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Severe manifestation of foetal alcohol syndrome – case report

Ciężka manifestacja alkoholowego zespołu płodowego – opis przypadku

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Abstract

Foetal alcohol syndrome is a term describing the set of conditions caused by maternal alcohol consumption in pregnancy. A complete evaluation includes an assessment of the three key diagnostic criteria – central nervous system damage, growth deficiency, and characteristic facial features (microcephaly, small palpebral fissures, low nasal bridge, smooth philtrum, thin upper lip). A premature one-day-old female infant with dysmorphic features, in severe general condition, was admitted to the Neonatal Intensive Care Unit due to respiratory distress and suspected heart defect. Hypotrophy, bronchopulmonary dysplasia, and both ventricular and atrial septal defect were diagnosed. The authors of this paper aim to raise awareness about the lifelong and irreversible nature of foetal alcohol syndrome, which significantly impacts various aspects of a child's life. However, it is also highlighted that foetal alcohol syndrome belongs to a small group of conditions that can be entirely prevented.

Keywords: neonatology, foetal alcohol spectrum disorders, pregnancy

Streszczenie

„Alkoholowy zespół płodowy" to termin określający zespół objawów spowodowanych spożywaniem alkoholu przez kobietę w ciąży. Kompletna ewaluacja zespołu obejmuje ocenę trzech głównych aspektów: uszkodzenia ośrodkowego układu nerwowego, zaburzeń wzrastania i obecności charakterystycznych cech dysmorfii (mikrocefalii, krótkiej szpary powiekowej, krótkiego i zadartego nosa, wygładzonej rynienki nosowej, wąskiej górnej wargi). Przedwcześnie urodzony noworodek z cechami dysmorfii został przyjęty do Oddziału Intensywnej Terapii Noworodka w pierwszym dniu życia w stanie ogólnym ciężkim, z niewydolnością układu oddechowego i podejrzeniem wrodzonej wady serca. Zdiagnozowano hipotrofię, dysplazję oskrzelowo-płucną oraz defekt przegrody międzykomorowej i międzyprzedsionkowej. Artykuł ma na celu podniesienie świadomości czytelników na temat nieodwracalnego alkoholowego zespołu płodowego, który znacząco wpływa na różne aspekty życia dziecka, a zarazem jest jednym z niewielu schorzeń, którym można całkowicie zapobiec. Należy powstrzymać się od spożywania alkoholu w czasie ciąży i w trakcie prób poczęcia.

Słowa kluczowe: neonatologia, alkoholowy zespół płodowy, ciąża

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INTRODUCTION

Foetal alcohol spectrum disorders (FASDs) are conditions that are a common cause of a congenital developmental disability acquired in the uterus as a result of the mother's alcohol use during pregnancy[1]. FASDs consist of five conditions including foetal alcohol syndrome (FAS), partial foetal alcohol syndrome (pFAS), alcohol-related neurodevelopmental disorder (ARND), neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE), and alcohol-related birth defects (ARBD). The goal of this classification is to categorise all of the wide-ranging physical and neurological effects that prenatal alcohol exposure can have on the foetus[2,3].

The main clinical manifestations of dysmorphic features and congenital abnormalities caused by FAS in infants are microcephaly, small palpebral fissures, low nasal bridge, smooth philtrum, thin upper lip, deformations of limbs, joints and fingers, and heart and kidney defects. In addition, the children's height or weight typically falls at or below the 10th percentile. Neurobehavioral changes include impaired fine motor skills, such as hyperactivity, limb tremors, breastfeeding problems, and disturbed sensory integration[4]. FAS is incurable, but treatment is possible. Early intervention from birth to the age of three is a crucial factor in the development of children suffering from FAS. Currently, behavioural therapies and psychoactive medications are being tested. Even though the condition is incurable, with the aid of supportive therapeutic interventions and management of psychiatric comorbidities it is possible to enhance FAS sufferers' independence and quality of life. In some cases, however, the disorders caused by alcohol consumed during pregnancy are life-threatening for the foetus[5].

The goals of this clinical case report are to remind every reader of the fact that maternal alcohol consumption during pregnancy has a dramatic effect on many developing foetal organs that are important for the survival of the child after delivery, and to highlight how easily FAS can be avoided. By discussing the therapeutic challenges, the authors aim to raise awareness regarding the daily struggles faced by children affected by a condition that could have been entirely prevented by abstaining from alcohol during pregnancy and when attempting to conceive. It is important to note that even a single unit of alcohol can be detrimental.

CASE PRESENTATION

A premature one-day-old female newborn with dysmorphic features was admitted to the Neonatal Intensive Care Unit due to respiratory distress and suspicion of a heart defect.

The girl was the second child of a mother with a history of alcoholism and nicotine addiction. Her first child has FASD syndrome and is currently under the custody of the grandmother. Our patient was born by spontaneous vaginal delivery at 34 weeks of gestational age, weighing 1,100 g (0 percentile), and measuring 37 cm (0 percentile), with a head circumference of 26.5 cm (0 percentile). The patient was in severe general condition with the Apgar scores of 3–5–7–8. Prenatal cardiomegaly, ventricular septal defect (VSD), and hypotrophy were diagnosed. Therefore, amniocentesis was performed – the results of array comparative genomic hybridisation was within normal limits.

After birth, the newborn needed resuscitation due to respiratory distress. Afterwards, she stayed on respiratory support. Physical examination revealed features of prematurity, hypotrophy, and distinct characteristics of dysmorphia in the form of hypertelorism, low hairline on the temples, triangular nose, smooth philtrum, narrow upper lip, gothic palate, low set auricles, transverse palmar crease, overlapping toes of the left foot, and sacral dimple. Two-vessel umbilical cord (with a single umbilical artery) was also detected in our patient.

Respiratory problems

Exposure to ethanol *in utero* reduces the surfactant content in foetal pulmonary alveoli. The toxic effect of ethanol on type I and II pneumocytes has been well documented, resulting in foetal/neonatal lung hypoplasia[5].

In the first months of life, due to respiratory distress syndrome, our newborn patient required respiratory support as well as one dose of endotracheal surfactant. Chest X-ray revealed a congenital defect and an atypical position of the diaphragm. Consequently, on the 45th day of life, a laparotomy was performed, during which diaphragm with maintained continuity was found on the right side, closely fused with the liver capsule. There was improvement in both clinical condition and in lung ventilation. However, the child still required mechanical ventilation. Bronchopulmonary dysplasia (BPD), also known as chronic lung disease, was diagnosed, and inhalations with Pulmicort and Berodual (budesonide, ipratropium bromide, fenoterol hydrobromide) were used. Due to ventilator-associated pneumonia (VAP), antibiotic therapy was repeatedly administered. In this context, it is also important to have in mind that patients with FAS have a strong need for respiratory syncytial virus (RSV) prevention because of an elevated risk of heart defects and BPD.

Neurological problems

During the first week of the child's life, increased upper limb tremors were observed along with meaningful anxiety and periodical pain (due to alcohol withdrawal syndrome). Phenobarbital and paracetamol were then added to the treatment, and improvement was achieved. On physical examination, the patient's poor spontaneous motility and neonatal reflexes, increased muscle tone, tendency for deflexion/bend-back position, and abnormal movement pattern stood out. All of the above-mentioned symptoms are linked to alcohol intoxication *in utero*. Fontanelle ultrasound revealed minor signs of prematurity, with two

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small hyperechogenic areas around the lateral ventricles. Magnetic resonance imaging (MRI) showed small areas of bleeding in the white matter of the right and left hemispheres of the brain as well as features of central nervous system damage caused by maternal alcohol consumption, including small posterior fossa of the skull with reduced volume of the cerebellum and brainstem. Another distinctive FAS feature is the presence of a bone block of the C5/C6 diaphysis (Klippel–Feil syndrome). MRI of the spine revealed numerous spinal and skeletal defects including cervical ribs, lack of coccyx and S5 formation, and sacral lipoma. Flattened spinal cone was located at the level of the L3 shaft. Defects of the spine and spinal cord proved to be characteristic of FAS.

Cardiovascular problems

The patient has a congenital defect of the cardiovascular system – VSD and atrial septal defect (ASD), causing increased pulmonary flow. Due to the advancement of pulmonary hypertension, banding of the pulmonary artery on the 83rd day of life was performed. At the end of the first year of life, the girl required heart defect correction with the use of extracorporeal circulation. Both ASD and VSD are the recognised as the most common heart defects diagnosed in infants with FAS.

Infectious problems

After birth, the child was diagnosed with intrauterine infection and treated with empirical antibiotic therapy. During further hospitalisation, our patient suffered from recurrent ventilator-associated pneumonia and bacterial conjunctivitis (*Staphylococcus haemolyticus*). Alcohol is known to worsen the foetal immune response (impairing the function of T cells, B cells, and macrophages), increasing the risk of death due to pneumonia or sepsis. The risk of infection in low birth weight infants born to mothers who occasionally consume ethanol increases 2.5 times. Excessive alcohol consumption during pregnancy elevates the risk by 3–4 times[5].

Haematological problems

Due to anaemia, which is common in pre-term newborns, several infusions of irradiated leukocyte-depleted red blood cell concentrate were transfused.

Nutritional problems

Our patient was born with signs of severe hypotrophy (0 percentile), which is characteristic of FAS. The baby never had a sucking reflex. Consequently, total parenteral nutrition with a feeding tube was used during the first days of life. In the fourth month of life, gastrostomy was performed. Intragastric feeding was implemented into percutaneous endoscopic gastrostomy (PEG). Both nasogastric tube and gastrostomy further increased the girl's risk of infections. Despite body weight gain, in the sixth month of life she weighed only 4,100 g (0 percentile).

Nephrological problems

Imagining tests revealed S-shaped kidney and crossed renal ectopia on the patient's right side. Diuresis was maintained within normal limits only with the use of diuretics. Lack of orifice of urethra in its typical position was discovered on physical examination. Kidney defects can also be associated with alcohol intoxication *in utero*.

Ophthalmic problems

MRI scan revealed optic nerve atrophy, significant myelination delay, and several narrow structures including optic chiasma and corpus cerebellum. During the patient's physical examination in the 9th month of life, target fixation, following with one eye, reaction to sounds (clapping, rattles) were not observed, and pupillary light reflex was questionable. All the above-mentioned complications are linked to alcohol intoxication *in utero*.

Laryngological problems

On physical examination, the only visible defect was the patient's cleft hard palate on the right side, with indentation on the alveolar process in the oral cavity and incomplete cleft palate. Ear, nose and throat specialist (ENT) consult also confirmed obstruction of the posterior nasal apertures. Due to the constant need for invasive ventilation, it was not possible to conduct an audiological examination to assess hearing. It is important to emphasise that cleft palate is a characteristic feature of FAS.

Genetic problems

Genetic consultation confirmed that because of the history of maternal alcohol consumption during pregnancy and the presence of characteristics associated with FAS there were no indications for extended genetic testing.

DISCUSSION

The four areas of clinical features that constitute the diagnosis of FAS include selected facial malformations, growth retardation, central nervous system abnormalities, and maternal alcohol consumption during pregnancy. Our patient presented almost all the symptoms attributable to intrauterine alcohol intoxication that can be diagnosed in infants. Alcohol consumption by pregnant women has significant consequences for the course of pregnancy and can cause preterm birth. In the case of premature infants intoxicated with ethanol in the uterus, the risk of perinatal death increases significantly. In this group of children, there is an over 15-fold increase in the risk of sepsis[5].

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Foetal alcohol exposure adversely effects the lungs. Exposure to ethanol *in utero* reduces the surfactant content in foetal pulmonary alveoli and can cause neonatal lung hypoplasia[5]. Despite being born at 34 weeks of gestation, our patient developed respiratory distress syndrome (RDS). During the first months of her life, she was alternately mechanically ventilated under pharmacological sedation or breathing with the help of non-invasive support. Furthermore, the patient suffered from repeated episodes of pneumonia, which resulted in multiple antibiotic treatments. Overall, the girl received antibiotic therapy with amoxicillin–clavulanic acid, ampicillin, gentamicin, and piperacillin–tazobactam for 30 days.

Our patient is now over a year old and has spent all her life in various medical wards. She has undergone a range of procedures, including pulmonary artery banding (PAB) as a consequence of ASD and VSD, plication of the diaphragm, tracheostomy, debanding and VSD closure, and PEG tube insertion. Diseases from which she suffers include exacerbation of chronic respiratory failure and circulatory insufficiency. To date, the patient continues to present severe neurological disorders that typically occur in severe cases of FAS.

The first child of our patient's mother also has FASD syndrome. The diagnosis of FASD in an elder sibling was a reinforcing factor in the diagnostic work-up of our patient, because in about 70% of such cases, the next child is also born with FASD[6]. FAS is the most severe form and accounts for 10% of FASD cases. Not all of the disorders that are present in FAS occur in FASD. However, major or minor brain damage is required to make a FASD diagnosis. Crucially, estimates indicate that 7% of children with FASD have never received a diagnosis and another 7% have been misdiagnosed[7].

CONCLUSIONS

The presented case shows how severely FAS can affect a child's life, causing a constant struggle with lifelong disabilities. The goals of this clinical case report are to remind every reader of the fact that maternal alcohol consumption during pregnancy has a dramatic effect on many developing foetal organs that are important for the survival of the child after delivery, and to highlight how easily FAS may be avoided. By discussing the therapeutic challenges, the authors hope to raise awareness regarding the daily struggles faced by children affected by a condition that can be entirely prevented by abstaining from alcohol during pregnancy and when trying to conceive. It is important to note that any amount of alcohol consumed by a pregnant mother carries the potential risk of brain damage to the foetus. It is crucial for our society, particularly healthcare professionals including doctors, nurses, and midwives, to acknowledge that no amount of alcohol has been proven safe at any time during pregnancy. There is a high level of probability that every single defect or abnormality that the child reported here is burdened with is a consequence of prenatal exposure to alcohol. FAS disturbs the lives of millions of individuals all around the world and is related to a broad array of physiological, psychiatric, and neurological consequences that can significantly impair behavioural, emotional, developmental, and social functioning.

Conflict of interest

The authors do not report any financial or personal affiliations to persons or organisations that could adversely affect the content of or claim to have rights to this publication.

Author contributions

Writing of manuscript: AM, LD. Critical review of manuscript: EGI. Final approval of manuscript: BKN.

References

1. Wagner JC, Tergeist M, Kruse B et al.: [Fetal alcohol spectrum disorders in adults] [Article in German]. Nervenarzt 2020; 91: 1069–1079.
2. Bukiya AN: Fetal cerebral artery mitochondrion as target of prenatal alcohol exposure. Int J Environ Res Public Health 2019; 16: 1586.
3. Adebiyi BO, Mukumbang FC, Beytell AM: To what extent is Fetal Alcohol Spectrum Disorder considered in policy-related documents in South Africa? A document review. Health Res Policy Syst 2019; 17: 46.
4. Del Campo M, Jones KL: A review of the physical features of the fetal alcohol spectrum disorders. Eur J Med Genet 2017; 60: 55–64.
5. Dębski R, Paszkowski T, Wielgoś M et al.: Stanowisko Grupy Ekspertów na temat wpływu alkoholu na ciążę: stan wiedzy na 2014 rok. Gin Pol Med Project 2014; 2 (32): 66–78.
6. Banach M, Matejek J: „W trosce o zdrowie dziecka i twoje". Płodowy Zespół Alkoholowy (FAS) – kompendium wiedzy. Scriptum, Kraków 2016.
7. Ergun G, Schultz MS, Rettig EK: Fetal alcohol spectrum disorder – issues of misdiagnosis and missed diagnosis in Black youth: a case report. Innov Clin Neurosci 2021; 18: 20–23.

DOI: 10.15557/PiMR.2023.0041