FAS/FASD: Screening, Diagnosis, and Treatment

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• Diagnosis
• Screening
• Treatment
FAS Diagnostic Criteria

• Fetal Alcohol Syndrome is a *clinical* diagnosis
• CDC criteria (2004) are used as they are based upon the most current data and the definitions put forth in the FAS Guidelines for Referral and Diagnosis report.
FAS Diagnostic Criteria

• Clear diagnostic criteria can help health care providers identify children
• With diagnosis, children can get the care and services they need
• Early identification can help prevent secondary disabilities
• Documenting maternal alcohol exposure is critical, but often difficult to obtain
• Maternal alcohol use is not essential to making FAS diagnosis
• Can make diagnosis 2 ways:
  ● Confirmed prenatal alcohol exposure
  ● Unknown maternal alcohol exposure
FAS Diagnostic Criteria

- Rule out other possible diagnoses
- There are facial features not unique to FAS, so differential diagnosis is important
Facies in Fetal Alcohol Syndrome

Discriminating Features
- short palpebral fissures
- indistinct philtrum
- thin upper lip

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

In the Young Child
FAS Diagnostic Criteria

• Documentation of all 3 facial abnormalities
  • Smooth philtrum
    — Lip philtrum guide 4 or 5
  • Thin vermilion (upper lip)
    — Lip philtrum guide 4 or 5
  • Small palpebral fissures
    — $\leq 10^{th}$ percentile
• Documentation of growth deficits
• Documentation of CNS or neurobehavioral disorders

*CDC/NCBDDD Scientific Working Group, 2004*
Philtrum and Vermillion
Philtrum and Vermillion Demonstration

- Lips gently closed
- No smile
- Examiner’s eyes in line with patient’s
- Match to ethnic photos

Photo: http://www.bbc.co.uk/insideout/west/series2/images/fetal_diagnosis_150.jpg
Palpebral Fissures

- OC (Outer Canthus)
- IC (Inner Canthus)
- PF (Palpebral Fissure)
Measuring Palpebral Fissures
FAS Diagnostic Criteria

• Documentation of growth abnormalities
  ● Prenatal or postnatal weight and/or height \( \leq 10^{\text{th}} \) percentile
  ● Adjusted for age, gender, gestational age, race, and ethnicity
Length & Weight
Growth in FAS - Males
Growth in FAS - Females
FAS Diagnostic Criteria

- Documentation of Central Nervous System or Neurobehavioral Disorders
  - Structural
  - Neurological
  - Functional
FAS Diagnostic Criteria

• Structural Disorders
  - Head circumference (OFC) ≤ 10th percentile
  - Brain abnormalities observed via
    - Imaging
    - Seizures
    - Impaired motor skills
Head Circumference
FAS Diagnostic Criteria

• Neurological Disorders
  ● Seizures not due to postnatal insult
  ● Impaired motor skills
  ● Sensorineural hearing loss
  ● Memory loss
  ● Poor hand-eye coordination
FAS Diagnostic Criteria

• Functional Disorders
  • Below average scores on standardized instrument or clinical impression of functional deficit in one of the following domains:
    • General Cognitive Deficits
    • Executive Functions
    • Motor Functions
    • Social Skills
  • Attention Deficit/Hyperactivity
  • Mental Health Problems
  • Other
FAS Diagnostic Criteria

• 70% of children diagnosed with FAS will also be diagnosed with ADD/ADHD.
FAS

- Diagnosis
- **Screening**
- Treatment
FAS Screening

• Morphological examination
  • Height
  • Weight
  • Head circumference
  • Palpebral fissure measurement
  • Philtrum assessment
FAS Screening

• When in doubt, suspicious, or screen positive, consult:
  • Dysmorphologist or clinical geneticist
  • Neuropsychologist
  • Developmental pediatrician

• Diagnosis best made by a dysmorphologist or clinical geneticist with experience in FASD
FAS Screening

- Cognitive
- Adaptive/Functional
- Language
- Motor
  - Gross
  - Fine
- Social Skills
- Emotional Development
- Academic Achievements
- Most will be on an IEP

Choosing the proper type of testing is best performed by a developmental physician, pediatric clinical psychologist or neuropsychologist.
Considerations for Initiating Referrals

- Confirmed heavy prenatal alcohol exposure
- In the following instances, with or without maternal alcohol exposure confirmation:
  - Any report of concern by a parent or caregiver
  - When all three facial features are present
  - When one or more facial features are present along with growth deficits in height and/or weight
Considerations for Initiating Referrals

In the following instances, with or without maternal alcohol exposure confirmation:

- When one or more facial features are present along with one or more CNS or neurobehavioral deficits
- When one or more facial features are present along with growth deficits and one or more CNS or neurobehavioral deficits
Most children exposed to high levels of alcohol in the womb do not develop the distinct facial features seen in fetal alcohol syndrome, but instead show signs of abnormal intellectual or behavioral development, according to a study by researchers at the National Institutes of Health and researchers in Chile. These abnormalities of the nervous system involved language delays, hyperactivity, attention deficits, or intellectual delays. The researchers used the term functional neurologic impairment to describe these abnormalities. The study documented an abnormality in one of these areas in about 44 percent of children whose mothers drank four or more drinks per day during pregnancy. In contrast, abnormal facial features were present in about 17 percent of alcohol exposed children.
FAS

- Diagnosis
- Screening
- Treatment
FAS/FASD – Role of Health Providers

- Primary care provider
  - Manage routine issues related to health care and FAS including
    - Behavior
    - Pharmacotherapy
    - Preventive medicine/anticipatory guidance
  - Educate/refer mother to prevent recurrence
FAS/FASD – Role of Health Providers

• Dysmorphologist
  - Aid in diagnosis, differential diagnosis
  - Monitoring of issues related to FAS

• Developmental pediatrician
  - Evaluate over time the developmental needs of the individual
FAS/FASD – Role of Health Providers

- **Psychologist**
  - Neurodevelopmental testing on individual
  - Family counseling regarding diagnosis

- **Social Worker**
  - Helping family to deal with stress of disorder
  - Access to services
FAS/FASD – Role of Health Providers

• Therapists
  ● Maximize potential through early and persistent intervention
  ● Use of adaptive techniques to overcome disability

• Patient/family advocates
  ● Provide respite opportunities for family
  ● Ensure that proper referrals are made for family and child within the resources of their community
  ● Provide long-term foresight and planning
Pharmacotherapy – Neuropsychiatric Issues

- Attention problems
- Depression and mood swings
- Sleep
- Aggression and impulse control
Attention Deficit Hyperactivity Disorder (ADHD)-Related Behavioral Problems

- **Dextroamphetamine (Dexedrine)**
  - 2.5-5 mg/day (max 40 mg/day)

- **Mixture of dextroamphetamine and levoamphetamine salts (Adderall)**
  - 2.5-5 mg/day (max 40 mg/day)

- **Methylphenidate (Ritalin, Concerta)**
  - Ritalin immediate release, 5-20 mg BID (max 72 mg/day), 10-60 mg/day adults
  - Concerta extended release, 18 mg/day (max 54 mg/day) children and adults
Attention Deficit Hyperactivity Disorder (ADHD)-Related Behavioral Problems

- Adderall (hyperactivity) and Strattera (concentration) together have been beneficial for some children diagnosed with FAS and ADHD.
ADHD-Related Behavioral Problems

• Pilot study (2000):
  ● 22% positive clinical response to methylphenidate
  ● 79% positive clinical response to dextroamphetamine

• These medications have differing effects on cerebral metabolism.

ADHD-Related Behavioral Problems

- Atomoxetine (Strattera)
  - 0.5 mg/kg/day in children (max 1.4 mg/kg/day)
  - 40 mg/day in adults (max 100 mg/day)
  - Lower side effects than stimulants
  - Anecdotally beneficial when combined with extended release Concerta.
Pharmacotherapy - Depression & Mood Swings

SSRIs

- Fluoxetine (Prozac)
  - Children 5-10 mg/day (max 20 mg/day)
  - Adults 20-80 mg/day (max 80 mg/day)

- Sertraline (Zoloft)
  - Children 25-200 mg/day
  - Adults 50 – 200 mg/day

- Paroxetine (Paxil)
  - Children 10 mg/day
  - Adults 20-50 mg/day
Pharmacotherapy - Depression & Mood Swings

• Zoloft is the only antidepressant approved by the FDA for children.
Pharmacotherapy - Depression & Mood Swings

SSRIs

• Fluoxetine (Luvox)
  • Children 25-200 mg/day
  • Adults 50-300 mg/day

• Citalopram (Celexa)
  • Children – no established dosages
  • Adults 20-40 mg/day (max 60 mg/day)

• Bupropion (Wellbutrin)
  • Children – no established dosages
  • Adults 100-450 mg/day
Pharmacotherapy - Sleep

• Melatonin – Natural & Not Addictive
  - Children 0.5-10 mg qHs
  - Adults 3-30 mg qHs

• Lorazepam (Ativan)
  - Children 0.5 mg qHs
  - Adults 2-4 mg qHs

• Zolpidem (Ambien)
  - Children—no established dosages
  - Adults 5-10 mg qHs

• Trazodone (Desyrel)
  - Children—no established dosages for sleep
  - Adults 50 mg qHs
Pharmacotherapy – Neuropsychiatric Issues

• All of these medications may have significant side effects
• Patients must be monitored closely by prescribing physician
• Must be aware of continually changing FDA public health medication advisories
Fetal Alcohol Spectrum Disorders

We see what we look for…. - and –
we look for what we know!
• The best practices in the care of a child with a FASD are....

Early recognition and
Early intervention!