MODULE SEVEN

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Referral and Diagnosis

WHEN TO REFER

For individuals who are not medical professionals (psychiatrists, doctors or nurses), please refer children for follow up related to possible FASD with their family physician or other specialists in your area. Please remember that the diagnosis of FASD is a specialized, team based process which may take awhile to accomplish. In the meantime, accommodations may be put in place to support the individual and their family.

Medical professionals who do not specialize in the diagnosis of FASD or belong to a team that performs these assessments should assess the child carefully and refer for diagnosis in the following situations: (Chudley et al., 2005, pp S5)

- Presence of the three characteristic facial features
- Evidence of significant prenatal exposure to alcohol at levels known to be associated with physical or developmental effects, or both
- Presence of one or more facial features with growth deficits plus known or probable significant prenatal alcohol exposure
- Presence of one or more facial features with one or more central nervous system deficits plus known or probable significant prenatal alcohol exposure
- Presence of one or more facial features with pre- or postnatal growth deficits, or both, and one or more central nervous system deficits plus known or probable significant prenatal alcohol exposure
- Individuals with learning or behavioural difficulties, or both, without physical or dysmorphic features and without known or likely prenatal alcohol exposure should be assessed by appropriate professionals or specialty clinics to identify and treat their problems

It is important to note that while most of the items on the previous list include physical features, many people affected by FASD do not exhibit obvious physical symptoms (Wemigwans, 2008).

APPROACHING PEOPLE ABOUT THE POSSIBILITY OF REFERRAL/DIAGNOSIS

Making a referral is more than writing a letter or connecting the individual/family with an expert in the field. Before referring anyone for diagnosis, it is important to first develop a trusting relationship with the individual and/or family so that they can be prepared and supported throughout the process (Chudley et al., 2005; Wemigwans, 2008). Building this relationship can take time. It can be an emotional time with many mixed feelings experienced by the individual/family; they may need information on FASD or be connected to community resources before agreeing to the diagnostic process (Wemigwans, 2008).
Once the individual/family is ready to discuss the possibility of an FASD diagnosis, they will continue to need support and information throughout the assessment process. It is vital that they are aware of and agreeable to pursuing a diagnosis. The individual/family can be prepared for the diagnostic process by (Wemigwans, 2008):

- informing them that the process will involve a wide range of professionals and will take an extended period of time
- telling them that you only suspect FASD, and that, even if FASD is not diagnosed, the process can be beneficial as the individual/family will know more about their challenges and how to address them
- discussing the potential psychosocial consequences of an FASD diagnosis

If the individual in question is a child or youth, the caregiver needs to give permission to initiate the process. When talking with the birth parent, it is important to be sensitive, asking questions in a skillful and careful manner. See Module Five: Supporting Women Who Drink During Pregnancy for more information on Motivational Interviewing. The objective of this conversation is not to place blame but to assure the caregiver that obtaining an early diagnosis can make a positive difference in the child's life (Wemigwans, 2008).

Once the individual/family is ready to proceed with diagnosis, they can be referred to either their family doctor for further assessment or directly to an assessment clinic.

CANADIAN GUIDELINES FOR THE DIAGNOSIS OF FASD

In 2005, a subcommittee of the Public Health Agency of Canada's National Advisory Committee on Fetal Alcohol Spectrum Disorder was created. The goal of this group was to review all of the available information on diagnosis and to integrate this information to create a standard for diagnosis to be used in Canada. A document, entitled *Fetal Alcohol Spectrum Disorder: Canadian Guidelines for Diagnosis,* was developed in consultation with experts from throughout Canada and published in the *Canadian Medical Association Journal*. (Chudley et al., 2005). There is a copy of the Guidelines in the accompanying Resource Kit.

These guidelines emphasize the importance of a multidisciplinary team approach to FASD diagnosis, and go far beyond merely listing diagnostic criteria. The Guidelines describe the entire diagnostic process beginning with screening and referral, and ending at treatment and follow-up.

This module is adapted from the *Canadian Guidelines for Diagnosis* (Chudley et al., 2005). It will examine and elaborate on each step of the diagnostic process and discuss recommendations made in the document.

The Diagnostic Process

The diagnostic process consists of a number of steps:

- screening and referral
- physical examination and differential diagnosis
- neurobehavioural assessment
- treatment and follow-up

The Guidelines emphasize the importance of using a multidisciplinary team approach. The core team should consist of a:

- coordinator for case management (e.g., nurse or social worker)
- physician specifically trained in FASD diagnosis
- psychologist
- occupational therapist
- speech-language pathologist

Depending on the unique needs of the individual, additional team members from a variety of professionals and individual supporters should be included in the process.

The use of a multidisciplinary team approach is crucial, as each member contributes an important dimension to the process. The multidisciplinary team plays a key role in creating recommendations covering all aspects of a person's life.

The Guidelines state that, in order to ensure the assessment is comprehensive, documentation from a variety of sources should be collected. This includes social services records, previous assessments, birth and pregnancy records, medical and hospital records, adoption records, academic records, achievement tests, developmental assessments, psychological and psychometric assessments, legal reports and documentation of the family history.

The Guidelines emphasize the importance of *readiness* (Chudley et al., 2005). Specifically, they state that for the best results, both the community and the family must agree with and be prepared to participate in the diagnostic process. Furthermore, they note the importance of *disclosure*. That is, the family must be made aware of the potential consequences that a diagnosis of FASD may have, both positive and negative. Members of the multidisciplinary team help guide the individual and his/her caregivers through the diagnostic process, offering support and services both during and after diagnosis.

Stage 1: Screening and Referral

The Guidelines recommend a number of different procedures for screening. This includes screening of pregnant and post-partum women as well as individuals who may have been affected by alcohol use. The purpose of screening is to identify those who should be referred for an assessment.

Stage 2: Physical Examination and Differential Diagnosis

There are two goals for this step:

- complete a physical and neurologic examination
- differential diagnosis

During a physical exam, the physician will measure growth (height and weight) and head size as well as looking for the characteristic features of FASD. The physician should record any other existing anomalies (e.g., cleft palate and congenital heart defects).

Although these anomalies may not relate specifically to FASD, they may be useful in excluding other possible disorders with features similar to FASD. This process is known as differential diagnosis which involves distinguishing between diseases of similar character by comparing their signs and symptoms.

Although many children with FASD show impaired growth at some point (in utero or throughout the lifespan), some issues have been raised about the criterion for growth impairment (Hoyme et al., 2005; Chudley et al., 2005).

- If alcohol is not consumed during third trimester there may be no growth deficiency
- Presence of illnesses like gestational diabetes can result in larger fetal size making it difficult to detect growth deficiency
- Parental size, genetic potential and other conditions (e.g., nutrition) can create or hide growth deficiency
- Growth deficiency may decrease with age
- "Growth norms" are based on the general population, and may not reflect differences found in subpopulations (e.g., ethnic or racial groups)

Stage 3: Neurobehavioral Assessment

The neurobehavioral assessment looks for evidence of CNS damage on three levels: structural, neurological, and functional. This information will be important when developing a treatment and follow-up plan that meets the unique needs of the individual.

Individuals who have been affected by prenatal exposure to alcohol display a wide range of neurobehavioural characteristics that vary from individual to individual. As a result, there is currently no standardized tool (or set of tools) that can confirm an FASD, and many tests and measures must be used. To further complicate the issue, the strengths and challenges faced by the individual with FASD can change throughout the lifespan, and/or be influenced by the environment or genetics.

The following areas should be assessed:

- **Neurological Level**
  - hard and soft neurological signs
- **Structural Level**
  - brain structure
- **Functional Level**
  - cognition (IQ)
  - communication: receptive and expressive
  - academic achievement
  - memory
  - executive functioning and abstract reasoning
  - attention deficit/hyperactivity
  - adaptive behaviour, social skills, social communication

Evidence of CNS damage must be found in three domains.

Many people incorrectly believe that mental handicap (low intelligence) is a defining characteristic of FASD. In fact, only 25% of individuals with full FAS and less than 10% of those with pFAS have an IQ of less than 70 (Streissguth, Barr, Kogan & Bookstein, 1996). Individuals with IQ scores in the normal range may still have specific cognitive and neuropsychological impairments that can be mistaken as a low IQ.

Stage 4: Treatment and Follow-Up

During this phase, a comprehensive report detailing the assessment and diagnosis should be completed including recommendations for treatment and follow-up. This is a useful tool for caregivers and professionals who are working with the newly diagnosed individual.

Rather than ending the process with the diagnosis, the Guidelines emphasize the importance of using the information gathered to improve the quality of life for the individual affected with FASD, their family, and their community. Education of all involved is a crucial element of this step in order to understand the diagnosis and begin to develop strategies for working with the individual and their caregivers.

The Guidelines stress that the post-diagnosis report is the most crucial piece of the assessment, acting as a "blueprint" for intervention. This is also the time when the multidisciplinary team should begin to establish connections between the individual and their caregivers, and community resources and services. These links with appropriate supports improve outcomes and decrease the likelihood of secondary behavioural characteristics.

BENEFITS OF DIAGNOSIS

There are many benefits to diagnosis, especially in the early years of childhood. They include (Chudley et al., 2005; Streissguth et al., 1996; Astley & Clarren, 2000):

- provides the information that is needed to develop interventions designed around this particular type of brain damage
- leads to the acceptance of the individual for who the individual is which allows individual strengths to emerge
- can validate an individual's problems and support the intervention needed to maximize his or her abilities
- provides a "name" for a condition that gives everyone a common language that is useful in discussion
- allows for adaptation to occur
- facilitates communication between clinicians and clients as well as their caregivers and families
- helps with understanding the individual's needs and challenges and assists in establishing realistic expectations
- assures parents/caregivers that the resulting behaviours are not the result of "bad parenting" or a "bad child"
- provides access to appropriate services, support groups and training specific to FASD
- ensures careful monitoring of the individual's health issues as they develop - and attention to issues more specifically related to prenatal alcohol exposure
- can prevent or lessen the impact of "secondary disabilities" such as mental health problems, alcohol and other drug problems, or other issues. Early diagnosis is an important protective factor

CONSEQUENCES OF DIAGNOSIS

There is a possibility of negative psychosocial consequences of confirming a diagnosis. They include (Chudley et al., 2005; Wemigwans, 2005):

- feelings of shame and guilt especially for the birth mother
- stigmatization of the individual who is diagnosed including labelling, low expectations of the individual's intelligence and performance by others
- being stereotyped
- having to admit that there is something 'wrong or different' about their brain and/or body

The professionals involved in the diagnostic process can be a valuable support to the individuals at this time by either providing counselling, support, or referrals to other appropriate services.

Co-occurring Medical Problems and Physical Birth Defects

There are many medical problems and physical birth defects that commonly co-occur with FASD. Note that alcohol use during pregnancy is not the only cause of these problems. These include, but are not limited to:

- high pain tolerance (Saskatchewan Learning, 2004)
- hearing problems (Saskatchewan Learning, 2004)
- heart defects/murmurs (Stratton et al., 1996)
- cranio-facial defects (Stratton et al., 1996)
- vision problems (Stratton et al, 1996)
- kidney defects (Stratton et al., 1996)
- skeletal abnormalities (Stratton et al., 1996)

OVERLAPPING DIAGNOSIS

Individuals with FASD tend to collect numerous diagnoses during their lifetime. In the early years, these diagnoses tend to reflect primary behavioural characteristics, e.g., speech and language delays and learning disabilities. As the individuals move towards adolescence (and having experienced years of "poor fit with the environment"), the diagnoses start to reflect secondary disabilities, e.g., Conduct Disorder, Oppositional Defiant Disorder, and others. (Malbin, 2008). Misdiagnosis can lead to inappropriate interventions and therefore increased frustration, which in turn can increase the risk of secondary disabilities (Astley & Clarren, 2000).

FASD is not in the Diagnostic and Statistics Manual (DSM). This is the manual used by psychologists and psychiatrists to diagnose mental illness. This results in the accumulation of diagnoses, and typically greater confusion (Malbin, 2008). These diagnoses may not be wrong. However, they may not truly capture the individual's challenges or take brain dysfunction into account (Malbin, 2008).

CASE STUDIES

Case Study 7.1: Jeremy

Jeremy has been referred to you in relation to his hyperactivity and an inability to get along with other children his own age. Jeremy says he has very few friends and has a hard time focusing on what the teacher is saying to him. His facial features are similar to a photo you have seen of a child affected by FASD. You suspect that he may have been exposed to alcohol prenatally.

For this case study, reflect on the questions below:

1. What are some other factors that may indicate the child may have been exposed to alcohol prenatally?
    - *Has difficulty understanding cause and effect*
    - *Language skills may seem to be deteriorating as he ages*
    - *May have trouble generalizing information*
    - *Has challenges learning the concept of time*
    - *Is a concrete thinker – has challenges with abstract concepts*
    - *Has issues with emotional regulation and reading social cues*
    - *Has delayed motor development and control*
    - *Is small for his age*
    - *May be sensitive to light, sound, texture or stimulation*

2. What are your next steps in the referral process?
    - *Developing a trusting relationship with the individual/family*
    - *Once the family is ready to discuss the possibility of a diagnosis, continue to provide support and information*
    - *Sharing with the individual/family that you only suspect FASD and that even if it isn't, the diagnosis process can be beneficial to help understand the individual and their needs*

3. How can you support this family through the diagnosis process?
    - *By providing support and information throughout the process*
    - *By informing them that the process can take a long time and involves many different professionals*
    - *By discussing the potential benefits and consequences of diagnosis*

DISCUSSION QUESTIONS

1. **What are some of the benefits to obtaining a diagnosis?**
    - *Helps to identify appropriate interventions and accommodations needed to support the individual*
    - *Can validate an individual's challenges and the need to accommodate them*
    - *Provides a 'name' for a condition that gives everyone a common language for discussion*

- *Assures parents/caregivers that the resulting behaviours are not a result of "bad parenting" or a "bad child"*
- *Provides access to appropriate services, support groups, and training specific to FASD*
- *Can prevent or lessen the impact of secondary disabilities*

2. **What are the possible downsides to a diagnosis?**
   - *Feelings of shame and guilt, especially for the birth mother*
   - *Stigmatization of the individual who is diagnosed including labelling, low expectations of the individual's intelligence and ability to perform tasks*
   - *Being stereotyped*
   - *Having to admit that there is something "wrong or different" about their brain*

3. **Why is it important to build a relationship with the individual/family before proceeding with a referral for diagnosis?**
   - *Families that feel supported and not judged will be more willing to proceed with the diagnostic process*

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