Module 7

Referral and Diagnosis of FASD

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Introduction

Ever since Jones and Smith (1973) introduced the diagnostic term "Fetal Alcohol Syndrome", work has been done to develop the clinical diagnostic criteria for this disability. FASD is a complex disability that affects each individual differently. Due to its complexity, FASD requires a multidisciplinary team to accurately assess, diagnose, and provide appropriate management strategies. Like other syndromes and disabilities, diagnosis is an ongoing and complicated process. Despite this complexity, evidence-based guidelines for diagnosis of FASD have been developed in Canada.

The first Canadian FASD diagnosis guidelines were created in 2005. Since then, new evidence, knowledge, and experience emerged, demanding a revision of the guidelines to enhance diagnosis and outcomes. In 2015, new guidelines were published to provide greater clarity and uniformity in FASD diagnosis. According to the Canadian diagnostic guidelines, the three key areas for diagnosis are: (Cook, et al. 2016):

- evidence of prenatal alcohol exposure
- sentinel facial features (i.e., facial features characteristic of FASD)
- evidence of impacts on central nervous system

Referral for Assessment and Possible Diagnosis

Due to a lack of effective treatment and management some professionals have been hesitant to refer children for an FASD assessment. Diagnosis can occur throughout the lifespan; however, early diagnosis is associated with positive outcomes and improves the options for interventions. It also helps the individual access age-appropriate supports and services that help prevent adverse impacts such as mental health challenges, alcohol and addictions, homelessness, and inappropriate sexual behaviour. According to Streissguth et al., (2004), early diagnosis is defined as getting a diagnosis before the age of six. However, it is never too late to receive a diagnosis (Pei et. al., n.d).

In Canada, the diagnosis of FASD is a specialized, team-based process that starts with referral (Cook et. al., 2016). It can be a very lengthy process and an individual may need care and support while waiting for a formal diagnosis.

Referral for a possible FASD diagnosis should be made whenever there is evidence of, or suspected, prenatal alcohol exposure at levels associated with physical or developmental effects (Cook et al., 2016). Individuals should be referred for assessment in the following cases (Chudley et al., 2005).

1. Presence of facial features characteristics of FASD along with impairment of three or more neurodevelopmental domains, or evidence of microencephaly among infants and young children.
2. Evidence of alcohol exposure during pregnancy along with impairment in three or more neurodevelopmental domains.
3. Evidence of alcohol consumption during pregnancy, as well as some indication of neurodevelopmental issues.

Wemigwans (2008) cautions that although some of the items on this list include physical features (e.g., facial or growth), many people with FASD do not show obvious physical symptoms. Qualified professionals can assess people with learning and/or behavioural difficulties who do not have sentinel features or known/probable prenatal alcohol exposure. In addition, it is recommended that referrals should include as much information as possible regarding other potential teratogen exposure as well as genetic factors to help with diagnostic accuracy (Loock, et al., 2005). The need for psychosocial support for clients should also be taken into consideration, to assist individuals and families through screening, referral, and if necessary, the diagnostic processes (Cook et al., 2016).

Making Referrals for Assessment

Referrals can be initiated by a variety of sources including the individual, their family, community service agencies, medical service providers, and government departments and ministries such as Mental Health and Addictions Services, as well as Justice and Children's Services (Cook et al., 2016). The referral source is often a reflection of the type of challenges the individual is experiencing, and this can provide important information for the management plan to ensure maximum success (Cook et al., 2016).

Making a referral is more than writing a letter or connecting the individual/family with an expert in the field. It is best if the referring professional have a trusting relationship with the individual/family so that they can be prepared and supported throughout the process (Chudley et al., 2005; Wemigwans, 2008). For example, an individual or family may need more information or a better understanding of FASD. Some may also benefit from being connected to community resources before agreeing to start the diagnostic process (Wemigwans, 2008). Building this relationship can take time and be a very emotional experience for the individual and family.

Once the individual/family is ready to discuss the possibility of pursuing a diagnosis of FASD, continued support and information throughout the assessment process are beneficial. They should be aware of, and agree to, each of the steps required in getting a diagnosis. The individual/family can be prepared for the diagnostic process by being informed that:

- the process involves a wide range of professionals and takes a long time
- FASD is only suspected and even if FASD is not diagnosed, the process can be helpful as the individual/family will know more about their challenges and how to address them
- it is important to be aware of the potential social and psychological consequences of a diagnosis of FASD

(Wemigwans, 2008)

The caregiver must give permission to start the diagnostic process for children or youth. Professionals should be sensitive and ask questions in a skilful, careful, and non-stigmatizing manner. Professionals can reassure the caregiver that an early diagnosis can make a positive difference in the child's life and not place blame (Wemigwans, 2008). After receiving a diagnosis, the child can be referred to appropriate therapies and supports.

The Impact of Diagnosis

Cook et. al developed new guidelines for the diagnosis of Fetal Alcohol Spectrum Disorder in 2016. Based on these guidelines, FASD should now be used as a diagnostic term when prenatal alcohol exposure is considered to be a significant contributor to observed deficits that cannot be fully explained by other etiologies (Cook et al., 2016). All other known relevant contributors (e.g., trauma, known genetic anomalies) should be documented with the FASD diagnosis as they can have significant impact on the functional and neurological challenges of the affected individuals (Cook et al., 2016). A diagnosis of FASD can come with both benefits and negative consequences. Consequences can be for the individual, as well as the support network of parents, caregivers, and professionals. These potential positive and negative consequences are outlined below.

<u>Benefits of Diagnosis</u>

Accurate diagnosis can:

- help clarify the specific FASD-related challenges experienced by the individual
- guide appropriate post-diagnosis interventions and strategies for support
- reduce the risk for adverse impacts (i.e., mental health problems, trouble with the law, homelessness, addictions issues, etc.)
- link birth parents with supports and interventions to prevent future affected pregnancies
- support individuals and families with applications for disability tax credits, financial supports, and other services for the individual and/or their caregivers
- provide important information regarding supports for youth transitioning to adulthood, such as guardianship, trusteeship, and legal representation agreements

(Canada FASD Research Network, n.d.)

<u>Benefits for the Individual</u>

A diagnosis of FASD can:

- provide the individual with the knowledge they need to function successfully, making use of their strengths and acknowledging their limitations
- help the individual understand themselves
- motivate the individual and/or family to seek out supports and services that will assist with day-to-day living challenges
- validate the individual's experiences, supporting self-awareness and growth, and maximizing changes for a better life
- help provide the understanding that certain behaviours are not the fault of the individual, while still maintaining that the individual is responsible for their behaviour
- help support people and caregivers to be more understanding and helpful

(Helgesson et al., 2018)

Benefits for Parents, Caregivers, and Professionals

A diagnosis of FASD can:

- provide the information that is needed to develop interventions designed around child's need[1]
- allow families to set realistic expectations[1]
- lead to the acceptance of the individual for who they are, which allows the individual's strengths to emerge[1]
- provide a "name" for the condition, giving everyone a common language[1]
- facilitate communication between clinicians and clients, as well as their caregivers and families[1]
- help with understanding the individual's needs and challenges[1]
- assure parents/caregivers that the resulting behaviours are not due to either 'bad parenting' or a 'bad child'[1]
- provide access to appropriate services, support groups, and training specific to FASD[1]
- provide information regarding incidence and prevalence rates of FASD in Canada[3]
- ensure careful monitoring of the individual's health issues as they develop, and can draw attention to specific issues related to prenatal alcohol exposure[2]
- prevent or lessen adverse impacts (e.g., mental health problems, alcohol, and drug problems) This is key because, as will become evident, early diagnosis is a critical protective factor[2].

Early diagnosis is a protective factor for adverse impacts.

([1]Doak et al., 2019, [2]Helgesson et al., 2018, [3]Canada FASD Research Network 2019)

Negative Consequences of Diagnosis

There are also a number of potential negative consequences of confirming a diagnosis of FASD that should be considered. These consequences can include:

- feelings of shame and guilt, especially for the birth parent
- stigmatization of the individual by others (e.g., labelling, low expectations of the individual's intelligence, low expectations of the individual's performance ability)
- stereotyping
- the individual having to admit that there is something 'wrong or different' about their brain and/or body

(Helgesson et al., 2018)

This may be a time when certain members of the multidisciplinary team (e.g., psychologists or counsellors) will be particularly valuable. These team members can work to help lessen the potential negative impact by providing counselling, support, and referrals to other services that may help the individual to work through these difficulties.

Early Diagnosis

Earlier diagnosis of FASD (i.e., before the age of 6) can be protective against the development of adverse impacts (See Module 9 for more details; Streissguth et al., 1996; Streissguth et al., 2004). However, early diagnosis is not common, and is not always possible. Research shows that many children with FASD do

not get diagnosed until they are above the age of six. In the United States for instance, the Washington FAS Diagnostic and Prevention Network (FAS DPN) data shows that the average age of referral for FASD diagnosis is 9.5 years (Olson et al., 2007). The North Dakota Registry data reported an average age of diagnosis of 7.1 years (Bagheri et al., 1998 as cited in Burd et al., 2003).

The most effective time to diagnose FASD is when a child reaches a developmental stage that allows for an accurate assessment of delays in cognitive development, speech and language development, attention, fine and gross motor skills, and other aspects of delayed or abnormal neuropsychological functioning (Paintner, et al., 2012). Based on research, the best time window when these factors can be assessed is thought to be between ages 2 and 16 years (Paintner et al., 2012). However, for children under age six, assessments may be inconclusive because some domains of brain functioning cannot be effectively assessed until the child is older (Cook et al., 2016). Other (non-age-related) reasons for inconclusive assessments can include the presence of confounding factors, such as concurrent life stress or illness (Cook et al., 2016).

Another benefit of early diagnosis of FASD is that it can allow for the identification of problematic alcohol use in the birth parent, providing the opportunity for interventions to prevent the occurrence of prenatal alcohol exposure (PAE) in future pregnancies (Chasnoff et al., 2015; Paintner et al., 2012). Unfortunately, as indicated, making a diagnosis of FASD in young children is difficult, resulting in many missed diagnoses and misdiagnoses (Chasnoff et al., 2015). It is reported that children with FASD are often misdiagnosed with other developmental disorders, such as autism spectrum disorder (ASD), asperger's syndrome (AS), attention deficit hyperactivity disorder (ADHD), or obsessive-compulsive disorder (OCD) (O'Malley, 2007 as cited in Carpenter, 2011). Another reason why diagnosing FASD is challenging is because the diagnosis of FASD involves complex physical and neurodevelopmental assessments. These assessments are conducted by a multidisciplinary team which generally includes a:

- pediatrician
- occupational therapist
- speech-language pathologist
- clinical psychologist

(Cook et al., 2016)

Other barriers to early and accurate diagnosis of FASD include the lack of apparent physical features; the confusion of diagnostic terminology associated with FASD; difficulty in performing facial features measurements during routine clinic visits; lack of access to multidisciplinary teams; and difficulty in establishing a history of alcohol use, especially because many children with FASD are adopted or raised in foster care (Chasnoff et al., 2015; Lange et al., 2013; Weyrauch, et al., 2017). Perceived and real stigma regarding alcohol use during pregnancy is also a barrier for early diagnosis (Lange et al., 2013). The co-occurrence of multiple risk factors (such as developmental delays or psychiatric disorders) and negative environmental exposures that may contribute significantly to the impact of FASD on the child further complicates the diagnostic process (Cook et al., 2016; Olson et al., 2007).

Misdiagnosis

The most important reason for making sure that FASD diagnostic criteria are valid and reliable is that there are serious implications for misdiagnosis. Some of the serious implications of misdiagnosing FASD include (Astley & Clarren, 2000):

- clinical implications (e.g., inappropriate patient care, increased risk for adverse impacts for the individual**,** and missed opportunities of connecting birth mothers with services and supports for primary prevention of FASD in future pregnancies)
- public health implications (e.g., inaccurate estimates of rates)
- research implications (findings are not meaningful if diagnoses are not accurate)

There are serious implications for misdiagnosis so diagnostic criteria must be valid and reliable.

Diagnosing FASD: Three Key Areas

Many systems for diagnosing FASD exist across the world. In Canada, three key areas are recognized for FASD diagnosis, according to Canadian FASD diagnostic standards (Cook et al., 2016).

#1: Alcohol Exposure During Pregnancy

For FASD to be diagnosed, the fetus must have been exposed to alcohol at some point during development. This can be hard to assess because of factors such as:

- the individual may not share about their alcohol use during pregnancy due to stigma, judgement
- the individual may not be able to remember specific information about their alcohol use (e.g., timing, amount, and frequency)
- information about the biological mother may not be available
- shame or fear may stop the birth parent from revealing their alcohol use during pregnancy (Stratton et al., 1996)

Alcohol exposure can be rated in one of three ways: 1) confirmed exposure; 2) unknown exposure; or 3) confirmed absence of exposure.

#2: Sentinel Facial Features (Phenotype)

Sentinel facial features refers to the unique pattern of facial features associated with FASD. Phenotype refers to the visible characteristics of a person. When a fetus has been exposed to alcohol, changes in facial features may occur. The presence of some features, discriminating features, helps to *rule out* the possibility of other disorders, while other features, associated features, are those that often occur as a result of prenatal alcohol exposure but are *not unique* to individuals with FASD. In other words, the presence of associated features alone does not allow for a diagnosis of FASD.

It is important to note that only about 10% of individuals with PAE have these sentinel facial features (CanFASD, 2019). However, these facial features are often the most outwardly observable sign of FASD. This is why FASD is often considered an invisible disability. Another factor which complicates using

physical features as key diagnostic criteria is the fact that the presence or absence of specific facial and physical features differ across age groups and populations (Fang et al., 2008; May et al., 2010; Moore et al., 2007). In other words, some features are normal in some populations and some features change when a child grows.

#3: Central Nervous System Impacts

The third criterion examined in the FASD diagnostic process are central nervous system (CNS) impacts. These impairments are assessed in ten neurodevelopment domains (Table 7.1) (Cook et al., 2016). A diagnosis of FASD can be made when there is evidence of severe impairment in three or more of the ten domains (Cook et al. 2016).

Note – detailed information about the impacts on CNS caused by prenatal alcohol use can be found in Module 4: Brain Anatomy, Development, and Function.

Table 7.1: Ten Brain Domains for Assessment of Central Nervous System Impairment (Cook et al., 2016; Lang, 2006)

Brain Domain	Testable Areas
Academic Achievement	• Skills in core academic areas (e.g., reading, math, and written language)
Adaptive Behaviour, Social Skills, and Social Communication	• Degree to which the person can meet the challenges of daily life • Ability to communicate appropriately and effectively in social situations • Ability to relay verbal information coherently and cohesively
Affect Regulation	• Includes anxiety, depression, and mood imbalance
Attention	• Processing capacity for selective, focused, sustained, and flexible attention • Behavioural abilities (e.g., concentration, hyperactivity, and impulsivity)
Cognition	• General level of verbal and non-verbal thinking ability
Executive Function, Impulse Control, and Hyperactivity	• Capacity for goal-directed behaviour including self-regulation, initiation, working memory, planning, organization, and self-monitoring
Language	• All aspects of expressive and receptive language • Ability to integrate language skills (e.g., sentence structure and grammar) • Use of words to convey meaning
Memory	• Ability to consolidate, store, and retrieve both short- and long-term information
Motor Skills	• Ability to use and coordinate large and small muscle groups

Brain Domain	Testable Areas
	• Gross motor skills including walking, running, hopping, and climbing • Fine motor skills including handwriting and eating • Hand-eye coordination
Neuroanatomy and Neurophysiology	• Presence of smaller head (microcephaly), brain size, seizure disorder and/or abnormal findings on a scan consistent with prenatal alcohol exposure

Growth deficiency used to be part of the key domains assessed in making a diagnosis of FASD. However, the 2015 Canadian clinical guidelines for the diagnosis of FASD no longer include growth deficiency as a diagnostic criteria as recent evidence and clinical experience suggests that growth is neither sensitive nor specific enough to indicate an FASD diagnosis (Cook et al., 2016; O'Leary, et al., 2009). Current diagnostic guidelines recommend that associated features that are not diagnostic should, however, be routinely assessed due to their high prevalence and importance in management planning. These include sleep problems, sensory sensitivities, physical/congenial anomalies, growth, attachment, proprioception, and vestibular problems (See table 7.2) (Cook et al., 2016). Other diagnostically important information to document includes a confirmed genetic condition, as well as a history of adverse childhood experiences such as violence, neglect, and abuse (e.g., trauma), as they can be contributing factors to cognitive and behavioural deficits (Cook et al., 2016).

Table 7.2: Features Commonly Associated with FASD (Cook et al., 2016)

Associated Features	Symptoms
Sleep Problems	Nightmares, wakefulness, inability to fall asleep and/or stay asleep
Sensory Sensitivities	Hypo/hypersensitive to one or more of the five senses (i.e., sight, hearing, taste, smell, and touch)
Physical Findings/Other Congenital Anomalies	Physical anomalies beyond the three sentinel facial features, both major and minor
Growth	Intrauterine growth restriction, small stature
Attachment	An aversion to touch and physical affection
Proprioception	Motor clumsiness, problems moderating grip (e.g., unintentionally breaks objects)
Vestibular	Balance problems, over/under reactive to head movement

Development of Current Canadian Guidelines for Diagnosis of FASD

In 2005, the first Canadian guidelines related to prenatal alcohol exposure were developed through an international, collaborative, evidence-based process. The 2005 Canadian guidelines were built on the strengths of the earlier Institute of Medicine (IOM) and 4-Digit systems, while working to avoid their limitations. Since 2005, there has been new evidence, expertise, and experience, and in order to improve both diagnoses and outcomes, the guidelines required revisions. In 2015, new guidelines were published which improve on the 2005 guidelines with the aim of providing clarity and consistency in FASD diagnoses (Cooke et al., 2016).

The new Canadian guidelines for the diagnosis of FASD titled "Fetal Alcohol Spectrum Disorder: A guideline for diagnosis across the lifespan" was first published electronically on epub in December 2015. The guidelines were then published in the Canadian Medical Association Journal (CMAJ) in February of 2016. The development was led by a steering committee, who relied on extensive clinician consultations, a thorough review of literature evidence, and used evidence-based tools to standardize the evaluation of evidence and development of recommendations (Cooke et al., 2016). The 2015 guidelines include new recommendations for diagnostic terminology and improve on the description of brain domains to be considered in assessment.

The 2015 guidelines also provide additional recommendations on the diagnostic process, and additional considerations for diagnosing infants, young children, adolescents, and adults (Cook et al., 2016).

Canadian Guidelines Diagnostic Criteria

Table 7.3 summarizes the 2015 Canadian guidelines for the diagnosis of FASD based on two diagnostic categories and one non-diagnostic category.

Table 7.3: Canadian Guidelines: Diagnostic Criteria for FASD (Cook et al., 2016, adapted from table 1 p. 193-4)

Diagnosis	Diagnostic Criteria
Diagnostic Category - I FASD with Sentinel Facial Features*	Although this diagnosis can be made in the absence of confirmed prenatal alcohol exposure due to the high specificity of the facial features, an accurate and reliable maternal alcohol history is still the recommended standard.
	Simultaneous presentation of the 3 sentinel facial features: a) Palpebral Fissure – shorter than usual space between the inner corner of the eyes b) Indistinct Philtrum – smoother or flattened groove between the nose and lips c) Thin upper lip
	Prenatal alcohol exposure (PAE) confirmed or unknown.

Diagnosis	Diagnostic Criteria
	This diagnosis should not be made when PAE is confirmed absent or at a level definitely below that known to be associated with physical and/or developmental effects.
	Evidence of impairment in 3 or more of the identified neurodevelopmental domains (see The Neurodevelopmental Assessment Section), or, in infants and young children, evidence of microcephaly.
	In addition, the following should also be documented if present: • Growth impairment and other alcohol-related birth defects • Hereditary, prenatal, and postnatal factors that may influence developmental outcomes
Diagnostic Category - II FASD without Sentinel Facial Features**	Evidence of impairment in 3 or more of the identified neurodevelopmental domains (see the Neurodevelopmental Assessment Section).
	Confirmation of prenatal alcohol exposure, with the estimated dose at a level known to be associated with neurodevelopmental effects.
	In addition, the following should also be documented if present: • Growth impairment and other alcohol-related birth defects • Hereditary, prenatal, and postnatal factors that may influence developmental outcomes
Non-Diagnostic Category **At Risk for Neurodevelopmental Disorder and FASD, Associated with Prenatal Alcohol Exposure**	This is a new category that was created to describe individuals who have confirmed prenatal alcohol exposure and some indication of neurodevelopmental concerns, but who do not meet the criteria for either of the above FASD diagnostic categories. As such, it should be noted that this category is not a diagnosis.
	This is a designation that should be given to individuals when: • There is confirmation of prenatal alcohol exposure, with the estimated dose at a level known to be associated with neurodevelopmental effects (see section on PAE) • Evidence of impairment in 3 or more of the identified neurodevelopmental domains (see the Neurodevelopmental Assessment Section) • There is some indication of neurodevelopmental disorder in combination with a plausible explanation as to why the neurodevelopmental assessment results failed to meet the criteria for significant impairment (e.g., patient was too young; assessment was incomplete; etc.)
	In addition, the following should also be documented if present: • Growth impairment and other alcohol-related birth defects • Hereditary, prenatal, and postnatal factors that may influence developmental outcomes
	This designation may also be considered for individuals with all 3 sentinel facial features of FASD, who do not yet have documentation or evidence for the requisite 3 or more neurodevelopmental domain criteria or true

Diagnosis	Diagnostic Criteria
	microcephaly. This designation should, however, never be considered when PAE is confirmed absent.

* This diagnostic category has replaced the term Fetal Alcohol Syndrome ("FAS") as described in the 2005 diagnostic guidelines.

** This diagnostic category has replaced the terms partial Fetal Alcohol Syndrome ("pFAS") and Alcohol Related Neurodevelopmental Disorder ("ARND") as described in the 2005 Guidelines.

The Diagnostic Process

The diagnostic process has four main steps or stages:

1) Screening, Referral, and Support
2) Physical Examination and Differential Diagnosis
3) Neurodevelopmental Assessment
4) Management and follow-up

These steps are described in further detail in the next section.

The guidelines highlight the importance of using a multidisciplinary team approach for diagnosis of FASD. The members of the team vary depending on the age of the individual being diagnosed. The core team for each age group should include:

Infants (<18 months)

- Paediatrician/Physician
- Child development specialist who has the skill set to conduct physical and functional assessments (i.e., Speech-Language Pathologist, Physiotherapist, Occupational Therapist, Clinical Psychologist)

Preschoolers (18 months-5 years)

- Paediatrician/Physician
- Occupational Therapist
- Speech-Language Pathologist
- Psychologist

School-aged children (6 years-age of majority)

- Paediatrician/Physician with expertise in FASD and differential diagnosis
- Occupational Therapist
- Speech-Language Pathologist
- Psychologist

Adults

- Physician
- Psychologist

- Speech-Language Pathologist/Psychologist with expertise in language assessment

Depending on the unique needs of the individual, additional team members could include addiction counsellors, childcare workers, cultural interpreters, mental health workers, parents or caregivers, probation officers, psychiatrists, teachers, vocational counsellors, nurses, geneticists or dysmorphologists, neuropsychologists, and family therapists (Cook et al., 2016).

The use of a multidisciplinary team approach is crucial, as each member contributes something unique to the process. After completing the assessment, the team creates a report that describes the unique strengths and challenges of the individual and makes useful recommendations for follow-up. This report can be used as a resource for the individual, caregivers, educators, and other people who may work with the individual.

In addition to the use of a multidisciplinary team, the guidelines state that documentation from a variety of sources is necessary in order to ensure a comprehensive assessment. These sources can include:

- academic records
- achievement tests
- information from social services
- birth and pregnancy records
- medical and hospital records
- developmental, psychological, and/or psychometric assessments
- legal reports
- documentation of the family history

The guidelines also emphasize the importance of *readiness* (Chudley et al., 2005). For the best results, the individual, family, and community must be prepared to participate in the diagnostic process. *Disclosure* by the diagnostic team (e.g., to the family) is vital. The family must be made aware of the potential positive and negative implications that may accompany a diagnosis of FASD.

The Four Steps of the Diagnostic Process

Step 1: Screening, Referral, and Support

The purpose of screening is to identify individuals who should be referred for an assessment. The primary healthcare providers should screen all individuals for alcohol use in a respectful, non-judgemental, and culturally sensitive way. The Canadian guidelines provide a number of recommendations for screening, which are summarized in Table 7.4.

Table 7.4: Recommendations Regarding Alcohol Use Screening and Referral in the Canadian Diagnostic Guidelines (Cook et al., 2015, adapted from p. 10-11)

All pregnant and post-partum women should be screened for alcohol use with validated screening tools (e.g., T-ACE) by relevant healthcare providers. Women at risk for heavy alcohol use should also receive early brief interventions (i.e., counselling and/or other services). If alcohol use is believed to be an issue, women should be offered support and referrals to appropriate counselling and treatment providers. The FASD screening can also play a preventative role, as it identifies those with a history of alcohol consumption during pregnancy, who may become pregnant in the future. These individuals can then be provided with education and prevention information. (Refer to Module 6: Prevention of FASD for more information about validated screening tools for alcohol use.)
Abstinence from alcohol should be recommended to all women during pregnancy. It is important to let women know that stopping drinking at any point during pregnancy will improve outcomes for the child, and pregnant individuals should be connected with the appropriate supports and services.
Referral of individuals for a possible FASD diagnosis should be made whenever there is evidence of or suspected prenatal alcohol exposure at levels associated with physical or developmental effects.
Determination of prenatal alcohol exposure requires confirmation that the mother consumed alcohol during the index pregnancy **(the pregnancy in question).** Confirmation of prenatal alcohol exposure must be based on: • reliable clinical observation • self-report • reports by reliable sources or medical records documenting positive blood alcohol • alcohol treatment during the pregnancy • other social, legal, or medical problems related to drinking during pregnancy If available, information concerning the number and type of alcoholic drinks consumed, pattern of use, and frequency of drinking should also be documented.

For an FASD diagnosis, the general consensus is the confirmation of more than 7 standard drinks per week or more than 4-5 drinks per occasion (within 2 hours) on at least 2 occasions. Given that the effects of a single binge episode may be relatively small, a minimum of 2 binge episodes is recommended to make an FASD diagnosis (Canada FASD Research Network, 2018). In Canada, a standard serving of alcohol contains 0.6 oz (17.7 mL) of pure ethanol, which approximates the amount of ethanol in a 12-ounce serving of regular beer at 5%; a 5-oz glass of wine at 12%, and a 1.5-oz glass of a spirit at 40% spirit (Canada FASD Research Network, 2018). See Module 5: Role and Impact of Alcohol for more information on standard drink sizes.

Step 2: Physical Examination and Differential Diagnoses

The goal of the physical assessment is to distinguish the specific physical features associated with prenatal alcohol exposure from those that arise due to other causes (Cook et al., 2016). The physician will look for the characteristic facial features of FASD. It has been noted that the sentinel facial features tend to become less evident throughout the lifespan, making it more difficult to detect in older children, adolescents, and adults. For this reason, the guidelines suggest that when assessing older clients, it may be useful to look at childhood photographs (Chudley et al., 2005).

The physician should also record any other existing anomalies. Several structural deficits and/or birth defects have been associated with FASD, such as defects in the ears, eyes, palmar creases, digits, elbow, joints, as well as oral facial clefts and congenital heart defects (DeRoo et al., 2008; Jones et al., 2010; O'Leary et al., 2013). Although these anomalies may not be specific to FASD, they may be useful in differentiating from other possible disorders with features similar to FASD. This process is known as differential diagnosis, which involves distinguishing between similar conditions by comparing and contrasting their signs and symptoms. The importance of differential diagnosis, and some of the disorders with features that are similar to FASD, will be discussed later in this module.

Growth should be monitored to detect pre- and/or postnatal growth deficiency and disproportionately low weight-to-height ratio. Although many children with FASD show growth deficiency at some point (i.e., in utero or after birth), growth deficiency is no longer considered a criterion for diagnosis according to the new guidelines (Cook et al., 2016). Some issues with using growth deficiency as diagnostic criteria include the following (Chudley et al., 2005):

- Growth deficiency may not be present if alcohol is not consumed during the third trimester.
- Certain illnesses (e.g., gestational diabetes) can result in larger fetal size, making it difficult to detect growth deficiency.
- Parental size, genetic potential, and other factors (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., racial, or ethnic groups).

Step 3: Neurodevelopmental Assessment

The neurodevelopmental assessment involves looking for evidence of CNS impairment on structural, neurological, and functional levels (Chudley et al., 2005). Even though the guidelines only require evidence of impairment within three of the ten neurodevelopmental domains (e.g., motor skills; neuroanatomy/neurophysiology; cognition; language; academic achievement; memory; attention; executive function; affect regulation; and adaptive behaviour, social skills, or social communication), it is important that all the domains are assessed. The assessment should look at and compare each of the basic and complex tasks in each domain. Although there may be possible overlap between domains, each domain should be assessed independently. This information will be useful when developing a treatment and follow-up plan that meets the unique needs of the individual.

Step 4: Management and Follow-Up

Step 4 occurs after an individual has received a diagnosis. During this phase, a comprehensive report detailing the assessments and examinations is completed. Recommendations for treatment and follow-up are also included. This can be a useful tool for caregivers and professionals who are working with the newly diagnosed individual.

The guidelines emphasize the importance of using this information to improve the quality of life for the individual affected with FASD, their family, and their community. Using a culturally appropriate approach, the family and individual can be educated about FASD and prepared for potential psychosocial tensions that may develop. Psychosocial tensions are a combination of psychological and social factors that may cause difficulties in the environment.

Another key element of this step is to help with understanding the diagnosis and using this knowledge to develop effective strategies. The Canadian guidelines stress that the post-diagnosis report is the most crucial piece of the process, acting as a "blueprint" for intervention.

Members of the diagnostic team should follow up and ensure that treatment plans/recommendations are followed. It is also important to provide the family and individual with information about accessing appropriate resources and services.

Establishing connections between the individual, caregivers, and community resources/supports is essential. Since it can be a complicated process, extra support from team members can provide valuable awareness at this stage by helping with the implementation of the treatment plan or identifying and finding relevant information, supports, and services. Creating links with appropriate supports will certainly improve outcomes and decrease the likelihood of adverse impacts.

Concurrent Problems, Differential and Overlapping Diagnoses

Concurrent Problems

Because of the far-reaching effects of FASD, there are many mental and physical concurrent (co-occurring) problems that are often linked to FASD. Some of these are adverse impacts or challenges, while others are thought to be primary disabilities that are directly related to FASD. It is important to note that maternal alcohol use is not the only cause of adverse impacts, as they are a result of a complex interplay between psychobiology and environmental factors.

Some medical problems and physical birth defects that are commonly associated with FASD are (Saskatchewan Learning, 2004; Stratton et al., 1996):

- hearing problems
- heart defects/murmurs
- craniofacial (head and face) differences
- vision problems
- kidney defects
- skeletal abnormalities

More recent research shows that children and adults with FASD are more likely to have many health conditions than members of the general population, and more than 400 health impacts have been found to co-occur with FASD, including a wide range of birth defects (Popova et al., 2016). The most common physical conditions in individuals with FASD include abnormalities of the peripheral nervous system, and chronic serious otitis media (ear infections) (Popova et al., 2016). For more details on co-occurring conditions, see Module 8.

Differential Diagnosis

FASD may affect several domains and shares many characteristics with other diagnoses; therefore, it is important to conduct a differential diagnosis (where other similar disorders are ruled out) (Cook et al., 2016). This is especially true because the implications of misdiagnosis can be severe. For example, an individual who has been misdiagnosed could receive treatment and/or medication that is inconsistent with effective interventions for FASD (Astley & Clarren, 2000).

A differential diagnosis rules out other disorders.

Differential diagnosis is especially important to consider before giving an FASD diagnosis to a child under the age 6 years, given the inability to conduct a comprehensive neuropsychological assessment in young children (e.g., cannot assess Academic Achievement; Cook et al., 2016). It is, therefore, important that these children receive appropriate investigations to exclude any underlying structural brain malformation or a genetic/metabolic disorder that share symptoms similar to FASD (Cook et al., 2016).

The combination of CNS impairment and craniofacial (head and face) differences are not unique to FASD (Leibson et. al., 2018). This means that it is important to rule out disorders with related symptoms prior to diagnosing FASD. Although it is not common, it is possible for a person with FASD to also have another genetic or substance-use related disorder (Chudley et. al., 2007).

Overlapping Diagnosis

Individuals with FASD tend to acquire numerous diagnoses during their lifetimes. In their early years, these diagnoses often reflect primary behavioural characteristics (e.g., failure to thrive, speech and language delays, and learning disabilities; Subramoney et al., 2018). As the individuals move towards adolescence, the diagnoses start to reflect adverse impacts (e.g., Depression, Conduct Disorder, Oppositional Defiant Disorder; O'Connor & Paley, 2009). According to Malbin (2008), this is due to years of being a "poor fit with the environment" and the constant stress of dealing with frequent misunderstandings in the social environment.

The recent Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-V), which is used by psychologists and psychiatrists to guide the diagnosis of mental health disorders, now includes proposed diagnostic criteria for FASD related conditions, and references FASD-related diagnoses under the section "Neurodevelopmental Disorders". For example, impairment in affect regulation is present when an individual meets the DSM-5 criteria for major depressive disorder (with recurrent episodes), persistent depressive disorder, disruptive mood dysregulation disorder (DMDD), separation anxiety disorder, social

anxiety disorder, panic disorder, or generalized anxiety disorder (Cook et al., 2016). Other relevant FASD-related conditions under the DSM-V classification include attention deficit hyperactivity disorder (ADHD), oppositional defiant disorder (ODD), intellectual disability, impulsive control disorder, conduct disorder, and specific learning disorder (Cook et al., 2016). Even prior to the DSM-5, the diagnosis of mental illness as a concurrent disorder with FASD has been well established. Frequently, individuals who do not yet have FASD as a primary diagnosis can accumulate many different diagnoses related to mental health. This, in turn, can lead to great confusion in terms of understanding and treatment (Malbin, 2008).

While these initial diagnoses may be correct, they may not truly capture the individual's fundamental challenges (e.g., brain-based deficits) or take their specific brain functions into account. When the brain impacts caused by prenatal alcohol exposure are not considered, the individual may face even more frustration and trauma because of inappropriate interventions and treatments. Accurate identification of the true causes of behaviours that consider *both* FASD and other diagnoses will allow for the development of the most effective and appropriate interventions/accommodations for the individual (Malbin, 2008).

Some of the common overlapping diagnoses that occur with FASD include (O'Connor & Paley, 2009; Weyrauch et al., 2017):

- Attention deficit hyperactive disorder (ADHD)
- Intellectual disability
- Oppositional defiant disorder (ODD)/conduct disorder (CD)
- Depressive disorder
- Anxiety disorder
- Psychotic disorder
- Insecure attachment/reactive attachment disorder

Because ADHD is a diagnosis that commonly co-occurs with FASD, many clinicians once believed that alcohol is a causal factor in ADHD (Coles et al. 1997). However, Coles et al. (1997) determined that children with FASD versus ADHD have *unique attention profiles.* These profiles are based on four distinct processes of attention which are: 1) focusing, 2) sustaining, 3) encoding, and 4) shifting.

Researchers found that children with ADHD struggle with focusing and sustaining attention, while children with FASD have deficits in encoding information, perception, and using flexibility in problem solving. This finding highlights two important points:

- FASD and ADHD are two independent diagnoses that can co-occur.
- Strategies that work for individuals with FASD will differ from those that work for individuals with ADHD.

Attachment for Children with FASD

Attachment is the deep and lasting connection that infants and children form with those they depend on for care (Cassidy, 1999). The development of attachment begins at birth and typically develops over the course of the first two to three years of an infant's life (Marvin & Britner, 1999; Zeanah, Keyes & Settles, 2003). The infant is very vulnerable during this time period and relies heavily on the caregiver for basic needs (Hardy, 2007).

Insecure attachment means that the bond between the child and mother or caregiver is not as healthy as it could be, impacting the child's long-term functioning.

Research has found that maternal alcohol consumption during pregnancy may be related to the development of insecure attachment (O'Connor et al., 2002; O'Connor et al., 1987). However, the link between prenatal alcohol exposure and attachment may be due to poor neurobiological outcomes for the child, such as negative affect and behaviours. The child's negative affect and behaviours may impact the mother's ability to provide the sensitive emotional support needed for the development of secure attachment (O'Connor, et al., 2002).

On the other hand, when mothers of children prenatally exposed to alcohol were able to provide high levels of emotional support, these children had better coping skills and more secure attachments (O'Connor et al., 2002). In fact, children prenatally exposed to moderate-to-high levels of alcohol who experienced high levels of maternal support, had coping skills equal to those of children with no or light exposure to alcohol and high levels of maternal support, and had better coping skills than children not exposed to alcohol with low levels of maternal support. These findings suggest that the link between prenatal alcohol exposure and attachment may be due to the difficulties mothers experience in providing sensitive emotional support which then impacts the attachment relationship. However, it appears that if the mother is able to overcome these difficulties and provide the child with the emotional support needed, this can contribute to the child developing coping skills and developing a secure attachment (see Module 8 Young Children with FASD for more details).

The mother-child attachment relationship has a strong impact on the child's social, emotional, and cognitive functioning throughout life, including:

- personality development and mental health (e.g., a child's ability to regulate their own emotions and behaviour, and the quality of their interpersonal relationships) (Fonagy, 2003; Weinfield, Sroufe, Egeland & Carlson, 1999)
- neurobiological development, which then impacts a number of other factors (e.g., self-regulation, physiological responses to stress, adaptability, ability to learn from experiences) (Schore, 2001a; Schore, 2001b; Schore, 2002)

It is important to provide mothers of children with FASD with education and support as early as possible. If mothers are made aware of the benefits of responding with sensitivity and high levels of emotional support, despite their child's negative affect and behaviour, and if given the supports needed, they may be able to have a positive impact on the attachment relationship and their child's long-term functioning.

Research has found that when mothers of children who were prenatally exposed to alcohol were able to provide high levels of emotional support, their children had better coping skills and more secure attachments (O'Connor et al., 2002).

The Future of Diagnosis of FASD

FASD diagnosis is a complex and time-consuming process. Early diagnosis, as beneficial as it is, can be challenging. As a result, various less complicated procedures for diagnosing FASD have been developed and are being investigated.

Screening for Prenatal Alcohol Exposure

One of the major challenges in diagnosing an FASD is confirming that a mother drank alcohol during her pregnancy (Caprara et al., 2007). To overcome this challenge, researchers have been developing different methods for determining if a fetus has been exposed to alcohol.

Biomarkers

Researchers have looked into PAE biomarkers that can be used to test either newborn hair or meconium (Benz, Rasmussen, Andrew, 2009). A biomarker is a substance/chemical found in the blood, other body fluids, or tissues that can be used to show the presence of exposure. Researchers have tested many different biomarkers for alcohol use during pregnancy. Promising research revolves around fatty acid ethyl esters (FAEE) which build up in organs damaged by high levels of alcohol use (Refaai et al., 2002). Findings show that FAEE harm the fetus (Bearer et al., 1992) but cannot cross the placenta barrier (Chan et al., 2004). One could conclude that if FAEE are found in a newborn, the fetus was exposed to alcohol (Caprara et al., 2007). That is because the alcohol forms FAEE in the fetus.

More recently, researchers have looked into the presence of various ethanol metabolites (biomarkers) in tissues of pregnant women and babies. The following ethanol metabolites have been found:

- Fatty acid ethyl ester (FAEE) in meconium of newborn and blood, hair of the mother and the newborn
- Ethyl glucuronide (EtG) in the placenta, nails, hair in adults and meconium, urine in newborn
- Phosphatidyl ethanol (Peth) in infant blood

(Jańczewska et. al., 2021)

Meconium Screening

Research has been conducted on detecting prenatal alcohol exposure through the screening of meconium (the first feces of a newborn child). Because meconium collects in the intestines beginning in the 13th fetal week, tests of meconium can indicate substance exposure throughout the second and third trimesters of pregnancy (Caprara et al., 2007, Goldberg & Aliani, 2018). FAEE is one of the

substances that has been found to accumulate in the meconium (Ostrea, 1999). More FAEE has been found in the meconium of children born to women who drink during pregnancy than those who abstained from alcohol during pregnancy (Moore & Lewis, 2001, Goldberg & Aliani, 2018). One of the major limitations of meconium screening is that meconium is only available during the first few days of an infant's life (Caprara et al., 2007, Goldberg & Aliani, 2018). It is also important that this test (and any test) be done with the consent of the child's legal guardian, which is typically the parents, or the child protection agency because of the heightened sensitivity toward detecting alcohol in pregnancy, and in deep respect to parents' rights (Public Health Agency of Canada [PHAC] & Canadian Association of Paediatric Health Centres [CAPHC], n.d.)

Hair Testing

Given that meconium screening can only be done during the first days of an infant's life, researchers have been looking for a type of screening that provides a longer time period for collection and assessment. One type of screening being examined is neonatal hair testing. This type of testing is being considered for a number of reasons, including the following:

- Chronic exposure to alcohol during pregnancy leads to increased levels of FAEE in neonatal hair (Caprara et al., 2007).
- Neonatal hair does not shed until approximately three months after birth (Caprara et al., 2007).
- FAEE has been found to accumulate in hair in amounts that distinguish between heavy alcohol abusers, social drinker, and abstainers (Pragst et al., 2001).

Although hair testing offers important advantages over other biological tissues, it comes with limitations. First, low concentrations of FAEE have been reported even in strict alcohol abstainers (Pragst & Yegles, 2008). Second, differences in hair growth and hair care can affect results (Bakhireva & Savage, 2011; Pragst & Yegles, 2008). Furthermore, it can be very expensive and technically challenging to extract biomarkers from hair which limits its widespread clinical use (Bakhireva & Savage, 2011). Moreover, due to aesthetic and cultural reasons, collection of hair, especially that of a neonate can be difficult (Bakhireva & Savage, 2011), alongside considerations for ethics and consent. More research is needed to confirm the appropriate cut-off values of FAEE and EtG (Pragst & Yegles, 2008). It is also noted that FAEE and EtG should not be considered as a stand-alone test, but as an important contribution within the context of all evidence available for an individual case (Pragst & Yegles, 2008).

The FASD Behavioural Phenotype

Research has also focused on establishing a behavioural phenotype for FASD. Researchers are trying to determine if there is a specific pattern of behaviours that distinguishes children with FASD from those without a diagnosed behavioural disorder, and from children with different related disorders (Nash et. al., 2006). The creation of such a tool would have a tremendous impact on FASD screening and diagnosis and intervention (Nash et al., 2008).

A study by Nash et al. (2006) took an important first step in identifying a behavioural phenotype for FASD. The study showed that there may be certain behavioural features that, when they occur in a

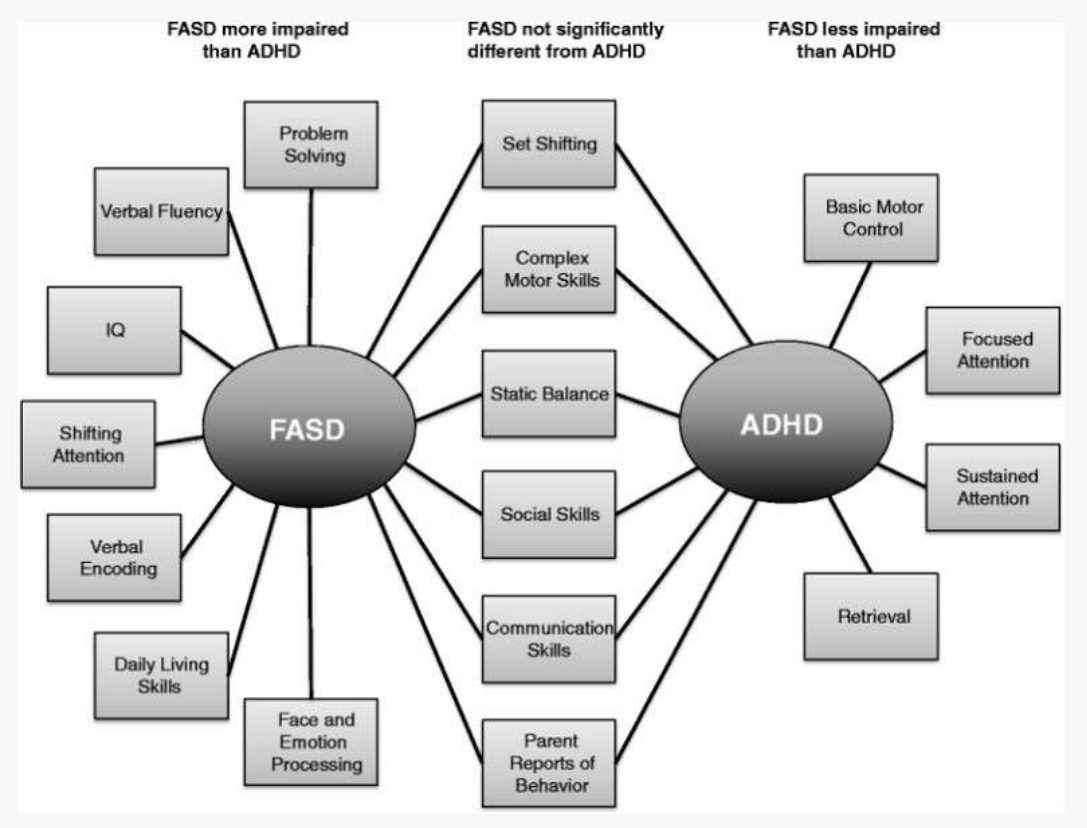
specific pattern, can indicate FASD while also making a distinction between FASD and other similar disorders (Nash et al., 2006).

In the study, caregivers gave ratings on the Child Behaviour Checklist. These ratings were compared for children with FASD, ADHD, and those with no behavioural disorder. The results showed that children with FASD and ADHD had distinct behavioural profiles. Due to the physiological changes in their brains because of prenatal alcohol exposure, children with FASD scored higher on the following items:

- Lack of guilt after misbehaving
- Cruelty
- Acting young for age
- Stealing from home or outside the home
- Lying and cheating

Additionally, research indicates that children with FASD experience halted development of socialization and communication skills with increasing age compared with those with ADHD whose skills improve over time at rates similar to controls (Crocker et al., 2009). Similarly, children with FASD also demonstrate poorer daily living skills (Crocker et al., 2009), social cognition, and facial emotion processing ability (Greenbaum et al., 2009) than children with ADHD and typically developing children.

Figure 7.1 Distinguishing and Overlapping Characteristics between FASD and ADHD (Mattson, et al., 2011, p. 94).



More recently, Nash et al. have extended their proposed screening tool to identify a set of behavioural characteristics that distinguished children with FASD from children with oppositional defiant/conduct disorder (ODD/CD) (2011). The aim is to develop a screening tool that allows children with FASD to be distinguished from children with other psychopathological conditions.

Nash et al., (2008) looked at the main issues in developing a behavioural phenotype for FASD. They reviewed all that is known about the cognitive-behavioural and socioemotional characteristics of FASD:

- IQ and academic achievement
- Language and communication
- Attention
- Executive functioning and working memory
- Learning and memory
- Socioemotional functioning (interplay between emotion and behaviour in the social world)
- Psychopathology (mental illness/conditions)

Nash et al., (2008) observed there has been overemphasis on cognitive factors and there are still many holes in a behavioural phenotype. This may be because cognitive factors are easier to "measure" and compare to norms. The high rate of co-occurring clinical mental health disorders also complicates creating a phenotype. A better and more concrete understanding of the full range of effects, including strengths and weaknesses, is needed.

Combining the phenotype with some of the new diagnostic procedures could have huge implications for the diagnostic process, early intervention, and lifelong well-being of individuals with an FASD. Further work in this area will provide insight and value to future research on diagnosis and screening. "In so far as it is possible, the primary goal of research efforts should be to provide hope for individuals with FASD and their families" (Nash et al., 2008, p. 880).

Other Research

As the research above has shown, technological advances have allowed researchers to examine FASD using a variety of methods. Other innovative methods include eye-tracking and ultrasound imaging of the corpus callosum (Module 4: Brain Anatomy, Development, and Function has more information on brain structures and FASD). Early results show that, when compared to controls, children with FASD:

- had longer reaction times, lower accuracy and control, and more direction errors on tests of eye movement (Green et al., 2007; Maurage et al., 2020)
- had a hook-like angle in the corpus callosum, shown by results from ultrasound images taken shortly after birth (Bookstein et al., 2007)

Although more research is needed in these areas, the preliminary research suggests that these tools could be useful for screening and diagnosis in the future.

Making a Referral for Diagnosis in Saskatchewan

Depending on the location of the individual and their family, there are different options for referral and diagnosis in three of Saskatchewan's urban centers.

For information on the services and supports available for individuals with FASD and their families, or other interested parties, see the Services for People with Disabilities section on the Government of Saskatchewan website https://www.saskatchewan.ca/residents/health/accessing-health-care-services/health-services-for-people-with-disabilities/fetal-alcohol-spectrum-disorder-services.

Conclusion

The creation of guidelines and diagnostic systems for FASD has been (and continues to be) an ongoing, complex, and lengthy process. Diagnosis is complicated by the fact that FASD is a complex condition, with a high degree of variability between individuals. Taking this variability into account, the Canadian diagnostic process covered in this module focuses on three key domains: evidence of prenatal alcohol exposure, central nervous system impairments, and sentinel facial features associated with FASD.

Individuals who have been affected by prenatal exposure to alcohol display a wide range of both primary disabilities and adverse impacts. As well, the strengths and challenges faced by individuals can change throughout the lifespan, and/or be influenced by the environment or genetics. As there is currently no standardized tool or set of tools that can confirm FASD, a series of tools and assessments must be used.

Obtaining a diagnosis can have negative consequences (e.g., feelings of maternal guilt and shame, stigmatization and stereotyping of the individual). However, the negatives can often be outweighed by the benefits to obtaining a diagnosis (e.g., identification of and access to appropriate interventions and accommodations).

Because the referral and diagnostic process can be lengthy and involved, it is important that individuals and families receive information and support from a multidisciplinary team of professionals who can meet their unique needs. Individuals and families who receive this support have been found to be more likely to complete the diagnostic process, and to follow the resulting recommendations.

Summary of 2015 Updates to Canadian Guidelines for the Diagnosis of FASD

- Fetal alcohol spectrum disorder (FASD) is a diagnostic term
- Recommendations are provided regarding the diagnostic criteria and process
- There are special considerations for diagnosing FASD in infants, young children, and adults
- Removed “growth” from the diagnostic criterion
- Added a new “at risk” category for those who do not fulfil diagnostic criterion but are at risk of FASD
- Specific changes and additions in the neurodevelopmental assessment:
  - “Hard and soft neurological signs including sensory motor” has been renamed “motor skills” and redefined
  - “Brain structure” has been renamed “neuroanatomy/neurophysiology” and redefined
  - “Communication” has been renamed “language”
  - “Attention deficit/hyperactivity” has been renamed “attention” and redefined
  - “Affect regulation” is added to the new guidelines
  - “Executive function” is expanded and clarified

(Cook et al., 2016)

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