“Wisdom of Practice”
in the Diagnosis and Treatment of
Fetal Alcohol Spectrum Disorders

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The Kennedy Krieger Institute
Learning Objectives

• List three diagnostic criteria of Fetal Alcohol Syndrome
• Describe Alcohol Related Neurodevelopmental Disorder
• Analyze newer concepts of FASD
• Identify the individual at risk for an FASD
• Describe long term cognitive, learning and behavioral implications for the individual with prenatal exposure to alcohol
• Discuss the range of interventions most commonly required by individuals with FASD
Alcohol is a potent neurotoxic substance when exposed to a developing brain.

No Amount of Alcohol is safe to use in pregnancy
FASD - Fetal Alcohol Spectrum Disorders

• An umbrella term describing the range of effects that can occur in an individual whose mother drank during pregnancy. These effects may include physical, mental, behavioral, and/or learning disabilities with possible lifelong implications.

• Bertrand et al. 2004
1. Individuals do not “grow out” of the central nervous system effects.
2. It is a diagnosis of exclusion, can appear like other disorders, and can co-exist with other disorders.
3. It is a serious life long developmental disability from the neurotoxic effects of prenatal alcohol on the developing brain.
FASD Mental Health Issues
Transcend DSM Criteria

• When meds are helpful there are still unexplained and seemingly unprovoked behavioral episodes
• Family or caretaker is exhausted and often angry, depressed or bewildered
• After years of psychotherapy and medication patient may still be unsuccessful or not progressing
1 in 100 individuals may have a FASD
Alcohol Diffuses to the Fetus during Gestation and in Breast Milk during Breastfeeding
<table>
<thead>
<tr>
<th>Period of the Ovum</th>
<th>Period of the Embryo (in weeks)</th>
<th>Period of the Fetus (in weeks)</th>
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<tbody>
<tr>
<td>1-2</td>
<td>3</td>
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<td>20-36</td>
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<tr>
<td>CNS Heart</td>
<td>Eye Heart Arm Leg Teeth</td>
<td>Brain</td>
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<tr>
<td></td>
<td>Eye</td>
<td>Palate Ear</td>
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<td>External Genitalia</td>
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<tr>
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<td>Ears</td>
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</table>

- Central Nervous System (CNS)
- Heart
- Arms
- Eyes
- Legs
- Teeth
- Palate
- External Genitalia
Deleterious effects appear multifactorial. Fetuses are differentially susceptible to the effects of alcohol exposure.
Fetal Alcohol Spectrum Disorders (FASD)

1. FAS - Fetal Alcohol Syndrome
2. pFAS - Partial Fetal Alcohol Syndrome
3. ARBD - Alcohol Related Birth Defects
4. ARND - Alcohol Related Neurodevelopmental Disorder
Fetal Alcohol Syndrome

• Low birth weight
• Central Nervous system effects
• Facial Dysmorphology

(Modern description of FAS published in early 1970’s)
Updated Criteria for Fetal Alcohol Syndrome

Criteria for Diagnosis:
1. Growth retardation-height and/or weight
2. 3 Dysmorphic facial features (short palpebral fissures, flattened philtrum, thin upper lip)
3. Cognitive Disability( at least 3 of the following: motor skills, speech and language, adaptive living skills problems, executive functioning, social skills disability, attention, hard neurological findings, LD, MR, etc.)
4. The presence of prenatal alcohol exposure helpful but not needed for this diagnosis

Canadian Criteria
The Facial Features of FAS in mouse fetus that was exposed to single binge of alcohol during 1\textsuperscript{st} trimester.

Sulik, 1996
Fetal Alcohol Syndrome
Examples of Physical Manifestations of FAS

- Growth- pre and/or post natal growth retardation
- Average IQ-65 range 20-105
- Poor eye hand coordination, tremulousness
- Irritability and hyperactivity
- Microcephaly, short palpebral fissures, maxillary hypoplasia, short nose, smooth philtrum and thin and smooth upper lip, micrognathia
- Joint abnormalities, abnormal position or function
- Cardiac abnormalities, ptosis
- Ophthalmic abnormalities
Partial FAS

- Confirmed prenatal alcohol exposure
- Evidence of 3 or more central nervous system domains:
  - Memory, brain structure, adaptive functioning, social communication, ADHD, soft neurological signs
- Simultaneous presentation of 2 facial anomalies at any age:
  - Short palpebral fissure length
  - Smooth or flattened philtrum
  - Thin upper lip
Alcohol Related Birth Defects

- Congenital anomalies
- Dysplasias
- Confirmed alcohol exposure
Alcohol-Related Neurodevelopmental Disorder

- Confirmed Alcohol Exposure; and
- Evidence of 3 or more central nervous system domains:
  - Memory, brain structure, adaptive functioning, social communication, ADHD, soft neurological signs, etc. (2 standard deviations below the mean)
Costs of FASD in the United States

- FAS costs US $5.4 billion in 2003
- An FAS birth carries lifetime health costs of $860,000 although can be as high as $4.2 million
- Including quality of life, FAS prevention may be “cost effective” at up to $850,000 per child

National Organization on Fetal Alcohol Syndrome
Russian prosecutors to investigate adoption procedure of boy who died in US

AP Worldstream; August 5, 2005; MARIA DANILLOVA, Associated Press Writer; 343 Words

... about the fate of Russian-born children adopted ... prompted a senior Russian lawmaker to call on halting adoptions by U.S. citizens ... Merryman was the 13th Russian-born child to die ... allowed foreign adoptions in the early 1990s ... said. Some 260,000 Russian orphans are ...
“….. the rate of Fetal Alcohol Syndrome was about 45 per 1,000 school entry children, in the first study. About 70 per 1,000 in the second study. It may be as high as 85 per 1,000 in the third study".

Professor Denis Viljoen, head of Human Genetics at Wits University in Johannesburg.
<table>
<thead>
<tr>
<th>Susceptibility (Risk) Factors</th>
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<tbody>
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<td>• Pattern</td>
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<td>• Timing</td>
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<td>• Dose</td>
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<td>• Genetic factors</td>
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<tr>
<td>• Parity</td>
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<tr>
<td>• Age of the mother</td>
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<tr>
<td>• Binge drinking</td>
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<tr>
<td>• Smoking</td>
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<td>• Other drug use</td>
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<td>• Constitutional factors</td>
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<tr>
<td>• Physical health</td>
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<td>• Poor nutrition</td>
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<td>• Trauma</td>
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<td>• Stress</td>
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</table>
How much alcohol is safe???

• Cannot ever be sure
• Risk is based on multiple factors
• Binge drinking may be more dangerous
• Genetic vulnerability across populations for both mother and child
• Environmental factors play an unappreciated role in affecting outcome
Animal models – Example of the comparability of effects

- Growth retardation
- Facial characteristics
- Heart, skeletal defects
- Microcephaly
- Reductions in basal ganglia and cerebellar volumes
- Callosal anomalies

- Hyperactivity, attentional problems
- Inhibitory deficits
- Impaired learning
- Perseveration errors
- Feeding difficulties
- Gait anomalies
- Hearing anomalies

Driscoll, et al., 1990; Samson, 1986
Alcohol is More Neurotoxic than Cocaine, Heroin, PCP or Marijuana
Alcohol Dose

- 1 drink/week-hyperactive and aggressive behaviors
- Moderate to heavy use-delinquent behavior and overall problem behavior
- Any alcohol use in pregnancy
  - 3.2X the risk for delinquent behavior

Sood et al. 2001
Brain-Behavior Principles

• The extent of damage is in a “dose-response” relationship although this is modulated by genetic variability (Binge drinking is worse)

• There appears to be a synergistic effect of certain added compounds like cocaine on the developing CNS
Brain-Behavior Principles

- Parts of the brain are affected differentially by alcohol
- Certain regions of the brain are damaged and other regions are spared
- Certain cell types are damaged whereas certain cell types are spared
- Most neurotransmitters systems appear to be affected
- The absence of dysmorphismology does not indicate that the individual is spared
Neuropsychological Findings

- Verbal learning
- Visual motor integration
- Memory
- Academic skills
- Fine motor skills and speed
- Language skills
- Mathematics skills
- Executive functioning

Mattson and Riley, 2000
Neuropsychological Performance

Mattson, et al., 1998
CNS Effects

• Depends upon developmental period the exposure occurs
• Depends upon the sensitivity of the region to alcohol’s toxic effects
• Cell types throughout the CNS and within the same structure are differentially sensitive to the toxic effects during certain times in gestation
Neuronal Effects

- neurogenesis
- neuronal differentiation
- neuronal migration
- arborization
- synaptogenesis

Miller, 1986
Regions of the Brain Most Commonly Affected By Prenatal Alcohol Exposure

- Frontal Lobes
- Parietal Lobes
- Corpus Callosum
- Basal Ganglia
- Cerebellar Vermis
Alcohol is a Midline Teratogen

- Key facial changes are related to midface hypoplasia.
- In 2001, Astley and Clarren evaluated the correlation of facial dysmorphology with brain dysfunction in a group of children with prenatal alcohol exposure.
- They found more children with more severe facial phenotypes demonstrated more impaired levels of cognitive, neuropsychological, and visual motor functioning.

Astley and Clarren, 2001
Corpus callosum abnormalities

Mattson, et al., 1994; Mattson & Riley, 1995; Riley et al., 1995
Corpus Callosum Abnormalities

Lockhart, P, Mahone, M., Mostofsky, S unpublished data
Alcohol and the Cerebellum

Purkinje Cell Layer

Pictures courtesy of James West
Suspected Mechanisms Implicated in CNS Damage

- Cell death modes (necrosis and apoptosis)
- Free radical damage
- Interference with growth factor functions
- Adverse effects on astrocyte formation
- Abnormal development of neurotransmitter system
- Altered glucose transport and uptake
- Abnormal cell adhesion molecules
- Altered regulation of gene expression
Individuals with FASD have a range of secondary disabilities that the individual is not born with and which could be ameliorated with appropriate interventions.
Secondary Disabilities

Streissguth, et al., 1996
Diagnosis of FASD

- Individuals with FAS and ARND will of course appear different
- But these individuals may be equally cognitively and behaviorally disabled
- Because individuals with ARND are usually not identified early they have endured more environmental distress and may have more secondary symptoms
Disorders that Resemble FASD

- Noonan’s Syndrome
- Williams Syndrome
- Dubowitz Syndrome
- Aarskog Syndrome
- Fetal anticonvulsant syndrome
- Other chromosome deletion and duplication syndromes
What are Problems Interfering with Health Professionals Providing “good enough services”

1. The disorder is not housed in the DSM
2. There is are few places to obtain a consultation
3. There is no text where this information is easily obtained in rapid style
4. These patients can often look happy and healthy thereby misleading the practitioner who has to make a rapid decision about treatment that the patient is stable
Caretakers

• Be ready to support the caretakers
• Require much time to absorb the reality of the situation of having a special needs child (may take years)
• Need understanding from helping professionals (therapist may need support from colleagues to avoid “burn-out”)
• Blame and or provoking guilt should be avoided in all interactions (most parents just don’t know that their behavior towards the child is unjustified)
• They have to be taught to understand and be more accepting
• Be ready to believe the fantastic stories they report
• These stories are generally true
• Need to evaluate the neurotic issues and stress behavior of caretakers
Caretaker-Child Problems

- “Goodness of Fit”
- Seriousness of the disability
- Intensity of the wish for a typical child
- Difficulties in obtaining adequate medical services
- Treatment planning
Patients Can Achieve Stability

• Despite the complexity of some of our patients with FASD many can achieve a certain level of stability over time when specific areas of their functioning are prioritized.

• In the more impaired individuals their problems are multifactorial and therefore a complex interplay between:
  − Cognitive factors
  − Environmental conditions
  − Susceptibility to behavioral and emotional reactivity
  − Genetic predisposition for psychiatric disorder or developmental disorders
  − Somatic disorders

• Tackling these problems requires an integrated context oriented approach.
Sample Diagnostic Work Up

• Dysmorphology evaluation
• Possible genetic testing
• Lead level
• MRI
• EEG
• Neurological evaluation
• Neuropsychology Evaluation
• Speech and Language Evaluation
• Occupational Therapy Evaluation
• Behavioral Psychology Evaluation
Assessment instruments

- BRIEF
- SIB-R
- Connor’s
- CBCL
- Sensory Processing
- Developmental history
- Parent Stress index
- BASC
- SNAP
FAS Facial Photographic Analysis Report

**IDENTIFICATION**

Name: Kinya  
First:  
Middle:  
Last: Marangu  
Subject I.D.: 10-41-16  
Source of Photo: Clinic  
Gender: Female  
Race: African American  
Birth Date: 11/7/1999

* Normal PFL Charts: Inset  
Lip-Philtrum Guide: African American

<table>
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<td>Photo Assessor</td>
<td>Boyle</td>
<td>Boyle</td>
</tr>
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</table>

PHOTO ASSESSMENT

Length of Real Internal Measure of Scale(sticker) placed on forehead (mm): 25.4
Length of Internal Measure of Scale in Frontal Photo (pixels): 226.7

| Left Palpebral Fissure Length | In photo (pixels): 183.0 | True Length (mm): 23.8 | Z-score: 3.34 |
| Right Palpebral Fissure Length | In photo (pixels): 204.0 | True Length (mm): 25.1 | Z-score: 2.97 |
| Mean Palpebral Fissure Length | In photo (pixels): 198.5 | True Length (mm): 24.5 | Z-score: 3.19 |
| Inner Canthal Distance | In photo (pixels): 237.0 | True Distance (mm): 26.6 |

Flat Philtrum (5-point rank): In Frontal Photo: 5  
In ¾ Photo: 5

Thin Upper Lip: Circularity (perimeter/area): 72.3  
5-Point rank (Circ): 5  
6-Point rank (Scale): 4

- clown eyebrows  
- ptosis  
- strabismus  
- epicanthal folds  
- flat midface  
- protruding ears  
- flat nasal bridge  
- hypertelorism

Other anomalies present: See (NOS)

Comments: slight head tilt toward patients right shoulder secondary to posture as right shoulder is a bit lower too. Attempts to correct posture. Other syndromes present: None reported

PHOTO QUALITY

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<thead>
<tr>
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<th>Frontal</th>
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<tr>
<td>Exposure (5-point rank)</td>
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<tr>
<td>Focus (5-point rank)</td>
<td>1 (good)</td>
<td>1 (good)</td>
</tr>
<tr>
<td>Facial Expression (5-point rank)</td>
<td>1 (Relaxed)</td>
<td>1 (Relaxed)</td>
</tr>
<tr>
<td>Reliability of ABC Score for palpebral fissure length (5-point rank)</td>
<td>2 (good)</td>
<td>2 (good)</td>
</tr>
<tr>
<td>Reliability of ABC Score for philtrum (5-point rank)</td>
<td>1 (very good)</td>
<td>1 (very good)</td>
</tr>
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</table>

OUTCOME

ABC-Score  
Data Used  

4-Digit Diagnostic Code for Face: FAS features severe

University of Washington FAS DPN FAS Facial Photographic Analysis Software © 2006
Diagnosis of FASD

• Diagnosis of Exclusion
• Can have major Axis I diagnosis/es but features of FASD may also appear like bipolar disorder, autism, conduct disorder, etc.
• Important to look at the quality of the symptoms and how close they are to DSM IV criteria
• Facial dysmorphic features are suggestive of FASD but also rule out presence of a genetic disorder
• Growth retardation needs to be ruled out (chart growth—are there any reasons for non-alcohol associated growth problems)
• Contribution of psychosocial problems to the symptoms
• What are the protective factors
Treatment of the Central Nervous System
Effects of Prenatal Alcohol Exposure

Hope derives from new concepts of treatment:
- Psychopharmacology (improving cognition, reduction of anxiety and mood problems)
- Psychotherapy (family support, repetitive messages)
- Environmental manipulation (structure, mentoring, etc.)
- Parenting therapy
- Speech and Language (social skills practice)
- Occupational Therapy (motor and sensory system treatment)
- Behavioral Therapy (reward systems)
Spectrum of Disability

Speech and Language
Motor Skills
Adaptive functioning
Executive functioning
Somatic problems
Environment
Axis I diagnoses

All of these areas of disability can negatively impact on the treatment of these patients if not factored in in a dynamic manner.
Treatment

- Most treatment protocols are not rigorously researched
- Medication treatment of Axis I diagnoses teasing out the cognitive from the major diagnoses decreases pain and suffering
- Structure, support, limits and close direction are a must
- Rewards built in are more helpful than punitive consequences
- Sexuality, drugs, victimization and boundaries must be carefully taught
- Talk therapy can be helpful to improve communication and decrease outbursts.
- Cognitive disability needs to be factored into the types of therapy used
The Psychiatrist in Partnership with other Health Professionals

- Internists and Neurologists
- Social Workers and care coordinators
- Occupational Therapists
- Speech and Language Pathology
- Behavioral Psychologist
- Dysmorphologist
- Respite agencies
- Behavioral Aide agencies
Presenting Complaints

Aggression, Hearing Voices, History of trauma

Cognitive Problems

Emotional/Behavioral symptoms

Environmental Factors

Multigenerational

Somatic

Sensory Integration dysfunction

Nightmares

Flashbacks

Attention problems

Auditory and Visual Hallucinations

Injuring sibling

Attachment issues

Multiple foster placements

Early and recurrent sexual abuse

Physical abuse

Lost for 3 days

Early neglect

Drug and alcohol abuse in parents

Psychiatric problems in mother

Traumatic brain injury

History of Seizures

Fine and Gross Motor skills

Expressive and Receptive Language Problems

IQ-Mild MR
Case Examples

• 15 year old that does not copy homework off the board, and has multiple tobacco violations
• 16 year old girl who can’t say “no” to boys
• 25 year old who has been homeless since his family put him out
• 37 year old who has “melt downs” and needs to live with someone to help organize her life
• 51 year old who needs to live with his significant other who no longer is interested in him
Personal Challenges of the Professional

- Patiently letting all the information unfold
- Being non-judgmental
- Avoiding demoralization
- Being a friendly supporter and objective at the same time
- Being able to step back from the situation
- Allowing the parent and patient to teach us
- Maintaining energy level in the face of disaster
- Knowing how to ask for help from colleagues
- Being consistent
- Being kind when under stress
Improving the Outcome of Individuals with FASD

• The non-medication therapies should be appropriate to the cognitive abilities of the individual
• The environment of the affected individual should be considered an extension of the therapy
Infant Screening

• Failure to thrive
• Small for gestational age
• Obvious dysmorphic features
• Developmental delays
• Unexplained medical complications
• History of substance or frank alcohol exposure
Early childhood

- Extreme hyperactivity and impulsivity
- Overwhelmed easily by sensory stimulation
- High pain threshold
- Does not learn from mistakes
- Intrusive
- Irritable; many meltdowns
- Not meeting developmental milestones
- Motor or language delays
- Mental retardation
- Prenatal substance exposure
Preadolescence

- Immaturity
- Poor social skills
- Inappropriate
- Tells “tall” tales
- Takes things
- Language and motor skills delays
- Melt downs

- ADHD
- LD (especially math)
- Poor peer relatedness
- Poor boundaries
- No friends
- Everyone is their friend
- Parents angry
Adolescence

- Immaturity
- ADHD
- Moody, temper tantrums
- Can’t take responsibility
- Few or no friends
- Substance exposure
- Using substances

- Poor generalization
- Poor cause and effect reasoning
- Doesn’t learn from mistakes
- Inappropriate
- Steals, tells “tall” tales
- Lack of independence for age
Adult

- Lack of independence
- Poor adaptive functioning
- Psychiatric disability
- Poor executive functioning
- ADHD
- LD

- Immature
- Does not learn from mistakes
- Cannot hold a job
- Still living at home
- Easy victim
- May have been arrested
Psychiatric Care

- All medications that are commonly used in psychiatric care should be considered
- Making certain that we safely prescribe is the important issue
- Getting proper medical work up may include EKG or EEG
- Monitoring vital signs, height and weight are very important when meds are prescribed
FASD ABC Checklist

• Appearance-Small eyes
  - Flattened philtrum
  - Thin upper lip
• Behavior- Does not understand consequences of behavior
  - Attention problems/Impulsivity
• Cognition- Mental retardation
  - IQ not commensurate with abilities

Any patient with known prenatal alcohol exposure should be screened for disability as early as possible.
Research demonstrates that there is no safe amount of alcohol to consume during pregnancy.
Prevention is the Key to Complete Elimination of this Very Serious Public Health Problem
Practice prevention in your own lives and that of family, friends and social contacts.
Thank you!!!