Fetal alcohol syndrome – causes, diagnostic criteria and prevalence

Alkoholowy zespół płodowy – przyczyny, kryteria diagnostyczne i prewalencja

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Słowa kluczowe: pełnoobjawowy płodowy zespół alkoholowy (FAS), spektrum poalkoholowych skutków rozwojowych (FASD), prenatalna ekspozycja na alkohol, diagnostyka FAS i FASD, 4-stopniowa skala diagnostyczna.

Abstract

Fetal alcohol syndrome (FAS) is the outcome of alcohol exposition in the prenatal period. It is irreversible. In Poland, FAS is becoming more and more common, the diagnostic tools are limited though. It is recommended to use the 4-Digit Diagnostic Code, which evaluates the 4 basic FAS symptoms: growth retardation, dysmorphic appearance, damage to the central nervous system and prenatal alcohol exposure. It has been confirmed that there is no safe amount of alcohol for a mother to drink while carrying a baby. To put it another way, only a complete lack of alcohol consumption is a guarantee that the baby will not suffer from FAS. It is necessary for society to know that even the smallest amount of alcohol is bad for the foetus. A number of people still believe that, for example, red wine is good and healthy for both the mother and child.

Streszczenie

Alkoholowy zespół płodowy (*fetal alcohol syndrome* – FAS) jest skutkiem prenatalnej ekspozycji na działanie alkoholu. Jest on nieodwracalny. W Polsce coraz częściej rozpoznawane są FAS i spektrum poalkoholowych skutków rozwojowych (*fetal alcohol spectrum disorders* – FASD), ale dostęp do specjalistycznych narzędzi diagnostycznych jest ograniczony. Wskazane jest posługiwanie się 4-stopniową skalą diagnostyczną (4-Digit Diagnostic Code), która umożliwia ocenę nasilenia 4 podstawowych objawów FAS, takich jak: opóźnienie wzrostu, cechy dysmorficzne charakterystyczne dla FAS, uszkodzenie ośrodkowego układu nerwowego oraz prenatalna ekspozycja na alkohol. Potwierdzono, że nie istnieje bezpieczna dawka alkoholu dla kobiety w ciąży i abstynencja w tym okresie gwarantuje niewystąpienie FAS u dziecka. Konieczne jest szerokie informowanie społeczeństwa o toksycznym działaniu nawet niewielkich dawek alkoholu w ciąży, ponieważ nadal pokutują przekonania o np. pozytywnym wpływie czerwonego wina na stan ciężarnej i płodu.

Introduction

Fetal alcohol spectrum disorder (FASD) is a wide term describing the whole spectrum of problems in the development of children whose mothers used alcohol during pregnancy. These problems may include defects of physical and mental development, bad behaviour and diminished learning capability. These symptoms may last for their whole life [1–3]. Fetal alcohol spectrum disorder may also be defined as a condition that includes neurobehavioural abnormalities and changes in the structure of the body and internal organs of the children whose mothers used alcohol during pregnancy [4].

General survey

The most dangerous outcomes of drinking during pregnancy are miscarriages or intrauterine fetal death and still birth. Less serious cases can result in fetal alcohol syndrome (FAS) or FASD.

Children with the complete phenotype of physical anomalies, growth retardation and central nervous system (CNS) development abnormalities are diagnosed with FAS. In daily practice we observe that the partial presence of these symptoms is much more common. Partially affected children are diagnosed with FASD and rarely with partial FAS (PFAS), alcohol-related abnormalities of neurodevelopment (ARND) or al-

cohol-related birth defects (ARBD) [5–7]. The research run by the Państwowa Agencja Rozwiązywania Problemów Alkoholowych (Polish Agency of Alcoholic Problems – PARP) states that 22–30% of pregnant women in Poland drink alcohol. Annually, there are about 900 children born in Poland with FAS and about ten times more born with FASD. In Southern Africa there are 39.2–46.4 children with FAS per 1000 births [8]. It is believed that there is no safe amount of alcohol to drink whilst pregnant [4]. The reaction to alcohol is specific to the individual, and there is no way to determine a safe amount to drink that applies to every woman. One of the studies that focused on this subject examined urine samples of pregnant women, who submitted them for routine testing, for the presence of alcohol. Thirty percent of the samples were found to be positive, which means that one in three of the pregnant women used alcohol [9].

Anonymous research run by PARP showed similar results; one in three women admitted to drinking alcohol while being pregnant [9]. The risk of negative effects of drinking while carrying a child grows with the amount of used alcohol, the frequency of drinking and the mother's age, and it is also greater if the mother is in bad physical shape, is addicted to nicotine or lives a stressful life [10]. The problem of drinking whilst pregnant is most common among women with average education, and least common among those with basic education. The majority of pregnant drinkers live in medium-sized cities. Women describing themselves as religious are less likely to drink while pregnant (12%), than are those who do not practice any religion (31%) [5]. Moskalewicz distinguishes two groups of major risk: the first group cossets of women who drink more than 7.5 l of grain alcohol annually, and the second group consists of women who drink more than 80 ml of grain alcohol at one time. Women from the second group make 10% of the population and usually are 20–49 years old [11]. Moreover, research on teenage drinking shows that girls drink more alcohol than they used to [12]. Another research project has revealed that the Polish population in fact know about the negative outcome of drinking whilst pregnant, but a lot of them, both men and women, believe that small amounts, mainly of red wine, may be good for both mother and foetus. People surveyed stated that this information came most often from their families and friends, less often from the press, radio, television or their doctors [5].

The term "FAS" was created by K.L. Jones and D.W. Smith in 1973. They focused on the similarities of the defects presented by pregnant drinkers' children, something which had previously been explained by malnutrition of the foetus [13]. In the eighteenth century, English doctors described children of alcoholic mothersas "weak, handicapped and lacking character". Sullivan (1899) recorded his observations of alcoholic women in prisons. Among the children of these women, he saw a large percentage of birth defects, and among the women themselves, frequent miscarriages. Interestingly, Sullivan realised that pregnancy spent in prison was less likely to end with problems or defects of the child because prisons are alcohol-free places [3].

The mechanism of alcohol's toxic influence on the foetus relies on causing abnormal migration of cells to the wrong parts of the brain and creating abnormal connections between neurons, and also activating apoptosis in CNS cells [1]. Alcohol goes through the placenta to the body of the foetus as early as in the second week of life. Forty–sixty minutes after consumption of alcohol by the mother, the concentration in alcohol the blood of the foetusis the same as that of the mother [2]. Two–three hours later its concentration is even greater than it is in the mother's bloodstream [14]. In the first trimester, the most vulnerable to alcohol are CNS, sight, hearing, kidneys, liver and heart. In the second trimester, CNS is the most vulnerable, along with the muscles, skin, endocrine system, skeletal system and teeth. Exposure to alcohol in this latter pregnancy period causes problems with concentration, short-term memory, difficulties with learning new information and difficulties with planning and foreseeing the outcome of actions [5, 10]. In the last trimester, alcohol retards weight gain and development of the lungs. It may also cause premature birth [2, 6, 15].

Fetal alcohol syndrome criteria

The diagnosis of FAS is a difficult task. People suffering from prenatal exposure to alcohol are affected by many symptoms, a large part of which is not specific exclusively for alcohol. They often manifest in various ways in the course of life. In Poland there is no access to diagnostic tools; there is also very little professional literature on the subject. The outcome of this is that the diagnosis is often based on a doctor's intuition. Incomplete diagnosis is also a common problem. Patients diagnosed with merely a symptom, such as ADHD, may not be treated accordingly for their condition.

In 1978 Clarren and Smith created the term "fetal alcohol effects" (FAE), and in 1980 the term was added to the more precise diagnostic criteria. This caused a rise in number of appropriately diagnosed patients. The criteria were thought to have been too wide because any patient prenatally exposed to alcohol could have been diagnosed with FAE [8]. In 1996 the Polish Working Group proposed a new classification of alcohol-related development defects [5]:

1) FAS with confirmed exposure to alcohol,
2) FAS without confirmed exposure to alcohol,
3) partial FAS,
4) alcohol-related birth defects,
5) alcohol-related abnormalities in neurodevelopment.

Currently, the most commonly used method in the diagnosis of FAS is the four-step diagnostic scale [8]. The four steps of this scale show the expression of four key characteristics for FAS in the following order:
1) growth retardation,
2) FAS dysmorphic features,
3) damage to the CNS,
4) prenatal exposure to alcohol.

The degree of expression of each of these characteristics is evaluated in the four-step Likert scale, in which one point means no FAS expression and four points means the classic presence of FAS symptoms [5, 15, 16].

Criterion I (growth retardation) addresses growth before and after birth. Growth retardation is connected to the slow weight gain of children born with FAS. Damaged CNS is also called static encephalopathy, which means that the condition of the brain is not deteriorating, but its progress in not satisfactory and sometimes is not present at all. What matters is the accurate evaluation of dysmorphic traits, especially on the face. These may not be easily seen immediately after birth, due to their lack of development, and in older patients too, since they may have regressed. The best time to evaluate them is the period between the 2nd and 10th years of life. The best way to determine how much women drink during their pregnancy is through an anonymous survey because most women deny the fact in conversation. The most commonly used questionnaires for screen drinking during pregnant are: CAGE (Cut down-Annoyed-Guilty-Eye-opener), T- ACE (Tolerance-annoyed-cut down-Eye-opener), TWEAK (Tolerance-Worried-Eye-opener-Amnesia-Cut Down) [1, 2, 4, 5, 7].

In summary, every child diagnosed with FAS needs a wide paediatric, psychological, pedagogical and neuropsychological profiling as well as neurological examination with neuroimaging such as magnetic resonance imaging and electroencephalography. Moreover, they require ophthalmic and laryngological care.

Conclusions

Currently the diagnosis of FAS is still developing in Poland. Knowledge about the problem is too scarce among medical personnel, psychologists and pedagogues. The full-symptom diagnosis is the most common because the visible dysmorphic characteristics and growth retardation make the parents seek help. Problems associated with FASD are most commonly discovered by teachers, who witness the child's inability to concentrate and remember the lessons.

The good thing about the latest classification is the ability to diagnose FAS without evidence of exposure to alcohol (FAS without confirmed exposure to alcohol).

Many of the children diagnosed with either FAS or FASD are raised outside their biological families. Their guardians often lack knowledge about the pregnancy, the birth or the earlier states of the child's development. Biological mothers are known to deny drinking, which makes the proper diagnosis harder to make and the treatment late or ineffective.

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