

FASD 101: Diagnosis and Support of FASD

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The FASD Learning Series is part of the Alberta government's commitment to programs and services for people affected by FASD and those who support them.

Session Goals

- History and terminology of FASD
- Biological impact of alcohol on prenatal brain development
- Diagnostic process with a multidisciplinary team to identify evidence of organic brain damage
- Presentations across the lifespan and supports that are needed

History of FASD

History of FASD

- Biblical, early artwork
- Initial medical reports in 1968 and 1973 but more awareness in 1990's
- Information still not included in training programs of professionals
- Animal research: biological mechanisms of alcohol as a teratogen to developing fetal brain cells; model of organic brain injury
- Clinical research - Streissguth, Clarren (University of Washington, Seattle)

History of FASD

- USA Institute of Medicine FAS, 1996
- Center for Disease Control US Guidelines 2004 (Bertrand)
- Canadian Guidelines for FASD Diagnosis CMAJ March 1, 2005 (Chudley)
- Mother Risk, Toronto Sick Kids
- Canada Northwest FASD Research Network
- International Collaborative on FASD (Riley)



FASD Challenges

FASD Challenges

- Confusion in terminology
- No consistency in diagnostic criteria leading to over and under diagnosis
- Lack of professionals and teams trained in diagnosis
- Lack of timely access to multidisciplinary teams

FASD Challenges

After diagnosis, lack of appropriate management and support resources that are evidence based

Lack of sustainable funding to meet needs of FASD population across their lifespan

Unrealistic goals set

- Prevent maternal drinking vs. understand why
- Change FASD individual vs. environment

FASD Strategies

Consensus conference on FASD across the lifespan with the Institute of Health Economics and Alberta Government, October 2009 (www.ihe.ca)

- Recommendations to address diagnosis, prevention and intervention better/best practices
- Stimulus for future research in FASD







Terminology

➤ **FASD**
Fetal Alcohol Spectrum Disorder

➤ **FASD is an Umbrella term**

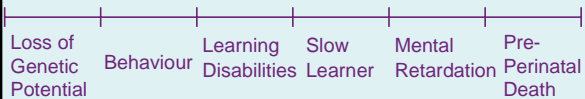


FASD

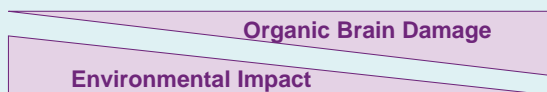
- F = Fetal** → Changes in normal development in utero
- A = Alcohol** → Teratogen; causes cell changes and damage
- S = Spectrum** → Damage/difficulties present from mild to severe
- D = Disorder** → Difficulty is inability to function and adapt as expected across life span



FASD Spectrum of Disability/Dysfunction



Continuum of effect of prenatal exposure to alcohol on brain function through life





Prevalence

- FAS = 0.5 - 3 / 1,000 in general population
- FAE = 3 - 5 X higher
- FASD from UA data 9 per 1000
- Increased in high risk populations
- Need to determine using consistent diagnostic criteria for true prevalence
- Cost to society, 1 to 2 million dollars plus the human cost
- Multigenerational impact

Prevalence

287 youths (average age 16 years)
alcohol related diagnoses = 67 (23.3%)

- 54 males + 13 females
 - Full FAS = 3 (1%)
 - ARND = 64 (22%)

Juvenile Justice System Burnaby, BC (Fast, Conroy, Look)

Prevalence

Data from children in foster care:

- Children with FASD enter earlier and stay longer in care, 17% diagnosed as FASD (Manitoba data, Fuchs and Burnside)
- Photographic analysis for the face of FAS of children in foster care 1 per 100 (Astley, Seattle, Washington)

Complexity in FASD

Complexity in FASD

- Need **confirmation** of **alcohol** exposure in that pregnancy
- Alcohol exposure **identifies risk** and is **not** diagnostic
- Need evidence of organic brain **damage**
- Need assessment by **multi/interdisciplinary** team to define spectrum of disability
- **Interventions** (educational, social, medical, mental health) guided by the assessment

Complexity in FASD

- Consider all other contributing **pre and post** natal factors
- Differential diagnosis and comorbidities
- Compounded or supported by **caregiving** situation
- **Increasing gaps** over time with need for longitudinal follow up
- **Secondary** disabilities

Mechanism of Alcohol

Why Alcohol Causes a Spectrum of Damage

- Alcohol = neurobehavioral teratogen
- Impact depends on:
- Amount and pattern of maternal drinking, binge impact (standard drink = 0.5 oz absolute alcohol)
 - Time in gestation (face day 19 to 21) brain develops throughout
 - Other maternal and fetal factors (genetics, nutrition, drugs, twin discordance)

Alcohol = Teratogen

Animal research on biological mechanisms:

- Direct effect on neuron maturation, migration, organization and function (Sulik)
- Oxidative stress, glutathione depletion in mitochondria, frontal area more vulnerable, neuronal death (Brien)
- Alteration in neurotransmitter and neuroendocrine function (Weinberg)

Alcohol = Teratogen

Indirect effect of alcohol:

- Effect on placenta vasoactivity via prostaglandin increase (Cook, Olsen)
- Indirect effect on maternal lifestyle and health
- Role of birth father and his alcohol use (stress mediated through cortisol)
- It is not just a woman's issue

Alcohol = Teratogen

Human studies:

- MRI findings: cerebellum, corpus callosum (Mattson), clinical MRI usually normal
- Functional MRI: less activation prefrontal area with increasing complexity of task (Chudley)
- DTI MRI: differences in white matter pathways (Sowell and Matson; Bealieu, Rasmussen, Andrew)
- Decrease in frontal lobe volume with severity if facial dysmorphology (Astley)

Impact of Postnatal Environment

Human studies:

- Direct effect on early brain development by deprivation, abuse, malnutrition
- More vulnerable if already brain damaged in utero
- Critical time periods to optimize brain development vs missed opportunities
- “Inter-related complexities”

Who Is At Risk?

- Across all cultures
- All socioeconomic groups
- Ask all women about drinking history
- Address WHY women drink
- Prevention: primary, secondary, tertiary
- Multigenerational - break the cycle
- Risk to fetus – no safe amount

Alcohol History

- Indirect by records - often not accurate, secondary gain, need reliable source
- Direct interview of birth mother
 - Nonjudgmental
 - Supportive for change
 - “Diagnosis for two”
 - Mentorship programs for high risk
 - Women specific rehab

Alcohol History

- Meconium analysis of fatty acid ethyl esters (Bearer), population studies, need for follow up of “at risk”
- Biological markers: saccadic eye movements in exposed individuals (Reynolds)
- No valid tools as yet for screening

Diagnosis

- Diagnostic Process
- Diagnostic Criteria
- Defining FASD: Hallmarks
- Other Alcohol Related Birth Defects
 - Multi-Factorial Risks
 - Differential Diagnosis
 - Comorbidities
 - Secondary Disabilities
- Comorbidities and Secondary Disabilities

Diagnostic Process

Who to refer for assessment:

- Need confirmed alcohol exposure
- No screening tools (PHAC Screening Workshop 2008)
- Need evidence of difficulties in function
- Community can often provide the basic testing and begin to connect to resources as part of process of referral to FASD Diagnostic Team

Diagnostic Process

Who to refer for assessment:

- **Community support person can be key prior to, during and after the assessment**
- **Without community readiness, diagnosis can become a negative label**

Diagnostic Criteria

Prenatal / postnatal growth retardation

- **Third trimester impact**
- **Consider other factors**
- **Cut off 10th percentile**

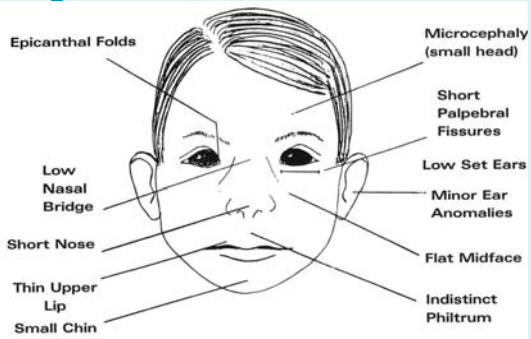
Diagnostic Criteria

Characteristic facial features

- **Day 19 - 21 gestation impact**
- **Fetal forebrain development**
- **Redefining population norms**

= Sentinel physical findings

Diagnostic Criteria



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Diagnostic Criteria

Not “Face” of FASD, but “Function”

Neurobehavioral pattern

- Disordered learning
- Behavior regulation problems
- Spectrum of minimal brain damage



Diagnostic Criteria

- Developmental delay, disorganization
- Impulsivity, inattention, hyperactivity
- IQ deficits
 - Not always
 - Concept of function below tested IQ level
- Learning deficits (especially for math, abstract processing, memory impact)
- Functional communication deficits



Diagnostic Criteria

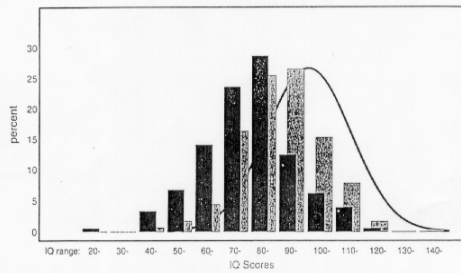


Figure 6.2. IQ distributions for FAS and FAE compared with the normal curve (N = 473). (Source: Streissguth, Barr, et al., 1996.)

Diagnostic Criteria

Impaired adaptive and executive function that affects daily living skills and socialization:

- Poor judgment and memory
- Difficulty generalizing and organizing
- Inability to learn from experience or connect cause and effect

= Primary Disabilities from organic brain damage

Defining FASD: Hallmarks

- Disordered pattern in development and acquiring expected skills
- Out of keeping with measured IQ
- Increasing difficulty in functioning with age
- May not be evident at early age (skill not expected)

Defining FASD: Hallmarks

- May be masked by supportive environment
- Most often “invisible disability”
- Pass tests
- Flunk life

Other Alcohol Related Birth Defects

- Embryogenesis - miscarriage
- Fetal development of all organs not just the brain
- Impact depends on timing:
 - Face develops day 19 to 21
 - Brain develops throughout
 - Vulnerable periods for each organ (heart, kidneys, skeletal, cleft palate)

Multi-Factorial Risks

- Need to consider all factors:
- Prenatal alcohol exposure
 - Other teratogens in utero
 - Genetic load
 - Adverse postnatal factors
 - Worse in more vulnerable damaged brain
 - Leads to secondary disabilities

Differential Diagnosis

What else could be the cause??

- Familial pattern or genetic syndromes
- Other teratogen exposure
- Small stature syndromes
- ADHD
- Acquired brain injury (perinatal, abuse)
- Mental handicap from other etiologies
- Other psychiatric disorders (genetic effect)

Comorbidities

What else can co-exist?

- ADHD, ODD, Tourette's
- Conduct Disorder
- Anxiety
- Depression
- Other mental health disorders
- Psychosocial impact: PTSD, RAD
- Other secondary disabilities

Secondary Disabilities

Etiology:

- Repeated failures academically and socially
- Not being understood "can't vs won't"
- Impact of multiple homes and abuse
- Results in frustrations for child and caregiver
- Not organic brain damage
- Theoretically preventable by good support system

Secondary Disabilities

Presentation:

- Low self-esteem, withdrawal, shutdown
- Depression (suicidal)
- Attachment Disorder
- Relationship difficulties
- Behavioral and emotional difficulties (anxiety, oppositionality)

Secondary Disabilities

- Not capable of independent living (but appear more able, not getting supports)
- Maladaptive behaviors:
 - Substance abuse
 - Becoming victims of abuse, taken advantage of
 - Breaking the law (intentional, get set up)

Secondary Disabilities

- Protective factors - Dr. Streissguth
- Early diagnosis before age 6 years
- Stable home environment without multiple placements
- Access to early interventions
- Not being victim of abuse

Secondary Disabilities

- Often secondary disabilities are the presenting features in adolescents
- Need to shift back to primary disabilities to understand cause of behavior and reframe intervention strategies

Comorbidities and Secondary Disabilities

- New theory that some co-morbidities and secondary disabilities may be part of the primary brain damage
- Need to identify and address all factors in intervention planning

DPN

- DPN Tool for Diagnosis
 - DPN Overview
 - Brain Domains

DPN Tool for Diagnosis

- Diagnostic Prevention Network, Astley and Clarren, University of Washington
- Standard of assessment for the Canada Northwest FASD Research Network
- Objective and Quantifiable

DPN Tool for Diagnosis

- 4-Digit Diagnostic Code to rank
- Growth deficiency
 - FAS facial features
 - Brain dysfunction looking for evidence for brain damage
 - Gestational alcohol exposure
 - Other pre and post natal factors

DPN Overview

4-Digit Diagnostic Code Grid

Growth Deficiency	FAS Facial Features	CNS Damage	Gestational Alcohol	
Severe	Severe	Definite	High risk	4
Moderate	Moderate	Probable	Low risk	3
Mild	Mild	Possible	No risk	2
None	None	Unlikely	Unknown	1

DPN Overview

Pre and Post Natal Grid

4		
3		
2		
1		
	Pre	Post

Prenatal Factors

- > Genetic Factors
- > Other teratogenic exposures
- > Poor prenatal care
- > Adverse prenatal events

DPN Overview

Pre and Post Natal Grid

Postnatal Factors

- > Physical Injury (head trauma)
- > Poor home environment
- > Physical/Sexual abuse
- > Poor Nutrition
- > Multiple Placements

4		
3		
2		
1		
	Pre	Post

DPN Overview

Alcohol Risk

4	Confirmed, high risk (4-6 drinks per occasion, 45 drinks per month, binges)
3	Confirmed, but less or exact amounts unknown
2	Unknown or questionable reliability
1	Definitely no alcohol

Standard drink = 0.5 oz absolute alcohol

DPN Overview

- Growth and face ranked by Physician using standard norms
- Physician also responsible for looking at all health and mental health factors
- Caregiver interview to give past and current challenges and commitment to the child
- Interview and observation of the individual with prenatal exposure to alcohol

DPN Overview

Brain assessment:

No single biological or functional test

- Assessment by all team members in their respective disciplines
- Formulation of the “picture” of the brain dysfunction by integrating all the information from standardized in each discipline
- Need evidence for 3 areas of impairment (below – 2 SD)
- Need for ongoing research

Brain Domains

- Intellectual
- Academic achievement
- Attention
- Sensory, motor, neurological signs, visual spatial
- Communication – receptive and expressive, higher level of functional and social skills, perspective taking

Brain Domains

- Memory including encoding, retrieval, working memory
- Executive functioning: including judgment, inhibition, mental flexibility, problem solving, planning, sequencing, initiating, shifting and abstract reasoning
- Adaptive functioning: impacts independent living, employability and not being victimized
- Other measures (emotional status, behavioral regulation)

FASD Across the Lifespan

- How Can It Present?
 - What Assessment Is Needed?
 - What Are the Support Systems Needed?
- Change the Environment not the Individual
 - Need for Longitudinal Monitoring

Newborn and Infancy

- Monitor for and treat NAS and ARBD
- Assess care giving situation
- Optimal time to reach out to birth Mom
- Birth mom supports (First Steps)
- Caregiver training and counseling
- Environment supports (feeding, sensory)
- Confirmed prenatal exposure to alcohol identifies child at risk for difficulties and needing developmental monitoring

Toddlerhood and Preschool

- Possible delays, look at quality of movement and play, disorganization
- Behavior regulation, activity level, “busy”, response to over stimulation, overly tactile
- Assessment of basic language and fine motor skills by community therapists
- Not specific for FASD diagnosis
- Paradox as it is the optimal time for interventions

Toddlerhood and Preschool

- In good environment may look “okay” but is not predictive of no problems later on and cannot be used for adoption counseling
- Recommend early intervention services, often do not qualify
- Counsel caregivers on optimizing the environment, structured play opportunities, stimulating language, supervision for safety
- Prevent attachment disorder (foster care)

Toddlerhood and Preschool

- Interventions based on individual needs
- Sensory supports are critical
- Strategies for deficits in social function
- Communication between program and home on what is working
- Respite for caregivers (Elves, Coaching Families)
- Treatment for attachment (Foster Pilot)

Kindergarten: Ages 5 to 6 Years

- Social difficulties more evident, overly friendly, not getting cues or limits, not listening based on individual needs
- Language “pink flag” chatty, talkative but understanding less
- Behavioral regulation difficulties with ADHD pattern, tantruming, easily over stimulated

Kindergarten: Ages 5 to 6 Years

- Transition to group learning setting with less one to one help shows early gaps in learning
- Assessment of basic language, fine motor and learning pattern by SLP,OT, Psychologist
- Too young to assess higher level skills
- Caregiver interview gives insight into the level of support needed in day to day living

Kindergarten: Ages 5 to 6 Years

- Diagnosis of FASD is often “deferred” at this age unless “hard” signs of brain damage
- Need for individual educational strategies
- Need for supportive environment in all settings
- Avoid over stimulation

Grades 3 to 4: Ages 8 to 10

- Societal expectations to be more independent, self-organized, shift from concrete to more abstract concepts
- FASD “cannot” related to difficulties in:
 - Executive functions
 - Social communication skills
 - Self-regulation and adaptive function

Grades 3 to 4: Ages 8 to 10

Presentation:

- Not keeping up academically, especially in the language demands of learning
- ADHD pattern
- Immature, cannot make or keep friends, not getting social cues, poor perspective taking
- Cannot organize without help in school and daily living
- More problems in unstructured times

Grades 3 to 4: Ages 8 to 10

Presentation:

- Talkative but tangential, lacking content and connections
- Not learning from consequences
- Impulsive, poor judgment, cannot generalize from one situation to another
- Needs daily routines with constant reminders
- Lying and stealing in spite of repeated teaching

Grades 3 to 4: Ages 8 to 10

Presentation:

- Mood swings and poor regulation, cannot self calm, sensory triggers
- Problems in motor planning: cannot put together all steps for a task

Grades 3 to 4: Ages 8 to 10

Assessment:

- All domains can be assessed by the team members using standardized tools
- Move from basic to more complex tests
- Direct testing of memory and executive functions with the child as well as indirect
- Functional assessments are much lower that predicted by cognitive ability from the IQ score

Grades 3 to 4: Ages 8 to 10

Assessment:

- Looking for evidence of severe brain impairment in 3 areas
- Observational information during the assessment and information from the caregiver interview and school also considered
- Assess for secondary disabilities

Grades 3 to 4: Ages 8 to 10

Recommendations:

- **Need individual educational strategies**
- **Do not expect independence but provide support across all settings**
- **Address ADHD with strategies and medications**
- **Address secondary disabilities but not by cognitive behavioral methods**
- **Supports for the caregivers (model of Coaching Families)**

Early Adolescence

Early adolescence:

- **Increasing behavioral and emotional difficulties often related to struggles in learning and day to day life = “can’t”**
- **More secondary disabilities**
- **Need educational shift from academics to concrete practical life skills**
- **Caregiver to continue to support concept of “stretch toddler”**

Adolescence: Late Teen to Adulthood

- **Presenting symptoms are the secondary disabilities**
- **“Invisible” disability of FASD (birth moms)**
- **Need to confirm the prenatal alcohol exposure**
- **Vocational assessment and training**
- **Assess independent living skills, provide supports**

Adolescence: Late Teen to Adulthood

- Self esteem, often not understanding or accepting their limitations and need for supports
- Mental health treatment, substance abuse risk

FASD Across the Lifespan: Key Points

- Expression of disability from prenatal exposure to alcohol
 - Varies at different ages
 - Is impacted by positive or negative environment
 - May be an invisible disability
- Increasing gaps in function over time
- Anticipatory guidance for caregivers

Developmental Timelines in FASD

Actual age of individual 18 years

Skill	Developmental Age Equivalent
Expressive Language	20
Comprehension	6
Money, time concepts	8
Emotional maturity	6
Physical maturity	18
Reading ability	16
Social skills	7
Living Skills	11

Chronological Age 0 5 10 15 20

Diane Malbin

Next Steps

- Why Assess FASD?
- Next Steps

Why Assess FASD?

- Diagnosis is not just a label for funding
- Understand difficulties as brain damage
- Need to move beyond diagnosis to intervention (advocate if gaps)
- Need to develop a community of supports through partnerships
- Need more evidence for what is best practice

Why Assess FASD?

- Prevention
- Secondary disabilities in FASD individual
- Birth mothers – future FASD births

Next Steps

- Are we making a difference by this diagnostic and intervention process?
- What is the impact on the individual with FASD in function and quality of life?
- What is the impact on the caregivers?
- What is the evidence that support specific programs?
- How to insure supports across the lifespan?

Next Steps

- Need training of multidisciplinary teams for consistent diagnosis that is accessible
- Need sustainable funding from diagnosis through supports and transitions with a lifespan focus
- Need research into best/better practices

Next Steps

- Need to focus on prevention strategies at all levels of primary public awareness and more targeted populations
- Especially need to focus on the women at highest risk and with the lens of social determinants of women's health and on women and their children (PCAP model, First Steps, Breaking the Cycle)

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