FASD in motion: 
Case surveillance and diagnosis in Australia in the 21st century

Dr Marcel Zimmet on behalf of APSU FASD Investigators
Stretch break
Learning objectives

- To map changes in FASD diagnosis over time, in relation to national initiatives and changes in diagnostic criteria
- To inform discussion about national approaches to FASD diagnosis and data collection
- To generate debate regarding opportunities and limitations of specialised FASD diagnostic clinics
- To identify clinician-related barriers to FASD diagnosis
There has been an increase in the diagnosis of FASD in Australia over the last decade coinciding with:
- development of specialist FASD clinics
- national diagnostic guidelines
- broadening of the FASD spectrum to include children with no sentinel facial features
The increased diagnostic reporting has been primarily driven by 7 FASD expert paediatricians, suggesting significant limitations in diagnosis and/or reporting of FASD by Australian paediatricians in general.

This may reflect ongoing concerns and misconceptions about FASD diagnosis.
Improving awareness of diagnostic rationale, methodology and case surveillance remains a priority, in conjunction with ongoing public health prevention measures.
The first national data set for the entire FASD spectrum aimed to capture emerging diagnostic and demographic patterns, in order to inform:

- Clinical practice
- Education
- Service delivery
- Public health initiatives
- Ongoing surveillance
Data captured

- Incidence rates (new cases per year)
- Who is diagnosing FASD and where
- Demographics of children diagnosed with FASD
- Types of FASD
- Other neurodevelopmental and clinical features
- Other factors – e.g. other prenatal drug exposure
- Health service provision
What
• Prospective national case-finding
• Active surveillance

When
• Jan 2015 – Dec 2017

Who
• Children and adolescents < 15 y o

How
• Paediatrician completes online case notification and then report

Case definition
• FASD +/- 3SFF (Australian guidelines)

Comparison
• To 2001-4 study
Verified cases: Jan 2015-Dec 2017

Notifications
N = 433

Case reports received
N = 392

Verified Cases
N = 280

Excluded n = 112
Diagnostic clinical criteria not met 16
Duplicates 41
>15 years old 32
Diagnosis outside study period 19
Incomplete records 4
FASD in motion
FASD surveillance in Australia in the 21st century

2001–2004
FAS / PFAS
Criteria: IOM 1996

FASD (+/- 3FF)
Criteria: Australian/Canadian 2015
2015–2017
Diagnostic trends over time
Incidence * per 100,000 children <15 yo (new cases)

2001-2004
Incidence* 0.58
Median Age 3 years

2015-2017
Incidence* 2.04
Median Age 8.5 years
Diagnostic trends over time: Cases

2001-2004
92 cases over 4 years
23 Cases per year

- FAS n = 25 (27%)
- Suspected FAS n = 2 (2%)
- PFAS n = 65 (71%)

2015-2017
280 Cases over 3 years
93 Cases per year

- FASD + 3 SFF n = 61 (22%)
- FASD < 3 SFF n = 219 (78%)
Diagnostic trends over time
Sentinel Facial Features (SFF)

2001-2004
- 3 SFF (FAS) 9 cases per year
- Ratio 3SFF:<3SFF 1:2.4
- 3 SFF (Total) 14 cases per year
- Ratio 3SFF:<3SFF 1:1.4

2015-2017
- 3 SFF 20 cases per year
- Ratio 3SFF:<3SFF 1:3.6
Profile of FASD diagnosis in Australia 2015-2017
A child diagnosed with FASD in 2014-7 in Australia is most likely:

- Male (63%)
- 8.5 years old
- In foster/adoptive care (54%)
- Birth parent (20%)
- Grandparent (15%)

To be diagnosed in:
- A specialist FASD clinic
- 3 states/territories (WA, QLD, NSW)

To have:
- <3 sentinel facial features
- No other prenatal nicotine/other drug exposure

To be involved with child protection services (past/current 74%)

To have:
- Middle range SES (26-75th percentile)
- Indigenous (59%)
Specialist FASD clinicians more likely to diagnose FASD <3 SFF vs +3SFF

- 82% vs 57%  $p < .001$

Clinical diagnostic patterns
Large majority of cases diagnosed/reported by FASD champions in multidisc. teams

- 84% by 7 paediatricians
- 49% by 1 paediatrician

Small number of paediatricians/clinicians diagnosing/reporting FASD

- FASD (n=34) vs Total APSU (n=1500)
  - reporting paediatricians/clinicians
    - Paediatricians ~95%:
      - General 40-51%, Devel. 10-15%, Neonat. 10-20%
    - Clin. Geneticists 2-5%
    - Child Psychiatry 1%

Who is diagnosing and reporting FASD in Australia?
Interpretation
Paediatricians diagnosing FASD in Australia

Positive trends

Paediatricians are diagnosing and reporting FASD
Increasing rates of FASD diagnosis
Are increased rates of drinking in pregnancy a factor in increased FASD diagnosis in Australia?
Data on pre-pregnancy awareness drinking

Figure 8.10: Pregnant women who drank more, less or the same amount of alcohol compared with when they were not pregnant, pregnant women aged 14–49, 2007 to 2013 (per cent)

Note: Base is only pregnant women or women pregnant and breastfeeding at the same time.
Source: Online Table 8.10.
Paediatricians diagnosing FASD in Australia
Positive trends

Better understanding of FASD spectrum, incl. children without physical features
Establishment of specialised FASD diagnostic clinics
Use and availability national diagnostic guidelines
Reporting mechanism in place, national registry established
Paediatricians diagnosing FASD in Australia
A work in progress

The main drive comes from a small group of FASD informed and motivated paediatricians/clinics

More paediatricians could be diagnosing/reporting FASD

There is significant variation across states, not proportional to state populations

There may be ongoing concerns and misconceptions about FASD diagnosis among paediatricians
Priorities

- Improve clinicians’ skills & confidence in discussing / assessing drinking in pregnancy and FASD
- Increase awareness of FASD diagnostic rationale, methodology and surveillance
- Enhance awareness of drinking in pregnancy risks and rates across society
- Promote consistent diagnostic approach using Australian Guide
- Continue public health prevention measures
- Facilitate epidemiological monitoring, research, education, and advocacy
- Enhance awareness of drinking in pregnancy risks and rates across society
Thank you
Discussion
EXTRAS
## Assessment rates & methods

<table>
<thead>
<tr>
<th>Category</th>
<th>Details</th>
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</thead>
<tbody>
<tr>
<td>Prenatal alcohol exposure</td>
<td>16% used standardised tool (typically AUDIT-C)</td>
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<td>- of those, 61% had high-risk exposure (score &gt;5)</td>
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<tr>
<td>Facial features</td>
<td><strong>Photo analysis software used in 37%</strong></td>
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<td><strong>Lip-philtrum guides 80%</strong></td>
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<td></td>
<td><strong>Direct PFL measurement 73%</strong> (Stromland charts used 60%)</td>
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<td>37% Short PFL, 54% Smooth philtrum, 45% Thin upper lip</td>
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<tr>
<td>Neurodevelopment</td>
<td><strong>Domain assessment rates varied 58-94%</strong></td>
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<td><strong>Standardised testing rates varied 48-90% between domains</strong></td>
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<td>(e.g. Motor Skills low, Cognition high)</td>
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<td>Growth impairment</td>
<td>Reported ‘unknown’ in 56%</td>
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<td>Genetics</td>
<td>Microarray 30%, karyotype in 8%, the vast majority normal.</td>
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Figure 6: Proportion of females exceeding the lifetime risk guidelines\(^a\), by age, 2007 to 2016 (per cent)

- Females under 40 years
  - 12-17
  - 18-24
  - 25-29
  - 30-39
- Females 40 years or older
  - 40-49
  - 50-59
  - 60-69
  - 70+

\(^a\) on average consumed more than 2 standard drinks per day

Source: NDSHS 2016 preliminary findings (Data tables).
NUMBER OF NEURODEVELOPMENT DOMAINS ASSESSED

FASD < 3, 33.8
FASD + 3, 27.9

% cases
<table>
<thead>
<tr>
<th>Neurodevelopmental Domains</th>
<th>Brain structure/neurology</th>
<th>Motor skills</th>
<th>Cognition</th>
<th>Language</th>
<th>Academic Achievement</th>
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<td>Memory</td>
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<td>Executive function, Impulse control Hyperactivity</td>
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<td>Affect Regulation</td>
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<td>Microcephaly 23% Structural 7%</td>
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<td>73%</td>
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<tr>
<td>Memory</td>
<td>Attention 79%</td>
<td>Executive function, Impulse control Hyperactivity 81%</td>
<td>Affect Regulation</td>
<td>Adaptive behaviour, Social skills, Social comm. 73%</td>
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Prevalence studies: community specific

FASD 20%  
*Fitzpatrick et al*

Banksia Hill: Youth in detention (2018)  
FASD 36%  
*Bower et al*

Incidence studies: national case surveillance

1. APSU FAS/PFAS (2001-2004) *Elliott et al*

2. APSU FASD (2015-2018)
Diagnostic patterns: State/territory variation

Possible reasons:
- Drinking patterns different in different regions
- Real differences in prevalence
- Under vs over diagnoses
- Access to diagnostic services difference
- Interests and biases of paediatricians
- Other clinicians diagnosing, and not reporting to APSU (e.g. geneticists, psychiatrists)