel Facial Features
  - No FASD diagnosis

*Commonwealth ‘ACANZS’ Collaborative: Australia, Canada, New Zealand, Scotland
Collaborative Initiative on FASD (CIFASD)

FIGURE 1
FASD diagnostic algorithm. See text for complete discussion. A positive dysmorphology facial evaluation requires 2 of the 3 cardinal facial features of FASD (short palpebral fissures, smooth philtrum, and a vermillion border of the upper lip). Cutoffs for neuropsychological testing are -1.5 SD. Cutoffs for stature, weight, and head circumference are at the 10th percentile.

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## Comparing FASD Diagnostic Systems

Examples of the interface with other diagnostic systems

<table>
<thead>
<tr>
<th>System</th>
<th>FASD with Sentinel Facial Features</th>
<th>FASD without Sentinel Facial Features</th>
<th>At Risk for Neurodevelopmental Disorder and FASD, Associated with Prenatal Alcohol Exposure</th>
</tr>
</thead>
<tbody>
<tr>
<td>2005 Canadian Diagnostic Guidelines</td>
<td>FAS</td>
<td>pFAS ARND</td>
<td></td>
</tr>
<tr>
<td>Institute of Medicine (IOM)</td>
<td>FAS pFAS</td>
<td>ARND</td>
<td></td>
</tr>
<tr>
<td>4-Digit Diagnostic Code (4DDC)</td>
<td>Growth 2, 3, or 4, Face 4, Brain 3 or 4, PAE 2, 3 or 4, FAS</td>
<td>Face 1, 2 or 3 Brain 3 or 4 PAE 3 or 4 ~SE-AE or Sentinel Physical Findings SE-AE</td>
<td>Face 1, 2, 3 or 4 Brain 2 (or untestable at time of assessment) PAE 2 (For Face 4 ~NB-AE), 3 or 4</td>
</tr>
<tr>
<td>CIFASD</td>
<td>FAS with/without Confirmed Maternal Alcohol Exposure</td>
<td>Partial FAS with/without Confirmed Maternal Alcohol Exposure</td>
<td>ARND</td>
</tr>
<tr>
<td>DSM-5</td>
<td>315.8 Neurodevelopmental Disorder, associated with Prenatal Alcohol Exposure Neurobehavioral Disorder, associated with Prenatal Alcohol Exposure (Appendix 3)</td>
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<td></td>
</tr>
<tr>
<td>ICD-10</td>
<td>Q86.0*</td>
<td>Q86.8** Q86.99***</td>
<td>Q86.8** Q86.99***</td>
</tr>
</tbody>
</table>

Guidelines are not laws!

All more alike than different.
# Simplified Common Terminology

<table>
<thead>
<tr>
<th>DIAGNOSIS</th>
<th>DESIGNATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>FASD with Sentinel Facial Findings</td>
<td>FASD without Sentinel Facial Findings</td>
</tr>
<tr>
<td><strong>PAE</strong></td>
<td></td>
</tr>
<tr>
<td>Not required</td>
<td>Confirmed</td>
</tr>
<tr>
<td><strong>FACE</strong></td>
<td></td>
</tr>
<tr>
<td>3 Facial Features</td>
<td>None required</td>
</tr>
<tr>
<td><strong>BRAIN</strong></td>
<td></td>
</tr>
<tr>
<td>3 domains of impairment (or microcephaly for infants)</td>
<td>3 domains of impairment</td>
</tr>
</tbody>
</table>

*CIFASD DX

*ad hoc Commonwealth ‘ACANZ’ Collaborative: Australia, Canada, New Zealand, Scotland…

ELLIOTT & LOOCK 2019
Shared common domains of brain function

1. Neuroanatomy /Neurophysiology
2. Motor Skills
3. Cognition
4. Language
5. Academic
6. Memory
7. Executive Functioning
8. Attention
9. Affect (Mood Regulation)
10. Adaptive

CIFASD: Significant and Prevalent (>1.5 SD) in ~2 domains
Commonwealth: Severe and Pervasive ( > 2 SD 3 domains)

ELLIOTT & LOOCK 2019
DSM-5
Three “Super” Domains

1. Neurocognitive
2. Self-Regulation
   e.g. attention, mood
3. Adaptive
4. Onset in childhood
5. Prenatal alcohol exposure (PAE)
6. IF unknown, add physical biomarkers
   – Facial features
   – Growth parameters (Ht, Wt, Head Size)

Elliott & Loock 2019
How often is FASD associated with historical or continued adverse Social Determinants of Health?

- Often: 64%
- Sometimes: 18%
- Never: 0%
Encircling Our FASD Diagnostic Systems: Medicine Wheel

“Two Eyed Seeing”
Why make a diagnosis? Real People, Real Lives, Real Futures!

Labels are for jars cans, and bottles.

Diagnoses are blueprints for intervention & support.

ELLIOTT & LOOCK 2019
Why make a diagnosis?

DISEASE (ICD)
• Prognosis
• Prevention
• Interventions
• Supports

FUNCTION (ICF)
• Activities
• Participation
• Resiliency & Strengths
• Meaningful Inclusion
Real People, Real Lives, Real Futures!

Balancing Sensitivity & Specificity

Beneficence & Non-Maleficence

ELLIOTT & LOOCK 2019
Where do we want to be in 20 years time?

- Universal diagnostic criteria/guideline that is valid, inclusive and allows comparison internationally
- How do we get there?
  - Action: Accurate recording of patient data – individual clinician
  - Agreement on common data set - researcher
  - International summit to develop an internationally agreed standard - clinicians and researchers