

PedsCases Podcast Scripts

This is a text version of a podcast from PedsCases.com on “Approach to Fetal Alcohol Spectrum Disorder .” These podcasts are designed to give medical students an overview of key topics in pediatrics. The audio versions are accessible on iTunes or at www.pedcases.com/podcasts.

Approach to Fetal Alcohol Spectrum Disorder

Developed by Ying Ling and Dr. Gail Andrew for PedsCases.com.
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Hello everyone. My name is Ying Ling, a medical student at the University of Alberta. With the help of Dr. Gail Andrew, a professor in the Department of Pediatrics at the University of Alberta and a medical director of the FASD Clinical and Research Services at the Glenrose Rehabilitation Hospital, we have developed this podcast to describe an approach to Fetal Alcohol Spectrum Disorder in the clinical setting.

Objectives

After listening to this podcast, we hope that learners will be able to define the important clinical features of FASD, understand the importance of preventing and educating against the use of alcohol in pregnancy, and describe the evaluation of and diagnostic criteria for FASD. Finally, we hope that this presentation helps listeners understand the basics of supporting a family that has a child with FASD in a primary care setting.

Case Scenario

Let’s start off with a quick case scenario. Two worried parents come into your primary care clinic with concerns regarding their 5-year-old foster son. Their son began kindergarten several months ago and they are concerned that their son has difficulties with language and is smaller in height when compared to other children in his kindergarten class. The mother also states that the teacher has mentioned that he does not sit still in class, has a hard time listening to instructions, and doesn’t get along well with his peers, as he doesn’t wait his turn and doesn’t share toys. The parents have heard that children in foster care have an increased chance of having had prenatal exposure to alcohol and consequently have FASD. They are wondering if their son has FASD and if so, what they can do to help him function better in school.

Definition

To start answering the parents’ questions, we need to first define the term Fetal Alcohol Spectrum Disorder. FASD is an umbrella term that includes Fetal Alcohol Syndrome

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(FAS), Partial Fetal Alcohol Syndrome (pFAS), and Alcohol Related Neurodevelopmental Disorder (ARND). These conditions represent a spectrum of neurodevelopmental, behavioural, and physical findings associated with prenatal alcohol exposure (PAE), which is a leading preventable cause of birth defects and developmental disabilities.

Epidemiology

In the United States and some Western European countries, prevalence of FASD can be as high as 2-5%. Unfortunately, in women of childbearing age, the prevalence of alcohol consumption is increasing, and as many pregnancies are unplanned, it is important to educate all students, women, expecting mothers and their partners in a respectful and compassionate manner. As there has been no “safe level” of alcohol consumption during pregnancy identified it is crucial to promote alcohol abstinence while trying to conceive and during pregnancy. In addition, tools such as the CAGE and AUDIT-C questionnaires should be used to screen high risk mothers, and if an expecting mother is found to be consuming alcohol, it is important to be supportive and offer referrals to intervention, harm reduction, and treatment programs.

Pathogenesis

There are various ways in which prenatal alcohol exposure leads to the clinical findings seen in FASD. Specifically, it is a teratogen that can cause irreversible changes in the developing brain both structurally and chemically. Fetuses are particularly vulnerable because they can only eliminate at 3-4% the maternal rate and amniotic fluid is constantly being recycled. Most organ systems have critical developmental times in gestation and if PAE occurs during these times, there can be abnormalities. In contrast, the brain development is affected throughout pregnancy. In the first trimester, PAE can cause structural CNS changes such as a thinner corpus callosum, an abnormal amygdala, and decreased volume in the frontal lobe, striatum, caudate, thalamus, and cerebellum. In the second and third trimesters, alcohol can affect neuronal migration and differentiation. Facial abnormalities are associated with PAE on days 10 to 21 of pregnancy while growth parameters such as weight and length are more affected in the third trimester.

Clinical Features

The clinical features seen in FASD can be grouped into structural changes, CNS findings, and co-morbid mental health conditions. When suspicious of FASD, it is important to ask about these features on history, or look for these findings on physical exam.

First on the list of structural changes include the three *facial dysmorphisms* commonly associated with FASD: short palpebral fissures, a thin vermilion border, and a smooth philtrum. It is important to remember that not all children with FASD have these facial findings, so absence of these features does not rule out FASD, especially if there is a

history of PAE. Another key consideration is that changes in the palpebral fissure, vermilion border, and philtrum can be seen in certain genetic disorders, including but not limited to Trisomy 21, Williams Syndrome, and Prader-Willi Syndrome, and these must be ruled out accordingly.

Other findings include *cardiac abnormalities* such as atrial or ventricular septal defects, and cardiac outflow tract defects. *Skeletal findings* including flexion contractures, scoliosis, and clinodactyly can also be seen. There may be *problems with vision* from congenital strabismus or ptosis and difficulties with conductive or sensorineural *hearing loss*. There may be associated congenital *renal defects* such as an aplastic, dysplastic, or hypoplastic kidney, a horseshoe kidney, ureteral duplications, and hydronephrosis. Lastly, it is common for children with FASD to have *growth retardation* and *decreased head circumference* below the 10th percentile.

The CNS findings can be further broken down into *neurologic features* that can be brought out in a comprehensive neurological physical exam and *functional consequences* that can be asked about on history. The neurologic features include abnormal motor, cranial nerve, or coordination findings, seizures that can be distressing for the family to witness, and sensory dysregulation. The functional consequences can present as behaviours that the parents or teachers may have noticed such as difficulties with planning, judgement, organization, learning from consequences, memory, cognition, concentration, hyperactivity, and communication. These consequences may manifest in problems behaving at school and socializing with peers.

Finally, co-morbid mental health conditions can be found in over 90% of individuals affected by FASD. Specifically, attention deficit hyperactivity disorder (ADHD) is co-morbid in over 65%; depression, anxiety, and substance abuse are found in high numbers as well. Children with FASD often also have difficulty falling and staying asleep. This can be from a combination of medications, the described mental health conditions, adverse life events, school, and social frustrations.

Evaluation & Diagnosis

Now that we've gone over the epidemiology, pathogenesis, and clinical features typically seen in FASD, we can focus on when to be concerned about FASD in a child and how to evaluate for it. As we have previously described, there are a large number of findings that can be associated with prenatal alcohol exposure. Often, the symptoms that are first detected are the ones that become apparent when the child reaches school age, such as deficits in adaptive functioning, learning, attention, and social communication, just like in the case scenario we described at the start. There are also certain risk factors to be aware of that can make FASD more likely when evaluating a child presenting with behavioural difficulties. For the child, a diagnosis of FASD can be more likely if they have a sibling with confirmed FASD, if they have a history of being in an orphanage or foster care, or if they've had involvement with child protective services.

There are also maternal and psychosocial risk factors, the majority of which are rooted in social determinants of health. These risk factors consist of a maternal history of a mental health condition, lower education, older age, increased gravidity and parity with a history of miscarriages, stillbirths, or other children with FASD. From the family perspective, if there are abusive relationships in the household and history of substance abuse in the mother or the father, it can also be an indicator that a child may be at risk for FASD. However, it is important to remember that even in the absence of any of these risk factors, all women who consume alcohol during pregnancy are at risk.

When there is any form of suspicion that a child may have FASD, whether it is from a history of behavioural challenges, or from clinical features consistent with FASD like the presence of facial dysmorphisms, or from the presence of the risk factors listed above, evaluation for FASD should be initiated as soon as possible. Earlier evaluation leads to earlier intervention, better outcomes, and can even prevent subsequent children from alcohol exposure if the birth mother can be reached to provide her with positive supports. A multidisciplinary team typically consisting of a paediatrician, a psychologist, a social worker, an occupational therapist, and a speech language pathologist will complete this evaluation. They will check for prenatal alcohol exposure from a reliable source such as birth records, child welfare reports, or direct interview with the birth mother if possible. They will also conduct a thorough physical exam, neurocognitive assessment, and behavioural assessment looking for the clinical findings that we discussed before.

After this evaluation is complete, the team can see if the child meets the diagnostic criteria for the three conditions that fall under the FASD umbrella. For *Fetal Alcohol Syndrome (FAS)*, he or she needs to have the three facial dysmorphisms consistent with FASD, evidence of growth retardation as well as CNS involvement in structural, neurologic, and functional domains. For *Partial Fetal Alcohol Syndrome (pFAS)*, the child needs two facial features, either evidence of growth retardation or CNS involvement, as well as confirmed alcohol exposure. And lastly, for *Alcohol Related Neurodevelopmental Disorder (ARND)*, the child needs to have confirmation of prenatal alcohol exposure and demonstrate CNS involvement in the functional domain causing impairment in neurocognition, self-regulation, and adaptive function. In ARND, presence of facial features, growth retardation, and CNS changes in the structural and neurologic domains are not required.

Management

Many systems are required to support an individual once a diagnosis of FASD is made, including schooling, welfare, healthcare, justice, as well as mental health supports and social supports. The role of the primary care provider is an extremely important one to coordinate access to these systems, to provide long-term management, and to provide continuous support for families with children diagnosed with FASD. As we had mentioned before, early evaluation for FASD is important, so as a primary care provider, it is crucial to provide early identification, screening, and referral to the multidisciplinary teams that can properly evaluate for and diagnose FASD.

A diagnosis of FASD results in a large financial and emotional burden being placed on the caregivers. For this reason, it is important for the primary care provider to provide family support in the form of education, guidance, encouragement, and connection to resources and support services. Families should be taught to readjust their expectations to be appropriate for their child, and provide a secure, stable, and safe home environment. Unstable environments with exposure to neglect and abuse can negatively impact early brain development that can worsen the prognosis of FASD.

As we had discussed previously, there are other organs that can be affected by prenatal alcohol exposure, so primary care providers should screen for other medical conditions and refer to appropriate specialists when indicated. Often, these medical conditions take the form of co-morbid neuropsychiatric conditions, which can be found in over 90% of individuals diagnosed with FASD, so screening for conditions like ADHD, anxiety, and depression should be done, with medication initiated and mental health services consulted when appropriate.

To help families manage behavioural difficulties and promote basic language, self-care, learning, and executive functioning skills, primary care providers can consult targeted special education programs for children with FASD, and connect to speech and language, occupational, and physical therapies when indicated, as early as possible. As FASD is a life long condition, caregivers need to plan for transition to the adult system. Primary care providers can connect and refer to social skills, adaptive or daily living skills, employment skills, housing, and financial supports when this transition begins. Some adverse outcomes associated with FASD that can be avoided are problems with the law, detention in institution, substance abuse problems, and homelessness. Therefore, care providers should also be mindful of substance abuse counselling if necessary and helping the individual avoid situations that can lead to victimization and trouble with the law, as individuals with FASD can be especially vulnerable to these situations.

Summary

Before we finish this podcast, let's summarize what we've learned by going back to our main learning objectives:

- Define and list the important clinical features of FASD. Fetal Alcohol Spectrum Disorder is an umbrella term that includes Fetal Alcohol Syndrome, Partial Fetal Alcohol Syndrome, and Alcohol Related Neurodevelopmental Disorder, each with their own diagnostic criteria. The clinical features that can be seen in FASD include structural changes in facial features, growth, and other organs; CNS findings in structural, neurologic, and functional domains; and co-morbid mental health conditions.
- Understand the importance of preventing alcohol use in pregnancy by educating all students, women, expecting mothers and their partners in a respectful and

compassionate manner. The CAGE and AUDIT-C questionnaires can be used as a screening tool.

- Describe that earlier evaluation by a multidisciplinary team leads to earlier intervention and better outcomes. This is made possible by the early identification, screening, and referral by a primary care provider that is familiar with the clinical features and risk factors for FASD.
- Understand that the primary care provider plays a crucial role in providing long-term management for medical co-morbidities, coordinating referrals to support services and therapies appropriate for the age of the individual, and providing continuous support and education for families of individuals with FASD.

That marks the end of our presentation on Fetal Alcohol Spectrum Disorder! Thanks for listening to PedsCases Podcasts.

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