

WI CAN Educational Series  
August 18, 2017

FETAL ALCOHOL SPECTRUM  
DISORDERS: SCREENING,  
ASSESSMENT AND DIAGNOSIS

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Disclosure Information for:  
David Wargowski, MD  
FASD Screening, Assessment and Diagnosis

- There are no relevant financial relationships related to this presentation/program
- There is no sponsorship/commercial support of this presentation/program
- The content being presented will be fair, well-balanced and evidence-based
- Learners who wish to receive Continuing Education Credit (CME/CLE/CE) must complete and turn in evaluations to successfully complete this program

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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 genetic and behavioral 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 clinic.



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## 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

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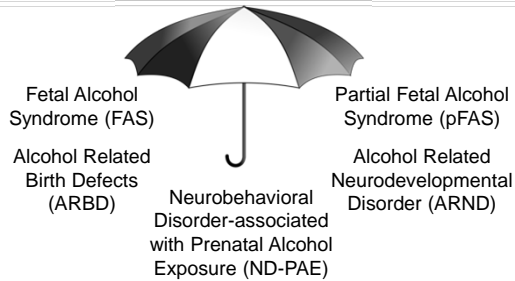
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## The Umbrella of FASD




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## FASDS – Brief Descriptions

### **Fetal Alcohol Syndrome (FAS) and Partial FAS (PFAS)**

- Facial features and CNS dysfunction or anomalies, with or without growth deficits

**ARBD** - Congenital anomalies without neurodevelopmental-behavioral effects (rare)

**ARND** - Neurodevelopmental-behavioral effects without major dysmorphic features (common)

**ND-PAE** - Neurodevelopmental-behavioral effects regardless of dysmorphic features; for use by behavioral health providers




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### Prenatal Alcohol Exposure: Relevance to Public Health

- Most common *preventable* cause of intellectual disability and behavior problems – likely seen in most practices.
- Effects can be *lifelong*.
- Affects 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)

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### Areas of the Brain Affected By Prenatal Alcohol Exposure

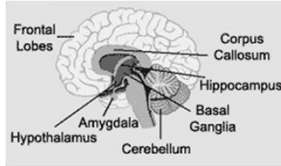
**Frontal Lobes** – impulses and judgment; controls executive function

**Hypothalamus** - appetite, emotions, temperature, and pain sensation

**Amygdala** - emotions

**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

Source: Dr. Sarah Mattson, University of San Diego

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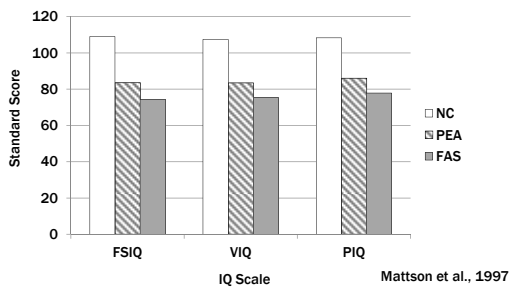
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### General Intellectual Performance




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**FASD: Relevance to Public Health**  
*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%)

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**FASD:**  
**Perspectives on Prevalence**

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

\*per 1000 school age children

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*Clues that a child may have an FASD*

- Developmental, cognitive, or behavioral concerns
- Growth deficits
- History of maternal alcohol or drug use
- A sibling diagnosed with a FASD
- Dysmorphic facial characteristics or other birth defects associated with FAS

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*Clues that a child may have an FASD*

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- Risk factors:
  - Other exposures associated with alcohol use (eg, tobacco, marijuana, cocaine, methamphetamine)
  - Adoption, particularly from endemic region (eg, Russia, Eastern Europe)
  - Foster care
- Parental concern

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Potential Benefits of a Diagnosis

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- Parental relief in the knowledge that the child's problems have a biological basis
- Facilitates access to evidence-based interventions which improve outcomes
- Avoids unnecessary additional testing and non-specific referrals or interventions

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Potential Benefits of a Diagnosis

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- Access to professional expertise, a range of medical, developmental, community and financial resources, and supportive counseling
- Discussing the cause with a biological mother may reduce recurrence risk in future offspring

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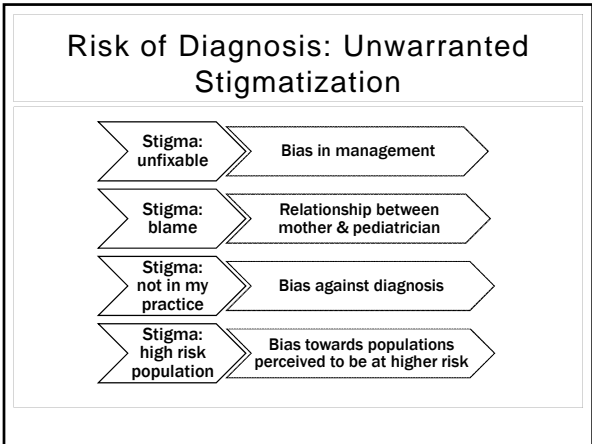
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### Who Drinks During Pregnancy?

- Women who drink during pregnancy come from all social, economic and ethnic groups
- Nationally, 1 in 10 women report alcohol use during pregnancy
- Many pregnancies are not recognized until the period of risk to alcohol exposure is well established

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### Who Drinks During Pregnancy?

- Among pregnant women, the highest prevalence of reported alcohol use was among those who were:
  - Aged 35-44 years (18.6%)
  - College graduates (13.0%)
  - Unmarried (4.6 x married)
    - Tan, Denny, Cheal, Sniezek & Kanny, 2015

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**Question Break**

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**Neurobehavioral Effects**

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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



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**Neurobehavioral Effects**

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Self-regulation problems

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



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## 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




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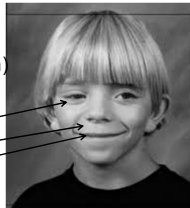
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## Physical Effects

- 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 vermilion border



PEDIATRICS Vol. 115 No. 1 January 2005

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## Available FASD Diagnostic Guidelines

### 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)
- CDC National Task Force on FAS and FAE (2004)
- FASD 4-digit diagnostic code (Astley and Clarren, *Alcohol*, 2000)

### Historically available guidelines:

- A practical clinical approach to diagnosis of fetal alcohol spectrum disorders: clarification of the 1996 Institute of Medicine criteria (Hoyme et al, *Pediatrics*, 2005)
- Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis (Chudley et al, *CMAJ*) 2005

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

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**2016 Revised IOM Criteria: FAS**

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I. FAS (requires all of A–D)

A. **Minor facial anomalies**, including  $\geq 2$  of the following:

1. Short palpebral fissures ( $\leq 10$ th percentile)
2. Thin vermilion border of the upper lip (score 4 or 5 on a racially normed lip/philtrum guide)
3. Smooth philtrum (score 4 or 5 on a racially normed lip/philtrum guide)

B. **Prenatal and/or postnatal growth retardation**  
Height or weight  $\leq 10$ th percentile, corrected for racial norms, if possible

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**2016 Revised IOM Criteria: FAS  
(cont)**

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C. **Deficient brain growth, abnormal morphogenesis, or abnormal neurophysiology**, including 1 of the following:

1. Structural brain abnormalities
2. Head circumference  $\leq 10$ th percentile
3. Recurrent nonfebrile seizures

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**2016 Revised IOM Criteria: FAS  
(cont)**

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I. FAS (requires all of A–D) (cont.)

D. **Neurobehavioral Impairment**

- **< 3 years:** Evidence of developmental delay  $\geq 1.5$  SD below the mean
- **$\geq 3$  years:** General conceptual ability, verbal or performance IQ  $\geq 1.5$  SD below the mean, OR
- Cognitive deficit  $\geq 1.5$  SD below the mean in executive function, learning, memory, or visual-spatial domains, OR
- Behavior deficit  $\geq 1.5$  SD below the mean in self-regulation (mood, attention, impulse control)

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**2016 Revised IOM Criteria:  
Partial FAS**

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**II. Partial FAS**  
(requires A and B and either C or D)

- A. Dysmorphic facial features (as for FAS)
- B. Neurobehavioral impairment (as for FAS)
- C. Confirmed prenatal alcohol exposure
- D. Growth impairment (as for FAS)

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**2016 Revised IOM Criteria:  
ARND**

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**III. Alcohol-related Neurodevelopmental Disorder:**

- A. Documented prenatal alcohol exposure, and
- B. Neurobehavioral impairment (as in FAS)

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**Revised IOM Criteria: ARBD**

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**IV. Alcohol-related birth defects**  
Documented prenatal alcohol exposure and one or more of the following congenital anomalies:

cardiac: atrial septal defects, aberrant great vessels, ventricular septal defects, conotruncal heart defects;  
skeletal: radioulnar synostosis, vertebral segmentation defects, large joint contractures, scoliosis;  
renal: aplastic/hypoplastic/dysplastic kidneys, "horseshoe" kidneys/ureteral duplications;  
eyes: strabismus, ptosis, retinal vascular anomalies, optic nerve hypoplasia;  
ears: conductive or neurosensory hearing loss;

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### Spectrum of FASD

FAS	<ul style="list-style-type: none"> <li>FAS with confirmed maternal exposure</li> <li>FAS w/out confirmed maternal exposure</li> </ul>					
PFAS	Partial FAS with or w/o confirmed exposure	+/-	OR	OR	OR	
ARBD	Alcohol-related birth defects (ARBD)					
ARND	Alcohol-related neurodevelopmental disorder (ARND)			OR		
ND-PAE	Neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE)				As Defined in DSM-5	

**A** Confirmed Exposure to Alcohol    **B** Facial Anomalies    **D** Growth Retardation    **E** CNS Abnormalities    **F** Birth Defects  
**C** Cognitive Abnormalities

Adapted from Neuroscience and Biobehavioral Reviews (2007); 31:218-238  
 PEDIATRICS Vol. 106 No. 2 August 2000

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### Steps Toward Diagnosis: Record Review/History

- History of prenatal alcohol exposure
- Birth records (weight, length, head circumference)
- Medical history/records (birth defects?)
- Postnatal growth records
- Developmental/behavioral history
- Psychological testing, including cognitive and behavioral assessments

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### The Diagnostic Assessment: Exposure history

- Awareness/acknowledgement of stigma, bias, and potential impact of questions on provider/parent relationship
- Approach with empathy
- Find common ground
- Work toward details (eg, number of standard drinks, timing during pregnancy, pattern of drinking)

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### Exposure History in Context

- Screening all children for PAE before suspicion or recognition of problems is ideal
- Introduce review of family, social, and pregnancy histories as part of assessment
- Work up through general exposures, any that caused concern, medications, recreational drugs, then alcohol
- When true, introduce alcohol as commonly consumed before pregnancy is recognized
- Assurance that purpose is not to blame but to help child

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### AAP Bright Futures

- 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.

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### AAP Bright Futures

- Suggested contact points:
  - All prenatal visits
  - The earliest well child visits
  - All new patient visits
  - Whenever a related concern is observed or raised




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## Question Break

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## Physical Assessment

- Growth measurements
- Palpebral fissure length
- Eye/vision
- Midface recession/hypoplasia
- Philtrum, lip (lip/philtrum guide)
- Palate
- Heart
- Elbows, digits
- Examination for other minor anomalies characteristic of FAS, and those that could lead to the suspicion of another genetic syndrome.

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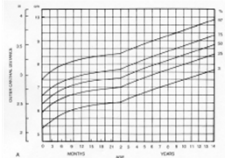


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### Correct and Objective Measurements are Key: Palpebral Fissure Length PFL

- Measure from inner to outer canthus following the angle of the face
- Plot measurement on graphs available (i.e. in Smith's *Recognizable Pattern of Human Malformation*)
- Short PFL defined as  $\leq 10^{\text{th}}$  centile



Adapted from Smith's Recognizable Patterns

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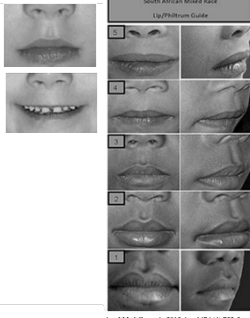
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### Lip and Philtrum Assessment

- Ensure the patient has a neutral expression
  - Smiling will falsely increase the score
- Place guide alongside face
- Score lip and philtrum separately
  - 4 or 5 is considered positive



Am J Med Genet A. 2015 Apr;167(A4):793-4

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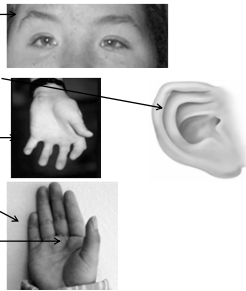
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### OTHER CHARACTERISTIC FEATURES OF FASD

- Ptosis
- "Railroad track" ear (prominent helical crus)
- Finger contractures
- "Hockey stick" palmar crease
- Limited elbow pronation/supination
- Heart murmur/defect



Fetal Alcohol Spectrum Disorders: Extending the Range of Structural Defects Am J Med Genet A. 2010 Nov;152A(11):2731-5

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### Neurobehavioral Assessment

- Developmental screening and routine behavioral history *may* be sufficient if it shows clear cognitive and behavioral or global deficits
- Most will require referral for psychological assessment of neurocognition, self regulation and adaptive function
  - Template referral letters available
  - Request that the psychologist use tests that will examine the affected domains in ND-PAE
- Review data – do deficits fit the diagnosis?
  - Data review checklist available

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### Potential Differential & Comorbid Diagnoses

#### Behavioral disorders examples

- ADHD
- Intellectual disability
- Early Trauma
- Conduct disorder/Oppositional defiant disorder
- Parenting issues

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### 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

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### PATHWAY TO A DIAGNOSIS

- The AAP FASD Toolkit ([www.aap.org/fasd](http://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.



#### Toolkit

The Fetal Alcohol Spectrum Disorders (FASD) Toolkit was developed to raise awareness, promote surveillance and screening, and ensure that all affected children receive appropriate and timely interventions.




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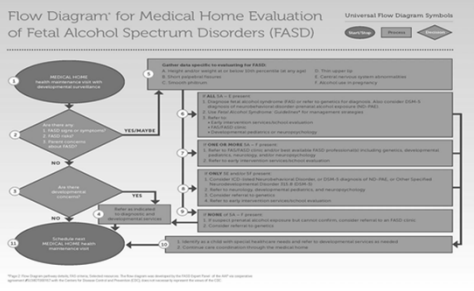
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## Establishing a Diagnosis




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## Indications for Referral

- For diagnosis: If uncertain whether findings satisfy criteria, depending on available resources:
  - FASD diagnostic clinic
  - Genetics and dysmorphology clinic
  - Neurodevelopmental/behavioral pediatrician (gaining expertise in this area)
  - Neuropsychologist or behavioral psychologists who can work through the ND-PAE route
- For determination of needs for education and behavioral management:
  - Neuropsychologist, clinical psychologist, school psychologist as available

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## Delivering the Diagnosis

- Approach should be similar to delivering other diagnoses of disorders that have significant impact on a child's future.
- Be sensitive to the possibility of stigma, particularly when the biological mother is involved.
- Only in the most clear-cut cases of FAS should the diagnosis be made despite the mother's adamant denial of alcohol use during pregnancy.
- Reference to intent/benefit of diagnosis (to enhance services) is helpful.

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**Diagnosis Established**  
*What now?*

- Counsel re: Natural history (risk of neurodevelopmental manifestations over time)
- Medical Home:  
<http://www.aafp.org/practice-management/transformation/pcmh.html>
- AAP toolkit: [www.aap.org/fasd](http://www.aap.org/fasd)

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**Take Home Messages**

- FASD are more common than recognized.
- Obtaining history of prenatal exposure to alcohol is good pediatric practice and should be routine for all patients
- Children will most likely present with neurodevelopmental/neurobehavioral problems
- A comprehensive physical and behavior assessment is best to establish the appropriate diagnosis and care plan

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**Resources**

- Centers for Disease Control and Prevention (CDC):  
[www.cdc.gov/ncbddd/fasd/index.ht](http://www.cdc.gov/ncbddd/fasd/index.ht)
- National Institute on Alcohol Abuse and Alcoholism: [www.niaaa.nih.gov](http://www.niaaa.nih.gov)
- American Academy of Pediatrics (AAP):  
[www.aap.org/fasd](http://www.aap.org/fasd)
- National Organization on Fetal Alcohol Syndrome: [www.nofas.org](http://www.nofas.org)

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### References

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