Fetal Alcohol Spectrum Disorders: The Basics

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Learning Objectives

By the end of this Webinar, attendees will be able to:

1. List diagnostic terms used under the umbrella term fetal alcohol spectrum disorders (FASD);
2. Discuss the brain basis of FASD;
3. Identify cognitive issues and behaviors that are often seen in individuals with an FASD.
Who is at risk for having a baby with brain damage from alcohol exposure?

- 28-year old unemployed, Native American woman, living on a reservation
- 17-year old A-student from a small Midwestern town, no boyfriend
- 34-year old marketing executive from a wealthy suburb, newly married
Who is at risk for having a baby with brain damage from alcohol exposure?

28 year old unemployed Native American woman, living on a reservation

   Chronic alcoholism, drinks hard liquor, poor access to prenatal care, may expose fetus throughout pregnancy

17 year old A-student from a small Midwestern town, no boyfriend

   Having casual sex without birth control, unaware that she is pregnant for first two months, parties on weekends – drinks beer

34 year old marketing executive from a wealthy suburb, newly married

   Unaware that she is pregnant for first month, drinks wine at home with dinner, bachelorette party, happy hours.
   After learning of pregnancy, obstetrician tells her “a single glass of wine with dinner is OK.”
Alcohol is capable of damaging a fetus at a point in development when most women are not aware of a pregnancy.

Even a single, binge episode can cause profound damage to the developing brain.
Human development from day 17-32 post-fertilization corresponds to day 7-11 in the mouse.

<table>
<thead>
<tr>
<th>Human</th>
<th>Mouse</th>
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<tbody>
<tr>
<td>Day 17</td>
<td>Day 7</td>
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<td>Day 22</td>
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<td>Day 26</td>
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<td>Day 29</td>
<td>Day 10</td>
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<tr>
<td>Day 32</td>
<td>Day 11</td>
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</tbody>
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Images courtesy of Kathy Sulik, Ph.D. University of North Carolina
The fetus would fit inside the zero on a penny at this point in development.
• Day 7: equivalent to week 3 (day 17 or 18) in humans
• Craniofacial effects
• Holoprosencephaly
  • Forebrain, midline brain anomalies
  • Callosum, hippocampus, basal ganglia
• Day 8.5: equivalent to week 4 (day 21-24) in humans

Despite clear research, evidence, and the high risk of drinking during pregnancy, public perception remains very inaccurate.
The popular media frequently draws invalid conclusions from research and presents distorted messages.


The SATURDAY ESSAY

Take Back Your Pregnancy

Modern pregnancy comes with a long list of strict rules, but does it have to? An economist examines the data and finds room for choice amid the familiar limits.

By EMILY OSTER
Aug. 9, 2013 6:31 p.m. ET

Modern pregnancy comes with many rules. Model pictured here, F. Martin Ramani, for WORLDCOM. Journal

NEW DAY

NEW STUDY ON PREGNANCY AND ALCOHOL

BRAIN DEVELOPMENT LIKELY UNHARMED BY MODERATE INTAKE

LIVE CNN
The reality: Fetal Alcohol Spectrum Disorders (FASD) represent a **serious**, public health problem

- In the US, 13% knowingly drink some alcohol while pregnant
- 1% drink heavily while pregnant
- 3-4% binge drink during pregnancy (SAMHSA)

- 50% of pregnancies are unplanned
- Fetal Alcohol Syndrome: 2 per 1000 live births
- Fetal Alcohol Spectrum Disorders (includes all conditions) is estimated to be 2% to 5%
- Comparison: Central Italy 2.3% to 6.3%
Drinking Patterns

- “Binge”: Four or more standard drinks in two hours
- 15% of women (child-bearing age) binge drink
- Ages 18-24: highest binge drinking
- The peak level of alcohol is associated with severity of FASD; binge drinking is most dangerous
- Ages 35-44: most likely to continue drinking even after learning of pregnancy
- College-educated: likely to continue drinking
Apoptosis from ethanol exposure (single “binge” model)

- Millions of neurons are inadvertently signaled to self-destruct (apoptosis)
- These animals have significant behavioral and learning deficits

Binge Drinking in Context: What Is a Standard Drink?

- 60% of women “over-pour” or underestimate the size of a drink, especially when pouring hard liquor.
- In a review of past-year alcohol use, 30% of women of childbearing age (average, 26 years) were classified as “risky” drinkers.
What Is Not a Standard Drink?

- Martini/Appletini
- Long Island Iced Tea
- Margarita or Daiquiri

- A **mixed drink** made with two or more liquors **is not** a standard drink.

- The alcohol content is **2- to 5-times more than a standard drink.**

- A “40” bottle of beer or malt liquor or a beer stein **is not** a standard drink!

- A balloon glass of wine contains **2- to 3-times more alcohol than a standard drink.**
An Unfortunate Confluence

Culture of Drinking → Fetal Brain Damage → Unprotected sex

Timing → Pregnancy unawareness
Fetal Alcohol Terminology

- **Fetal Alcohol Syndrome (FAS)**
  - FAS was first used in 1973 by Dr. David Smith and Dr. Ken Lyons Jones at the University of Washington.
  - Diagnosed on the basis of:
    - Facial dysmorphia (measurement / formal evaluation)
    - Growth impairment (height / weight)
    - Brain abnormalities (structural / cognitive)
    - Alcohol exposure
Other FASD Terminology used over the Years in Various Contexts

- Prenatal Exposure to Alcohol (PAE)
- Fetal Alcohol Effects (FAE)
- Partial fetal alcohol syndrome (pFAS)
- Alcohol-related neurodevelopmental disorder (ARND)
- Alcohol-related birth defects (ARBD)
- Static encephalopathy (an unchanging injury to the brain)
- Neurodevelopmental Disorder... (DSM-V)
Fetal Alcohol Spectrum Disorders (FASD)

- **FASD**: an **umbrella** term describing the range of effects that can occur with prenatal alcohol exposure.
- **FASD** may include physical, intellectual, behavioral, and/or learning disabilities with lifelong implications.
- **FASD** is not a clinical diagnosis.
Most Commonly Used FASD Diagnoses

• Fetal Alcohol Syndrome (FAS)
  • Growth deficient, three facial features, brain impairment, alcohol exposure known or unknown

• Partial FAS
  • Known alcohol exposure, 2+ facial features, brain impairment, growth deficiency may or may not be present

• Alcohol Related Neurodevelopmental Disorder (ARND)
  • Known alcohol exposure, brain impairment (based on cognitive results usually), facial features may or may not be present, growth deficiency may or may not be present
  • There is not a strong consensus on the precise definition of ARND as a diagnosis. Some question its validity.
Cognitive Consequences of Prenatal Alcohol Exposure

- Impaired intellectual functioning
- Sensory Integration difficulties
- Dysregulation of mood and behavior
- Poor working memory
- Impaired judgment
- Impaired language reasoning/processing
- Impaired executive functioning
- Impaired social adaptive functioning

Typically, three domains of cognitive impairment are required for diagnosis.
A Typical Neuropsychological Battery for an FASD evaluation

- Intelligence (IQ)
- Adaptive functioning (independent living skills)
- Attention
  - Vigilance
  - Shifting
- Executive functioning
  - Planning / organizing
  - Adjusting to feedback
  - Flexibility
- Language functioning
  - Especially “pragmatic language”
- Motor skill
- Visual-perceptual skill
- Behavior / Psychological functioning
  - Hyperactivity
  - Emotion regulation
  - Anxiety / Depression
FASD: a Pre-cursor to Psychopathology / other problems

- Substance abuse disorders: 60%
- Major Depression: 44%
- Bipolar Disorder: 20%
- Anti-social personality: 16%
- ADHD: 73%
- Learning Disability: 36%
- Sleep Disorder: 12-21%
- Poor fit with societal expectations: 92%

Famy, Streissguth, Unis (1998); Burd, Klug, Martsolf, Kerbeshian (2003)
Screening and Diagnosis
Screening for Prenatal Alcohol Exposure

• Settings
  • Ob-Gyn (pregnant and non-yet-pregnant patients)
  • Family practitioner (women of childbearing age)
  • Pediatrician (subsequent pregnancies)
  • Substance-abuse treatment programs (high risk)
Screening for FASD

• Important to incorporate a screening tool into Electronic Medical Records (EMR)
• Staff training on FASD and screening process
• A plan of action is necessary: clinic flow chart
• Develop a resource directory
• Find other ways clinics can give the message (e.g., when giving pregnancy test results)
Assess the Full Pattern of Alcohol Use

- The pattern of drinking before the mother knew of her pregnancy is critical.
  - This pattern predicts drinking during pregnancy.
  - It acts as a “lead in” to the next questions.
- A history of chemical dependency diagnosis/treatment is also predictive.
The Alcohol Screening Questionnaire

• Simple Yes/No questions are ineffective. A discussion is more fruitful.
  • How often do you drink?
  • How much do you drink at a time?
  • When was your last drink?
Screening Tool Components

- 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, and future plans
- Ask **every woman** **every** time
- Respect and support, not guilt or shame
- Regardless of answer, provide an educational message and/or written material
Why is it important to ask at every visit?

• A woman may not be ready to share information at the first visit. She needs to build a trusting relationship with the provider.

• She may not be drinking at the first visit, but a crisis happens later in the pregnancy that leads to her drinking.

• 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.
Automate it in the Electronic Medical Record

Before you knew you were pregnant...
- How often, on average, do (did) you drink?
- On a day or night when you did drink, about how many drinks did you have? (a drink equals a bottle of
Since you knew you were pregnant...
- How often, on average, do (did) you drink?
- On a day or night when you did drink, about how many drinks did you have? (a drink equals a bottle of
- When was the last time you had a drink?
- Alcohol message given?

Before you knew you were pregnant...
- Do (did) you use street drugs?
- Do (did) you use prescription pain medications?
- How often, on average, do (did) you use drugs?

Since you knew you were pregnant...
- Do (did) you use street drugs?
- Do (did) you use prescription pain medications?
- How often, on average, did (do) you use drugs?
- Drug message given?

Add 1 Information
- Have you ever been in treatment for alcohol or drugs?
- If yes, when?
Diagnosing an FASD

• A multidisciplinary team of trained professionals can diagnose a disorder from the FASD spectrum. The core diagnostic team should include:
  • Physician (ideally a dysmorphologist or developmental pediatrician)
  • Psychologist/neuropsychologist
  • Speech and language pathologist
  • Occupational therapist
  • Social worker
• A key part of the diagnostic process will be to connect a person with an FASD to a system of support.
Diagnosing FAS

**Diagnostic Criteria:**
1. Documentation of pre-natal alcohol exposure
2. Growth deficiency
3. Dysmorphic facial features
4. Brain abnormality and/or cognitive impairment

**Three most significant facial features**
- Short palpebral fissure (small eye opening)
- Thin vermilion (upper lip)
- Hypoplastic (smooth) philtrum

*Clarren, SK Ped, 1995*
Facial Features of FAS

Critical features: 1) short palpebral fissures; 2) indistinct philtrum; 3) thin upper lip
(The other features shown are sometimes seen, but are not the definitive features)
Other dysmorphic features of FAS

- “Railroad-track” ear
- Hockey-stick palmar crease
Benefits of Early and Accurate Diagnosis and Intervention in FASD

• Early and accurate diagnosis of an FASD helps to—
  • Lessen secondary disabilities such as substance abuse, mental health issues, or school failure.
  • Improve life outcomes.
  • Achieve success for people with an FASD by focusing on their strengths and needs rather than their “weaknesses” and what they have done “wrong.”
  • Prevent future births of children with an FASD.
Alcohol is a teratogen with system-wide effects

Teratogen: a drug or other substance capable of interfering with the development of a fetus and causing birth defects and/or developmental disabilities.
“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
Report to Congress, 1996
Alcohol is a Uniquely Dangerous Teratogen

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Maternal alcohol consumption is the **sole cause** of FASD
- But other factors add to the risk.
  - Maternal age, health, nutrition
  - Maternal and fetal genetics
  - Paternal drinking is not a factor in FASD

FASDs are **100% preventable**

Prevention is a complex mix of:
- Education and public policy
- Screening
- Substance abuse prevention efforts
We have moved well beyond “blaming” the woman.  
**FASD is a complex societal problem.**

- Public still misunderstands risks from drinking and does not recognize the high prevalence of FASD.
- Society condones drinking in women of childbearing age.
- FASD can occur from binge drinking, which is generally highly socially-accepted (e.g., college, e.g., bachelorette party).
- FASD can occur when a mother does not know of a pregnancy.
- Doctors implicitly give permission by not discussing alcohol use with their patients or downplaying the risks.
- Alcoholism is a highly challenging condition to overcome.
• FASD occurs across levels of SES
  • Not strictly a problem of poverty / substance abuse
• FASD occurs in “social” drinkers
• Safest approach: no alcohol during pregnancy or while trying to become pregnant
Risk Factors for Alcohol-exposed Pregnancy

Substance Abuse and Mental Health Factors:
- History of alcohol consumption.¹
- Family history of alcohol use.²,³
- History of inpatient treatment for drug, alcohol, or mental health issues.⁴

Personal, Sexual, and Family Factors:
- Previous birth of child with an FASD.³,⁵
- Non-use of contraceptives, unplanned pregnancy.⁶
- Physical, emotional, and/or sexual abuse.⁶
- Alcohol use by partner.²,³
The Brain in FASD
Neuroimaging in FASD

• Not yet a diagnostic tool, but a research tool
• Non-invasive, relatively quick
• Allows for new insights into the range of effects of prenatal alcohol exposure
Our understanding of the effects of prenatal alcohol has evolved since these pictures were first presented.
Extreme structural abnormalities in FAS
(12-year old, male subjects)
No gross abnormalities in most cases
**White matter:** The fibers that connect different regions of the brain is also affected by prenatal exposure to alcohol.

Pictured are two, 12-year old boys (1 FASD; 1 unexposed control)
- Here, Diffusion Tensor MRI reveals the **full extent** of the damage
- It can also **reveal subtle damage** not identifiable by other techniques
- With newer methods, more children with subtle damage will be identified and we will better understand how to rehabilitate the damage / accommodate to the damage

Mean Diffusion in the corpus callosum
(a measure of integrity of the white matter)

A simple MRI study shows disruption in basic right-left brain communication in a child with FASD.

This inefficiency in communication occurs 24 hours a day, 365 days per year.
Cognitive Consequences of Prenatal Alcohol Exposure

**FAS:** Associated with impaired intelligence (IQ < 70) and developmental disability

**FASD** (whole spectrum): IQ ranges from 40s to 140s

Mean IQ in FASD is often reported in the mid-80’s (low average, but not impaired)

**Commonly-seen deficits in FASD**
- Impaired intellectual functioning
- Sensory Integration difficulties
- Dysregulation of mood and behavior
- Poor working memory
- Impaired judgment
- Impaired language reasoning/processing
- Impaired executive functioning
- Impaired social adaptive functioning
Neurobehavioral Impairments Typical in Persons with an FASD

• **Executive function** involves abstract thinking, problem-solving, planning, and flexibility in thinking. People with an FASD may have difficulty—
  – Generalizing —applying information from one situation to another.
  – “Reading” people and situations—properly connecting the words, body language, and actions of average daily activities.
• Understanding time
• Understanding money
• Anticipating future consequences of actions
Neurobehavioral Impairments Typical in Persons With an FASD

• **Working memory**—alcohol injury to one portion of the brain results in less efficient encoding of verbal and nonverbal information.¹
  - Thus information from the last few minutes or hours may not be stored. Working memory is necessary for higher order concept learning.

• People with an FASD may “forget” learned material or skills, but often just don’t “get it.” Unless they are told repeatedly, the information may not be encoded and stored properly in the brain.
Related Behavioral Issues for Persons with an FASD

Impaired Executive Function may cause persons with an FASD to:

- Not learn from own experience, mistakes, or natural consequences.
- Fail to respond to reward and consequence systems (e.g., points, levels, stickers).
- Be unable to imagine the consequences of their words or actions.
- Have difficulty entertaining themselves.
- Have difficulty understanding the meaning of jokes, idioms, or sarcasm.
- Give in to peer pressure.
- Be naïve, gullible.
- Be unable to identify dangerous people or situations.
- Confess to a crime they did not commit.
Related Behavioral Issues for Persons with an FASD

- Persons with impaired working memory may—
  - Not complete tasks or chores.
  - Be unable to follow **multiple** directions or rules.
  - Appear to be oppositional or defiant.
  - Not know to ask questions.
  - Have trouble with changes in tasks and routine.
Less well-understood deficit: pragmatic language deficits

- Not necessarily deficits in grammar, syntax, vocabulary, fluency, etc.
- Probably contributes to “literal thinking”
- “Irrational” speech
- Misunderstandings
- Frustration with communication
- Ultimately, problems in daily living
Frequently Seen Behaviors in FASD

Conroy et al (1999): From a study of British Columbia youth involved in the criminal justice system
Common Strengths of Persons with an FASD

• **Friendly, likable**: May be outgoing and sociable and have little anxiety about strangers.

• **Verbal, chatty**: May be very socially interested (but not necessarily socially-skilled).

• **Helpful, hard-working**: If you ask, they will do it. They can be very good workers with the right job and training.

• **Determined, resilient**: They don’t hold grudges and will come back, if rejected. Every day is a new day!

• **Want to be liked**: They will do whatever they can to have friends.

• However, these strengths also may get them into difficulty.
Case Example #1

- “Mary”, a 13 year-old girl referred by county case worker
- Residential treatment for nine months
- Long-standing attention problems and learning difficulties
- Aggression, extremely poor judgment, unsafe behaviors
- Medications generally minimally effective
- History of pre-natal alcohol exposure (as with three prior siblings)
- Neurocognitive Evaluation:
  - IQ in the range of 58 (mean = 100, SD=15): More than 2.5 SD low
  - Impairments in reading and math skills
  - Verbal memory functioning: 24 (mean = 50, SD = 10); More than 2.5 SD low
  - Impaired self-care skills
- Physical evaluation:
  - OFC 10%ile (small head)
  - Palpebral fissures <5%ile (small eyes)
  - Flat philtrum (abnormal)
  - Thin upper lip (abnormal)
  - Height 3%ile (abnormal)
  - Weight 60%ile (normal)
- Diagnosis: FAS
- Prognosis: lifelong impairment, very limited educational and work capacity, psychiatric comorbidity
- Needs: residential placements / group home living, psychiatric medication, Educational programming for intellectual impairment, behavioral therapies
Case Example #2

- “Zach”, a 18-year-old young man referred by the court
- Reason for referral: charges pending – sex with a 15-year old girl
  - Now registered as a sex offender
- Long history of social difficulties, few friends, received social skill training in school
- Judgment problems – poor decision making, easily influenced by peers (1 shoplifting)
- Questions of autism posed when he was five or six; ruled-out
- Mild learning difficulties (Reading, Math) but IQ was relatively normal (FSIQ=94)
- Domestic adoption; mother used alcohol heavily as well as cocaine and marijuana
- Physical evaluation:
  - OFC 65%ile (normal head)
  - Palpebral fissures 75%ile (normal eyes)
  - Philtrum (mildly flattened)
  - Upper lip (normal)
  - Height 60%ile (normal)
  - Weight 85%ile (normal)
- Diagnosis: ARND
- Neurocognitive testing:
  - Attention deficit
  - Poor adaptive functioning (SS=76); Much lower than his IQ
  - Mildly low academics
- Clinical issues: new-onset Depression and Anxiety (restricted from seeing children in family)
- Needs: Specialized legal assistance, therapy and psychiatric care, plan for semi-independent living, transitional planning / continuation of high school / work assistance
### Economic Costs of FAS/FASD

**In the United States**—

- $1.6 to $2.5 million in lifetime costs per individual with FAS or other FASD.\(^1\)
- The estimated economic cost of drinking during pregnancy 2006 was $5.2 billion in 2006.\(^2\)
- Children diagnosed with FAS incur Medicaid costs nine times as high as children without FAS.\(^3\)

**Costs are incurred in**—

- Medical care
- Social services
- Housing
- Criminal justice
- Specialized education
- Substance abuse treatment
- Decreased or lost productivity
Public Awareness

- Fetal Alcohol Syndrome. Cured.
  - Effects like brain and heart damage are 100% preventable. Simply don’t drink while pregnant. www.mofas.com

- Fetal Alcohol Syndrome. Cured.

- Fetal Alcohol Syndrome. Cured.
  - Effects like learning and memory problems are 100% preventable. Simply don’t drink while pregnant. www.mofas.com
Giving your unborn child a beer is just as ridiculous.

The U.S. Surgeon General Advisory says no amount of alcohol is safe during pregnancy.
Share 049: Zero Alcohol For Nine Months.
www.mofas.org
Giving your unborn child a beer is just as ridiculous.

No amount of alcohol is safe during pregnancy.

www.mofas.org
Lessons Learned from Broad Public Campaigns

• Women do know about Fetal Alcohol Syndrome
  • >80% reported seeing previous media campaigns

• The public misperceives the relative risks
  • Alcohol risks downplayed relative to other drugs
  • Believe that “moderation” will protect the fetus from harm
  • OB/Gyns / family doctors still give the wrong message or at the least, a very mixed message

• Needed:
  • Not just more education
  • Social norm change is necessary
  • Consistent prevention message from all sources
    • Resulted in adding media campaign targeting physicians
Conclusion

- FASD is 100 percent preventable.
- FASD can occur when a fetus is exposed to alcohol in the womb.
- FASD may cause physical, intellectual, behavioral, and/or learning disabilities with lifelong implications.
- These disabilities are related to alcohol damage to specific areas of the brain.
- FASD cannot be cured. However, it can be prevented.