Fetal Alcohol Spectrum Disorder: From Screening to Assessment

Objectives

- Describe how to screen for prenatal alcohol exposure during pregnancy
- Describe how to screen for FASD in children
- Identify the key components of an FASD diagnostic assessment.
- Describe how to develop a system-wide plan to incorporate screening into the standard of care.

What are MN women hearing about alcohol during pregnancy?

- Wilder Foundation conducted a statewide survey with a representative sample of women age 18 through 44 who live throughout Minnesota
- 352 women completed the surveys
- Goal: Help MOFAS better understand women’s attitudes and perceptions toward alcohol consumption during pregnancy.

Wilder Study Findings

Minnesota women are still getting mixed messages about alcohol use during pregnancy from their doctors and health care providers.
- 63% of women reported their doctor told them not to drink alcohol at all while pregnant
- 18% said their doctor asked them about drinking, but did not warn them, because they do not drink
- 11% said their doctor told them to drink only lightly, or in moderation
- 7% said their doctor did not mention alcohol to them at all

Mixed Messages

Collectively, nearly 1 in 5 women did not receive any messages at all about alcohol use, or were told they could drink lightly, or in moderation

How much alcohol does it take?

It depends on many factors:
- Amount of alcohol
- Timing of drinking during pregnancy
- Nutrition of the woman
- Woman’s ability to metabolize alcohol
- Resiliency of the fetus
- Parity – the number of times the woman has given birth
FASD crosses all cultures, races and socio-economic levels
If you’re not looking for it, you won’t see it
– Women from all backgrounds can struggle with alcohol consumption during pregnancy
– There is no “typical” case of prenatal alcohol exposure
Provider bias
– It can be easy to assume a woman knows not to drink during pregnancy
– It can be easy to assume we can tell when a woman is drinking during pregnancy

1. Screening Tool
Don’t ask a simple Yes/No question.
Have a discussion.
• How often do you drink?
• How much do you drink at a time?
• When was your last drink?

Why Ask at Every Visit?
• She may not be ready to share that information at the first visit
• She may not be drinking at the first visit but ends up drinking later in pregnancy
• Each time is an opportunity for the patient to talk about it and for her to hear the message of the dangers of drinking during pregnancy

Two Main Elements of the Prenatal Screening
1. Incorporate screening all pregnant patients for alcohol use into the standard of care
2. Create a plan of action to have in place when a woman needs more support to not drink during pregnancy

1. Screening Tool
• Ask about the woman’s alcohol use before she knew she was pregnant
• Ask about current alcohol use since she found out she was pregnant
• Ask every woman every time
• Respect and support, not guilt or shame
• Regardless of her answer, give an education message and a brochure
2. Action Plan for your clinic

- Create an action plan for your location that clearly illustrates the protocol to be followed if a woman is using alcohol during pregnancy.

- Be prepared to provide a different source of support if the original referral was unsuccessful.
2. Individualized action plans

Consider creating individualized action plans for women using alcohol during pregnancy

- Each woman is in a unique situation
- Each woman has strengths that she can utilize during this time period

2. Action Plan

- Support for Sobriety
- Who is in the woman’s social circle that will support her?
  - Partner
  - Family
  - Friends
  - Groups

Educational Tools

- Having physical tools to show women can help generate conversation
- Keep tools in exam rooms so that they are readily available

Fetal Development Wheel
Screening for Alcohol Use During Pregnancy: Four Different Scenarios

http://www.youtube.com/watch?v=qlwqTQh_Hls

Pediatric Screening

Needs Assessment

- Minnesota Institute of Public Health (MIPH) conducted a needs assessment in 2010
- Interviewed Clinic and Nurse Managers of 15 family practice or pediatric clinics (76 clinics were contacted)
- Key informant interview on the phone, 13 questions

Needs Assessment Findings

Reason(s) for Not Screening for Developmental Delays in Children Related to Prenatal Alcohol Exposure
1. Administration has never discussed/decided on criteria
2. I don’t really think it’s a problem with children who are seen within our clinic
3. Not enough time

Top 3 answers from 11 different options
American Academy of Pediatrics Algorithm

- AAP FASD Toolkit online April 2013
- The FASD Toolkit was developed to raise awareness, promote surveillance and screening, and ensure that all affected children receive appropriate and timely interventions.
- This comprehensive toolkit serves as the framework for the medical home management of a child with an FASD in the medical home.
- This toolkit was developed by FASD Expert Panel members under the auspices of a 5-year cooperative agreement with the CDC’s National Center for Birth Defects and Developmental Disabilities.

Early consensus on screening

FASD Diagnostic Assessment
- Facial features
- Growth deficiencies
- Prenatal alcohol exposure
- Cognitive Functioning

Are not common, do not rely on these indicators

Focused on screening for these indicators in primary care

Asking about prenatal alcohol exposure

- Who should do the screening? Nurses? Physicians?
  - All clinics decided the nurses would do the screening
Asking about prenatal alcohol exposure

- How should PAE be assessed?
  - In person? On paper?
  - All asked in person except one clinic that used a paper form
  - All agreed that “maybe” answers were coded as positive PAE (“alcohol may have been consumed”)
  - The screening is done only once, not at every visit. At successive well child visits the EMR shows that the screening has or has not been completed

Examples of question on paper

Screening for Prenatal Exposure to Alcohol

We are committed to the health and wellness of your child and are asking the following questions. This information will be used to make the appropriate referrals to alcohol during pregnancy so that we can provide developmentally sensitive for early interventions.

Do you know if your child was exposed to any alcohol BEFORE pregnancy was known?

<table>
<thead>
<tr>
<th>Yes</th>
<th>No, Do Not Know</th>
<th>May have been consumed</th>
<th>Know for sure NO alcohol was consumed</th>
</tr>
</thead>
</table>

Do you know if your child was exposed to any alcohol AFTER pregnancy was confirmed?

<table>
<thead>
<tr>
<th>Yes</th>
<th>No, Do Not Know</th>
<th>May have been consumed</th>
<th>Know for sure NO alcohol was consumed</th>
</tr>
</thead>
</table>

Person completing this form: ____________________________________ Date: ____________

Your relationship to child being seen today: __________________________________________

(Person print your name)

Example from Minneapolis Public Schools

Were there any problems during labor and delivery? 

*If___ Yes ___ No ___ May have been problem with least skin color ___ Breast ___ Tension ___ Back ___ Others: __________________________

Did your child need medical care following birth? 

*If___ Yes ___ No ___ May have been problems with prenatal care ___ Heart ___ Apnea ___ Other: __________________________

IMMUNIZATIONS: Please bring a copy of your child’s immunizations at the time of your child’s screening appointment. 

My child is up to date with his/her immunizations. 

My child is not up to date with his/her immunizations. 

I don’t know if my child needs immunizations. 

I am a pediatrics/obstetrician/gynecologist.

Example from Minneapolis Public Schools

How the information is pieced together

- The provider is responsible for connecting the dots between PAE and cognitive delays to make a referral for an FASD diagnostic assessment
- Children with PAE but no cognitive delays were put on positive monitor status
- One clinic had the provider’s nurse identify which children had PAE during pre-visit planning and bring it to the provider’s attention before the clinic visit.

Connect the dots

Positive PAE + Elevated ASQ = FASD Diagnostic Referral

Once the information about prenatal alcohol exposure is recorded in the EMR it stays in the child’s chart. If developmental delays are detected at any time, the provider can connect the dots and make a referral for an FASD diagnostic assessment.
How the information is pieced together

- Nurse asks question
- Documents in chart
- Prenatal Alcohol Exposure
- Developmental Delay
- Scored ASQ-SE reveals developmental delays
- Pediatrician sees both are present in chart, makes referral
- FASD Diagnostic Assessment

Recording the information

- How is the information recorded in the Electronic Medical Record?
  - Where is it located?
  - What do we call it?
  - How do we find it for future visits?

This is specific to each EMR system and clinic.

Recording the information

- What information is shared with parents/caregivers? How is it worded?
  - Some, but not all, of the clinics put the information on the After Visit Summary
  - Some, but not all, of the clinics put the information on the problem list in a way that wasn’t shown to the parent at every visit.

Examples from clinics

- A clinic found that reports that clinical informatics were running were pulling the information in a way that made the system think that the child was drinking; not the mother during the pregnancy with the child
- A clinic changed their wording to say “FASD screening done” because “FASD screening-positive” was misinterpreted by a parent as a positive diagnosis of FAS
- A clinic was able to find a way to transfer information about the mother’s alcohol use during pregnancy from the mother’s chart into the baby’s chart using the Stork Report

Fetal Alcohol Spectrum Disorders

- are related congenital conditions caused by exposure to alcohol and its metabolites during fetal life.
- involve multiple organ systems, especially the brain and central nervous system.
- presentation both physical and behavioral.
Fetal Alcohol Spectrum Disorders

- are **chronic medical** conditions
- can lead to **permanent morbidity, disability, and mortality**.
- early diagnosis and **treatment** can prevent sequela.

Congenital Anomalies can include:

- macro/microscopic brain defects
- growth defects
- dysmorphic facial and other features
- other organ system defects.

White Matter Abnormalities in FASD

- Alcohol also interferes with myelination
  - Delayed
  - Abnormal patterns
- Interferes with myelin basic protein expression and oligodendrocyte activity

FASD Clinical Presentation

Figure 9. fMRI time-series from one FASD subject illustrating low correlation between BOLD signal change in right and left medial orbital frontal cortex.

Figure 9. fMRI time-series from one control subject illustrating high correlation between BOLD signal change in right and left medial orbital frontal cortex.
### Diagnosis and Screening

- **Screening**
  
  discerns those likely to have a condition in a given population, through a validated test

- **Diagnosis**
  
  determines if a condition is or is not present in a given individual

### Diagnostic Process

- **Screening and Referral**

- **Physical Exam and Differential Diagnosis**

- **Neuropsychology and Rehabilitation Testing**

- **Treatment/Recommendations/Referrals**

- **Follow-up**

### The FASD Diagnosis Evaluation Team

At minimum: experienced

- physician or nurse practitioner
- psychologist

Preferred:

- pediatrician or pediatric nurse practitioner
- pediatric neuropsychologist or pediatric psychologist
- pediatric occupational therapist
- pediatric speech and language therapist

Before and after the diagnosis evaluation:

- clinic nurse and/or care coordinator
- support person to family
- social worker, case aide, home health worker, able family member

### Health History

#### Prenatal Alcohol Exposure History

- Alcohol exposure during pregnancy?
- How much?
- How often?

- Who gave the exposure history?
- Note any past documentation.

### Physical Exam

#### Patient Check-in

- Growth including height, weight, OFC
- Vision and hearing screening
- Photo of face
- Vital signs

#### Complete physical exam

- Emphasize Neurology Exam
- Measure facial features (eyes, philtrum, lip)
- Consider lab testing: Chromosomes/CGH/Fragile X, TSH cascade, CBC/diff, iron studies, zinc, Vitamin D level, Infectious disease
Significant FASD Clinical Features

- **Growth impairment**
  - Shorter stature (at or below 10th percentile)
  - Lower weight (at or below 10th percentile)

- **Dysmorphic facial features**
  - Short palpebral fissure (length)
  - Hypoplastic nasolabial philtrum (degree of prominence: flatness)
  - Hypoplastic upper lip (degree of circularity: thinness)

- **Microcephaly**
  - OFC < 10th percentile

- **Central Nervous System (CNS) Abnormality**
  - Neurologic (i.e. Seizures)
  - Structural (i.e. MRI)
  - Functional (evidenced on neuropsychology testing)

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Growth

- Centers for Disease Control (CDC), WHO and CDC Growth Charts
- Get most recent charts at: [www.cdc.gov](http://www.cdc.gov)
- Use charts that range from 3-97th percentile
- WHO 0-2yrs
- CDC 2-20 yrs

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Lip/Philtrum Guides

- Reprinted with permission, Susan Astley, Ph.D.

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Rating of Lip and Philtrum


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FASD: eye measurements

<table>
<thead>
<tr>
<th>Palpebral Fissure Length (PFL) Z-score Calculator</th>
<th>January 1, 1960</th>
<th>January 1, 2000</th>
<th>January 1, 2020</th>
<th>January 1, 2028</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ethnicity, Birth Date</td>
<td>OFC, PFL, PFA</td>
<td>OFC, PFL, PFA</td>
<td>OFC, PFL, PFA</td>
<td>OFC, PFL, PFA</td>
</tr>
<tr>
<td>Caucasian Male or Female (Hall, 1989) 0-16 yrs</td>
<td>28.69</td>
<td>10.00</td>
<td>28.00</td>
<td>28.20</td>
</tr>
<tr>
<td>Hispanic Female (Clarren et al., 2010) 6-16 yrs</td>
<td>26.03</td>
<td>10.00</td>
<td>26.03</td>
<td>26.03</td>
</tr>
<tr>
<td>Caucasian Male (Clarren et al., 2010) 6-16 yrs</td>
<td>26.49</td>
<td>10.00</td>
<td>26.49</td>
<td>26.49</td>
</tr>
<tr>
<td>Scandinavian Female (Stromland et al., 1999) 0-18 yrs</td>
<td>26.84</td>
<td>10.00</td>
<td>26.84</td>
<td>26.84</td>
</tr>
<tr>
<td>Scandinavian Male (Stromland et al., 1999) 0-18 yrs</td>
<td>27.43</td>
<td>10.00</td>
<td>27.43</td>
<td>27.43</td>
</tr>
</tbody>
</table>

* The PFL Z-score reflects how many standard deviations (SD) the patient’s PFL is above or below the normal population mean. For example, if a 1-year-old child had a PFL = 20 mm, that child’s PFL would be 1.27 SDs below the population mean on the Stromland male PFL charts.
Neurologic Features

- Microcephaly with brain structure anomalies: decreased brain volume, agenesis of the corpus callosum, decreased frontal lobe and cerebellar size, microstructural changes
- Decreased coordination and balance
- Positive neurologic soft signs (tremor, decreased fine motor, sensory problems)
- Seizures
- Optic nerve hypoplasia
- Hearing defect

Physiologic Features

- Central Nervous System function
- Mood
- Behavior
- Hormonal dysregulation
- Growth
- Stress response
- Sleep
- Other neuro-endocrine functions

Occipital Frontal Circumference (Head Size)

- Use WHO and CDC; Nelhaus OFC charts
- 3-97th percentile
- Measure 3 times, take largest number
- Video of proper OFC measurement: [www.peds.umn.edu/iac/](http://www.peds.umn.edu/iac/)
  Click on "Topics", then on "How to Measure a Child's OFC".

Neurologic Exam Components

- DTR’s
- Gait
- Coordination
- Balance
- Strength
- Cranial Nerves
- Soft signs (finger pursuit, RAM, sequential finger motion, etc.)
- Adjust for age

Order CNS Functional Evaluations

- Order comprehensive assessments:
  - Neuropsychology
  - Occupational therapy
  - Speech and language therapy
  - Physical therapy (if indicated)
- Attention
- Executive functioning
- Behavior
- Sensory processing
- Speech and language, especially figurative language
- Memory
- Visual-motor integration
- Social-adaptive functioning
- Cognition
- Academic Achievement
## Musculoskeletal Features

- Radioulnar Synestosis
- Clinodactyly and campylocdactyly
- Hockey-stick palmar crease
- Micrognathia

## Other Clinical Features

- Heart murmur and related defects
- Hypoplastic fingernails or toenails, especially the 5\textsuperscript{th} digit
- Kidney defects
- Stunted growth
- Ear defects

## Fetal Alcohol Syndrome (FAS) Minimum Criteria

- **Prenatal Alcohol Exposure**: Confirmed or unconfirmed (either)
- **Growth**: height or weight at or below the 10\textsuperscript{th} percentile for sex and age at any time from birth on, not from other causes (either) **AND**
- **Facial Dysmorphology**: thin upper lip, flat philtrum, and short palpebral fissures for age and racial background (all 3 features) **AND**
- **CNS**:
  - * Head size at or below the 10\textsuperscript{th} percentile for age and sex
  - * at or below -1.5 SD on 3 or more neuropsychology tests
  - * structural brain anomalies
  - * unexplained neuro problems or signs (i.e. seizures)
  - * global cognitive or IQ at or below 3\textsuperscript{rd} %tile or -2 SD (<70)

## Partial FAS Minimum Criteria

- **Prenatal Alcohol Exposure**: Confirmed **AND**
- **Facial Dysmorphology**: 2 of 3 features **AND**
- **CNS**:
  - Head size at or below the 10\textsuperscript{th} percentile for age and sex
  - at or below -1.5 SD on 3 or more neuropsychology tests **or**
  - structural brain anomalies **or**
  - unexplained neuro problems or signs (i.e. seizures) **or**
  - global cognitive or IQ at or below 3\textsuperscript{rd} %tile or -2 SD (<70)
  - **Growth defect**: Not applicable

## Alcohol-Related Neurodevelopmental Disorder (ARND): Minimum Criteria

- **Prenatal Alcohol Exposure**: Confirmed **AND**
- **CNS**:
  - At or below -1.5 SD on neuropsychology tests measuring brain functioning in 2 or more areas of development (3 is a severe case). **AND**
- **Growth Deficit and Facial Dysmorphology**: Not applicable

## Future of Diagnosis

- Development of Neuropsych Profile for FASD.
- Provide services to all screening positive for Prenatal Alcohol Exposure (PAE) and developmental delays, not waiting for a diagnosis.
- Ongoing surveillance in health care home for PAE-positive screens, who do not have any developmental, behavioral, mental health manifestations.
- Screen for FASD (Growth, Facial Features, and Maternal History) at all Routine Pediatric visits, all new pediatric patients, and all visits for neurologic, developmental, behavioral, mental health issues.
- Refer all FASD-positive screens for an FASD Diagnosis Evaluation.
- Biomarkers – not yet on the horizon, but would be easiest.
- School/clinic/family collaboration imperative for all with an FASD.
- Change name to reflect actual brain changes.
Contact Information:
MOFAS
www.mofas.org
Email:
info@mofas.org
Phone: 651-917-2370