Fetal Alcohol Syndrome and Fetal Alcohol Effects

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A.M.B.E.R. clinic
Albuquerque Multidisciplinary Behavioral Evaluation for Recovery and Resiliency

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Overview

- **Diagnostic criteria**
- **Phenotypic variation**
- **Clinical assessment**
- **Multidisciplinary approach**
- **Lifespan issues:**

  secondary complications

Diagnosis

- **Fetal Alcohol Spectrum Disorders**
  - **FAS: Caused by maternal ingestion of alcohol during pregnancy, resulting in physical and intellectual disorders.**
- **Alcohol-Related Neurodevelopmental Disorder (ARND)**
- **Alcohol-Related Birth Defects (ARBD)**

Diagnosis

- **Prevalence: 0.2 -1.5/1,000 births**

  0.5 -2.0/1,000 live births
- **1,000 – 6,000 est. affected/4 M born (US); increases in high risk groups**
- *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. The term FASD is not intended for use as a clinical diagnosis*

Diagnosis

- **Criteria:**
  - **Documentation of all 3 facial abnormalities (smooth philtrum, thin vermillion border, small palpebral fissures)**
  - **Documentation of growth abnormality**
  - **Documentation of CNS abnormality**
- **Common symptoms: chaos!**
  - **Small head circumference**
  - **Deficits: cognitive fx; executive fx; motor delays; attention; social skills; sensory problems…**

Phenotypal Variations…

- **Dysmorphia**
  - interference with nerve cell development and functioning; increased formation of cell-damaging free radicals; altered pathways of biochemical signals within cells; altered expression of certain genes and genetic information.
  - (Jones, 1973) **short palpebral fissure, maxillary hypoplasia (with prognathism), and the presence of epicanthal folds; noted for some patients: altered palmar flexional crease patterns (i.e., hockeystick crease), cardiac anomalies, joint disability, overlapping fingers, ear anomalies, hemangiomas, ptosis, hypoplastic nails, and pectus deformities. Additional features described included: microcephaly, short nose, smooth philtrum with thin vermillion border, cleft lip, micrognathia, protruding auricles, short or webbed neck, vertebra and rib anomalies, short metacarpal bones, menigomyelocele, hydrocephalus, and hypoplastic labia majora** (Jones et al, 1997)

Phenotypal Variations

- **Clinically significant brain changes**
  - **decr. Corpus callosum, cerebellum, basal ganglia**
- **Function:**
  - **Global reduction in IQ**
  - **3 domains of function impaired**
    - **cognitive skills**
    - **executive functions**
    - **motor fx (gross; fine)**
    - **attention & hyperactivity**
    - **social skills**
    - **sensory defensiveness**

Phenotypal Variations

- **Conduct disorder**
- **Oppositional defiant disorder**
- **Anxiety disorders**
- **Sleep disorders**
- **Depressive disorders**
- **Adjustment disorders**

Clinical Assessment

- **Obtain history from the mother**
- **Physical examination for physical signs and symptoms**
- **History of behavior and developmental milestones; neurodevelopmental concerns**
- **Referral to specialist for confirmation**

Clinical Assessment

- **Referral to specialist:**
  - **specific measurements of dysmorphic development**
  - **standardized assessment using imaging techniques**
  - **standardized measurement of cognitive function**
  - **baseline for response to interventions**

Multidisciplinary Approach

- **Ongoing supports for developmental stage to enhance learning and development**
- **Environmental modulations so that the child is not living in perpetual overwhelmed state**
- **Communication with primary care provider; within team.**
- **Anticipate changing needs**
- **Education and support of family**

Lifespan Issues

- **Mental illnesses**
  - **broad spectrum**
  - **idiosyncratic responses to medication**
  - **may need life-long support**
- **Reactivity to environmental stressors may not diminish over time – more like a static injury**

Challenges

- **Under-recognition of syndrome**
- **Lack of supports across disciplines**
- **Social factors supporting substance abuse in potential mothers**
  - **Drug access; poverty; lack of social supports…**

Summary

- **FASD is under-recognized**
- **Education is a major step toward prevention**
- **Prevalence is comparable to DS or spina bifida**
- **Lifetime supports make a difference in prevention of secondary complications: injuries, arrests, violence…**