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## FASD – Fetal Alcohol Syndrome Disorder

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### ABSTRACT

Consumption of alcohol during pregnancy is a highly undesirable phenomenon, often causing miscarriages or many abnormalities of the child's development. Fetal Alcohol Spectrum Disorder (FASD) describes physical and mental disorders resulting from the effects of alcohol on health and behavioral disorders of the child, which are usually secondary to changes in the central nervous system caused by fetal alcohol intoxication during pregnancy. These changes are usually irreversible and evident throughout the life of a child affected by FASD, but if appropriate therapies are implemented, it is possible to minimize the symptoms of these disorders. Their nature and severity depend on several factors, including the time of exposure to alcohol and the health status of a pregnant woman. Clinical features of FASD include craniofacial anomalies, central nervous system disorders and growth retardation. Alcohol-induced structural changes include anomalies in the cardiovascular, skeletal, renal and urinary systems as well as in the organs of sight and hearing. Time of initial diagnosis plays an important role, because it allows the introduction of appropriate therapy for a child with FASD and the introduction of appropriate education of parents and other members of the family. It is possible to minimize the symptoms and disorders resulting from the Fetal Alcohol Disorders Spectrum, to the extent enabling them proper functioning of the child and his family.

**Keywords:** FAS, pregnancy, alcohol, FASD, disorders

## **1. INTRODUCTION**

Consumption of alcohol by pregnant women is a highly undesirable phenomenon, resulting in dead births or a series of developmental disorders of the child.

The disease syndrome describing the consequences of prenatal exposure of a child to alcohol, is FASD - the fetal alcohol spectrum disorders. The term includes not only health effects, both in the physical and mental context, but also the behavioral ones.

FASD consists of the following groups of disorders [5, 7]:

- FAS – Fetal Alcohol Syndrome
- pFAS – partial Fetal Alcohol Syndrome
- ARND – alcohol-related neurodevelopmental disorders
- ND-PAE – neurobehavioral disorders associated with prenatal alcohol exposure
- ARBD – alcohol-related birth defects

The purpose of this chapter is to present the health problems and diagnostic difficulties of children with FASD and the possibilities of medical and diagnostic assistance.

## **2. PATHOGENESIS**

Alcohol is a toxic substance that has irreversible effects on the central nervous system. The performed imaging studies prove the occurrence of brain structure and function disorders in people whose mothers consumed alcohol during pregnancy [1, 4, 7, 11]. The abnormalities include: reduced volume of the brain with a reduced volume of centers extremely important for health and proper functioning: frontal lobe, striatum, caudate, thalamus, cerebellum, corpus callosum and hippocampus [5, 6].

Damage to the above anatomical structures is manifested by improper transmission of impulses between the centers of the cerebral cortex, disturbances of memory and remembering, weakening of perception and impaired body motility [9, 14, 21].

The teratogenic effects exerted by alcohol on the fetus are dependent on the amount of alcohol consumed by a pregnant woman, the time of exposure to teratogen and the state of health of a woman. Attempts have been made to determine a "safe" dose of alcohol for pregnant women, but it does not exist. Even small amounts of alcohol have harmful effects on the fetus, especially in the first trimester of pregnancy.

In the first three months of fetal development, a high percentage of alcohol-related developmental disorders of the face and brain was observed. Exposure to teratogen in the second trimester of pregnancy increases the risk of spontaneous abortion. Consumption of alcohol by the future mother during the last three months of pregnancy influences the birth weight and length of the fetus, as well as the development of the brain [2, 4, 5, 9, 11].

In addition, it has been observed that systematic consumption of small amounts of alcohol and one-off intake of large amounts of alcohol is the most harmful.

The research carried out shows that the child of a pregnant woman who regularly consumes a minimum of five standard doses of alcohol (where 10 g of pure ethyl alcohol is considered per dose, i.e. the equivalent of 200 g of 5% beer or 100 g of 10% wine or 25 g of 40% vodka) on a weekly scale, will suffer from Fetal Alcohol Syndrome (FAS).

### **3. FASD EPIDEMIOLOGY IN POLAND AND IN EUROPE**

The first studies on the frequency of alcohol consumption by pregnant women were conducted in Poland in 2002, under the direction of Professor Tomasz Niemec, by doctor Piotr Raczyński at the Institute of Mother and Child in Warsaw. The research methodology was based on the analysis of urine sample of pregnant women, which were provided for routine testing to the laboratory. The obtained results indicated alcohol consumption by at least every third pregnant woman [23].

Another studies on the frequency of alcohol consumption by pregnant women were carried out by doctor Marek Banach in 2011. The survey covered 878 students of various Krakow universities who were pregnant. The results of published studies indicate that over 70% of surveyed women consumed alcohol during pregnancy [23].

The latest studies on the frequency of prenatal exposure were conducted by PARPA in the ALICJA program, which is still at the implementation stage. The results obtained so far indicate the frequency of occurrence in Poland of children with full Fetal Alcohol Syndrome (FAS) of at least 4 per 1000 live births. This statistic is comparable with other European countries [23]. The occurrence of some maternal and environmental risk factors for FASD development has been demonstrated [7, 10, 11, 12].

These are, among others:

- low level of mother's education,
- mother's age (over 45 years of age),
- miscarriage and abortion in an interview,
- nutritional deficiencies of the mother during pregnancy,
- occurrence of FASD in pre-born children,
- use of psychoactive substances,
- mother's mental illness, especially depression,
- physical or sexual abuse experience by the mother in the past,
- consumption of alcohol and other psychoactive substances by the closest persons of a pregnant woman,
- low economic status of the mother.

Women who do not meet the above factors who consume alcohol during pregnancy are at the same risk of giving birth to a child with FASD as women from other risk groups [12].

### **4. CLINICAL FEATURES**

The basic clinical features of people with FASD concern the face, central nervous system abnormalities and growth disorders. One should also bear in mind the number of co-occurring developmental disorders of other systems and organs [5, 6, 8, 9, 16, 17, 22].

1. The face of people suffering from FASD is characterized by short palpebral fissures, a smooth medial cleft and a thin red zone of the lip (Figure 1).
2. Structural congenital defects resulting from prenatal alcohol exposure to persons with Fetal Alcohol Spectrum Disorder (FASD), include symptoms from:

- **cardiovascular system:** atrial heart septal defect, ventricular septal defect and defects of the arterial cone and arterial trunk (e.g. anomalies of large vessels, Tetralogy of the Fallot).

Clinical symptoms of these diseases begin to appear in childhood, often taking the form of shortness of breath and chest pain during physical exertion and respiratory infections.

During adolescence and adulthood, life-threatening symptoms of right ventricular heart failure (limbs edema, swelling of the abdomen, enlargement of the liver, jaundice, blood coagulation disorders) and symptoms of left ventricular heart failure (dyspnoea, worsening during exercise and lying down, sleep apnea, wheezing, dizziness, cold hands and feet)
  - **skeletal system:** considerable flexion cramps of the joints, funnel chest, pigeon chest, Klippel-Feil syndrome (congenital short neck, due to cervical vertebra adhesions or undevelopment of the vertebrae), vertebral segmentation defects, hemivertebra, scoliosis (lateral curvature of the spine), radial-ulnar synostosis, undeveloped nails, short toes of the small fingers, clinodactyly of the fifth finger of the hand (the medial curvature of the little finger of the hand), camptodactyly (flexion contraction of the fingers).

The above diseases hinder the proper movement of the upper and lower limbs, and at the same time the basic activities of everyday life (walking, running, writing, cooking, cleaning, driving motor vehicles, etc.). In addition, they are often accompanied by pain.
  - **renal-urinary system:** dysplastic kidney (with disordered structure), hypoplastic kidney (with a reduced number of nephrons, i.e. basic functional and structural units), horseshoe kidney (right and left kidney are connected by lower poles); ureteral proliferation, hydronephrosis (urinary incontinence, no urinary leakage to the bladder for expulsion).
  - The above abnormalities can be manifested by abdominal and lumbar pains, frequent urination, urinary tract infections with diarrheal symptoms (pruritus, burning, pain during urination).
  - **Organ of sight:** squint (non-parallel eyeballs position), ptosis, retinal vascular anomalies, optic nerve hypoplasia, refractive problems (difficulty in refraction of light rays passing through eye structures) secondary to microcephaly.

The above-mentioned diseases manifest primarily in vision disorders and require treatment by an ophthalmologist.
  - **Hearing organ:** conductive hearing loss or receiving hearing loss.

These diseases prevent proper acoustic sounds and are an indication for treatment by an ENT doctor.
3. Central nervous system abnormalities affect about 70% of people exposed in the prenatal period to mother's intake of large amounts of alcohol (over 14 drinks during the week).

Clinical manifestation depends on age, getting worse over time and on other individual factors. Specific characteristic symptoms of a given age group are listed below.

- **Infancy period** – irritability, tremors, disorders of the autonomic system (innervating internal organs), disturbances of state regulation (e.g. sleep, concentration, agitation) and developmental delay
- **Childhood period** – hyperactivity, lack of concentration, cognitive impairment, emotional reactions, learning disabilities, hypotonia (too low blood pressure), impaired hearing and vision, convulsions, memory deficits and understanding and assimilation of new information
- **Puberty and early adulthood** – disorders resulting from the primary deficit in skills of living in society, impaired adaptation and executive functions (e.g. cessation of education, inability to maintain employment, inappropriate sexual behaviors).

Changes in the central nervous system caused by alcohol can be divided into:

- **Structural** – anomalies involving a reduced head circumference (usually defined in the cases below  $\leq 10$ th percentile, for a given age and gender, or if height and weight are  $< 10$  percentile, and head circumference  $\leq 3$  percentile) or structural abnormalities seen in neuroimaging (e.g., reduction in size and change in the shape of the corpus callosum, cerebellum or basal ganglia)
- **Neurological** – disorders composed of:
  - „**hard**” disorders – abnormal reflexes, abnormal muscle tension, cranial nerve defects
  - „**light disorders**” – impaired motor coordination and a sense of body balance, oculomotor disorders, nystagmus (involuntary eye movements), difficulty in performing complex activities or fast response in stressful situations and confusion
  - **Recurrent epileptic seizures** not caused by factors in the postpartum period (including infections)
- **Functional** – disorders that translate into IQ and cognitive functions of the individual. Scientific research indicates that children of mothers who have consumed alcohol during pregnancy have a lower IQ than their peers. People with FASD have impaired visual perception, difficulty in focusing and learning, problems with emotional stability, they are often overactive and nervous.



**Figure 1.** Examples of facial phenotype of children with FASD.

Difficulties in the proper personal and interpersonal functioning of the individual differ depending on the structure of the brain which has been damaged (Table 1) [20].

**Table 1.** Other possible difficulties for children with FASD due to CNS damage.

| <b>Damaged part of the brain</b>                   | <b>Possible disorders</b>   |
|--|---|
| Frontal lobes<br>Executive functions               | <ul style="list-style-type: none"> <li>- problems with decision making and planning, which is associated with the appearance of the task</li> <li>- ability to act according to the planned goal</li> <li>- creative and abstract thinking; assessment of own needs, desires and effectiveness of their satisfaction, evaluation of their own desires and procedures for their fulfillment</li> </ul> |
| Somatosensory cortex                               | <ul style="list-style-type: none"> <li>- sensory disorders</li> <li>- processing disorders</li> </ul>   |
| Hippocampus  | <ul style="list-style-type: none"> <li>- memorization processes</li> </ul>  |
| Cerebellum   | <ul style="list-style-type: none"> <li>- fluidity of movements</li> <li>- speech melody</li> <li>- in combination with the frontal lobes, fluidity of word production</li> </ul>  |
| Basal ganglia                                      | <ul style="list-style-type: none"> <li>- understanding of social behaviors</li> </ul>   |
| Right-hemisphere language functions                | <ul style="list-style-type: none"> <li>- problems with understanding information given indirectly, understanding metaphors, figurative speech, allusions</li> </ul>   |
| Corpus callosum<br>AKA: <i>Collosal commissure</i> | <ul style="list-style-type: none"> <li>- data transmission between hemispheres</li> </ul>   |

It is extremely important to realize that people with FASD require diagnosis and treatment throughout their lives. At an early age, primary disorders appear, which in the case of lack of diagnosis and treatment may result in the development of secondary disorders. (Table 2) [23] It is worth emphasizing that it is possible to eliminate secondary symptoms if they are diagnosed and treated in a timely manner [1, 3].

**Table 2.** Disorders that can come from CNS damage and secondary problems if the child does not receive help.

| <b>Primary disorders associated with CNS</b>   | <b>Secondary disorders</b>   |
|--|--|
| <ul style="list-style-type: none"> <li>- Sometimes drug withdrawal</li> <li>- Sleep disorders</li> <li>- Nervousness - manifested by restless sleep, too shallow sleep or hypersensitivity to stimuli</li> <li>- No or troubled sucking, at the same troubles during feeding</li> <li>- Postnatal growth impairment</li> <li>- Tearfulness</li> <li>- Disorders in the development of reflexes</li> <li>- Hyperactivity of the HPA axis</li> <li>- Attachment disorders related to the family situation, as well as poor development of the child, e.g. lack of sucking reflex, disturbance of feeling, changed tone of crying, which may affect the behavior of the caregiver</li> <li>- Health problems</li> </ul> | <ul style="list-style-type: none"> <li>- Anger outbursts</li> <li>- Aggression</li> <li>- Attempts to regulate using psychoactive drugs</li> <li>- Socially undesirable behavior</li> <li>- Personality disorder</li> </ul>  |
| <ul style="list-style-type: none"> <li>- Disorders in language development: delay in speech development or other difficulties such as speech defects, poor vocabulary</li> <li>- Disorders associated with sensory development: hypersensitivity to stimuli and/or hypoaesthesia</li> <li>- Hyperactivity</li> <li>- Problems with feeling the distance, e.g. due to frontal damage</li> <li>- Eating disorders</li> <li>- Disorders related to height and weight</li> <li>- Problems with food tolerance</li> <li>- Excessive mobility</li> </ul>   | <ul style="list-style-type: none"> <li>- Lack of pain and involvement in risky situations</li> <li>- Getting into the wrong relationship due to the lack of feeling of distance</li> <li>- Problems at school</li> <li>- Eating</li> <li>- Problems with adapting in a group of peers</li> <li>- Problems with memory</li> <li>- Problems with learning to write and read</li> <li>- Problems with mathematics: understanding of the number, space, time, value of money</li> <li>- Emotional instability</li> </ul> |

|   |   |
|---|---|
| <ul style="list-style-type: none"> <li>- Postnatal growth impairment - underweight, low growth or excessive weight gain</li> <li>- Subcortical nuclei damage</li> <li>- Frontal damage</li> <li>- Problems with understanding the context of social situations</li> <li>- No distance to oneself</li> <li>- Disorders in the development of executive functions such as: planning, decision making, anticipation, organization</li> </ul> | <ul style="list-style-type: none"> <li>- Problems with understanding social situations</li> <li>- Worse learning through experience</li> <li>- Problems with abstract thinking and understanding of concepts</li> <li>- School problems</li> <li>- Problems with independent functioning</li> </ul> |
|---|---|

**Table 3.** Compilation of FASD diagnostic criteria.

| <b>FASD – Fetal Alcohol Spectrum Disorder</b> | <b>Diagnostic criteria</b>   |
|---|--|
| <b>FAS – Fetal Alcohol Syndrome</b>           | <ul style="list-style-type: none"> <li>➤ Minimum two characteristic features within the craniofacial region</li> <li>➤ Growth delay</li> <li>➤ Clear evidence of the impact of the disease on the brain</li> <li>➤ Neurobehavioral disorders</li> <li>➤ Prenatal alcohol exposure (whether documented or not)</li> </ul>   |
| <b>p-FAS – partial Fetal Alcohol Syndrome</b> | <p>In documented cases of prenatal exposure to alcohol:</p> <ul style="list-style-type: none"> <li>➤ Minimum two characteristic features within the craniofacial region</li> <li>➤ Neurobehavioral disorders</li> </ul> <p>In cases of undocumented prenatal exposure to alcohol:</p> <ul style="list-style-type: none"> <li>➤ Minimum two characteristic features within the craniofacial region</li> <li>➤ Growth delay or clear evidence of disease impact on the brain</li> <li>➤ Neurobehavioral disorders</li> </ul> |

|  |  |
|--|--|
| <p><b>ARND</b> – alcohol-related neurodevelopmental disorder</p>                             | <ul style="list-style-type: none"> <li>➤ Documented prenatal exposure to alcohol</li> <li>➤ Neurobehavioral disorders</li> </ul> <p><b>Note:</b> It is not possible to confirm the diagnosis unequivocally in children under 3 years of age.</p>                   |
| <p><b>ARBD</b> – alcohol-related neurobehavioral disorders</p>                               | <ul style="list-style-type: none"> <li>➤ Documented prenatal exposure to alcohol</li> <li>➤ At least one major characteristic malformation associated with prenatal alcohol exposure</li> </ul>  |
| <p><b>ND-PAE</b> – Neurodevelopmental Disorder associated with Prenatal Alcohol Exposure</p> | <ul style="list-style-type: none"> <li>➤ Documented prenatal exposure to alcohol</li> <li>➤ Neurobehavioral disorders in childhood</li> <li>➤ Facial signs of disease, growth retardation and concomitant clear evidence of disease impact on the brain</li> </ul> |

The above clinical features, depending on the severity, are the basis for the separation of diagnostic differences between the Fetal Alcohol Syndrome (FAS), partial Fetal Alcohol Syndrome (pFAS), alcohol-related neurodevelopmental disorder (ARND), alcohol-related neurobehavioral disorders (ARBD), Neurodevelopmental Disorder associated with Prenatal Alcohol Exposure (ND-PAE). (Table 3) [5, 16].

## 5. DIAGNOSTICS

Due to the multifaceted nature of symptoms, the diagnostic process should involve a team consisting of at least a doctor (often a treatment by several doctors is necessary - specialists in various fields of medicine) and a psychologist. During the analysis of each case, the presence and nature of dysmorphism and other losses in the child's health discussed above (Table 1, 2 and 3) are assessed - these aspects are assessed by the doctor.

The psychologist's competence includes assessment of IQ (child's IQ) and assessment of language development, memory, ability to concentrate, cognitive functions, mathematical skills, motor development and a range of other factors, including environmental ones [1, 2, 7, 11-13].

## **5. 1. Therapy and care over children with FAS/FASD**

It should be noted that the sooner the diagnosis is made, the sooner steps can be taken to overcome the disorder resulting from the Fetal Alcohol Spectrum Disorders (FASD). Due to the highly individualized nature of central nervous system abnormalities and developmental deficits, in order to achieve the best therapeutic effects it is necessary to adjust the therapy model individually for each child.

Work on disorders resulting from the central nervous system includes a series of activities aimed at their greatest possible leveling. Based on over 23 years of experience and research, doctor of social sciences Teresa Jadczyk-Szumilo from the University of Social Sciences and Humanities in Katowice has developed a model that includes [23]:

- 1. Work on the regulation of basic brain processes** – the paper includes stimulation exercises to support regulation in areas such as:
  - ⇒ Hyperactivity of the HPA axis – basic process that blocks the development of children with FASD
  - ⇒ Sleep disorders
  - ⇒ Sensory disorder
  - ⇒ Disturbances in sensory-motor processing
  - ⇒ Working on the early-birth trauma
  - ⇒ Emotions regulation, including work on modifications related to changed emotions regulation in connection with the attachment style
- 2. Stimulation of motor patterns**, including reflex patterns
  - ⇒ Motor patterns of reflexes
  - ⇒ Visual habits
  - ⇒ Automation of large and small praxis
  - ⇒ Work on auditory processing - neurodevelopmental basics
- 3. Work on the development of higher psychical functions**
  - ⇒ Language therapy, including language training
  - ⇒ Memory exercises
  - ⇒ Concentration exercises
  - ⇒ Searching for individual, alternative ways to learn to read and write
  - ⇒ Development of mathematical thinking
  - ⇒ Hearing training
  - ⇒ Environmental accommodations

In some cases, pharmacotherapy may be included as complementary therapy, which primarily aims to treat co-existing neuropsychiatric disorders, such as Attention Deficit Hyperactivity Disorder (ADHD), mood swings (including depression), adaptive behavioral inabilities (e.g. aggression) or feelings of anxiety.

## **5. 2. Education and family support in the context of education and upbringing of a child**

As part of a comprehensive therapeutic approach towards a child with the Fetal Alcohol Spectrum Disorder (FASD), both the physician and the psychologist, after prior early diagnosis, should also conduct a parallel educational policy towards the parents and the sick child's family.

Awareness of the caregivers of a person with FASD will enable them to understand the disorders and difficulties resulting from the disease entity, in addition, adjusting expectations and educational models to one that is more appropriate for the child [1, 5, 7, 22].

Another aspect is the support of the family, which in appropriate conditions allows to reduce or eliminate the stigmatization of parents of a child with FASD, which has a direct impact on their better well-being and better functioning. Not without value in this case is also the issue of helping such a family, which includes parents' training and advice of a given health care provider.

### **5. 3. Prognosis**

Children with the Fetal Alcohol Spectrum Disorder (FASD), especially those who have not been diagnosed, and at the same time not receiving proper care and treatment, are at increased risk of developing secondary-disorder symptoms in adulthood. In addition, it should be noted that as a result of these disorders, it is also possible that accidental side effects will be experienced in the family of a child with FASD, especially in parents. Such effects include financial problems (resulting from the costs of running appropriate therapies), stress or secondary effects on the increased amount of alcohol consumed (e.g. liver diseases, suicides) caused by not dealing with problems related to the Fetal Alcohol Spectrum [13].

Taking into account all the above aspects allows to understand the importance of early diagnosis of the disease and introduction of appropriate therapies, thanks to which the above-mentioned disorders can be eliminated, giving the child and his family the chance to function properly.

### **5. 4. Prevention**

As part of prevention, particular attention should be paid to educating the society about the effects of mother's alcohol consumption during pregnancy. In addition, abstinence should be promoted both before and during pregnancy, which can protect against the onset of Fetal Alcohol Spectrum Disorder (FASD) in a future child [12, 13].

## **6. CONCLUSIONS**

Fetal Alcohol Spectrum Disorder (FASD) is an extremely undesirable phenomenon, both medically and socially, nevertheless still common, taking into account the statistics of research conducted around the world regarding pregnant women who consume alcohol. The number of disorders, their character and severity is individual for each child with FASD, depending on the individual brain structures damaged by alcohol. Irregularities appear throughout the child's life and include, among others, anatomical anomalies, behavioral disorders secondary to defects of coordinating brain structures, reduced IQ as well as broadly defined developmental delay (including intellectual, emotional, psychosocial, linguistic) as well as growth delay.

Due to the multilevel nature of the problem: health (for a child) and social (effects of the influence of certain disorders in the environment of a person affected by FASD), it is extremely important to conduct a broad social education, which would reduce the incidence of alcohol consumption by pregnant women. In the case of children with the Fetal Alcohol Spectrum Disorder, the most important role is played by making an early diagnosis, which in turn will

enable the implementation of appropriate therapeutic programs individually for each child, thanks to which s/he will have the chance to level the disorder to a degree that does not make him stand out from his peers.

At the same time, it should be remembered that adequate education of the sick child's family is of considerable importance - properly trained family members are better able to understand the FASD issues, which has a direct impact on creating better environmental conditions for the child. This makes it possible for both the sick person and their family to function properly (to the extent in which the disease entity allows it).

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