Fetal Alcohol Spectrum Disorder: The Real Deal, Imitators, and What Lies Ahead
Judith Eckerle Kang

Goals
- Why do we care about prenatal alcohol use?
- Trends in alcohol use and Minnesota risks
- Define FASD
- Discuss a differential diagnosis of FASD
- Diagnosis of FASD and subtypes
- How are children doing who have the diagnosis of FASD? Research and studies

Fetal alcohol spectrum disorders
- Fetal alcohol spectrum disorders (FASD) is an umbrella term describing the range of effects that can occur in an individual whose mother drank alcohol during pregnancy.

- These effects may include physical, mental, behavioral, and/or learning disabilities with possible lifelong implications.
Why do we care about prenatal alcohol use?

• 45% of pregnancies in the USA are unplanned

• Each year, as many as 40,000 babies are born with FASD, costing the Nation about $4 billion.

• Leading cause of preventable mental retardation and developmental disability in the USA and probably, worldwide

Facts about Fetal Alcohol Spectrum Disorders (FASD)

• Most people with FASD have average IQs
• Rate of FAS (Syndrome) is 1:500 births
• Rate of FASD (spectrum) is 1:100 births
• FASD is 100% preventable

On any given day in the United States 10,500 babies are born:

• 1 of these babies is HIV positive
• 7 of these babies are born with Spina Bifida
• 12 of these babies are born with Down Syndrome
• 40 of these babies are born with Autism Spectrum Disorder
• 100 of these babies are born with FASD
Other populations: FASD

- Foster care system: 1.5 per 100
- Communities with high alcohol consumption, S. Africa: 9 per 100
- Eastern European Adoptees (W. Sweden): 52 per 100

Is FASD a “New” Problem?

- “Behold, thou shalt conceive and bear a son and now drink no wine or strong drink” Judges 13:7
- Aristotle said, “Foolish, drunken, and hare-brained women most often bring forth children like unto themselves, morose and languid.”

- “Of all the substances of abuse including cocaine, heroin, and marijuana, alcohol produces by far the most serious neurobehavioral effects in the fetus resulting in life-long permanent disorders of memory function, impulse control and judgment”

— IOM Report to Congress, 1996
Alcohol Definitions

- What does moderate drinking mean?
- There is no one definition of moderate drinking, but generally the term is used to describe a lower risk pattern of drinking.
- Dietary Guidelines for Americans: no more than 1 drink per day for women (amount consumed on any single day and is not intended as an average over several days)
CDC definitions: Alcohol

• Heavy drinking
  - For women, more than 1 drink per day on average.
  - For men, more than 2 drinks per day on average.
• Binge drinking
  - For women, 4 or more drinks during a single occasion.
  - For men, 5 or more drinks during a single occasion.

• http://www.cdc.gov/alcohol/terms.htm#2

FASD in Minnesota

• FASD occurs at least three times more often than Down Syndrome and almost eight times more often than spina bifida in Minnesota.

• Minnesota has the nation’s seventh highest rate of alcohol consumption by women of child-bearing age (58%) coupled with a 50% rate of unplanned pregnancy.

FASD in Minnesota

• According to the Prenatal Questionnaire, the percentage of pregnant woman that drank ranged from 24.7% (North Dakota) to 28.5% (Minnesota).

• Of the 1100 chart reviews conducted on persons screened for FASD in Minnesota 21.4% received no diagnoses, 7.3% received a FAS Diagnosis, and 71.4% received a FAE diagnosis.
The FASD Umbrella

Factors that affect FASD Risk

The degree of damage is dependent on:
1. Dose during pregnancy – the bigger the dose, the greater the risk
2. Timing of exposure during pregnancy
3. Individual factors of the mother and child (genetic factors, metabolism and resiliency)

FASD: An Underdiagnosed Spectrum

- Data from more than 400 FASD patients in the University of Minnesota’s Fetal Alcohol Clinic database show that the majority of children (68%) with documented prenatal exposure to alcohol do not meet full diagnostic criteria for FAS (unpublished clinical data).
- Jeffrey R. Wozniak, Ph.D., Assistant Professor of Psychiatry, University of Minnesota
FASD: A Underdiagnosed Spectrum

• Fetal Alcohol Effects or FAE was coined by Clarren & Smith in 1978
• Alcohol-Related Neurobehavioral Disorder or ARND (Institute of Medicine, 1996).
• This FASD continuum includes, by some estimates, 5 to 15 times as many children as those diagnosed with full-criteria FAS


Diagnosis of FASD (Spectrum)

How is FASD Diagnosed?

1) Growth deficiency (height or weight ≤ 10th percentile).
2) A unique cluster of minor facial anomalies (small eyes, smooth philtrum, thin upper lip).
3) Central nervous system damage (structural, neurological, and/or functional impairment).
4) Prenatal alcohol exposure
1) Growth
• Centers for Disease Control (CDC) Growth Charts 2000

2) Facial Features
• “Face” of FAS forms during the 3rd week of gestation, most likely days 19-21

Lip/Philtrum Guides
• [Link to Lip/Philtrum Guides](http://depts.washington.edu/fasqipn/htmls/lip-philtrum-guides.htm)
• Reprinted with permission, Dr. Susan Astley
Rating of Lip Philtrum


FASD: eye measurements

Photographic software
3) CNS: Central Nervous System

- Microcephaly (<10%)
- Structural brain abnormalities
- Neurological clinical signs (seizures, spasticity, paralysis etc)
- 3 functional domains (neuropsych testing)
These two images are of the brain of a 9-year-old girl with FAS. She has agenesis of the corpus callosum, and the large dark area in the back of her brain above the cerebellum is essentially empty space.


4) Alcohol Exposure

- Confirmed/Unconfirmed depending on the classification system

- What do we mean by confirmed alcohol exposure?

Partial Fetal Alcohol Syndrome (PFAS)

- Most, but not all, of the growth deficiency and/or facial features of FAS.
- Central nervous system damage (structural, neurological, and/or functional impairment).
- Prenatal alcohol exposure
ARND: Alcohol Related Neurodevelopmental Disorders

• (ARND) is a diagnostic classification, coined by the Institute of Medicine in 1996, for patients who present with:
  • Central nervous system damage (structural, neurological, and/or functional impairment).
  • Prenatal alcohol exposure.

ARBD: Alcohol Related Birth Defects

• Congenital anomalies including physical malformations and dysplasia
  • Facial features
  • Cardiac, skeletal, renal, eyes, ears, minor anomalies

FASD – other clinical features

• Congenital heart defect (25 – 50%)
• Scoliosis (curvature of the spine) 10 – 15%
• Radioulnar synostosis (cannot rotate the forearms normally)
• Hypoplasia of nails (most commonly the 5th fingernails and 5th toenails)
FASD – other features

- Mixed hearing loss and otitis media – 90%
- Sensorineural (nerve damage) hearing loss – 30%
- Impaired vision (due to optic nerve hypoplasia – small optic nerves, or due to untreated strabismus)


Other diagnoses

- reactive attachment disorder
- autism
- ODD or conduct disorder
- pervasive developmental delay
- learning disabilities
- emotional/behavior problems
- ADHD


Diagnosis of FASD: Art or Science?

*growth, facial features, neurological deficits, history of alcohol exposure, *

Soft signs:
- Clinodactyly
- Maternal Age
- Multiparity
- Ears
- Hernia
- Genitourinary malformations
- Cardiac
- Cleft lip or palate
Diagnostic Systems

• 4 Digit Diagnostic Code - University of Washington, Seattle

  - 4 categories (growth, face, CNS, alcohol)
  - each on scale 1 to 4 based on severity
  - 256 possible 4 digit codes, 22 categories

IOM, revised

- The Institute of Medicine (IOM) guidelines for FAS, the first system to standardize diagnoses and revised by Hoyme in 2005
  - requires structural or neurological impairment for a diagnosis of FAS, but also has criteria for diagnosing PFAS, ARND and ARBD
  - Does not require confirmed history of alcohol exposure for PFAS


CDC

- The Centers for Disease Control's "Fetal Alcohol Syndrome: Guidelines for Referral and Diagnosis: established general consensus on the diagnosis FAS but only recognizes FAS as a diagnosis, though allows for a recognition of the spectrum.

- Fetal Alcohol Syndrome: Guidelines for Referral and Diagnosis (PDF). CDC (July 2004)

Minnesota System

- < or = to minus 1.5SD below mean in 3+ of Ten Brain Domains:
  - Achievement, adaptive behavior, attention, cognition, executive functioning, language, memory, motor skills, sensory integration or soft neurological problems, social communication

The imitators… A differential diagnosis

Cornelia de Lange syndrome (CdLS)

- distinctive facial features
- growth retardation (prenatal onset; <5th centile throughout life)
- Low-set posteriorly rotated and/or hirsute ears with thickened helices
- Long smooth philtrum, thin vermillion border of upper lip
Velocardiofacial Syndrome (VCF or DiGeorge Syndrome)

- Cleft lip and/or palate
- Distinctive facial features
- Cardiac septal defects
- Hypernasal speech
- Hypotonia
- Defective thymic development.

Valproate Syndrome

- Prenatal exposure to valproic acid (depakene, depakote) during the first trimester
- Distinctive facial features
- Neural tube defects
- Congenital heart disease
- Cleft lip and/or palate
- Genitourinary malformations
- Tracheomalacia
- Arm/hand defects
- Arachnodactyly/overlapping digits
- Abdominal wall defects
- Intellectual impairment

Why diagnose at all? What lies ahead for these children?
Why diagnose at all?

- Early diagnosis with proper intervention may decrease the appearance and attenuate the course of secondary disabilities. A diagnosis of full-blown Fetal Alcohol Syndrome before the age of 6 can help to prevent some secondary disabilities.
- The best time to identify characteristics begins around 3 to 4 years of age and continues to about 12 to 13. Facial features and growth changes in adolescence making it harder to recognize FAS.

FASD is the Invisible Disability

- Attention deficits
- Memory deficits
- Hyperactivity
- Difficulty with abstract concepts
- Inability to manage money
- Poor problem solving skills

- Difficulty learning from consequences
- Immature social behavior
- Inappropriately friendly to strangers
- Lack of control over emotions
- Poor impulse control
- Poor judgment

<table>
<thead>
<tr>
<th>Behavior</th>
<th>Misinterpretation</th>
<th>Accurate Interpretation</th>
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<tbody>
<tr>
<td>Noncompliance</td>
<td>Willful misconduct</td>
<td>Difficultly translating verbal directions into action</td>
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<td></td>
<td>Attention seeking stubborn</td>
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<tr>
<td>Often Late</td>
<td>Lazy, slow</td>
<td>Can’t understand abstract concept of time</td>
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<td></td>
<td>Poor parenting</td>
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<td></td>
<td>Willful misconduct</td>
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<tr>
<td>Stealing</td>
<td>Deliberate dishonesty</td>
<td>Doesn’t understand ownership (abstract)</td>
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<td>Lack of conscience</td>
<td>Immature thinking</td>
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<td>Lying (confabulation)</td>
<td>Deliberate Sociopathic behavior</td>
<td>Memory/sequencing problems</td>
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<td></td>
<td>Lack of conscience</td>
<td>Trying to please</td>
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“People who don't have the facial features are truly discriminated against in terms of services. When they don't have a classic FAS face, the tendency is to act as though there's nothing wrong. They are expected to perform normally, but they're goofing up all the time. They get blamed for being lazy or careless, yet these people have functional brain impairments.”
~Ann Streissguth

Secondary Disabilities

- Mental Health Problems
- Truancy at school
- Trouble with the Law
- Inappropriate Sexual Behavior
- Alcohol / Drug Problems
- Pregnancy
Protective Factors

- A diagnosis before 6 years of age
- Living in a stable, nurturing home
- Not being a victim of violence
- Having received developmental disabilities services
- Having a diagnosis of FAS rather than FAE
- Lower than 70 IQ
How do we figure out ways to help kids and adults with FASD?

Don’t Try Harder…
Try Differently
• If you’ve told a child a 1000 times and he still does not understand...

then it is not the child who is a slow learner.

Growth Endocrine Project:
FASD Facial features and Cognitive outcomes
• 122 Eastern European children, adopted between 7-59 mo of age on arrival.
• Mean age 20mo
• All from institutional care (85% entire life spent in institutional care– rest 0.87)
Measures

- Growth (Ht, wt, OFC), Neurocognitive and facial feature information was collected at all 3 time points.

- Time 1 (arrival): 122 Children
- Time 2 (6 mo post): 93 children
- Time 3 (30 mo post): 58 children: 8 children in the High Risk Face group (HRF) and 50 in the Low Risk Face group (LRF)

30 mo post adoption: Cognitive

- The HRF group had lower scores for verbal working memory (p<0.05)

Verbal Working Memory

- The verbal working memory subscale measures a child’s ability to transform, store, and retrieve verbal information in short-term memory. The subscale measures the cognitive ability to hold and sort through verbal information. In children, the ability to repeat a sentence measures short-term memory.
30 mo post adoption: Cognitive

- The HRF group had lower scores for nonverbal fluid reasoning ($p<0.02$)

Nonverbal Fluid Reasoning:

- This measures a child’s ability to solve novel problems without dependence on academic or cultural information.
- Uses visual sequences and analogical patterns to test inductive and deductive reasoning skills.
- In children, it measures the ability to identify shapes, colors and sizes in order to identify sequences and patterns.

30 mo post adoption: IQ

- No significant difference was seen in IQ and both groups scored within the normal range.
There are promising new treatment options on the horizon.

- Specifically, recent pre-clinical studies have shown that **dietary choline supplementation** prenatally and even postnatally as late as days 21-30 in the rodent (equivalent to human childhood) attenuates the memory and behavioral deficits associated with prenatal alcohol exposure.


**Online Training**

- [http://www.cme.umn.edu/cme/online/onlinefasd/home.html](http://www.cme.umn.edu/cme/online/onlinefasd/home.html)

- cmh.umn.edu

We are fortunate to have the resources to help children with special needs, and to be able to guide families to understand what the potential issues are, in order to help create healthy families here in the USA.
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