

Himmelfarb J et al. Kidney International 2002; 62: 1524

# FASD Overview – Why and How



# Chat Questions

- How many children in the last 6 months, did you think may have FASD
  - Possible X
- How many children in the last 12 months have you diagnosed with FASD
  - Diagnosed X
- What are your current concerns / barriers to considering an FASD diagnosis
  - Comments...



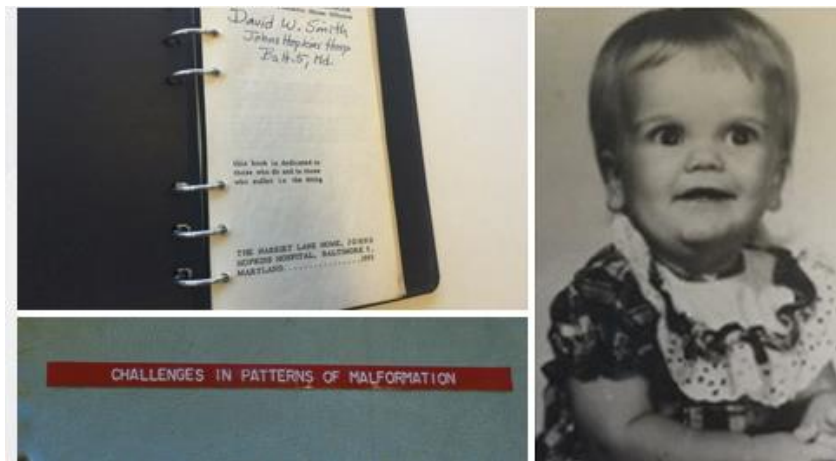
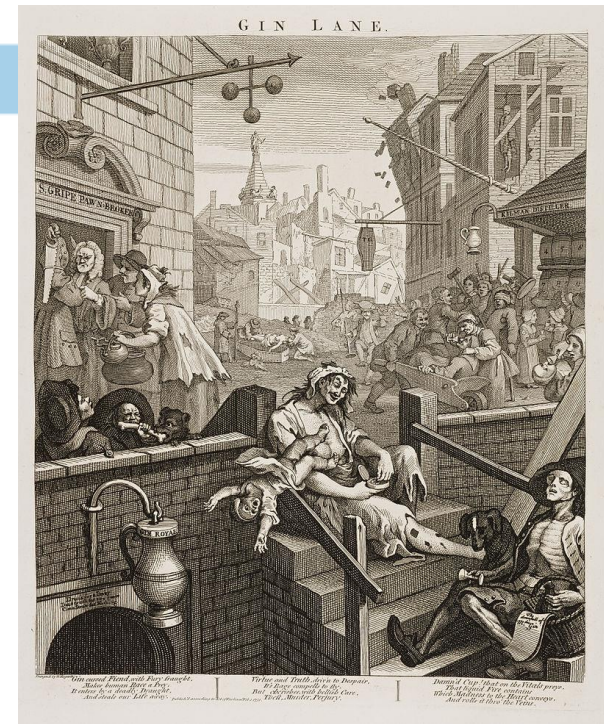


- **FASD is**
  - Common, chronic & expensive
  - In all our caseloads
  - Preventable & responsive to intervention
  - Return on investment is high
  - FASD is core business for all clinicians



## History of FASD –FASD is not new

- 1725 letter to UK parliament - *Gin babies* “too often the cause of weak, feeble and distempered children”
- 1899 higher Still birth rates in alcoholic prisoners
- 1968 Lemoine published his thesis on 127 cases of abnormalities observed in children of alcoholic mothers
- 1973 Smith and Jones coined the term ‘Fetal Alcohol Syndrome’





# Challenge your beliefs





## Excuses for not considering FASD as part of Dx

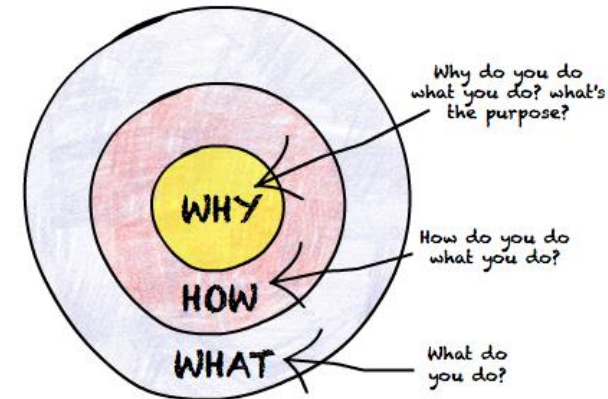
- Guilt it places on mother
- Stigmatising child, mother or family
- Harms the relationship between mum and child
- Labelling a child
- What's the point - doesn't change supports / funding
- How do you know it is the alcohol and not .....other drugs, early trauma, prematurity, genetic etc
- Inconsistencies in Diagnostic Frameworks
- Don't have the funding to do these assessments





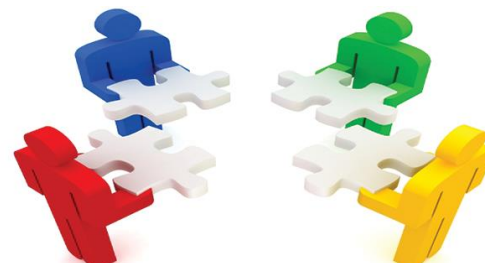
## Why the right diagnosis makes a difference...

- Guides intervention
- Informs prognosis
- Prevention
  - Secondary disabilities
  - Recurrence in siblings
  - Increased community awareness
- Accurate prevalence data
- Access to services - ECEI / NDIS, Support Groups
- Allocation of resources
  - Local level – Funding opportunities
  - State-Federal - Strategic Policy Planning





# FASD Diagnosis Overview







# FASD

The brain injury is invisible (83%)

Most people look “normal”

But behaviour is “abnormal”

Brain injury with a behavioural phenotype





# FASD Diagnosis

- Diffuse brain injury caused by fetal exposure to alcohol
- Face
- Brain - Evidence of neurodevelopmental injury
- (Growth)
- & every other organ / system in the body





# Disabilities

FASD is a disability with two parts

1. **Primary** disability is the brain injury due to PAE

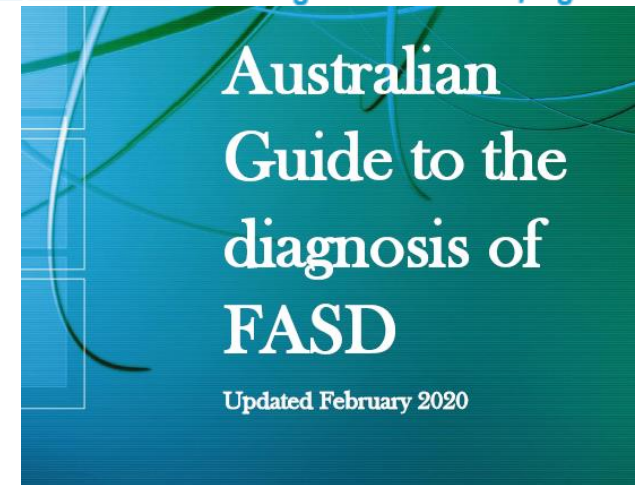
- Permanent
- Cannot be fixed

2. **Secondary** disabilities

- Develop as result of above
- Chronic, severe & expensive
- Responsive to interventions



# FASD Diagnosis



## ■ Australian Guideline

- Since 2016

<https://www.fasdhub.org.au/fasd-information/assessment-and-diagnosis/guide-to-diagnosis/>

- Guideline Review process recently commenced

**Importance of a National / International consistent framework for FASD diagnosis**

Australian Guidelines essentially consistent with Canadian Guidelines 2015



## Key components of FASD diagnostic Assessment - **Who**

- **Multidisciplinary team**
  - Ideal model for FASD diagnostic assessment
  - Accurately assess range of outcomes associated with PAE
  - Collaborative diagnostic formulation for both FASD and co-morbid diagnoses
  
- **“Help from your friends”**
  - Utilising other disciplines either internal or external to your workplace
  - Collating historical and current assessments - to identify function across the different brain domains.
  - Case conference or key person pulls diagnostic formulation together
  
- **Tiered Approach**
  - Mt Isa - Yapatjarrathati project
  
- **Sole practitioner**
  - Raising awareness of possibility of FASD
  - “testing to criteria”





## Key components of FASD diagnostic Assessment - **What**

- Confirmation of Pre-natal alcohol exposure - PAE
- Clinical History:
  - Presenting concerns
  - Obstetric, developmental, medical, mental, behavioural, social
  - Known medical conditions / current medications
- Identifying adverse pre-natal / post-natal factors
  - Co-morbid or confounding factors
- Medical Examination – Including FASD Facial Features
- Assessment of Neuro-development
  - 10 Brain Domains
  - Is there evidence of severe impairment



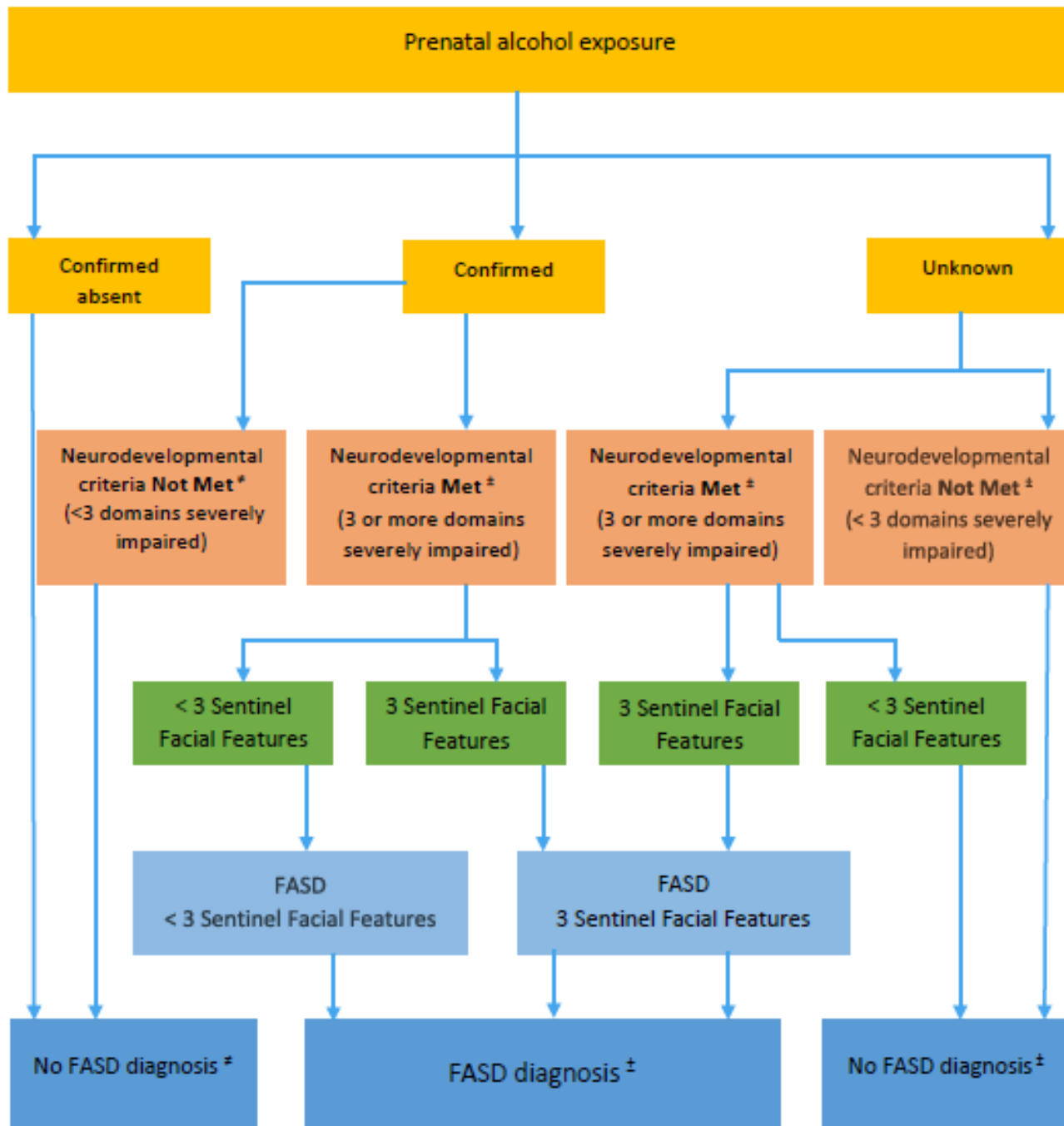
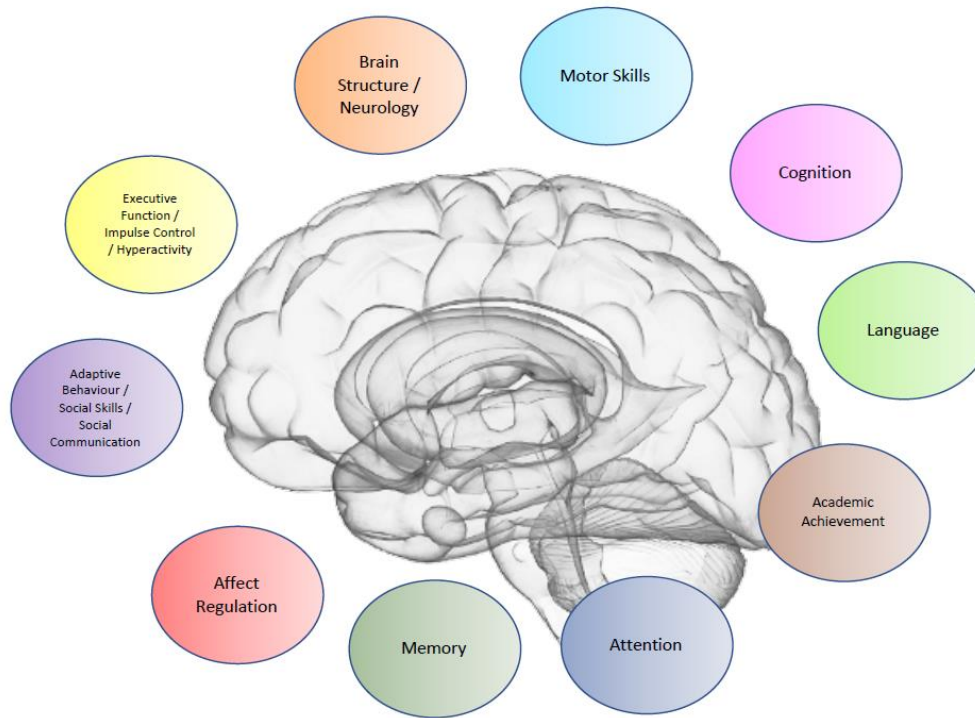




Table 1: Diagnostic Criteria and Categories for FASD

FETAL ALCOHOL SPECTRUM DISORDER		
Diagnostic criteria	Diagnostic categories	
	FASD with 3 Sentinel Facial Features	FASD with < 3 Sentinel Facial Features
Prenatal alcohol exposure	Confirmed or unknown	Confirmed
Neurodevelopmental domains <ul style="list-style-type: none"> <li>- Brain structure/Neurology</li> <li>- Motor skills</li> <li>- Cognition</li> <li>- Language</li> <li>- Academic Achievement</li> <li>- Memory</li> <li>- Attention</li> <li>- Executive Function, including impulse control and hyperactivity</li> <li>- Affect Regulation</li> <li>- Adaptive Behaviour, Social Skills or Social Communication</li> </ul>	Severe impairment in at least 3 neurodevelopmental domains	Severe impairment in at least 3 neurodevelopmental domains
Sentinel facial features <ul style="list-style-type: none"> <li>- Short palpebral fissure</li> <li>- Smooth philtrum</li> <li>- Thin upper lip</li> </ul>	Presence of 3 sentinel facial features	Presence of 0, 1 or 2 sentinel facial features



# BRAIN DOMAINS OVERVIEW



# Neurodevelopment Assessment

- 10 Brain Domains
  - Neuroanatomy/Neurophysiology
  - Motor
  - Cognition
  - Language
  - Academic Achievement
  - Memory
  - Executive functions
  - Attention
  - Adaptive Behaviour / Social Skills / Social Communication Pragmatics
  - Affect Regulation





# Defining Brain Domain

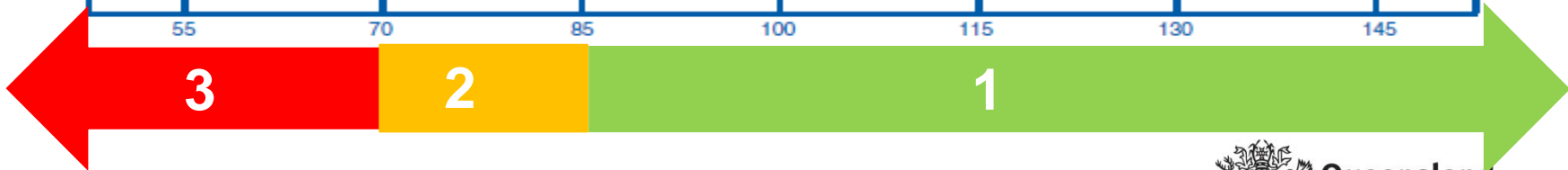
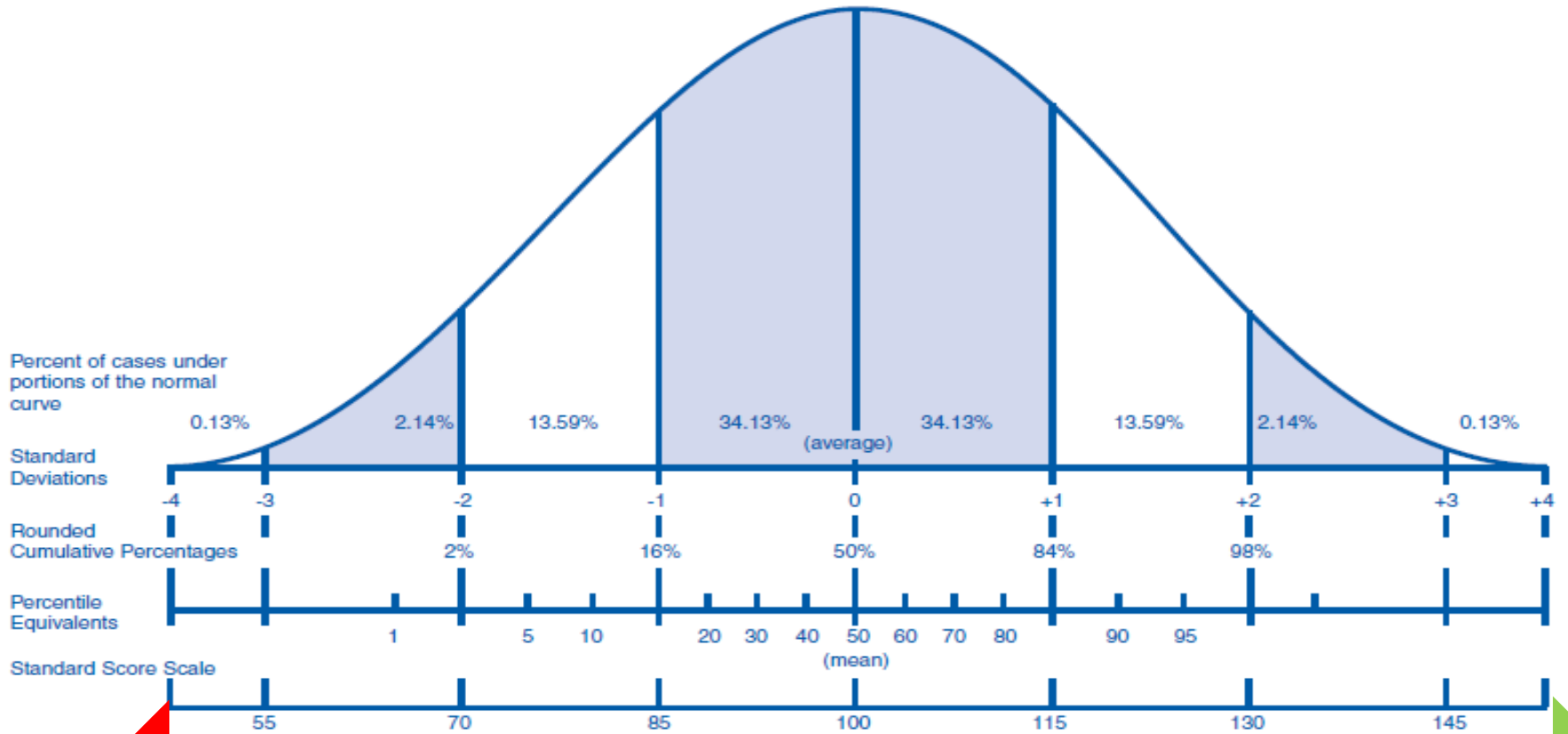
Rank	SD	Percentile	Standard Score	
1	< 1 below mean	> 16%	> 85	Normal
2	1 - 2 below mean	3 – 16%	70 - 85	Mild Moderate
3	> 2 below mean	≤2 %	< 70	Severe

The '**clinical cut-off**' for **severe impairment** is defined either as a *global score* or a *major subdomain* score on a standardised validated neurodevelopmental scale that is 2 or more standard deviations below the mean ( $\leq 2$  SD) or less than / equal to the 2<sup>nd</sup> percentile ( $< 2^{\text{nd}}$  PC). **Australian Guidelines refer to <3<sup>rd</sup> percentile, which means the same.**





### THE NORMAL CURVE, PERCENTILES, AND STANDARD SCORES





# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	3
Cognition	2
Language	3
Academic Achievement	2
Memory	2
Executive functions	2
Attention	2
Adaptive/ Social /Pragmatics	3
Affect Regulation	3





# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	3
Cognition	2
Language	3
Academic Achievement	2
Memory	2
Executive functions	2
Attention	2
Adaptive/ Social /Pragmatics	3
Affect Regulation	3







# Brain Domains

Neuroanatomy/Neurophysiology	1
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Language	3
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Memory	2
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Attention	2
Adaptive/ Social /Pragmatics	3
Affect Regulation	3

Severe  
Impairment in 4  
brain domains

Meets Criteria for  
FASD

If PAE confirmed  
Or has Facial Features

&

Pre-Post natal factors  
considered





# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	1
Cognition	1
Language	2
Academic Achievement	2
Memory	1
Executive functions	1
Attention	3
Adaptive/ Social /Pragmatics	1
Affect Regulation	1





# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	1
Cognition	1
Language	2
Academic Achievement	2
Memory	1
Executive functions	1
Attention	3
Adaptive/ Social /Pragmatics	1
Affect Regulation	1



# Brain Domains

Neuroanatomy/Neurophysiology	1	Severe Impairment only 1 brain domains
Motor	1	Does not meet criteria for FASD
Cognition	1	
Language	2	Possible diagnosis
Academic Achievement	2	<ul style="list-style-type: none"><li>• Inattentive ADHD</li><li>• Mild-moderate language disorder</li><li>• Possible Specific Learning Disorder</li></ul>
Memory	1	
Executive functions	1	
Attention	3	<ul style="list-style-type: none"><li>• Depending on age and other factors may be <b>At Risk of FASD</b></li></ul>
Adaptive/ Social /Pragmatics	1	
Affect Regulation	1	



# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	2
Cognition	3
Language	2
Academic Achievement	2
Memory	1
Executive functions	2
Attention	2
Adaptive/ Social /Pragmatics	2
Affect Regulation	3





# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	2
Cognition	3
Language	2
Academic Achievement	2
Memory	1
Executive functions	2
Attention	2
Adaptive/ Social /Pragmatics	2
Affect Regulation	3





# Brain Domains

Neuroanatomy/Neurophysiology	1
Motor	2
Cognition	3
Language	2
Academic Achievement	2
Memory	1
Executive functions	2
Attention	2
Adaptive/ Social /Pragmatics	2
Affect Regulation	3

Only 2 Severe Brain Domains

Doesn't meet criteria for FASD

But significant challenges

Depending on Cognitive testing possible Intellectual Impairment

Possible **“At risk of FASD”**







# Questions