Fetal Alcohol Spectrum Disorders: Identification and Diagnosis

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Objectives

• This presentation will review:
  – Terminology
  – Scope and significance
  – Alcohol Use in Pregnant Women
  – Prevalence and Course of FASDs
  – Clinical presentation
  – Evaluation and Diagnosis
Terminology, Scope, and Significance
Fetal Alcohol Syndrome

• Preventable birth defect caused by maternal alcohol consumption during pregnancy

• Characterized by physical, cognitive, and behavioral abnormalities

• Lifelong impairments
Fetal Alcohol Spectrum Disorders

- An umbrella term used to describe the full range of physical, cognitive, and behavioral problems seen in individuals who have been exposed to alcohol prenatally
  - Includes:
    - Fetal Alcohol Syndrome (FAS)
    - Partial or Atypical Fetal Alcohol Syndrome
    - Alcohol Related Neurodevelopmental Disorder (ARND)
    - Alcohol Related Birth Defects (ARBD)
Scope of the Problem

• Considered to be the most common known etiology of mental retardation in the U.S.

• FAS is an extremely costly disorder

• Common misconceptions persist among the public
Alcohol Use and Pregnant Women
Alcohol Use Among Women of Childbearing Age

- 53% of women of childbearing age report some alcohol use
- 12-23% of women of childbearing age report binge drinking (> 5 drinks/occasion)
Alcohol Use Among Pregnant Women

• 12% of pregnant women in the U.S. report some alcohol use (CDC, 2004)
  – 1 in 8 pregnancies is an alcohol-exposed pregnancy

• 1.9 – 3.9% of pregnant women report frequent or binge drinking (CDC, 2004; SAMHSA, 2005)
Factors Associated with Alcohol Use in Pregnant Women

- Younger age
- Caucasian
- Less education
- Single
- Unemployed
- Past sexual abuse
- Current or past physical abuse
- Using tobacco or other drugs
- Living with or having partners who are substance users
- Higher levels of depressed mood
- Higher tolerance of alcohol
Which of These Factors Moderate the Impact of Alcohol on the Developing Fetus?

• Type of Alcohol?
• Level of exposure?
• Pattern of exposure?
• Timing of exposure?
Are some types of alcohol safer than others during pregnancy?

• “Hard liquor would be bad…If it burns going down…” (Branco & Kaskutsas, 2001)
  – There appears to be a common misperception among the public and even some health professionals that some types of alcohol are less damaging to the developing fetus than others
Standard Drink Calculations

• 0.60 ounce of absolute alcohol is equivalent to one standard drink
  – To calculate # of standard drinks:
    • (% absolute alcohol x # ounces)/0.60

• Examples:
  – 12 oz Budweiser: (0.05 x 12 oz)/0.60 = 1 stand. drink
  – 40 oz Olde English: (0.075 x 40 oz)/0.60 = 5 stand. drinks
  – Long Island Iced Tea: (0.40 x 2.25 oz)/0.60 = 1½ stand. drinks
Alcohol Equivalents

Each of these contains about the same amount of alcohol

- Beer
- Table Wine
- Fortified Wine
- Hard Liquor
How much alcohol is it safe to drink during pregnancy?

- Clearly, there is a dose-response relationship between alcohol consumption and teratogenic effects
- However, also see effects with lower levels of chronic drinking
  - ½ drink 3 times per week: cognitive and growth effects
  - Recent animal studies indicate the one occasion of two drinks consumed during pregnancy is enough to kill some developing brain cells, leading to permanent neurological damage
- Women differ in how they’re affected by alcohol, so we cannot predict how any particular woman’s baby will be affected: what appears to be a safe amount of alcohol for one may be devastating for another
- The pattern of binge drinking particularly damaging to fetal development
Major effects of in utero alcohol exposure by trimester

Figure reprinted with permission from: Streissguth and Little, 1994
Prevalence, Course, and Clinical Presentation of FASDs
Rates for FASDs

- Birth prevalence of FAS: 0.5 - 2/1000 births
- All alcohol related disorders (FASDs): at least 10/1000 births
- Approximately 40,000 children born with FAS or a related disorder per year
- More common than some other more well-known birth defects
Course of FASDs

• FASDS are associated with a significantly increased risk for secondary disabilities, resulting in negative long-term outcomes for many individuals affected by prenatal alcohol exposure
  – Longitudinal study of adolescents and adults with FASDs:
    • 94% had mental health problems
    • 80% had employment problems
    • 60% experienced trouble with the law
    • 50% had experienced confinement
    • 49% had engaged in inappropriate sexual behavior
    • 35% had alcohol and/or drug problems

(Streissguth et al., 1996; 2004)
Clinical Presentation: Growth Deficiency

- Prenatal and/or postnatal growth deficiency
- May see growth problems in height, weight, or both
- Growth deficits may persist in many alcohol–exposed individuals, although may see some catch up in adolescence, more often in weight than in height.
Clinical Presentation: Facial Dysmorphology

The characteristic facial features of Fetal Alcohol Syndrome can be seen in both a child and a mouse fetus that were exposed to alcohol during development.

Pregnant mice were given alcohol when the embryos were at early gastrulation stages; i.e. at a time corresponding to the third to early fourth week of human development.

- Photos reprinted with permission from Sulik, Johnston, & Webb, 1981
Clinical Presentation: Facial Features of FAS

- Photo reprinted with permission from Susan Astley, University of Washington: www.fasdpn.org
Brain damage resulting from prenatal alcohol exposure

Photo: Clarren, 1986
Structural Brain Abnormalities

- Brain structures found to be affected by prenatal alcohol exposure include:
  - Overall brain volume
  - Cerebellum
  - Corpus callosum
  - Basal ganglia
FASDs and IQ

• Although FAS is the most common known cause of mental retardation, most children with FAS are not retarded

• Studies of this population have documented IQ’s ranging from the severe range of mental retardation to the above average range of intellectual functioning

• The majority of alcohol-exposed individuals typically fall below the average range of intellectual functioning

• Children with other spectrum disorders, such as ARND, on average, have somewhat higher IQs than those with full FAS.

• Even if a child affected by prenatal alcohol exposure has a normal IQ, he/she can still have profound cognitive and learning problems, as well as deficits in other domains
CNS Dysfunction

- Attentional problems
- Learning and memory
CNS Dysfunction

- Speech and language
- Executive functioning
CNS Dysfunction

- Quantitative and numerical concepts
- Visual-spatial skills
- Motor
Recognizing FASD: Infancy

- Negative affect
- Insecure attachment behavior
- Poor habituation and orientation
- Problems with regulation
- Increased activity level
- Less mature motor behavior
- Poor sucking, feeding difficulties
Recognizing FASD: Early Childhood

- Inattention, hyperactivity
- Impulsivity
- Easily dysregulated/poor frustration tolerance
- Difficulty with transitions and adapting to change
- Difficulty following directions
- Difficulty perceiving danger
Recognizing FASD: School-age

- Academic problems in multiple domains (but may have uneven performance)
- Externalizing and internalizing problems
- Difficulty anticipating consequences of behavior
- Socially indiscriminant (friendly with everyone, even strangers), but often have no real friends
- Easily influenced by others
- Difficulty reading social cues
Recognizing FASD: Adolescence

- School failure or withdrawal
- “Innocent delinquents”
- Alcohol/substance use problems
- Inappropriate sexual behavior
Recognizing FASD: Adulthood

– Poor organization, reasoning, decision making
– Difficulties with time management
– Employment problems
– Difficulty with finances
– Relationship problems
– Psychiatric problems
– High rates of legal confinement, trouble with the law
Assessment and Diagnosis
Challenges in Preventing and Identifying FASDs

- Lack of training or expertise
- Lack of resources
- Discomfort with inquiring about prenatal exposure
- Parents’ inability or unwillingness to provide information regarding history of exposure
- Discomfort with making the diagnosis
Importance of Identifying FASDs

- Increases likelihood of individual receiving services and treatment
- Increases likelihood of individual receiving appropriate services and treatment
- Behaviors may be interpreted more appropriately as a reflection of neurocognitive impairments associated with prenatal alcohol exposure, rather than as oppositionality or sociopathy
- Opportunity to prevent additional alcohol exposed pregnancies
Importance of Identifying FASDs

• *In families in which one child is diagnosed with FAS, the incidence of FAS among younger siblings is:*

  771 in 1,000

(Abel, 1988)
Evaluation

• Multidisciplinary Approach
  – These individuals are typically impacted across multiple domains of functioning
  – Functioning in one domain often affects functioning in other domains
  – Enhances both diagnostic and treatment planning process
Evaluation

• Components of FASD Evaluation
  – Physical examination (incl. dysmorphology exam)
  – Comprehensive history
  – Psychological testing
  – Behavioral observations
  – Collateral reports (e.g., teacher, therapists, etc.)
Assessment of Maternal Drinking During Pregnancy

- Non-blaming approach
- Don’t ask IF, ask HOW MUCH?
- Inquire how many weeks pregnant the woman was when she learned she was pregnant
- Assess prior to pregnancy, prior to pregnancy recognition, and post pregnancy recognition separately
Assessment of Maternal Drinking During Pregnancy

- Assess alcohol use in context of interview about health behavior during pregnancy
- Begin with more benign substances (e.g., caffeine)
- Ask about different types of alcohol separately (e.g., beer, wine, hard liquor)
- Assess pattern of drinking (both typical and maximum consumption)
Diagnostic Systems for Identifying Fetal Alcohol Spectrum Disorders

- Hoyme, May, Jones et al., 2005
- Astley, 2004
- CDC, 2004
- Institute of Medicine, 1996
**CDC Criteria for Diagnosing FAS**

- **Growth**
  - Confirmed prenatal or postnatal height or weight ≤ 10th percentile, documented at any one point in time.

- **Facial dysmorphismology**
  - Smooth philtrum (Ranking 4 or 5 on Astley lip-philtrum guide)
  - Thin vermillion border (Ranking 4 or 5 on Astley lip-philtrum guide)
  - Short palpebral fissures (≤ 10th percentile)

- **CNS Abnormalities**
  - Structural (OFC < 10th percentile or abnormalities observable through imaging studies)
  - Neurological
  - Functional

- **Maternal Alcohol Use**
  - Confirmed
  - Unknown
Measuring Palpebral Fissure Length

![Image of an eye with labels for measurement of palpebral fissure length.](Photo reprinted with permission from Susan Astley, University of Washington: www.fasdpn.org)

Palpebral Fissure Length = endocanthion to exocanthion
Upper Lip and Philtrum: Child with FAS and Non-Exposed Child

Photos reprinted with permission from Susan Astley, University of Washington: www.fasdpn.org
Partial FAS: Diagnostic Criteria

- Confirmed alcohol exposure **AND**

- Evidence of some components of the pattern of characteristic facial features, **AND**

- Evidence of:
  - Some growth retardation, **OR**
  - CNS abnormalities, **OR**
  - A complex pattern of behavioral or cognitive abnormalities that are inconsistent with developmental level and cannot be explained by familial background or environment alone
Alcohol Related Neurodevelopmental Disorder (ARND): Diagnostic Criteria

- Confirmed alcohol exposure **AND**

- Evidence of
  - CNS abnormalities **OR**
  - A complex pattern of behavioral or cognitive abnormalities that are inconsistent with developmental level and cannot be explained by familial background or environment alone
Alcohol Related Birth Defects (ARBD): Diagnostic Criteria

- Confirmed alcohol exposure \textbf{AND}
- One or more congenital defects
  - Cardiac
  - Skeletal
  - Kidney
  - Vision
  - Hearing
Key Points

• An individual with prenatal alcohol exposure can have none of the physical signs, but still have significant brain damage.

• An individual with prenatal alcohol exposure can have a normal IQ, but still have profound cognitive, learning, and behavioral problems.

• Early diagnosis and intervention are critical in preventing secondary disabilities associated with FASDs, such as delinquency and school failure.