Overview of FASD

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What is FASD and How is it Diagnosed: Overview

- History and terminology
- Prenatal exposure to alcohol as a teratogen and risk factor
- Complexity of the diagnosis of FASD
- Multidisciplinary approach to assess for evidence of organic brain damage
- Presentations across the lifespan
- Impact of diagnosis on individual, caregiver and community

History of FASD

- Biblical and early artwork
- Initial medical reports in 1968 and 1973
- Institute of Medicine, USA, 1996
- 4 Digit Code 1997, 1999, 2004 (Astley, Clarren)
- CDC Guidelines 2004 (Bertrand)
- Canadian Guidelines for Diagnosis 2005 (Chudley)
- International perspectives (Reily)

Terminology

- FASD - umbrella term referring to lifelong disability from prenatal alcohol exposure (PAE) and includes:
  - Full FAS
  - Partial FAS
  - ARND Alcohol Related Neurodevelopmental Disorder ("invisible disability")
  - Common features: confirmed history of PAE and evidence of organic brain damage by objective testing and history

Alcohol as a Teratogen

- Animal research on the biological mechanisms of alcohol in fetal brain damage
  - Direct effect on neuron maturation, migration and organization (Sulik)
  - Oxidative stress, glutathione depletion in neuron mitochondria (Brien)
  - Neuroendocrine and neurotransmitter (Weinberg)

Alcohol as a Teratogen

- Indirect effect
  - Effect on placenta vasoactivity via prostaglandin (Cook)
  - Alcohol impact on maternal health and other teratogen exposures
  - Role of alcohol use by male partner on stress factors

Alcohol as a Teratogen

- Spectrum of damage depends on:
  - Amount and pattern, binge impact
  - Time in gestation: face day 19 to 21, brain throughout
  - Other maternal and fetal factors (genetics, epigenetics, nutrition, drugs, health, twin discordance)
  - **KEY:** Alcohol **EXPOSURE** is a **RISK** but NOT a **DIAGNOSIS** for FASD

Alcohol as a Teratogen

- Neuroimaging evidence for brain damage
  - Structural MRI and Volumetric studies; corpus callosum, cerebellum, caudate (Matson)
  - Decrease in frontal lobe volume with severity of facial dysmorphology (Astley)
  - Fetal ultrasound (UCLA)
  - FMRI: less activation in the prefrontal area with increasing complexity of task (Chudley)
  - DTI MRI: differences in white matter pathways

Biological Markers for FASD

- Facial dysmorphology midfacial hypoplasia (day 19 to 21 PAE) <10% of most clinic populations; foster care data shows higher incidence
- Growth deficiency (differential, familial)
- Meconium analysis of fatty acid ethyl esters (Bearer, Koren) limitations
- Saccadic eye movements (Reynolds)

Impact of Postnatal Environment

- Direct effect on early brain development of deprivation, abuse, nutrition factors
- More vulnerable if already brain damaged in utero
- Critical time periods to optimize brain development (interventions)

Diagnostic Criteria

- Confirmed PAE
- Prenatal/postnatal growth deficiency <10% (population curves CDC)
- Facial features: thin upper lip, flat philtrum, short palprebral fissures (photographic analysis, population norms, ethnicity)
- Evidence of brain damage (need multidisciplinary team data)

Diagnostic Process

- Tool: 4 – Digit Diagnostic Code (Astley and Clarren) quantitative and objective
- Multidisciplinary Diagnostic Team:
  - Gather relevant historical and current data on function in different environments
  - Do direct testing with the individual for evidence of organic brain damage
  - Consider all other pre and postnatal factors in formulating a diagnosis (differential diagnosis and co-morbidities)

Diagnostic Process

- Need for consistent training and mentoring of multidisciplinary teams on diagnosis for:
  - Consistency of diagnosis across sites
  - Accurate data gathering
  - Monitoring of prevalence (surveillance)
  - Monitoring of impact of prevention programs on incidence
  - Monitoring of effectiveness of interventions into and through adulthood

Assessment of Brain Damage/Dysfunction

- No single biological or functional test
- Need to move from basic tests (often average) to more complex assessment of brain function for daily life with independence and safety
- Current standard is the need for evidence of impairment in function in 3 areas that are not influenced by each other (ongoing research!)

Assessment of Brain Damage/Dysfunction

- Work by the Canada Northwest FASD Research Network using a consensus format has identified the tools in each core discipline (Physician, Neuro/Psychologist, Occupational Therapist, Speech Language Pathologist, Social Worker/Coordinator)
- Formulation of the “Picture” of brain strengths and weaknesses of that individual by all team members

Assessment of Brain Damage/Dysfunction

- Key brain domains:
  - Intellectual: most often higher that function
  - Academic achievement
  - Attention
  - Sensory, motor, neurological signs,

    visual spatial
  - Communication at basic and higher level of functional and social communication

Assessment of Brain Damage/Dysfunction

- Key brain domains (continued):
  - Memory: encoding, retrieval, working memory
  - Executive function: including judgment, inhibition, mental flexibility, problem solving, planning, sequencing, initiating
  - Adaptive/day-to-day function that impacts independent living, employability, not being victimized

Assessment of Brain Damage/Dysfunction

- Assessment of mental health issues:
  - May be secondary to adverse life experiences, late diagnosis, not being understood (Streissguth)
  - May be part of the prenatal brain damage from the alcohol exposure
  - May be the presenting symptoms in adolescents and adults – need to get the history of PAE

FASD Across the Lifespan

- Newborn and Infancy: optimal window to work with birth mother and infant dyad but need history of PAE
- Toddlerhood and Preschool: may have delays and disorganization but need to look at the environmental factors; basic tests not diagnostic of brain damage but need services based on function (paradox)

FASD Across the Lifespan

- Kindergarten Ages 5 to 6: Social, attentional, communication, learning and regulational difficulties may be more evident but may not be sufficient for diagnosis (services based on needs and not diagnosis)
- School Age 8 to 10 years: More evidence for brain damage by objective testing; need for school, community and home based supports

FASD Across the Lifespan

- Adolescents and adults: increasing evidence that inability to function in life is out of keeping with intellectual ability with emerging mental health issues
- May not always have the PAE history if the individual is disconnected from birth family (complexity of Adult FASD Diagnostic Services)

Why Diagnose FASD

- Diagnosis needs to lead to understanding of that individual's supports in education, community living and employability for life
- Shift to acquired brain damage model
- Need for more research into strategies (memory training, social skills, environmental supports, caregiver training) including evaluations of outcomes

Why Diagnose FASD

- Need research into the longitudinal trajectory of individuals with FASD who receive evidence based supports
- Diagnosis of a child with FASD is a **Diagnosis for TWO**
  - Birth mother is at further risk to her own health and future childrens' exposure;
  - need to work with prevention services for women that address WHY women drink

Policy Recommendations

- Ongoing research into best assessment tools at each age group
- Training of multidisciplinary teams for consistency and to increase capacity
- Sustainable funding for diagnostic teams for rural and urban access
- Research to identify better/best practices for interventions across the lifespan

Policy Recommendations

- Implement interventions in local communities
- Funding for longitudinal follow up across the system of care, including transition planning
- Prevention strategies to include access to best practice models of care for high risk birth mothers as well as primary prevention and health determinants approach