FETAL ALCOHOL SPECTRUM DISORDER (FASD)

HOW AND WHY WE SHOULD MAKE THIS DIAGNOSIS

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FETAL ALCOHOL SPECTRUM DISORDER (FASD)

FASD is a lifelong but *preventable* neurodevelopmental disorder caused by *in-utero* or prenatal alcohol exposure (PAE).

Prenatal alcohol exposure causes diffuse, permanent and irreversible damage to the fetal brain, especially the frontal lobes, resulting in lifelong dysfunction and disabilities.

FASD is a type of acquired brain injury, and there is a spectrum of manifestations and disorder.

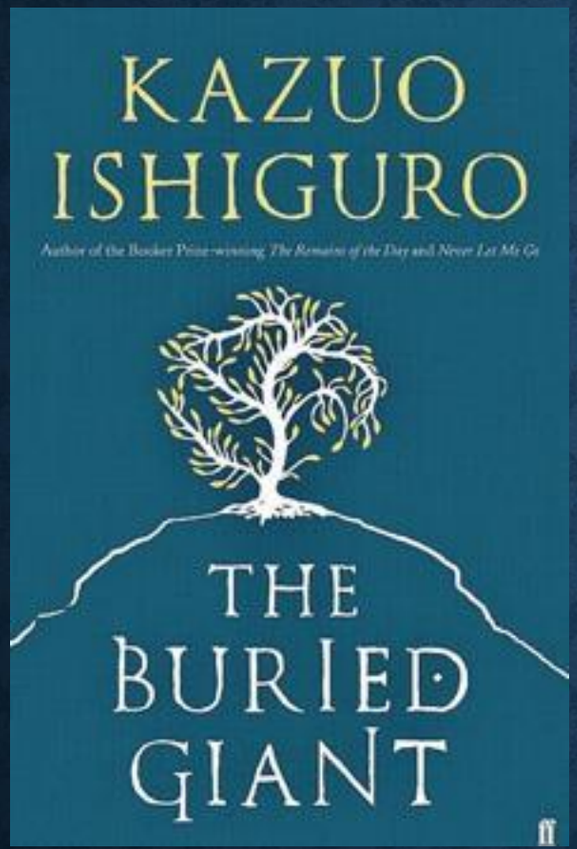
WHAT DO WE KNOW ABOUT FASD RATES?

- 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%)



FASD IS THE MOST COMMON CAUSE OF PREVENTABLE DEVELOPMENTAL DISABILITY

Kids with FASD are in everyone's caseload and in all ethnic/social groups

- Many are *missed or not diagnosed with FASD*
- Missed opportunities for *optimal management* and *prevention*
- Outcomes improved by *early diagnosis* and *intervention*
-

FASD DIAGNOSTIC CRITERIA

3 key aspects

DIAGNOSTIC CATEGORIES

FASD	
FASD + 3SFF	**FASD <3SFF**
PAE +/-	PAE
ND impairment	ND impairment
3 Facial features	0,1,2 Facial features

SFF = Sentinel Facial Features
PAE = prenatal alcohol exposure

Prenatal alcohol exposure

- Confirmed exposure

Neurodevelopmental impairment

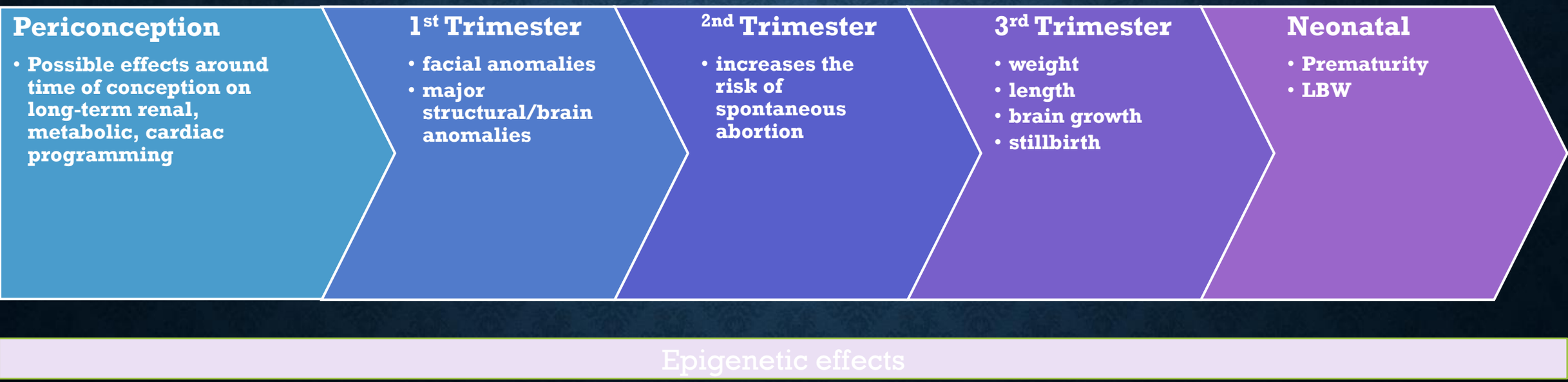
- 3 (out of 10) domains significantly impaired *<3rd %le*
  - Structural/neurology – e.g. microcephaly (HC <3rd %le)
  - Functional – e.g. Cognition, Communication, ADHD, Adaptive Behaviour)

Facial features

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

Effects of alcohol exposure during pregnancy on fetal development

Fetal brain damage may occur as result of exposure at any time
(leading to neurodevelopment problems in later life)



Feldman et al Prenatal Alcohol Exposure Patterns and Alcohol-Related Birth Defects and Growth Deficiencies: A Prospective Study Alcoholism: Clinical and Experimental Research 2012 Vol 36, 4

FACTORS THAT MEDIATE THE IMPACT OF PAE

- The effect of prenatal alcohol exposure on the embryo and fetus is influenced by a range of factors including the:
  - Timing, dose and frequency of alcohol intake
  - Maternal age, health and genetics
  - Fetal genetics

PRENATAL ALCOHOL EXPOSURE (PAE)

Standardised approach to asking about alcohol allows for assessment of exposure risk – use the AUDIT-C

Assessment of PAE requires clinical judgement and careful evaluation of a range of information that may provide confirmation of maternal alcohol use and quantification of intake

EXPOSURE / RISK LEVELS

- Evidence of exposure can be evaluated to estimate the overall level of risk using the following broad risk categories:
- **No exposure**
  - (confirmed absence), no risk of FASD
- **Unknown exposure**
  - (alcohol use is unknown)
- **Confirmed exposure**
  - (AUDIT-C score =1-4; or confirmed use, but exposure less than high risk level for FASD; or confirmed use, but not known if exposed at a high risk level for FASD)
- **Confirmed-high risk exposure**
  - (AUDIT-C score = 5+; confirmed use, exposure at high risk level for FASD).

HIGH RISK EXPOSURE

- Confirmed **high risk exposure** for FASD can be considered to include, *at any time during pregnancy*:
  - An AUDIT-C score of **5 or more**
  - Reported consumption of **5 or more standard drinks on one occasion**
  - **Other reliable evidence of high consumption**

STARTING THE CONVERSATION

Alcohol use in early pregnancy (if available)

Was the pregnancy planned or unplanned?	☐ Planned ☐ Unplanned ☐ Unknown
When did the birth mother realise that she was pregnant? _____ (weeks)	☐ Unknown
Did the birth mother drink alcohol before the pregnancy was confirmed?	☐ Yes ☐ No ☐ Unknown
Did the birth mother modify her drinking behaviour on confirmation of pregnancy? If Yes please specify:	☐ Yes ☐ No ☐ Unknown
During which trimesters was alcohol consumed? (tick one or more)	☐ None ☐ 1st ☐ 2nd ☐ 3rd ☐ Unknown

AUDIT C

AUDIT-C questions

Source of reported information on alcohol use: ☐ Birth mother ☐ Other (please specify)

1. How often did the birth mother have a drink containing alcohol during this pregnancy?

Unknown	Never [skip Q2+Q3]	Monthly or less	2-4 times a month	2-3 times a week	4 or more times a week
☐	☐$_0$	☐$_1$	☐$_2$	☐$_3$	☐$_4$

2. How many standard drinks did the birth mother have on a typical day when she was drinking during this pregnancy?

Unknown	1 or 2	3 or 4	5 or 6	7 to 9	10 or more
☐	☐$_0$	☐$_1$	☐$_2$	☐$_3$	☐$_4$

3. How often did the birth mother have 5 or more standard drinks on one occasion during this pregnancy?

Unknown	Never	Less than monthly	Monthly	Weekly	Daily or almost daily
☐	☐$_0$	☐$_1$	☐$_2$	☐$_3$	☐$_4$

AUDIT-C score this pregnancy: (Q1+Q2+Q3)=_____ Scores= 0=no exposure 1-4= confirmed exposure 5+= confirmed high-risk exposure

ASSESSING OTHER EXPOSURES – *PRE AND POSTNATAL*

It is also important to ask about other substances and medications that may affect fetal development.

- Other drugs – eg. cigarettes, opiates, amphetamines
- Anticonvulsants – eg. sodium valproate
- Other medications – eg. ibuprofen

Other prenatal risk factors also need to be discussed:

- Medical – eg. pregnancy complications, congenital infection, trauma, maternal ill health
- Psychosocial – eg. family violence, parental homelessness or transient accommodation, *early life trauma*

STANDARD DRINKS

ALCOHOL USE DURING PREGNANCY IN AUSTRALIA

The *Australian National Drug Strategy Household Survey (2013)* reported:

- **About 4 in 10 (42%)** Australian women consumed alcohol during pregnancy (**whether or not they knew they were pregnant**);
- **More than half (56%)** of Australian women had consumed alcohol while being unware of their pregnancy;
- **A quarter (26%)** of women continued to consume alcohol even once they knew they were pregnant

References

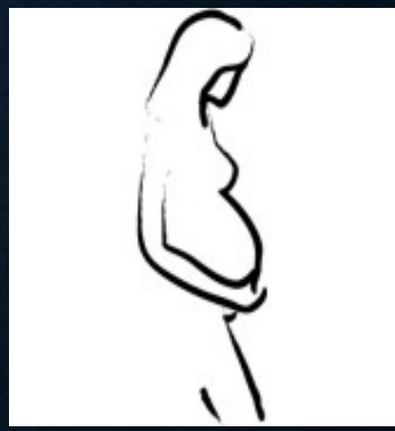
Australian National Drug Strategy Household Survey (2013)

http://www.aihw.gov.au/2013-national-drug-strategy-household-survey/

Link to paper? YES

UNSW STUDY OF ETOH IN EARLY PREGNANCY

- **Fetal alcohol exposure <u>early</u> in pregnancy is highly prevalent**
- *Heavy or binge-drinking* during early pregnancy was more common than low-level drinking
- Most women *reduce or cease* their alcohol consumption after becoming aware of pregnancy
- Factors associated with decision to cease, reduce or continue related to:
  - ETOH use prior to pregnancy recognition
  - Maternal age
  - Smoking status
  - Illicit substance use



PRACTICAL TIPS – STARTING THE DISCUSSION

Starting the discussion:

- Was the pregnancy planned or unplanned?
- When did the mother become aware of her pregnancy?
- What lifestyle changes did you make then?
- Ask respectfully
- Several times if necessary
- In a *compassionate and non-judgemental manner*

NEURO-DEVELOPMENTAL IMPAIRMENT

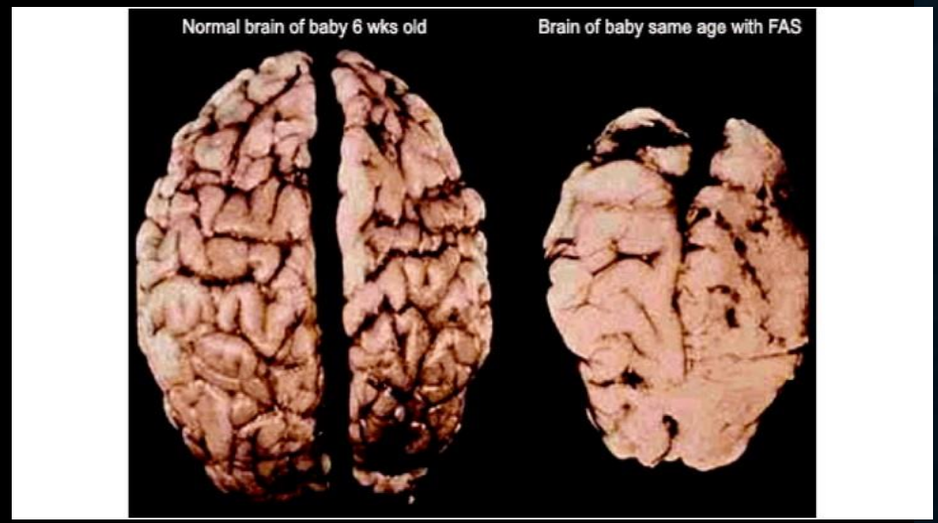
Neurodevelopmental impairment due to prenatal alcohol exposure is the hallmark feature of Fetal Alcohol Spectrum Disorder.

PRENATAL ALCOHOL EXPOSURE AND FETAL BRAIN DAMAGE

Fetal brain damage may occur as a result of prenatal alcohol exposure ***at any time during the pregnancy***.

with impaired facial development of the fetus which causes characteristic FASD facial abnormalities.

This *disruption of normal fetal brain development* may cause neurodevelopmental and behavioural problems in later life, as well as neurological abnormalities and structural brain abnormalities.



This brain damage is <u>not</u> necessarily associated

PRENATAL ALCOHOL EXPOSURE AND FETAL BRAIN DAMAGE - THE GLIAL PERSPECTIVE

Alcohol affects fetal ***brain cell function***, both ***neurons*** and supporting ***glial cells***, based on evidence from tissue and brain imaging research.

Alcohol-induced abnormalities in ***glial cells*** are thought to **contribute to**

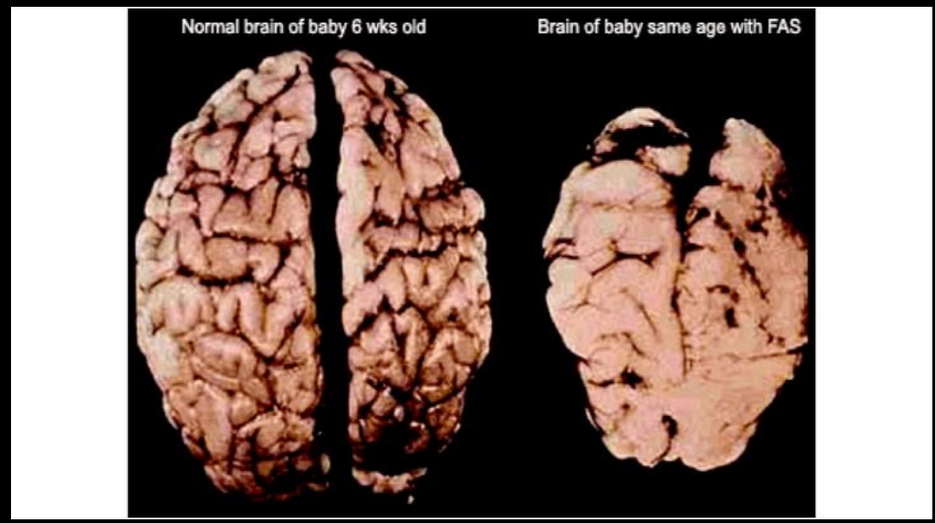
abnormal neuronal development

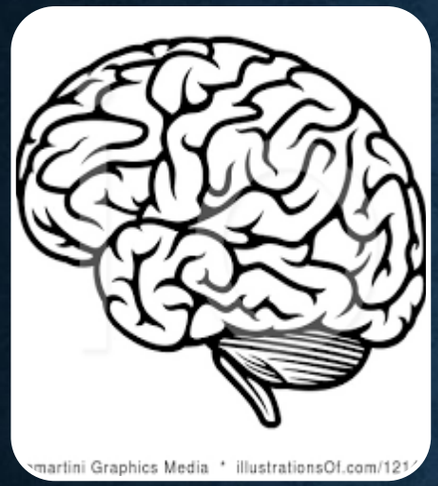
reduced neuron survival

disrupted brain architecture / connectivity

abnormal brain plasticity.

Microcephaly may be caused by reduced astrocyte proliferation and survival due to alcohol exposure. Astrocytes rapidly proliferate and mature during the brain growth spurt in the third trimester.





- Brain structure/ neurology
- Motor skills
- Cognition
- Language
- Academic achievement
- Memory
- Attention
- Executive function, impulse control and hyperactivity
- Affect regulation
- Adaptive behaviour, social skills or social communication

NEURODEVELOPMENTAL IMPAIRMENT - CRITERIA

A FASD diagnosis requires **objective evidence of *severe impairment* of central nervous system function** in *at least 3* of these 10 specified neurodevelopmental domains.

The rationale is that there should be evidence of PAE causing pervasive brain dysfunction.

Evidence of severe impairment may be attributed to prenatal alcohol exposure only when other possible causative factors have been considered and/or excluded.

NEURODEVELOPMENTAL IMPAIRMENT - PATTERNS

Patterns of neurodevelopmental impairment in individuals with PAE are ***complex and diverse.***

There is no typical pattern of impairment in FASD, most likely due to differences in the timing and level of PAE, and genetic and environmental factors that influence maternal blood alcohol level and fetal brain development.

Nonetheless, problems such as deficits in attention are frequently identified in individuals with FASD. ADHD occurs in over 60% of people with FASD.

ASSESSMENT PROCESS

Assessment of neurodevelopmental domains includes:

- Measurement of occipitofrontal head circumference (OFC)
- Neurological examination
- Developmental assessment
- Review of investigation results – e.g. Brain MRI or genetic screening

BRAIN STRUCTURE/NEUROLOGY

Impairment	Specifics
Microcephaly	Occipitofrontal head circumference is <3rd percentile or 2 or more standard deviations below the mean. This should be based on current measurement, and corrected age for ex-premature infants until 2 years of age.
Structural brain abnormalities	Structural brain abnormalities known to be associated with prenatal alcohol exposure are shown on brain imaging.
Significant neurological diagnoses	which are otherwise unexplained when other aetiologies have been excluded. e.g. Cerebral Palsy, visual impairment, sensorineural hearing loss.
Seizure disorder	which are not due to known postnatal causes

SEVERE IMPAIRMENT IN THE FUNCTIONAL DOMAINS

In most domains, **severe impairment** is defined by a **'clinical cutoff'** score on standardised validated assessment tools for that domain (e.g. intelligence test for Cognition domain). This indicates that the individual's *functional skills* in that domain are severely impaired.

This is defined as a score of 2 or more standard deviations below the mean (≤2 SD) *or* than the 3rd percentile (<3rd PC).

The score can be a *global score* (e.g. full scale IQ < 70 standard) or *Major subdomain score* (e.g. non verbal reasoning < 70 standard score)

Evidence of a **significant discrepancy** between major subdomain scores also constitutes evidence of severe impairment.

FASD TREATMENT RESISTANT ADHD

- **Problems with Executive Function**
- People who have been exposed prenatally to alcohol often exhibit deficits with "executive functions". Executive functions are a cluster of processes involved in "the ability to plan and guide behavior to achieve a goal in an efficient manner" (Kodituwakku, Kalberg, and May). In order to carry out an assignment at school, students must organize and order their behavior and actions.
- Executive functioning has been defined by Muriel Lezak (McCreight, 1997) as having four major components:
- 1. Goal formation
  2. Planning
  3. Carrying out goals
  4. Doing so effectively

FASD TREATMENT RESISTANT ADHD

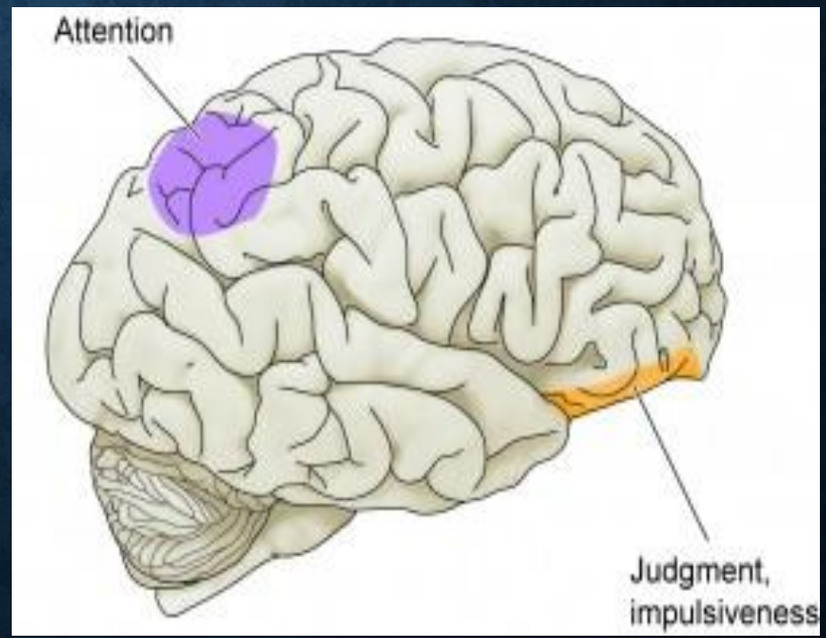
- *If the teacher tells the children to get out their math books and do questions one to ten, each pupil must understand this in terms of first getting out the textbook, the exercise book, the pencil, and eraser, and also clearing the desk of anything that is extraneous to the task or distracting. Then the pupil must understand the number of questions to be done in terms of a quantity with a beginning and an end and must make some judgment about how much time to spend on each question. After this, the pupil must begin the work, stay on task, use his or her knowledge of the subject, and finally, complete the task.*
  (McCreight, 1997).

FASD TREATMENT RESISTANT ADHD

- When an assignment is given, most students are able to get started without consciously thinking about it. However, when the FASD student with executive dysfunction is given an assignment, he/she does not know where to begin. Often, he reacts impulsively before determining his desired goal, planning, and organizing the steps he needs to obtain his goal.
- Executive functions are "higher order" processes and require the ability to:
- Delay responding
- Shift between activities flexibly
- Plan for the future

FASD TREATMENT RESISTANT ADHD

- Executive functions are "higher order" processes and require the ability to:
- Delay responding
- Shift between activities flexibly
- Plan for the future
- These higher order processes require the integration of several abilities, such as:
- Working memory
- Impulse control
- Organizational skills
- Temporal ordering and sequencing
- Internalizing language
- Mental flexibility
- Attention and effort

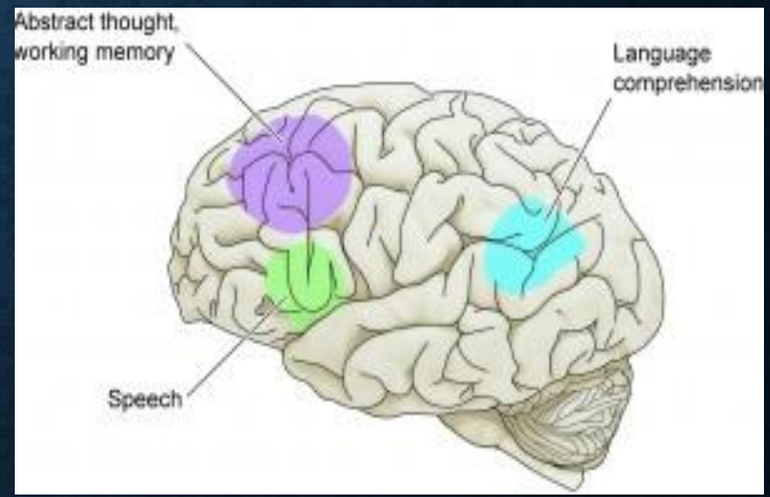


FASD TREATMENT RESISTANT ADHD

- Executive functions have been likened to a bicycle wheel, and the many skills (such as the ones mentioned above) that comprise it are the spokes. It is possible that only 1 or 2 spokes are damaged; however, the wheel is affected. FASD students whose executive functions are compromised may have only 1 or 2 of these skills affected, or they may have many skills affected. When executive functions are compromised, learning is disrupted.

FASD TREATMENT RESISTANT ADHD

- The following characteristics describe the FASD student who has executive function deficits in each of the areas listed above:
- **Working memory**
- Forgetfulness – these are the "I forgot" students. (They never had the information in the first place.)
- Difficulty holding information "in their heads"
- Missing chunks of information
- Difficulty remembering multi-step directions
- Can't keep track of their things
- Forgets supplies, assignments, and books
- Difficulty with mental math
- Difficulty with reading comprehension

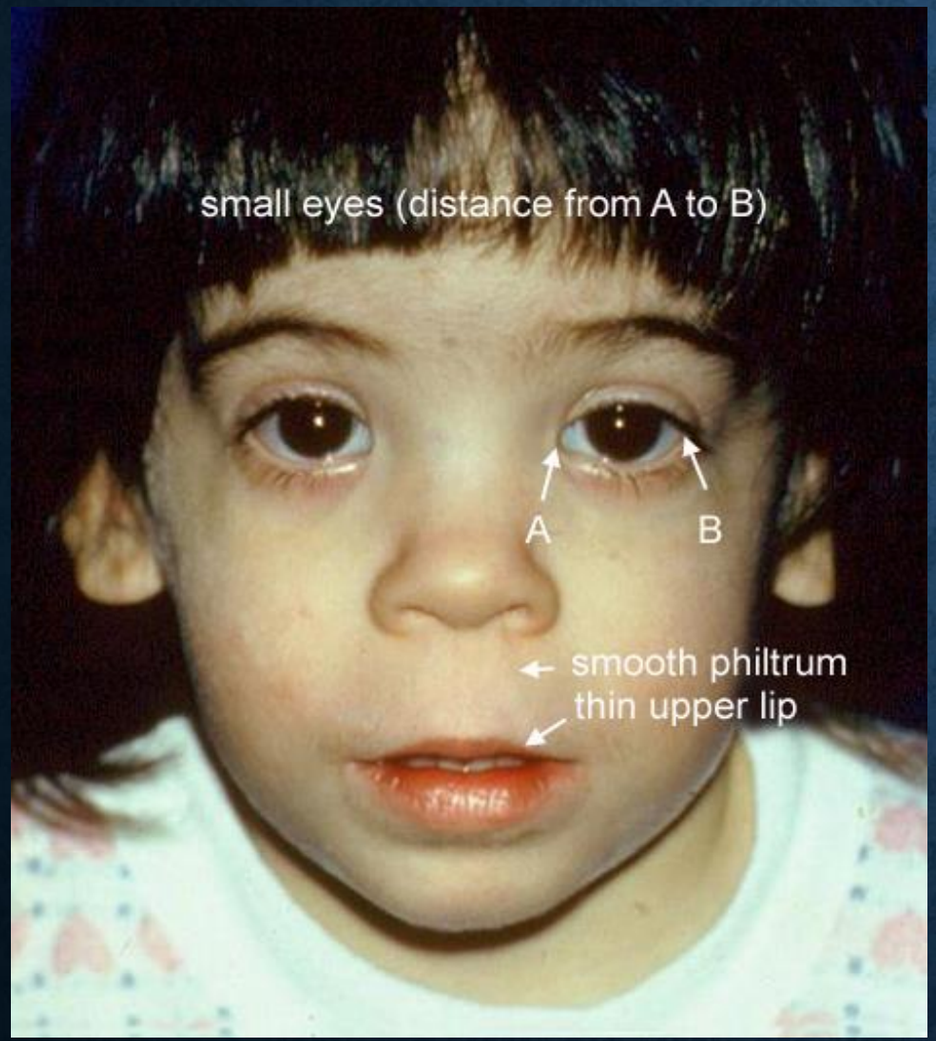


FASD TREATMENT RESISTANT ADHD

- **Impulse control**
- Responds quickly in class without thinking
- Calls out in class
- Acts out in class
- **Organization**
- Has difficulty knowing how to start an assignment
- Has difficulty organizing a plan of action to complete an assignment
- Has difficulty organizing or sequencing written information logically
- Has difficulty seeing the forest through the trees
- Looks anxious and overwhelmed
- **Time management**
- Difficulty judging the order in which assignments need to be completed
- Difficulty estimating how much time it will take to complete an assignment

FASD TREATMENT RESISTANT ADHD

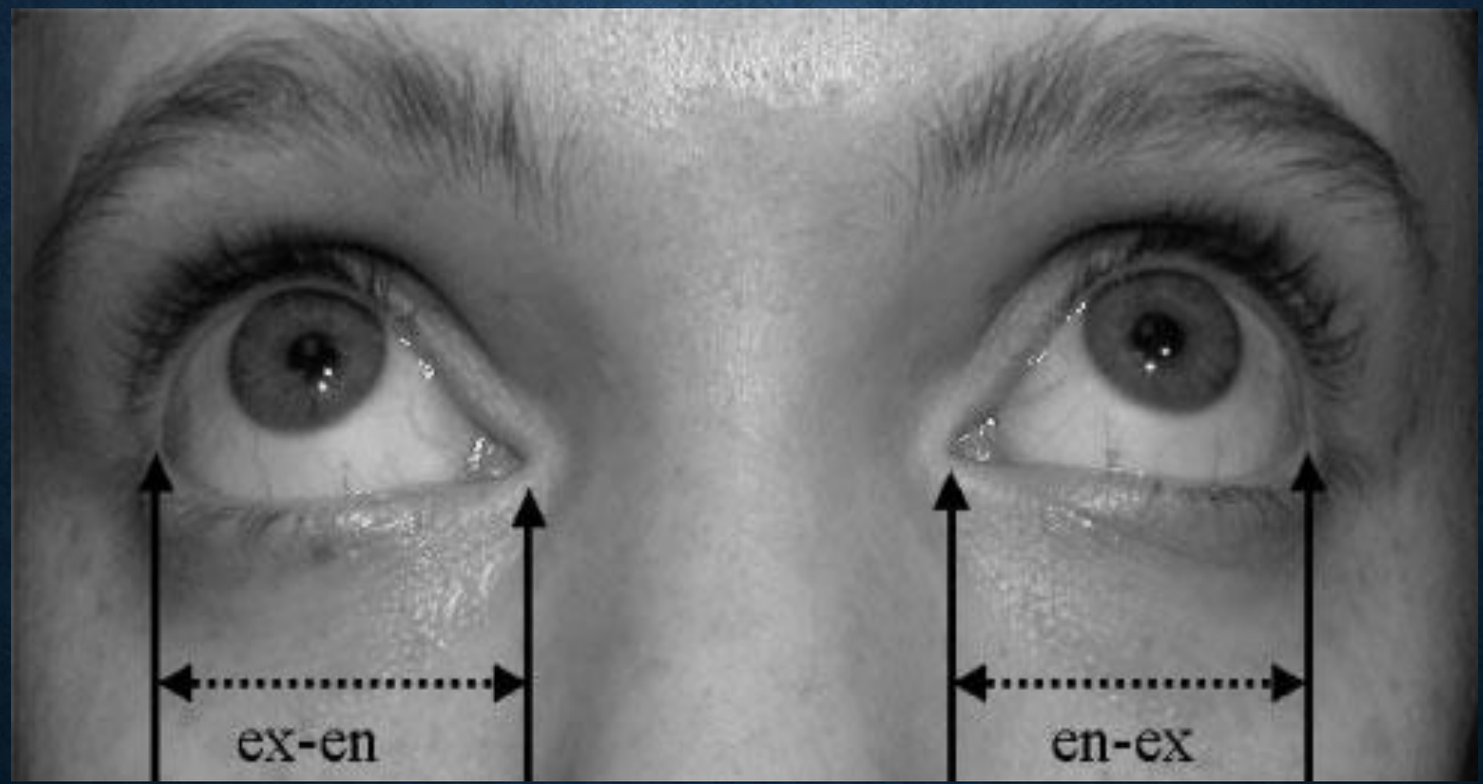
- Difficulty planning or preparing for future projects
- **Mental flexibility**
- Difficulty with transitions
- Difficulty shifting to a new strategy or a new way to look at something
- Perseverates; gets stuck
- **Internalizing language**
- Lack of using self-talk or reflective thinking
- Difficulty with problem solving
- Difficulty drawing from past experiences
- **Attention and effort**
- Easily distracted
- Difficulty paying attention and maintaining attention
- Difficulty getting started
- Assignments often incomplete



SENTINEL FACIAL FEATURES

FACIAL FEATURES

- *smooth philtrum*
- *thin upper lip*
- *short palpebral fissure length*
  -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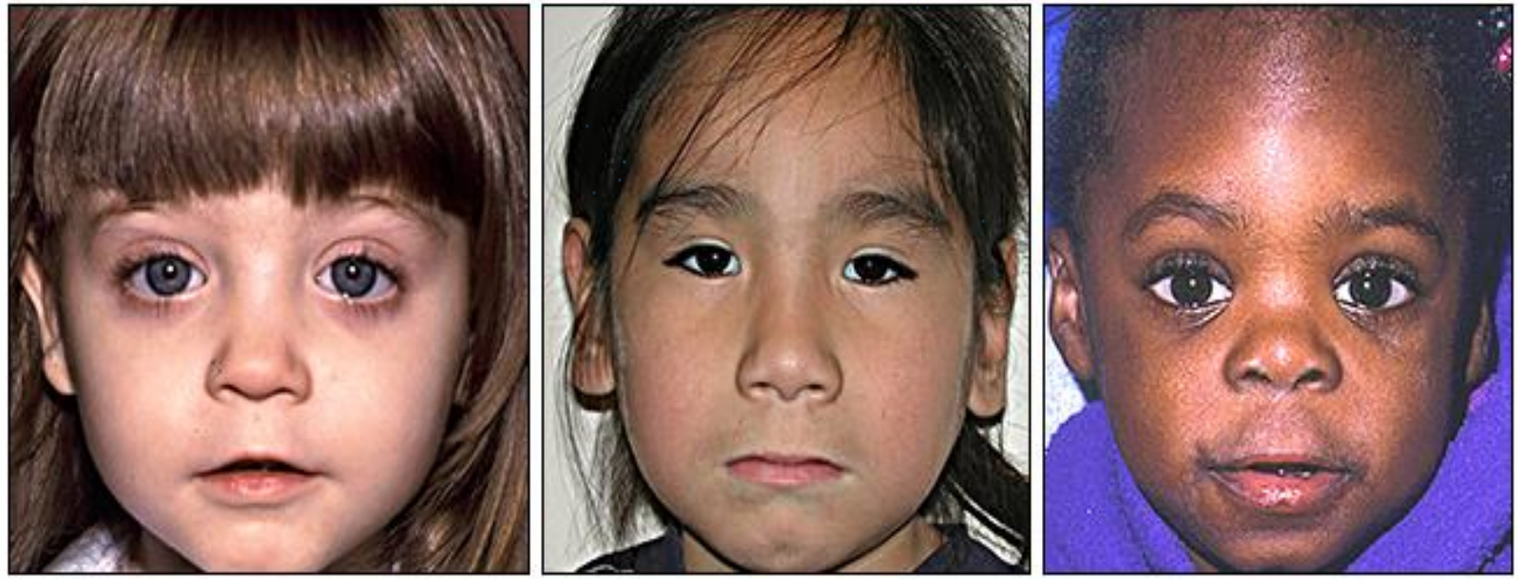
Characteristic facial features in a child with fetal alcohol spectrum disorder



Examples of the fetal alcohol syndrome facial phenotype across three races: Caucasian, Native American, and African American. Characteristic facial features include short palpebral fissure length, smooth philtrum, and a thin upper lip.

FACIAL FEATURES - ASSESSMENT

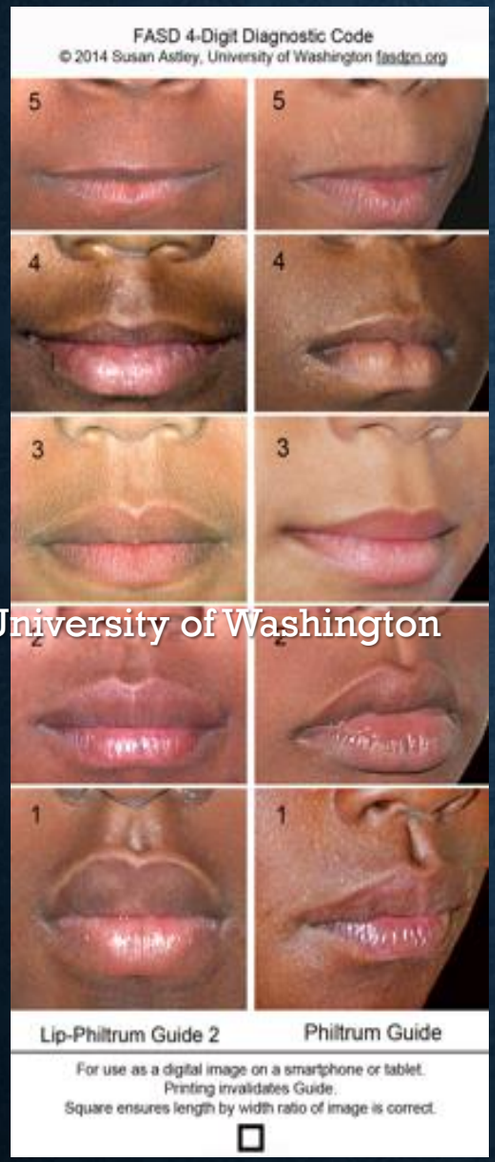
Assessment methods (can be multiple)

Direct PFL measurement ruler

Lip-philtrum guide

Facial photo analysis

LIP-PHILTRUM GUIDES



University of Washington

Frontal view ¾ view

Facial features of FASD

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

WHY DIAGNOSE?

Parents and caregivers say it helps them to understand the cause of their child's problems and to develop realistic expectations of their capacities

"We didn't know what was wrong with him. He was about four. I was hitting my head on a brick wall.

Everyone kept saying, it's parenting…

Once the diagnosis came in, you knew what to focus on and that helps so much having a diagnosis"

WHY DIAGNOSE?

Functional support vs *diagnostic 'label'*

As paediatricians, allied and mental health practitioners we diagnose conditions and provide functional support

Supports early intervention

Enhanced understanding of the child's strengths and needs by parents/teachers

Prevention/advocacy

Diagnosis may lead to prevention in future pregnancies;

Identifying/ 'counting' the problem;

Raising public awareness re: risk of harm from prenatal alcohol exposure

ALPHABET SOUP?

SCREENING FOR FASD

- There are currently *no validated* standardised screening tools specifically for the developmental, behavioural or physical features FASD (equivalent for example, to the Modified Checklist for Autism in Toddlers (M-CHAT-R).
- This is partly related to the wide spectrum of possible neurodevelopmental impairments in FASD and the variation in presenting symptoms.
- Some *non-validated tools* are available
- Screening of the general population for FASD is unnecessary and costly.
- Recognition of high risk groups for FASD is important (discussed further in next section).
- Further research is required to develop reliable validated screening tools.
- ***The best 'screening tool' is an accurate history of alcohol use in pregnancy***

DISCUSSING ALCOHOL USE IN PREGNANCY: THE BEST 'SCREENING TOOL'

Consideration of prenatal alcohol exposure and possible FASD should be part of standard 'mainstream' clinical practice for health and child protection professionals.

An accurate history of alcohol use in pregnancy to identify prenatal alcohol exposure is the best 'screening tool' for FASD:

- This should be conducted in a *sensitive and respectful manner.*
- Discussion of maternal drinking and associated risks should be integral to *all* prenatal and postnatal care of women and children by *all* health care professionals.
    - An obstetric history should *always* include discussion of alcohol consumption in pregnancy and assessment for prenatal alcohol exposure *as standard practice*, as for any other significant pregnancy complication or prenatal exposure e.g. other drugs, medications and infection.
- Validated screening tools for evaluating *alcohol use* should be used (e.g. AUDIT-C – viewed in the Tab below) (Include Audit-C TAB picture from other module).

REFERRAL: UNDERLYING PRINCIPLES

FASD as a differential diagnosis

- FASD should be part of the differential diagnosis for any significant developmental, behavioural, learning or mental health problems in an individual, *until prenatal alcohol exposure is excluded*.
- *Assessment for FASD should be considered in all children with a neurodevelopmental disorder of unknown aetiology who have a history of PAE.*

Support and consent as part of the referral process

- Supports should be provided for the individual, caregiver and/or family, as part of the referral process, including appropriate support and intervention if ongoing alcohol misuse is an issue.
- Consent for assessment should be obtained from the individual or their caregiver.

WHEN TO REFER

Clinicians should refer an individual for a FASD diagnostic assessment if:

- Prenatal alcohol exposure was at high risk levels*
- Neurodevelopmental impairment and PAE are present, whether or not they have distinctive facial features
- Microcephaly and PAE are present
- They or their parents or caregiver have concerns (regardless of above)

REFERRAL THRESHOLD FOR INDIVIDUALS AT INCREASED RISK OF FASD

The *threshold* for referring some people for FASD diagnostic assessment should be lower in the following **high-risk groups and/or settings**.

These include children, adolescents or adults:

- In out-of-home care (adoption/foster).
- In contact with justice system.
- Living in a community known to have high rates of drinking.
- Whose birth mother has known alcohol-related illness or dependency.
- With a family member with Fetal Alcohol Spectrum Disorder.

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*

WHAT TO DO ?

- 1. Define the neurodevelopmental disability and aim to do what we do with those with intellectual disability, autism, TBI and ADHD.
- 2. Explain behaviour and shape expectations based on the disability.
- 3. Treat medical comorbidity – chronic illness makes FASD problems worse and vice versa
- 4. Treat psychiatric co-morbidity – anxiety, depression and PTSD. 5. Address the context in which the child is living
- 6. Gently increase awareness to increase prevention in future pregnancies.

CONCLUSION / TAKE HOME MESSAGES

- This highlights *opportunities* and *need* for:

Improvement in:

- ✓ Clinical service provision
- ✓ Assessment services and procedures
- ✓ Post-diagnosis care

Ongoing development of:

- ✓ Diagnostic and support services
- ✓ FASD-informed clinician education

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QUESTIONS

THANK YOU