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.

**Fetal Alcohol Spectrum Disorder:**

**Screening and Diagnosis Implications for Two**

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**Date:** April 7, 2009

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**Session Goals**

Screening for FASD  
- Who?  
- What?  
- Why?  
- When?  
- Where?

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**The Range of Abnormality in Damage from In Uterine Ethanol**

Specific condition: Fetal Alcohol Syndrome  
- Ethanol exposure and  
- Atypical brain development  
- Specific set of facial abnormalities  
- +/- Growth delays  
- +/- Other organ malformations
The Range of Abnormality in Damage from In Uterine Ethanol

Other conditions in:
Fetal Alcohol Spectrum Disorder
- Ethanol exposure
And
- Atypical brain development
- +/- Some facial abnormalities
- +/- Growth delays
- +/- Other organ malformations

Screening for FASD: Who?
- Who Is Being Screened For FASD?
- Screening According To WHO
- WHO Criteria In The Context Of FASD

Who is being screened for FASD?
- FASD is a multigenerational diagnosis
- The implications are not necessarily the same for the mother and the offspring
- Identifying both is important
- The only way to identify the mother at this time is through recognition in her child
Screening According To WHO

According to the World Health Organization, to successfully implement a screening program the following conditions should be met:

- A suitable test should exist
- The disease or condition that is being screened for should be important medically, socially, or economically
- The natural history of the disease should be understood and the population at risk should be identifiable
- The test should be acceptable to the population

Continued

- The condition should be recognizable at an early stage
- There must be an accepted and effective treatment for the condition
- There should be facilities for assessment, diagnosis and rehabilitation

Continued

- Interventions should be acceptable to the population
- The cost of screening should not be disproportionate to the cost of caring for the affected individuals
- Screening programs should be a continuing process
WHO Criteria In The Context Of FASD

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WHO criteria in the context of FASD

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WHO criteria in the context of FASD

- The cost of screening should not be disproportionate to the cost of caring for the affected individuals
- Screening programs should be a continuing process
Screening for FASD: What?

- What Are We Screening For?
- The Brain In FAS Rarely Is This Obviously Abnormal
  - Corpus Callosum Abnormalities
  - Brain Mapping
- How Do We Discern That There is Structural Damage to The Brain at The Microscopic and Microcellular Level?
- Alcohol Effects Every Area of The Brain
  - ALARMMERS +

What Are We Screening For?

Brain Damage that is manifest as problems in ADAPTATION not necessarily linked to problems with
- Temperament and Mood
- Health
- Negative Experiences
- Adverse Environment

The Brain in FAS rarely is this obviously abnormal

From Clarren
How Do We Discern That There is Structural Damage to the Brain at the Microscopic and Microcellular Level?

- Current clinical imaging is normal in about 85% of cases of FASD
- The diffuse lesion is inferred from studies in animals
- The dysfunction resulting is detected in a battery of cognitive and performance measures
Alcohol Effects Every Area of the Brain

**Brain Stem:**
- Regulation of State

**Cerebellum:**
- Motor Skills and coordination/balance

**Limbic System+:**
- Attention

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**Cerebrum**

- **Left Temporal lobe +**
  - Speech and Language

- **Frontal Lobes**
  - Executive Functioning and Reasoning

- **Multiple Locations**
  - Learning, Memory, Cognition

- **Whole brain**
  - Adaptive Skills and Applications

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**Assessment of these areas can be remembered with the pneumonic:**

**ALARMMERS +**
ALARMERS +

- Adaptation
- Learning
- Attention
- Reasoning
- Memory
- Motor

- Executive function
- Regulation of State
- Speech/language
- Plus
- Brain anomalies
- Neurological signs

Screening for FASD: Why?

- This Global Brain Dysfunction Leads to Maladaptation
- But There Are Many Reasons for Maladaptation and We Think About Them Differently!
- The Dual Role of The Neurocognitive Evaluation

This Global Brain Dysfunction Leads to Maladaptation

- Poor educational and social experience
- Alienation/exhaustion of caregivers
- Then, among others:
  - Social isolation
  - Poor job performance…
  - Poverty
  - Mental Health and Substance Abuse Issues
This Global Brain Dysfunction Leads to Maladaptation

➢ Then, among others: (continued)
  • Homelessness
  • Victimization
  • Potential for antisocial behaviours

Early recognition should help to alleviate or prevent these long term adverse outcomes

But there are many reasons for maladaptation and we think about them differently!

Issues in Health or in the Environment
  ➢ Societal judgment: Disease
  ➢ Intervention goal: Treatment or Cure

Issues of Temperament or Mood
  ➢ Societal judgment: Disease or Disobedience
  ➢ Intervention goal: Treatment or Cure

But there are many reasons for maladaptation and we think about them differently!

Issues of Cognition and Performance
  ➢ Societal judgment: Disobedience or Disability
  ➢ Intervention goal: Acceptance or Treatment
The Dual Role of The Neurocognitive Evaluation:

- Establishes the component of the maladaptation that is due to brain dysfunction
- Is the template for the development of an individual multi-system intervention pathway

Screening for FASD: When?

- Why bother to screen for this diagnosis now?
- Finding the FASD fingerprint
- Screening for Alcohol Histories and Meconium Testing
- Screening for Growth Retardation
- Screening for the Neurodevelopmental Profile of FASD

Why bother to screen for this diagnosis now?

- The diagnosis of brain dysfunction due to brain damage is in itself therapeutic.
- This shifts the collective interpretation of the individuals problems from “he won’t to he can’t”
Why bother to screen for this diagnosis now?

- It allows for selecting the right individuals to new intervention programs
- It is one important way to identify high risk mothers for subsequent interventions

Finding the FASD fingerprint:

- Alcohol exposure:
  - Meconium testing
  - Reliable clinical history
- Growth assessment
- Facial analysis
- Neurocognitive profile

Screening for Alcohol Histories and Meconium Testing

- Getting an accurate alcohol exposure history has been found to be unreliable and is often not available at all
- Alcohol metabolites, fatty acid ethyl esters (faee) can be measured in meconium
- Tests can separate natural levels of faee from those achieved in women consuming alcohol in the second half of pregnancy

Development of Canadian Screening Tools for Fetal Alcohol Spectrum Disorder
Screening for Alcohol Histories and Meconium Testing
- High levels establish a pregnancy as “at risk” for FASD
-_misses all pregnancies exposed in the more dangerous first half of gestation
- May set up a difficult ethical dilemma if women choose to deny their use

Screening for Growth Retardation
- Utility of clinically obvious growth retardation overstated in early papers of FASD
- Clinical IUGR is only seen in massive exposures to alcohol in the third trimester

Screening for Growth Retardation
- Growth deficiency from early exposure is detectable only through large case controlled studies
- Studies of children with IUGR do not reveal ethanol as a common cause
Screening for the Neurodevelopmental Profile of FASD

- There is no specific profile for FASD
- DSM dx like
  - Intellectual Handicap
  - Specific Learning Disabilities
  - Attention Deficit Disorder
  - Oppositional Defiance Disorder
  - and many others
  - FUNCTIONAL diagnoses are seen in some FASD but overlap with other etiologic conditions

Screening for the Neurodevelopmental Profile of FASD

- Attempts are being made to develop a test that captures global maladaptation as the primary complaint in the context of other brain dysfunctions
- While these tests can separate normal from abnormal, their specificity for FASD or even for diffuse brain injury is not yet established
Screening for the Neurodevelopmental Profile of FASD

- Tests that the committee found to be worth further study included:
  - Asante Centre for Fetal Alcohol Syndrome Probation Officer Screening & Referral Form
  - Modified Child Behaviour Check List
  - The Medicine Wheel Tools were developed for the Esgenoôpetug Mi'gmaq First Nations community in New Brunswick
  - Fetal Alcohol Behaviour Scale (FABS)

Screening for FASD: Where?

- Screening For The Face of FASD
- The Face in FAS
- The Length of The Eye Slit and The Shape of The Upper Lip Were The Critical Features
- It Isn’t So Easy To Understand The Measure of The Palpebral Fissure

Screening For The Face of FASD

- Is specific for a history of alcohol exposure and detecting neurocognitive and performance deficits
- Only occurs in the fraction of the population that is called FAS perhaps 1/10 of FASD or less
- Is the feature that separates this syndrome from other specific dysmorphic conditions
The Face in FAS

The Length of the Eye Slit and the Shape of the Upper Lip Were the Critical Features

From Astley and Claren
The Length of the Eye Slit and the Shape of the Upper Lip Were the Critical Features

From Astley and Clarren

It Isn’t So Easy To Understand The Measure of The Palpebral Fissure

From Astley and Clarren
It Isn't So Easy To Understand The Measure of The Palpebral Fissure

- Grows with the face until adulthood
- Both eyes should be within a mm of the same size
- Little difference between males and females
- Correlates with head circumference
- Epicanthal tissues must be gently moved away!
- Normative data on Canadians in analysis

What is the screening procedure that is actually used by clinics in Canada actively seeing children for FASD?

This has been evaluated in:
Building Clinical Capacity for Fetal Alcohol Spectrum Disorder Diagnoses in Western and Northern Canada

Results of 1141 FASD assessments January 1, 2005 to April 30, 2006

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<th>FASD</th>
<th>Capacity</th>
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<td>112</td>
<td>233</td>
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<td>1141</td>
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So at the end of the day:

- Population based screening is not yet technically ready nor do we have the capacity to handle referrals
- Clinical screening or patient selection at this time means referring those with:
  - A history of mysterious maladaptation
  - AND
  - A history of alcohol exposure

WHEN in the life of the individual should FASD screening (referral) be done?

- At the youngest possible age at which the screening can yield a reliable final diagnosis
- This will rarely occur in infancy

“Static Encephalopathy”/ “Neurodevelopmental Injury”/ “Prenatal brain damage”

- These are diagnoses of location:
  - It’s in the brain!
- These are diagnoses of pathology
- THE PATTERNS OF ABNORMALITY ARE DIFFERENT IN EVERY PATIENT
- The presentation is maladaptation – often “mysterious maladaptation”
The mother says to her child with FAS: “Please don’t run in the street, you could be hit by a car. What did I just say?” 

The child replies: “I will not run in the street, I can be hit by a car.”

After many repetitions, the mother opens the door and lets the child play in the yard. 

The child promptly runs across the street.

Why Did The Child With FAS Cross The Road?

- Concrete language
- Memory disorder
- Impulsivity (ADHD)
- Risk taker
- Oppositional
- Depression
“Alcohol Related Prenatal Brain Damage” Means:

- Alcohol has altered the structure of the brain
- These alterations are on a continuum from mild to severe
- They are associated with functional problems that are on a continuum from mild to severe
- The identified alterations may or may not be the cause of a particular action of concern!
- It is easy make a mistake in interpreting behaviour without careful evaluation of the possible reasons

So In The Ideal World

- A professional would consider FASD as a possibility when worried about “mysterious maladaptation” and/or an alcohol history
- Tell the family about the concern
- Refer to an FASD diagnostic center that would determine a positive screen – mysterious maladaptation and an alcohol exposure
- Full assessment and treatment plan in the diagnostic center

There are two problems with this scenario in the real world

1. What does a family hear when they are told that FASD is suspected?
2. How long will they need to wait until they are diagnosed?
When People Are Told Their Child Has A Birth Defect Condition

- There is almost always a sense of responsibility and sometimes guilt as well.
- Generally these feelings are not really rational.
- We might imagine that these feelings are weightier when an action in pregnancy did, in fact, cause the problem.

When Birth Parents Are Told Their Child Might Have FASD

- Do they suddenly hear that they are “fetus abusers”?
- Do they believe that we think of them as “fetus abusers” and think less of them?
- Do they hear that their child may have a lifetime handicapping condition?
- What is our responsibility in this conversation?

The fact is: I have never met a birth mother who drank in pregnancy because she wanted to harm her unborn child.
It Is Very Interesting

- In Canada and in the United States the vast majority of patients diagnosed with FASD are in foster and adoptive circumstances
- Is this because that is where the patients are or is this an ascertainment bias because of our uncertainty in discussing this potential diagnosis with birth parents?
- What do we know about FASD birth parents?

Response to a proposal to prevent FAS from the Centers for Disease Control from the United States

“Using a clinic for the diagnosis of FAS as a method for identifying women at high risk to produce other children damaged by alcohol.”

Sterling K. Clarren, MD, Susan J. Astley, PhD

Birth Mothers

- Looking For The Birth Mothers
- Among The 80 Women That We Could Locate
Looking For The Birth Mothers

Among 160 children with FAS, 10 were living with their birth mothers as the primary caregiver.

70 other mothers were involved in their child’s life and could be contacted.

40 were known to systems that could not release the addresses to us.

40 were missing or dead.

Among The 80 Women That We Could Locate

All agreed to a half day interview for $75(US).

Still not easy to make contact.

We could not tell them what their child’s diagnosis was, but no one asked (but many spontaneously speculated that it was because their child had FAS).

Among The 80 Women That We Could Locate

Expected distribution of majority and minority peoples.

They had started life at all levels of society.

- As adults most lived in poverty.

They had a reasonable distribution of IQ’s but there was a blip of low IQ’s at the bottom.
Among The 80 Women That We Could Locate

- Nearly universal reports of lifetimes experiences with extreme physical, sexual and emotional abuse
- Nearly all had long standing battles with alcohol use and abuse

Among The 80 Women That We Could Locate

- All would be anticipated to have two Mental Health Diagnoses
  - Substance Abuse and Post Traumatic Stress
- But most had several more:
  - Depression
  - Manic depression
  - Phobias
  - Schizophrenia, etc
- The average patient had 4 DSM diagnoses in her record, two patients had 10

Among The 80 Women That We Could Locate

- There was social isolation: few friends, poor relationships with the family, limited activity
- We estimated that about half the women had FASD themselves
- Best access to services - social, mental health, medical - WAS DURING PREGNANCY
Building Capacity for Diagnosing “Mysterious Maladaptation”

- The diagnostic model exists
- It has not been a simple matter to identify qualified professionals and train them to function as teams doing this type of work
- It would be estimated that perhaps 30,000 individuals with FASD are in Alberta and our annual capacity for diagnosis is not 350 evaluations

In Conclusion

- Screening is not diagnosis
- Screening is both simple and complicated
- Care and sensitivity in understanding the power of the potential diagnosis needs to be considered
- Diagnosis is important for understanding the mother and the offspring
- The system will not be able to work well until there is more diagnostic capacity
Contact Information

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For Information on Upcoming Sessions in the Series:
www.fasd-cmc.alberta.ca

Please Take the Time to Fill Out the On-Line Evaluation

Thank You!