Fetal Alcohol Spectrum Disorders: Screening, Diagnosis and Management

The 22nd Annual Childhood Development and Behavior Conference

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Faculty Disclosure Information

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Learning Objectives

1. Understand the epidemiology of FASDs
2. Review diagnostic criteria for FASDs
3. Learn how to screen children and families for an FASD
4. Become aware of evidence-based intervention strategies
Alcohol Consumption

• 2011-13 CDC Behavioral Risk Factor Surveillance System (BRFSS)
  – Pregnant women:
    • 10.2% report drinking alcohol in the past 30 days, including
    • 3.1% reporting binge drinking (≥4 drinks).
  – Non-pregnant women of childbearing age:
    • 54% report any alcohol use
    • 18% binge drinking
  – Among binge drinkers:
    • Pregnant women had a higher frequency of binge drinking, larger alcohol consumption
  – Alcohol use among pregnant women has not decreased significantly
Weighted Prevalence Estimates of Binge Drinking† Among Women aged 18-44 Years – BRFSS 2015

Percentage of women aged 18-44 years

- 8.8 - 14.2
- 14.5 - 17.3
- 17.5 - 19.3
- 19.7 - 32.9

Territories
- Guam
- Puerto Rico

CDC

Yale School of Medicine
Prevalence of FASDs

- Variable prevalence rates reported
- The most common *preventable* cause of intellectual disability and behavior problems
- CDC Fetal Alcohol Syndrome Surveillance Network:
  - 0.2-1.5 cases of FAS per 1000 live births
  - CDC estimates FASD rate at least 3 times the rate of FAS
- 86% of foster and adopted youth with FASDs go undiagnosed
- Cross-Sectional study
- 13,146 children in 4 regions of the US (52% boys, mean age 6.7 years, white maternal race, 79%)
  - Rocky Mountain
  - Midwestern
  - Southeastern
  - Pacific Southwestern
- Prevalence: 11.3-50/1000 FASD - most conservative estimate
Alcohol Exposure and Embryonic Development
Effects on Neurogenesis

- Neuronal Formation
- Migration
- Myelination
- Synaptogenesis
- Even in normal appearing brains, functioning impaired
Areas of Brain Affected By Prenatal Alcohol Exposure

Impulses and judgment, Executive functioning.

Passes information from the left brain (rules, logic) to the right brain (impulse, feelings) and vice versa.

Memory, learning and emotion

Appetite, emotions, temperature, and pain sensation

Coordination and movement

Spatial memory and behaviors like perseveration and the inability to switch modes, work toward goals, and predict outcomes, and the perception of time
### Spectrum of FASD

<table>
<thead>
<tr>
<th>Category</th>
<th>With confirmed exposure</th>
<th>Without confirmed exposure</th>
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<tbody>
<tr>
<td><strong>Fetal Alcohol Syndrome</strong></td>
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<tr>
<td><strong>Partial FAS</strong></td>
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<tr>
<td><strong>Alcohol-related birth defects (ARBD)</strong></td>
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<td><strong>Alcohol-related neurodevelopmental disorder (ARND)</strong></td>
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<tr>
<td><strong>Neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE)</strong></td>
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- **Confirmed Exposure to Alcohol**
- **Facial Anomalies**
- **Growth Retardation**
- **CNS Abnormalities**
- **Cognitive or Behavioral Impairment**
- **Birth Defects**

- **As Defined in DSM-5**
Changes to Diagnostic Schemes

1. Updated Guidelines in partnership with National Institute Alcohol Abuse and Alcoholism
2. Neurobehavioral Disorder – Prenatal Alcohol Exposure
3. Canadian Guidelines
FASD diagnostic algorithm.

H. Eugene Hoyme et al. Pediatrics 2016;138:e20154256
Definition of Prenatal Alcohol Exposure (PAE)

- Can include drinking levels reported by the mother 3 months before her report of pregnancy recognition or a positive pregnancy test.
- The information must be obtained from the biological mother or a reliable collateral source:
  - ≥6 drinks/week for ≥2 weeks during pregnancy
  - ≥3 drinks per occasion on ≥2 occasions during pregnancy
  - Documentation of alcohol-related social or legal problems before or during the index pregnancy
  - Documentation of intoxication during pregnancy by blood, breath, or urine alcohol content testing
  - Increased prenatal risk as assessed by a validated screening tool
Facial Dysmorphisms

- Short Palpebral fissures
- Shallow philtrum
- Thin Upper lip

- Most stable features over time
- Although one can see other anomalies, these are more inconsistent
Lip- Philtrum

FAS

Lip-Philtrum Guide 1
Lip/philtrum guide for the white population, incorporating a 45-degree view.

H. Eugene Hoyme et al. Pediatrics 2016;138:e20154256
“Railroad Track” Ear

“Hockey Stick” Palmar Crease
Neuropsychology exam

- Evidence of global impairment
  - Performance, Verbal or Spatial IQ
  - Children < 3, developmental delay
- Cognitive Deficit in at least 1 neurobehavioral domain > 1.5 SD
  - Executive functioning
  - Specific learning impairment
  - Memory of visual-spatial impairment
- Behavioral impairment WITHOUT cognitive impairment
  - Impairments of self-regulation (mood or behavioral regulation impairment,
    - Attention deficit
    - Impulse control
Alcohol Related Neurodevelopmental Disorder

Requires:

A. Documented prenatal alcohol exposure
B. Neurobehavioral impairment

• Diagnosis cannot be made definitively in children <3 y of age
Introduction of Neurobehavioral Disorder associated with Prenatal Alcohol Exposure as a new diagnostic category

Intended to replace Alcohol Related Neurodevelopmental Disorder (ARND)

Included as “Condition for Further Study”

Can be used as a specified condition under “Other Specified Neurodevelopmental Disorder”

ICD-9 code 315.8, ICD-10, F88

Can make this diagnosis in the presence or absence of facial/physical dysmorphia
Why Another Diagnosis??

• No specific mental health code that adequately documented the cognitive and mental health impact of PAE
• Existing diagnostic codes do not adequately capture mental health needs
• Unifies areas of deficit within one diagnostic category
• Attempt to improve reimbursement for evaluation and treatment (for providers and families)
Neurobehavioral Disorder - With Prenatal Alcohol Exposure

ND-PAE

- Neurocognitive Impairment
  - Global IQ
  - Impairment in:
    - Executive Functioning
    - Learning
    - Memory
    - Visual Spatial Reasoning

- Impairment in Self-Regulation
  - Impairment in mood or behavioral regulation
  - Attention problems
  - Impairment in impulse control

- Deficits in Adaptive Functioning
  - Communication Deficit
  - Social Deficit
  - Daily Living Skills Impairment
  - Motor Impairment
Updated Canadian Guidelines

- The use of fetal alcohol spectrum disorder (FASD) as a diagnostic term
- The inclusion of special considerations for diagnosing FASD in infants, young children and adults
- The deletion of growth as a diagnostic criterion
- The addition of a new “at-risk” category that will capture individuals who do not meet the diagnostic criteria but are still at risk of FASD
- The revision and refinement of brain domains evaluated in the neurodevelopmental assessment
Diagnostic algorithm for Fetal Alcohol Spectrum Disorder (FASD).

Jocelynn L. Cook et al. CMAJ 2016;188:191-197

SFF=Sentinel Facial Features
Why Make This Diagnosis?

- Waze directions
- Step by step directions
- Identifies hazards and upcoming roadblock
- Alternative routes
Potential Benefits of a Diagnosis

• Parental relief in the knowledge that the child’s problems have a biological basis
• Facilitates access to evidence-based interventions
• Avoids unnecessary additional testing and non-specific referrals or interventions
Risks of Not Accurately Identifying and Treating FASDs

**For the individual:**
- Unemployment (79%)
- Loss of family
- Homelessness
- Juvenile Justice (60%)
- Jail and arrest (50%)
- Premature death
- Increased substance abuse (35%)
- Mental Health problems (94%)
- School dropout (43%)

**For the family:**
- Loss of family
- Increased substance use
- Premature death
- Financial strain
- Emotional stress

So.. Why aren’t FASDs Diagnosed More?

- Parental shame, guilt, fear of stigma
- Child Abuse Prevention and Treatment Act (CAPTA)
  - Maternal alcohol use is not reportable in almost all states
  - FASDs are reportable to Child Protective Services
- State reporting laws vary
- Birth mother inaccessible to obtain an accurate history
- Parental fear of legal action
Screening

• When did you first know you were pregnant?

• In the 3 months before you knew you were pregnant, how many times did you have 4 or more drinks in a day?

• During the pregnancy, how many times did you have any alcohol?
Communicating with Families about Suspected FASD

- Affirm families for talking about PAE
- Use a strengths-based, functional approach
- Avoid blaming
- Don’t challenge resistance
- Leave the door open for future discussion
- Don’t provide false reassurance
- Talk it out with colleagues
When to Consider a FASD?

- History of maternal alcohol or drug use
- A sibling diagnosed with a FASD
- Developmental, cognitive, or behavioral concerns
- Complex medical concerns (e.g., cardiac)
- Growth deficits
- Dysmorphic facial characteristics associated with FAS are present
- Child is in foster care or history of complex trauma
Managing ND-PAE in the Primary Care Family Centered Medical Home

- **Medical Home**
  - Pediatrician
  - Care coordinator
  - Social worker
  - Education specialists
  - Rehabilitation specialists
  - Therapists
  - Child Protective Services

- **Consultants**
  - Developmental Pediatrician
  - Psychologist
  - Psychiatrist
  - Genetics
  - Neurologist
Monitoring Changes Across Time

Infancy
- Problems with regulation – sleep, eating, irritability
- Developmental Delay

Toddler/Preschool
- Problems with self-regulation; prolonged tantrums
- Emergence of more significant behavior problems
- Difficulty maintaining focus, emergence of hyperactivity

School Age
- Poor school performance
- Cognitive limitations
- Poor peer relations
- Academic achievement less than expected for intellectual level
- Mood lability

Adolescence
- Trouble with law
- Promiscuous behaviors
- School dropout
- Easily taken advantage of
FASD Interventions

- Despite promise of reported interventions, sample sizes small, not widely tested and not widely available
- Limits to the extent of improvement of each individual intervention
- Larger studies needed to identify predictors and mediators treatment response
DEB EVENSON AND JAN LUTKE'S
EIGHT MAGIC KEYS
PLANNING FOR STUDENTS WITH FETAL ALCOHOL SPECTRUM DISORDER

- **Concrete**
  - Talk in concrete terms
  - Avoid abstract language

- **Consistency**
  - Parents & educators use the same words & strategies

- **Repetition**
  - Re-teach many times to retain in long-term memory

- **Routine**
  - Help reduce anxiety

- **Specific**
  - Say exactly what you mean
  - Give step by step directions

- **Structure**
  - The glue that makes the world make sense... their foundation

- **Simplicity**

- **Supervision**
  - Scaffold independence

**KISS**
- Keep it short & sweet

Keith Winks 2017
Psychopharmacology

- Often require targeted intervention to address specific symptoms
- Children with FASDs often prescribed higher number of psychotropic drugs
- Limited evidence base
  - Few studies
  - Small N
  - Few prospective
- Some support for use of medications, particularly stimulants
- May have less favorable or atypical responses, sometimes resulting in higher doses prescribed
Changes in Practice

- Consider an FASD when children present with complex neurodevelopmental presentation
- Screen for prenatal alcohol use
- Establish a medical home for children with FASDs
- Learn about potential community resources for children with FASDs
- Develop a “monitoring checklist” of areas of potential problem to use at follow-up visits
- Implement services even in the absence of a diagnosis
Resources

- American Academy of Pediatrics Fetal Alcohol Spectrum Disorders Toolkit: [www.aap.org](http://www.aap.org)
- SAMHSA FASD Center for Excellence: [fasdcenter.samhsa.gov](http://fasdcenter.samhsa.gov)
- Centers for Disease Control and Prevention FAS Prevention Team: [www.cdc.gov/ncbddd/fas](http://www.cdc.gov/ncbddd/fas)
- National Organization on Fetal Alcohol Syndrome (NOFAS): [www.nofas.org](http://www.nofas.org)