Fetal Alcohol Spectrum Disorders

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Causes of Birth Defects and Developmental Disabilities
Fetal Alcohol Spectrum Disorders: FASD

- Umbrella term for the complex range of brain injuries (organic brain damage) that can result from prenatal exposure to alcohol
- #1 known preventable cause of intellectual and behavioral impairment

Unless otherwise noted, content information contained in this PowerPoint presentation is referenced in the Fetal Alcohol Spectrum Disorders Competency-Based Curriculum Development Guide for Medical and Allied Health Education and Practice, Centers for Disease Control and Prevention, 2009 available at [www.cdc.gov](http://www.cdc.gov)
Fetal Alcohol Spectrum Disorders:

FAS ~20%  Invisible ~80%

- Fetal Alcohol Syndrome (FAS)
- Invisible end of the spectrum:
  - Partial FAS
  - Alcohol Related Neurodevelopmental Disorders (ARND) or current DSM5 terminology: Neurodevelopmental Disorders - Prenatal Alcohol Exposure (ND-PAE)
  - Alcohol Related Birth Defects
Prevalence:
Fetal Alcohol Syndrome: about 1 per 1000 children
Full spectrum of disorders may be as high as 9 to 10 per 1000 children
Under-recognition of FASD

• Difficulty in making the diagnosis, especially in the newborn period or in adolescents and adults

• Not on the radar (esp. after the newborn period) among general pediatricians; and/or poor access to expert diagnostic teams

• Lack of a clearly defined neurobehavioral phenotype

• Under-ascertainment of alcohol use by pregnant women

• Crosses all economic and racial categories

• Sensitivity of the subject

Contributing factors

• 50% of pregnancies are unplanned
• Unaware of pregnancy until 8 or more weeks
• Lack of awareness of the risks of drinking alcohol while pregnant.
• Women still get the wrong message
• Women don’t think of alcohol as a substance. May not think of beer or wine as ‘alcohol’. May not be aware or are in denial of the amount they drink
• Social or dependent drinking

Alcohol Use

- Alcohol is legal, socially acceptable, readily available, party and fun time, heavily advertised to women.
- Over 50% of U. S. women age 18 to 44 use alcohol.
- Nearly 33% binge drink (National Center for Health Statistics 2008).
- Binge drinking is the most common pattern of excessive alcohol use.
- Most who binge drink are not alcohol dependent.
- Women who binge drink are increased risk of an unintended pregnancy and alcohol-exposed pregnancy.
Binge Drinking

- An episode of “binge” or risky drinking for women of childbearing age (18 – 44) is defined as 4 or more standard drinks in about a two hour period.
What is a standard drink?
Definition: 0.6 ounces of pure or absolute alcohol (aa) = 1 standard drink

**Alcohol drink equivalents**

| 12 oz. of beer or cooler | 8–9 oz. of malt liquor (8.5 oz. shown in a 12-oz. glass that, if full, would hold about 1.5 standard drinks of malt liquor) | 5 oz. of table wine | 3–4 oz. of fortified wine (such as sherry or port) 3.5 oz. shown | 2–3 oz. of cordial, liqueur, or aperitif 2.5 oz. shown | 1.5 oz. of brandy (a single jigger) | 1.5 oz. of spirits (a single jigger of 80-proof gin, vodka, whiskey, etc.) Shown straight and in a highball glass with ice to show level before adding mixer* |

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Sensible Drinking Limits for Women

- Less than 4 standard drinks per occasion
- Less than 8 standard drinks per week
- No drinking if pregnant, breastfeeding, planning to become pregnant, or if sexually active and not using reliable contraception
Prevention of Alcohol-Exposed Pregnancies

• Provide all patients with information about alcohol, contraception, and FASDs

• *One of the strongest predictors of substance use during pregnancy is substance use before pregnancy*

• Provide alcohol screening to women of childbearing age
  • Risky drinking vs Dependent drinking

• Provide brief alcohol intervention for women at risk

• Provide targeted alcohol treatment and promote contraception use for women at highest risk:
  • Women who have a child with an FASD
  • Women with a history of alcohol abuse and/or dependence
  • CHOICES
Evaluation of prenatal alcohol exposure for FASD diagnosis

- One of the most difficult diagnostic issues
- Birth mothers
  - Important questions:
    - What was your alcohol consumption prior to becoming pregnant
    - When did you find out you were pregnant?
  - Difficulty remembering
  - Under-reporting
  - Stigma, judgment, blame
- Adopted or foster
  - Birth history often not available or incomplete
- No adequate test
  - Research in progress
Interviewing Techniques

• Ask a standard series of health questions that are directed to all patients in order to improve health
  • *Embed at least one question to identify prenatal alcohol use among other questions related to lifestyle e.g.*
    • How is your diet?
    • Do/did you take prenatal vitamins?
    • Were you on any prescription or non-prescription medications?
    • Do/did you ever enjoy an alcohol beverage or two?
    • When was the last time you had an alcohol beverage?
    • Do/did you smoke any cigarettes or were exposed to second hand smoke?
    • What about exercise?
    • When did you find out or suspect you were pregnant?
Interviewing Techniques

• Avoid questions that suggest you want a negative answer
  • You don’t drink alcohol do you?
  • You didn’t drink while you are pregnant did you?
  • Did you drink once you found out you are pregnant? Yes or no

• Positive:
  • Many women and men wind down the day with a glass of wine or have a few drinks on a night out. Do you sometimes enjoy a drink or two?
  • Can you tell me a bit about your drinking pattern before you knew you were pregnant?
  • How had you changed your alcohol consumption since finding out you were pregnant?
    • I continue my usual habit
    • I have cut down or stopped
    • I am trying to abstain but find it difficult
IMPACT OF ALCOHOL ON PREGNANCY OUTCOMES
Alcohol is the most abused substance

- Of all the substances of abuse --- including cocaine, heroin and marijuana--- alcohol produces by far the most serious neurobehavioral effects in the fetus
- During pregnancy, there is no safe time no safe amount, no safe type

*Institute of Medicine, Report on Fetal Alcohol Syndrome Diagnosis, Epidemiology, Prevention and Treatment, 1996*
Alcohol crosses the placenta within minutes, resulting in similar blood concentration in embryo and fetus as mother.

Alcohol is a teratogen, a substance that is toxic to the developing fetus.
Not everyone is affected by alcohol the same way
30-40% risk of adverse effects

Maternal
• Drinking Pattern: Amount, how often, binge drinking
• Individual differences in absorption, distribution
• Metabolic processes that modify blood alcohol level are highly variable among individuals
• Nutrition & vitamin status
• Age

Fetal
• Genetic differences
• Lack of, or immature metabolism
• Windows of development
• Timing of exposure
Alcohol interferes with Fetal Development
Mechanisms

- Numerous and complicated, still under investigation
- Interferes brain development
  - Causes excessive cell death
  - disrupts nerve formation
  - disrupts cellular migration and differentiation
  - disrupts hormonal and chemical regulatory systems
    - neurotransmitter signal and receptor disruption
    - nerve growth factors
- Interferes with fetal uptake of critical nutrients (amino acids, glucose, vitamins, etc.)
- Induces vasoconstriction within the placenta, umbilical arteries/vein
Potential Effects

- Premature Birth
- Pre- and Postnatal Growth Retardation
- Physical Malformations
  - Brain, face, heart, eyes, ears, kidneys, bones
- Microcephaly
- Cognitive and Behavioral Problems
Diagnostic criteria for Fetal Alcohol Syndrome

• Growth deficits
• Specific facial abnormalities
• Neurobehavioral Deficits
Growth Deficiency

Documented Height and/or Weight at ≤ 10%
- at any one point pre- or postnatal
- adjusted for age, sex, gestational age, race, ethnicity
Growth Deficiency
Microcephaly

- Head circumference ≤ 10\textsuperscript{th} percentile
FAS and the Brain:
Neurobehavioral Deficits: Alcohol can affect all parts of the brain.
A. Magnetic resonance imaging showing the side view of a 14-year-old control subject with a normal corpus callosum; B. 12-year-old with FAS and a thin corpus callosum; C. 14-year-old with FAS and agenesis (absence due to abnormal development) of the corpus callosum.
Facial Abnormalities

- Discriminating Features:
  - short palpebral fissures
  - flat midface
  - short nose
  - indistinct philtrum
  - thin upper lip

- Associated Features:
  - epicanthal folds
  - low nasal bridge
  - minor ear anomalies
  - micrognathia

In the Young Child
3 Cardinal Features of FAS

- Short Palpebral Fissure Length
- Smooth Philtrum
- Thin Upper Lip
Cardinal Features

- Facial Abnormalities
  - Smooth philtrum
    - Lip philtrum guide 4 or 5
  - Thin vermilion
    - Lip philtrum guide 4 or 5
Faces of FASD
Some Additional Physical Features

- Eye: Optic nerve hypoplasia, tortuous retinal vessels, vision problems
- Mouth: cleft lip/palate, narrow palate
- Heart: Atrial and ventricular septal defects, Tetralogy of Fallot, transposition of the great vessels
- Vertebrae: cervical spine anomalies, scoliosis, neural tube defects
- Kidneys: structural anomalies, tubular dysfunction
- Hearing difficulties
- Atopic dermatitis
Railroad Track Ear

11.8% FAS compared to 1.8% not FAS

Palmar Creases

hockey stick: 21.6% FAS; 5.3% not FAS
single traverse 15.5% FAS; 3.8% not FAS

Clinodactyly
Camptodactyly
36.7% FAS; 6.1% non FAS

The Spectrum Recap

- FAS is the most severe: growth deficiency; 3 cardinal facial features, central nervous system damage

- The rest of the spectrum has some degree of the symptoms described for FAS.
  - Partial FAS (pFAS)
  - Alcohol Related Birth Defects (ARBD)
  - Alcohol Related Neurodevelopmental Disorders (ARND) or
    - Neurodevelopmental Disorder associated with prenatal alcohol exposure (ND-PAE) the DSM-5 term describe this category and this term will be replacing the term ARND in the future.

- Individuals often have none of the physical characteristics but all those within the spectrum will have some degree of learning and behavioral dysfunction as a result of damage to the developing brain.
Benefits of Diagnosis

- Better management of behaviors when provided appropriate therapeutic techniques, structure and support system; improved outcomes
- Helps decrease anger and frustration for individuals, families, providers, juvenile justice, and communities by helping to understand that negative behaviors results from the disability
- Helps those with an FASD to succeed by focusing on their strengths and why they have trouble in certain areas
- Helps prevent future births of children with FASD
Questions?

Thank you