Facial dysmorphism across the fetal alcohol spectrum

Peter Hammond

Molecular Medicine Unit
UCL Institute of Child Health, London, UK
New objective techniques to assess facial dysmorphism in FASD

- FAS/PFAS and those without classic features
- young infants in 1st year of life
- disorders with overlapping facial features

RESULTS AND RELEVANCE HERE
TECHNICAL ASPECTS ELSEWHERE
Suttie et al: Pediatrics, on line Feb 26th 2013
### FASD Diagnostic Criteria

<table>
<thead>
<tr>
<th>FASD</th>
<th>FAS</th>
<th>PFAS</th>
<th>Heavy Exposure (HE)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Alcohol Exposure</strong></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Prenatal exposure of 1 oz AA/day OR ≥ 4 binges of at least 2 oz AA</td>
<td>✓</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Face</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Palpebral fissure width ≤10th %tile</td>
<td>≥ 2 of 3</td>
<td>≥ 2 of 3</td>
<td></td>
</tr>
<tr>
<td>Thin upper lip (4/5 on Astley scale)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Smooth philtrum (4/5 on Astley scale)</td>
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<tr>
<td>Growth</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Height/weight ≤10th %tile</td>
<td>✓</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Brain growth</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Head circumference ≤10th %tile</td>
<td>≥ 1 of 2</td>
<td>≥ 1 of 4</td>
<td></td>
</tr>
<tr>
<td>Structural brain anomaly</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Behaviour/Cognition</td>
<td></td>
<td></td>
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<tr>
<td>Behavioural or cognitive abnormalities</td>
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</tbody>
</table>
3D facial photographs
FACIAL DYSMORPHISM ACROSS THE FETAL ALCOHOL SPECTRUM

192 Cape Coloured Children South Africa

with Sandy Jacobsen & Joseph Jacobsen

FAS = 22 ; PFAS = 26 ; HC = 69;
non-syndromic HE = 75
FACES
COMPARISON OF FACE OF CHILD WITH FAS TO AVERAGE OF MATCHED CONTROLS

Front view
- smooth upper lip
- inner eye folds

Profile view
- short nose
- flat nasal bridge
- mid-facial flatness
- backward rotation of lower jaw

FACE SIGNATURE

FACE SHAPE
NORMALISED
AGAINST
CONTROLS

Red- 2 S.D. deflated
Blue- 2 S.D. inflated
Green- coincident
FACE SIGNATURES: CHIDREN WITH FASD
IDENTIFYING UPPER LIP SMOOTHNESS

Red - deflated
Blue - inflated
Green - coincident

±1.0 SD
±1.5 SD
2.0 SD
SIGNATURE GRAPH
clusters faces with similar shape differences from matched controls
SIGNATURE GRAPH: FAS, PFAS & HE
SIGNATURE GRAPH: FAS, PFAS & HE
SIGNATURE GRAPH: FAS, PFAS & HE

**HE2**
facial differences more control like than FAS/PFAS

**HE1**
facial differences more FAS/PFAS like than control
### SIGNATURE GRAPH: FAS, PFAS & HE

<table>
<thead>
<tr>
<th></th>
<th>HE2</th>
<th>HC</th>
<th>HE1 vs HE2 (t)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>WISC IQ</strong></td>
<td>73.3</td>
<td>73.3</td>
<td>-1.80†</td>
</tr>
<tr>
<td><strong>CVLT-C 1</strong></td>
<td>47.3</td>
<td>45.8</td>
<td>-2.02*</td>
</tr>
<tr>
<td><strong>CVLT-C 2</strong></td>
<td>93.7</td>
<td>93.2</td>
<td>-1.89†</td>
</tr>
</tbody>
</table>

† p < 0.08
* p < 0.05

<table>
<thead>
<tr>
<th></th>
<th>HE1</th>
<th>FAS</th>
<th>PFAS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>WISC IQ</strong></td>
<td>65.5</td>
<td>65.4</td>
<td>63.0</td>
</tr>
<tr>
<td><strong>CVLT-C 1</strong></td>
<td>40.0</td>
<td>42.7</td>
<td>41.5</td>
</tr>
<tr>
<td><strong>CVLT-C 2</strong></td>
<td>84.3</td>
<td>88.5</td>
<td>88.3</td>
</tr>
<tr>
<td>Differential Diagnoses for FASD</td>
<td></td>
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</tbody>
</table>
|---------------------------------
| Aarskog                        |
| Cornelia de Lange (n=50)       |
| Dubowitz                      |
| Fetal hydantoin               |
| Fetal valproate               |
| Maternal PKU fetal effects    |
| Noonan/CFC/Costello (50)      |
| Toluene embryopathy           |
| Williams (65)                 |

SCREENING FOR CDLS FACIAL FEATURES

- CONTROLS
- CdLS

Graph showing data points for CONTROLS and CdLS on a scatter plot.
SCREENING FOR CDLS FACIAL FEATURES

- CdLS

HC-CDLS

-2 -1 0 1 2
DIFFERENTIAL DIAGNOSES FOR FASD
DIFFERENTIAL DIAGNOSES FOR FASD

- EXPOSED-FAS/PFAS
- EXPOSED-HE
- CDLS
- RAS
- WS

HC-WS
HC-RAS
HC-CDLS
DIFFERENTIAL DIAGNOSES FOR FASD
DIFFERENTIAL DIAGNOSES FOR FASD

<table>
<thead>
<tr>
<th></th>
<th>FASD (n=60)</th>
<th>SYND (n=165)</th>
</tr>
</thead>
<tbody>
<tr>
<td>FASD</td>
<td>96%</td>
<td>4%</td>
</tr>
<tr>
<td>SYND</td>
<td>2%</td>
<td>98%</td>
</tr>
</tbody>
</table>

- EXPOSED-FAS/PFAS
- EXPOSED-HE
- CDLS
- RAS
- WS
IN SUMMARY

- CAMERA ISSUES – 3D cameras expensive & rare resource; BUT 3D webcams and smaller/cheaper 3D cameras are imminent
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Hein Odendaal

**WAYNE STATE**
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Joseph Jacobsen