The Revised Canadian Guidelines for Diagnosis of Fetal Alcohol Spectrum Disorder (FASD)

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February 2017

Some slides by Bruce Pipher, MD

Cook, Green, Lilley, Anderson, Baldwin, Chudley, Conry, LeBlanc, Loock, Lutke, Mallon, McFarlane, Temple, Rosales (2015). Fetal alcohol spectrum disorder: A guideline for diagnosis across the lifespan. Canadian Medical Association Journal.

Outline

1. A little about the big picture: Diagnostic schemes for FASD
2. Key changes in the new Canadian guidelines
3. New category names and what they mean
4. Domains and how each is assessed
5. Specific discussion of the new affect regulation domain
6. If time permits: a little about competing systems

Current landscape of diagnostic schemes

There is a patchwork of similar but not identical diagnostic schemes in use in different places:

-Seattle 4 Digit Code (1997, 2004)

-Canadian Guidelines (2005, 2016)

-Hoyme (2005, 2016)

-DSM-5 (2013)*different in intent than the others

Coles et al (2016)

Moderate convergence among the diagnostic schemes in a clinic population with a mean age of 5:

-Canadian Guidelines (2005): 25%

-Seattle 4 Digit Code (2004): 38%

-Emory Clinic (2005): 46%

-Hoyme (2005): 60%

Considerations

- Results likely affected by age of population (Canadian system prioritizes function and therefore makes it harder to diagnose young kids)
- Differences between original publication and current practice in clinics that use the system

Why is diagnosing FASD so hard?

- It's a hybrid between diagnosing a cause (like a genetic syndrome) and a behavioural outcome (like ADHD)
- No workable biomarker for alcohol exposure: always cases where the alcohol history is unknown
- While physical features add to certainty, most individuals affected by alcohol don't have them
- Lots of research shows relationships but doesn't say where to put a cutoff

What is there consensus on?

- Multidisciplinary assessment is needed (except for DSM)
- Key variables include facial features, neurodevelopmental function, and alcohol history
- There is a population of very impaired individuals who don't have the facial features

What is there disagreement on?

- How many facial features are relevant?
- How abnormal should facial features be?
- How should neurodevelopment be measured? (direct tests vs. questionnaires, how many domains)
- How abnormal should neurodevelopment be?

Canadian Guidelines (2005)

FAS	Partial FAS	ARND
Growth		
Face	Face	
Brain	Brain	Brain
Alcohol confirmed or unknown	Alcohol confirmed	Alcohol confirmed

Revisions: Easy Decisions

1. Growth and face should be handled differently.

a. Growth impairment removed as a criteria, although it should still be documented

-very rare and adds little to specificity

b. Facial features treated as an all-or-nothing variable

- Astley (2013)
- All 3 facial features have more than 95% specificity (that is, 95% of those with all 3 facial features have FASD, including CNS dysfunction and alcohol exposure)
- Specificity drops dramatically if you go to 2 facial features – if you look in non-clinic samples for those with 2 features, the majority were not alcohol exposed and have no CNS deficits

2. Depression and anxiety should be considered a symptom, not a confound

Strong research evidence showing that:

-very high comorbidity levels by adulthood

-animal data clearly show a direct causal pathway via the HPA axis

Clinic experience suggested that:

-in practice, many adults did not get a diagnosis because clinicians felt that they had to choose between attributing impairment to alcohol exposure or mood/anxiety symptoms

Revisions: Difficult Decisions

1. What's the best diagnostic term?

FASD? Preferred by parents and agencies for clarity – the term chosen

ARND or ND-PAE? (unwieldy but better expresses the limits of talking about causality in an individual, not a group)

2. What's the right threshold for continuous measurements?

- 4 digit code and old Canadian guidelines used a -2 sd threshold for most measurements and brain tests
- US attendees pushed for a relaxed threshold on brain tests (-1.5 or even -1 sd)
- Canadian attendees not strongly in favour
- Alternate systems tend to adjust the balance by assessing fewer domains

Decision:
Threshold to stay at -2 sd

At a Glance

	Face	Brain	Alcohol
FASD with Sentinel Physical Features	Complete	Impaired	Confirmed or unknown
FASD without Sentinel Physical Features	Absent or incomplete	Impaired	Confirmed
At Risk for Neurodevelopmental Disorder and FASD, due to PAE	Complete, incomplete or absent	Not able to be accurately tested (due to age or other factors)	Confirmed

At a Glance

FASD with Sentinel Physical Features	FASD without Sentinel Physical Features	At Risk for ND and FASD, due to PAE
Previously FAS	Previously ARND	Previously no label
Previously PFAS		

1. FASD with Sentinel Facial Features

A. Simultaneous presentation of the 3 sentinel facial features (short palpebral fissures, smooth philtrum, thin upper lip)

AND

B. Evidence of impairment in 3 or more of the identified CNS domains (or microcephaly in infants)

AND

C. Alcohol exposure confirmed or unknown

2. FASD without Sentinel Facial Features

A. Evidence of impairment in 3 or more of the identified CNS domains

AND

B. Confirmation of prenatal alcohol exposure, with the estimated dose at a level known to be associated with neurobehavioural effects

3. At Risk for Neurodevelopmental Disorder and FASD, due to Prenatal Alcohol Exposure

A. Confirmation of PAE

B. AND

B. CNS criteria are not met

AND

C. Some indication of neurodevelopmental disorder in combination with a plausible reason that the neurodevelopmental assessment should not be considered conclusive (e.g. young age, situational factors).

3. At Risk for Neurodevelopmental Disorder and FASD, associated with Prenatal Alcohol Exposure

This designation may also be considered for individuals with all 3 sentinel facial features, who do not yet have documentation or evidence for the requisite 3 or more neurodevelopmental domain criteria or true microcephaly. This designation should never be considered when PAE is confirmed absent.

Comments

Confirmation of Alcohol Exposure

- Confirmation of PAE requires confirmation that the biological mother consumed alcohol during the index pregnancy based on: reliable clinical observation; self-report; reports by a reliable source; medical records documenting positive BACs; alcohol treatment or other social, legal or medical problems related to drinking during the pregnancy.

Alcohol Threshold

- There is *confirmation* of PAE, with the *estimated* dose at a level known to be associated with neurobehavioral effects.

The Neurodevelopmental Assessment

- Read the long online appendix
- Much better detail and clarity than 2005 guidelines but too long to go into here

The Neurodevelopmental Assessment

A diagnosis of FASD is only made when there is evidence of severe and pervasive brain dysfunction, which is defined by impairment in 3 or more of ten specific domains

Domains

- Motor Skills
- Neuroanatomy/Neuro-physiology
- Cognition
- Language
- Academic Achievement
- Memory
- Attention
- Executive Function, including Impulse Control
- *Affect Regulation* (DSM-5 anxiety or mood disorder)
- Adaptive Behaivour, Social Skills, or Social Communication

Impairment

- Impairment is defined as a global score or major subdomain score on a standardized neurodevelopmental measure that is $\geq$ 2 sd below the mean (that is, at or below a standard score of 70 or about the 2.4%ile)

Impairment

- In some domains, large discrepancies among subdomains may be considered.
- In some domains, clinical assessment with converging evidence from multiple sources and DSM-5 diagnostic criteria may also be considered.

1. Motor Skills

- Composite or multiple subtest scores -2 SD on formal assessment of
  - -fine motor skills
  - -gross motor skills
  - -graphomotor skills
  - -visual-motor integration

Differences on neurological exam may be used in combination with scores.

2. Neuroanatomy/neurophysiology

-head circumference -2 SD

-seizure disorder not explained by postnatal causes

-imaging results associated with FASD by research

3. Cognition

- Composite or major subdomain score -2 SD on a standardized IQ test
- OR
- Large discrepancy among subdomains with a base rate <3% and the lower score -1 SD

4. Language

- Composite score -2 SD for
  - -core language
  - -receptive language
  - -expressive language

OR multiple scores -2 SD in higher-level language

OR discrepancy between RL and EL, with a base rate <3% and the lower score -1 SD

5. Academic Achievement

- Score -2 SD on standardized tests of reading, math or written expression OR large discrepancy between cognition and one of the above with a base rate <3% and the lower score -1 SD
- Not an instructional deficit

6. Memory

- Score -2 SD on a composite measure of overall memory, verbal memory, or visual memory
- OR large discrepancy between verbal and nonverbal memory with a base rate <3% and the lower score -1 SD
- Not working memory (that's EF)

7. Attention

- Multiple subtest scores -2 SD on direct neuropsych tests
- OR
- Converging clinical evidence of an attention deficit from multiple sources, including interviews, questionnaires, file review, and observation

8. Executive Function, including Impulse Control and Hyperactivity

- Multiple subtest scores -2 SD on direct neuropsych tests
- OR
- Converging clinical evidence of an attention deficit from multiple sources, including interviews, questionnaires, file review, and observation

9. Affect Regulation

- Meets DSM-5 criteria for a disorder of depression or anxiety
- Not a short term response to life circumstances

Adaptive behaviour, social skills, or social communication

- Composite score -2 SD in social language, social communication, or pragmatic language
- OR global composite or major subdomain -2 SD on adaptive rating scale
- OR (only if no suitable informant for adaptive scale) file review showing severe challenges with independent living and/or social competence

General considerations

- Use clinical judgment when scores are ambiguous.
- Make sure ratings are free of bias.
- Don't count the same score in 2 domains.
- Think pervasive disability, rather than completely independent domains.
- Rule out short term, reversible causes.
- There should be an overall pattern of severe and pervasive disability, based on multiple convergent sources of info.

Special Considerations for Infants and Young Children

At Risk category

The revision team really wanted to eliminate the 'age curve'; the difficulty in diagnosing younger kids. We did not find a technology that we thought could do that.

a. Scores seem to decline over time.
b. Some domains can't be tested or can't be tested well in young children.

What's the technology problem?

- Facial features alone are poor predictors of who will have neurodevelopmental disability later
- Neurodevelopmental tests tend to underpredict disability later
- The only consistent predictor is microcephaly plus all 3 facial features, but this is a very small group.

Caveat

- If a team uses the 'At Risk' description, the report should include a specific reason (most often young age) and a specific plan for reassessment

Microcephaly Exception

1. Infants and young children with sentinel facial features and microcephaly should be diagnosed with FASD with Sentinel Facial Features.
2. BUT microcephaly alone is not sufficient evidence of CNS dysfunction in a child old enough for a conclusive neurodevelopmental assessment.

1. Notice that a team *can* diagnose FASD in an infant who does meet the CNS criteria, either with or without facial features.
2. However, the At Risk designation is available for those who do not.

Special Considerations for Adults

CNS criteria adjusted slightly to remove barriers

- Mental health included as a domain – well supported by evidence but also done because of concern that MH symptoms were a barrier to diagnosis for adults.
- Adaptive behaviour "work-around" on the basis of file review for those without a good informant for a formal questionnaire.

Will the prevalence change?

- We never know until we try but the committee believes that these changes will remove barriers to diagnosis in adults but otherwise have a limited impact on prevalence.

Main change to assessment process is affect regulation

- Core team members unchanged (Physician, psychologist, in childhood SLP and OT)
- New domain: Affect regulation
- Screen + brief clinical interview model

AFFECT REGULATION

Domain - Affect Regulation

- Major Depressive Disorder, **Recurrent Episodes**
- Persistent Depressive Disorder (Dysthymia)
- Disruptive Mood Dysregulation Disorder (DMDD)

- Separation Anxiety Disorder
- Selective Mutism
- Social Anxiety Disorder
- Panic Disorder
- Agoraphobia
- Generalized Anxiety Disorder

Affect Regulation

- Does not include Unspecified Anxiety Disorder
- Does not include specific Phobia

FASD and Psychiatric Co-morbidities

- Anxiety Disorders (0-20%)
- Depressive Disorders (7-44%)

Famy et al (1998), Fryer et al (2007)

Consider Differential Diagnosis

- Oppositional Defiant Disorder
- Post Traumatic Stress Disorder
- Attachment Disorders
- Bipolar Disorder
- Obsessive Compulsive Disorder

Context

- Children and Adults being assessed may have experience significant difficulties due to abuse, neglect, trauma and instability
- Further complications such as alcohol/substance abuse, health issues/head injuries maybe confounding
- Complex psychosocial factors need to be considered and addressed as part of a client/family centered approach

Context

- Diagnostics should be contingent on clinical assessment and mental health evaluation
- Discern between relatively short term symptoms secondary to adverse environmental factors (ex/ unstable placements) versus persistent, recurrent issues with dysregulation

Evaluating for Anxiety/Depression

- Information gathered from outside reports/background information form
- General Screening Questions
- As clinically indicated – complete mental health assessment
- Standardized Interviews/semi-structured tools while "gold standard" but time consuming (K-SADS, ADIS). Shorter options: P-Chips, DAWBA. May improve diagnostic reliability and ensure comprehensive review of associated features

Clinical Implementation for assessing affect regulation

- Previous diagnoses from reliable source may be acceptable
- In complex presentations (ex/ multiple co-morbidities) independent mental health assessment may be warranted and subsequently proceed with FASD evaluation

Clinical Implications

- Clinicians must be prepared to enquire around suicidal ideation/plans particularly in cases of depressive disorder with appropriate management and follow through
- Youth identified with mental health conditions should be referred to appropriate local treatment resources/team

Updates on Other Diagnostic Systems

Hoyme et al (2016)

- Just out in Pediatrics
- Also multidisciplinary
- 4 categories:
  - FAS
  - PFAS
  - ARND
  - ARBD

Frustratingly different

- Laxer threshold for face: 2 features, 10th %ile
- Laxer threshold for Neurodevelopment: -1.5 SD vs. -2SD but fewer domains
- "With cognitive impairment" specifier if shown on direct rather than indirect tests
- Different neurodevelopmental criteria for different diagnoses

What's up with DSM-5?

- A very confusing situation
- In the back of the book, in Conditions for Further Study, there is a new category called Neurobehavioral Disorder Associated with Prenatal Alcohol Exposure (ND-PAE)

DSM Criteria

-Not explicitly multidisciplinary

-Not explicitly based on standardized testing and therefore no score cutoff discussed

-Requires more than minimal exposure to alcohol AND neurodevelopmental impairment

-Domains of impairment are defined and have to be multiple

In use or not?

- "Conditions for further study" are explicitly not supposed to be used clinically

BUT

In the front of the book, under Other Specified Neurodevelopmental Disorder (315.8), the example given is "Neurodevelopmental Disorder associated with PAE"

Hoyme et al (2016) on DSM-5 ND-PAE

"ARND is a complex medical diagnosis, best assigned as part of a multidisciplinary team assessment….vs. an experimental mental health diagnostic code, intended to be used by a variety of clinicians"

Most useful as a placeholder/treatment plan modifier??

What to do we need to move ahead?

- Field trials and predictive studies of the points on which the systems disagree
- Sensitivity and specificity numbers for different diagnostic decisions