FETAL ALCOHOL SPECTRUM DISORDERS: SCREENING, ASSESSMENT AND DIAGNOSIS

Presented by
Susan Buttross, M.D., FAAP
University of Mississippi Medical Center
Medical Director: Center for the Advancement of Youth
Disclosure Statement

- I have no financial disclosures related to this topic.
- I will not discuss off label drugs in this talk.
- I am an FASD Champion for AAP District VII
  (of course it is not a paid position ☹️)
The AAP and the Improving Fetal Alcohol Spectrum Disorders Prevention and Practice through National Partnerships collaborative thank the following experts for their help in developing this series of presentations:

- David Wargowski, MD, Kenneth L Jones, MD, Prachi Shah, MD, Yasmin Senturias, MD, Vincent C Smith, MD, MPH FAAP, H. Eugene Hoyme, MD, and Miguel Del Campo, MD (Screening, Assessment, and Diagnosis)
- Yasmin Senturias, MD, FAAP (ND-PAE)
- Yasmin Senturias, MD, FAAP, Vincent C Smith, MD MPH FAAP, and Prachi Shah, MD (Treatment Across the Lifespan for Person with FASD)

Funded by the National Center on Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention: U38OT0001, U84DD0011 & U84DD0010
Learning Objectives

1. Describe diagnostic criteria and approaches for conditions along the continuum of FASDs, including ARND, ND-PAE, FAS, pFAS, and ARBD.

2. Distinguish major physical and neurobehavioral features for differential diagnosis of FASDs from other behavioral and genetic disorders as well as relevant comorbidities.

3. Explain the importance of screening every patient for a history of prenatal alcohol exposure at birth and during their first visit to your practice and subsequent visits as appropriate.
FASD Terminology and Acronyms

- **PAE: Prenatal Alcohol Exposure**
- **FASD: Fetal Alcohol Spectrum Disorders**
  - Umbrella term for a range of effects that can result from prenatal alcohol exposure (not a diagnosis)
  - Encompasses a group of specific disorders (more on these later), including:
    - **FAS: Fetal Alcohol Syndrome**
      - The most widely known diagnosis in the spectrum
    - **ARND: Alcohol Related Neurodevelopmental Disorder**
      - Used in some dx schemes for individuals without physical characteristics
    - **ND-PAE: Neurobehavioral disorder associated with prenatal alcohol exposure.**
      - New category in DSM-5 Section III: Emerging Measures and Models
      - Defines more precisely the developmental and behavioral manifestations associated with PAE
The Umbrella of FASD

- Fetal Alcohol Syndrome (FAS)
- Alcohol Related Birth Defects (ARBD)
- Partial Fetal Alcohol Syndrome (pFAS)
- Alcohol Related Neurodevelopmental Disorder (ARND)
- Neurobehavioral Disorder-associated with Prenatal Alcohol Exposure (ND-PAE)
Fetal Alcohol Syndrome (FAS) & Partial FAS (pFAS) - Facial features, CNS dysfunction/anomalies, possible growth deficits

ARBD - Congenital anomalies only (rare)

ARND – Neurodevelopmental/behavioral effects without cardinal dysmorphic features (common)

ND-PAE – Neurodevelopmental/behavioral effects regardless of dysmorphic features (most likely common)
Prenatal Alcohol Exposure: Relevance to Pediatric Practice

- Most common *preventable* cause of intellectual disability and behavior problems – likely seen in most practices.
- Effects can be *lifelong*.
- Effect development and function more so than other drugs or teratogens.

"Of all the substances of abuse, including cocaine, heroin, and marijuana, alcohol produces by far the most serious neurobehavioral effects in the fetus."

- Institute of Medicine, 1996

- Can contribute to a range of growth deficits and structural anomalies (FASD)
When to Consider a FASD Diagnosis?

- Developmental, cognitive, or behavioral concerns
- Complex medical concerns (e.g., cardiac)
- Growth deficits
- History of maternal alcohol or drug use
- A sibling diagnosed with a FASD
- Dysmorphic facial characteristics associated with FAS are present
Areas of the Brain Affected By Prenatal Alcohol Exposure

- **Hypothalamus**: appetite, emotions, temperature, and pain sensation
- **Cerebellum**: coordination and movement
- **Basal Ganglia**: spatial memory, switching gears, working toward goals, predicting behavioral outcomes, and the perception of time
- **Corpus Callosum**: passes information from the left brain (rules, logic) to the right brain (impulse, feelings) and vice versa
- **Hippocampus**: memory, learning, emotion
- **Amygdala**: emotions
- **Frontal Lobes**: impulses and judgment; controls executive function

Source: Dr. Sarah Mattson, University of San Diego
General Intellectual Performance

IQ Scale Mattson et al., 1997

FSIQ  VIQ  PIQ

NC  PEA  FAS

Standard Score

IQ Scale

Mattson et al., 1997
FASD: Relevance to Pediatric Practice

High Prevalence

- Prevalence in a Midwestern city (May, 2014)
  - FAS: 6-9/1000 children
  - All FASD: 24-48 /1000 children (2.4% to 4.8%)

- Increased prevalence among children in child welfare (Lange, 2013)
  - FAS: 60/1000 children (6%)
  - All FASD: 169/1000 children (16.9%)
## FASD: Perspectives on Prevalence

<table>
<thead>
<tr>
<th>Birth defect</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down syndrome</td>
<td>1.2/1000 births</td>
</tr>
<tr>
<td>Cleft lip +/- palate</td>
<td>1.2/1000 births</td>
</tr>
<tr>
<td>Spina bifida</td>
<td>1/1000 births</td>
</tr>
<tr>
<td>Autism</td>
<td>12.5-14/1000*</td>
</tr>
<tr>
<td>Fetal Alcohol Syndrome (FAS)</td>
<td>6-9/1000*</td>
</tr>
<tr>
<td>All FASDs</td>
<td>24-48/1000* (May, 2014)</td>
</tr>
</tbody>
</table>

*per 1000 school age children
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
- Discussing the cause with a biological mother may reduce recurrence risk in future offspring
Why Pediatricians Do Not Routinely Screen for FASD

- Insufficient information and training

- STIGMA: Stereotypic views about mothers who might be exposing their baby to alcohol leading to prejudice, discrimination, and bias AND/OR stereotypic view about children with an FASD
Assessment Domains for Diagnosis

- Prenatal Exposure to Alcohol History
- CNS (structural, neurologic, functional)
- Growth
- Dysmorphic Facial Features
Exposure History in Context of Routine Care

- Obtaining a history of in utero exposure to alcohol before suspicion or recognition of problems is ideal.
- Introduce review of family, social, and pregnancy histories as part of assessment.
- Work through all potential exposures: any of parent concern, medications, environmental, recreational drugs, and alcohol.
- Present alcohol as commonly consumed before pregnancy is recognized and ask about that timeframe.
Exposure History in Context of Routine Care

- Use open-ended questions,
  EXAMPLE: “tell me about your alcohol use in the 3 months prior to finding out you were pregnant.”
  NOT: “You didn’t drink while pregnant – Did you?”

- Ask about partner’s drinking habits

- Provide assurance that to provide the best care possible it’s important to know all of the facts about the pregnancy
AAP’s Bright Futures, guidelines for health supervision of infants, children and adolescents, suggest three screening questions for the pediatric situation:

- How often do you drink beer, wine or liquor in your household?
- In the 3 months before you knew you were pregnant, how many times did you have 4 or more drinks in a day?
- During your pregnancy, how many times did you have 4 or more drinks in a day?

If a positive response is obtained additional questions about amount, frequency and timing may be appropriate for diagnostic purposes.

Suggested contact points:

- all prenatal visits,
- the earliest well child visits,
- all new patient visits,
- whenever a related concern is observed or raised.
Mothers want their pediatrician to value and like them. Be gentle, non-judgmental, ask, then listen to their story.

Stick to the facts, provide information and education on alcohol/drug use or dependency in a matter-of-fact manner.

Fear of judgment is one of the main reasons women may not disclose that they drank during pregnancy.

Remind her that you care about her child, HER and their family. Share relevant stories when possible.

Avoid words like “FASD Kid” rather use person-first language such as “child with an FASD”.
Best Definition of Cognition and Behavior in FASD: ND-PAE

- DSM-5 emerging diagnosis
- Criteria describes impairment in neurocognition, self-regulation and adaptive function.
- Diagnosis is made in the context of confirmed prenatal alcohol exposure.
- Criteria do not require the presence of physical features.
Neurobehavioral Effects

Neurocognitive deficits

- Low IQ or developmental delay
- Executive functioning deficits
- Impaired learning, memory or specific learning problems (esp. visual-spatial and math)
- Motor functioning delays for younger children
Neurobehavioral Effects

**Self-regulation problems**
- Self-soothing, sleep
- Difficulty managing mood
- Behavior management issues
- Attention problems (esp. shifting attention)
- Poor impulse control
Neurobehavioral Effects

**Difficulty learning/Delayed adaptive skills**

- Communication deficits, especially social communications such as understanding idioms or jokes
- Problems with social skills
- Problems with self care or daily living skills
- Motor issues in younger children
Physical Effects

- Weight and/or length growth deficiency (pre or post natal)
- Abnormal brain structures (esp. small cranium, corpus callosum)
- Dysmorphic facial features:
  - Short palpebral fissures
  - Smooth philtrum
  - Thin vermillion border
Currently available guidelines:
- Updated Clinical Guidelines for Diagnosing Fetal Alcohol Spectrum Disorders (Hoyme et al, Pediatrics, 2016)
- Canadian guidelines for diagnosis (Cook et al, CMAJ, 2015)
- National Task Force on FAS and FAE (2004)
- FASD 4-digit diagnostic code (Astley and Clarren, Alcohol, 2000)

Historically available guidelines:
- Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis (Chudley et al, CMAJ 2005)
- Fetal Alcohol Syndrome (The Lancet, 1973)

The Diagnostic and Statistical Manual version 5 published by the American Psychiatric Association also proposes criteria for neurobehavioral disorder associated with prenatal alcohol exposure.
### Spectrum of FASD

**FAS**
- FAS with confirmed maternal exposure
- FAS w/o confirmed maternal exposure

**PFAS**
- Partial FAS with or w/o confirmed exposure

**ARBD**
- Alcohol-related birth defects (ARBD)
- Alcohol-related neurodevelopmental disorder (ARND)

**ARND**
- Neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE)

<table>
<thead>
<tr>
<th>FAS with confirmed maternal exposure</th>
<th>OR</th>
<th>OR</th>
<th>OR</th>
</tr>
</thead>
<tbody>
<tr>
<td>FAS w/out confirmed maternal exposure</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Partial FAS with or w/o confirmed exposure</td>
<td>+/-</td>
<td>OR</td>
<td>OR</td>
</tr>
<tr>
<td>Alcohol-related birth defects (ARBD)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Alcohol-related neurodevelopmental disorder (ARND)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE)</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

---

Adapted from Neuroscience and Biobehavioral Reviews (2007): 31:230-238
PEDIATRICS Vol. 106 No. 2 August 2000
Record Review & History

- Exposure History
- Birth records (weight, length, head circumference)
- Medical history/records (birth defects?, exposures?)
- Postnatal growth records
- Developmental/behavioral history
- Psychological testing, including cognitive and behavioral assessments
The AAP FASD Toolkit (www.aap.org/fasd) is a comprehensive resource for identification, diagnosis and medical home management on patients with a FASD.

Toolkit includes information on common diagnostic approaches and tools, a flow diagram for evaluation of FASDs, and guidelines for referral and diagnosis.
Potential Differential & Comorbid Diagnoses

Behavioral disorders examples

- ADHD
- Intellectual disability
- Early Trauma
- Conduct disorder/Oppositional defiant disorder
- Parenting issues
Potential Differential & Comorbid Diagnoses

Genetic and growth disorders examples

- Williams syndrome
- Dubowitz syndrome
- Fetal valproate syndrome
- Maternal PKU fetal effects
- Nutritional insufficiency – growth
- Prenatal smoking - growth
Delivering the Diagnosis

- Approach should be similar to delivering any diagnoses that may have significant impact on a child’s future.

- Sensitivity to the possibility of stigma is critical, particularly when the biological mother is involved.

- Explanation of how the diagnosis leads to treatment options and better parent-child interactions is helpful.

- Only the FAS diagnosis can be made without confirmed in utero exposure to alcohol – for other dx’s revisit as appropriate.
Diagnosis Established

What now?

- Counsel re: Natural history risk of neurodevelopmental manifestations over time, need to intervene to avoid secondary conditions.
- AAP toolkit: www.aap.org/fasd
Referrals

- **For diagnosis:** If uncertain whether findings satisfy criteria, depending on available resources:
  - FASD diagnostic clinic
  - Genetics and dysmorphology clinic
  - Neurodevelopmental/behavioral pediatrician
  - Neuropsychologist or behavioral psychologists for ND-PAE or ARND

- **For treatment/care needs and care planning or management:**
  - Neuropsychologist, clinical psychologist, school psychologist, early intervention
  - SpL, OTR or PT as indicated
  - Social work
  - Medical specialists as indicated (e.g., Otorhinolaryngologist to address frequent ear infections)
# Interprofessional Care for Patients with FASD

## Health Care is Changing

- Move towards outcomes-based, value-based and **team-based** care.
- Reimbursement with the Medicare Access & CHIP Reauthorization Act of 2015 (MACRA) will support values-based care and reimburse **team-based** care.

More information on potential team members is available in the AAP FASD Toolkit

## Interprofessional Care

- Create and develop a team from different specialties to collaborate in planning & care.
- Establish a common goal
- Care should be interdependent, complimentary, coordinated.
- The patient and family should be considered team members if practicable.
American Academy of Pediatrics (AAP): www.aap.org/fasd

Centers for Disease Control and Prevention (CDC): www.cdc.gov/ncbddd/fasd

National Organization on Fetal Alcohol Syndrome: www.nofas.org

National Institute on Alcohol Abuse and Alcoholism: www.niaaa.nih.gov
FASD are more common than recognized. You most likely have children in your practice with a FASD,

Obtaining history of prenatal exposure to alcohol is good practice and should be routine for all patients,

Children will most likely present with neurodevelopmental/neurobehavioral problems,

But a comprehensive physical and behavior assessment is best to establish the appropriate diagnosis and care plan
The primary facial dysmorphic features associated with FAS are:
- Short palpebral fissures, smooth philtrum, thin upper lip.

The diagnosis of ND-PAE (DSM-5) requires confirmed alcohol exposure and
- Neurocognitive impairment, self-regulation impairment and deficits in adaptive functioning.

Traditional behavioral therapies may not work best for children with an FASD.
The following approaches are recommended for children diagnosed with an FASD:

- Regularly scheduled follow-up in the medical home
- Evaluation by a psychologist to assess neurocognitive functioning, self-regulation, and adaptive functioning
- Medication management for co-occurring conditions if necessary
References

- Hagen, et al., *Pediatrics* 2016, 138 (4) e20151553; DOI: 10.1542/peds.2015-1553
- Hoyme et al., *Pediatrics* 2016, 138 (2) e20154256; DOI: 10.1542/peds.2015-4256
- May et al., *Pediatrics* 2014;134(5);855-66
- Lange et al., *Pediatrics* 2013;132(4):e980-95
- Tan, Denny, Cheal, Sniezek & Kanny, 2015: [http://www.cdc.gov/mmwr/preview/mmwrhtml/mm6437a3.htm](http://www.cdc.gov/mmwr/preview/mmwrhtml/mm6437a3.htm)
- Wisconsin statute 146.0257: [http://docs.legis.wisconsin.gov/statutes/statutes/146/0257](http://docs.legis.wisconsin.gov/statutes/statutes/146/0257)
Questions?