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CASE REPORT

VACTERL ASSOCIATION: CASE REPORT

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Abstract. Background: *VACTERL is a rare association of congenital abnormalities with incidence 1/10 000 to 1/40 000 live births. VACTERL association is typically defined by the presence of at least three of the following congenital malformations: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities. We are presenting a case report of natural delivery of live born full-term fetus with VACTERL association. Our aim is to emphasize the role of prenatal diagnostics of fetal malformations in order to avoid deliveries of polymalformative fetuses with poor prognosis and mental trauma of the parents.* ***Methods:*** *A 18-year-old primiparous woman in 38th gestational week was admitted in the department because of spontaneous rupture of amniotic membranes and irregular uterine activity. History of the patient indicated insufficient monitoring during the pregnancy. Ultrasound examination showed hypotrophic and polymalformative fetus. The amount of amniotic fluid was increased despite of definite rupture of amniotic sac. Presence of only two umbilical vessels was established.* ***Results:*** *Regardless of the manifestation of fetal distress during the delivery, medical council decided that cesarean section was unjustified because of unsecure prognosis of the newborn and the labor should be completed vaginally. Hypotrophic male with multiple congenital malformations was born that required surgical intervention in the postpartum period. Lethal outcome occurred due to multiple organ failure.* ***Conclusions:*** *The prognosis of VACTERL is determined by the number, severity, and nature of abnormalities present. The ultrasound can identify not only the defects but also specify the prognosis of the fetus.*

Key words: *VACTERL association, delivery, ultrasound, prenatal diagnosis*

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INTRODUCTION

VACTERL represents an acronym for broad spectrum of congenital disorders, which can be found prenatally, perinatally or later in life. Diagnostic criteria include at least three of the following features: vertebral defects, commonly accompanied by rib anomalies; anal atresia; cardiac defects such as ventricular septal defects, atrial defects and tetralogy of Fallot; tracheo-esophageal fistula, with or without esophageal atresia; renal anomalies including renal agenesis, horseshoe kidney, and cystic and/or dysplastic kidneys; and limb abnormalities classically defined as radial anomalies that can occur in unilateral or bilateral form [1, 2, 3]. The annual incidence has been reported to be 1/10 000 to 1/40 000 live births without specific geographic distribution [4]. The majority of cases are sporadic but some risk

factors have been described. Pregestational maternal diabetes, chronic obstructive pulmonary disease, pregnancy conceived after assisted reproductive techniques, in utero exposures to estrogen/progesterone, statins, doxorubicin have been implicated as environmental risk factors for this association. Different mutations and chromosomal anomalies are also included in the etiology. Primiparity seemed to be associated with VACTERL as well as with isolated VACTERL component features [4]. Clinical presentation is diverse, ranging from mild cases undiagnosed until adulthood to severe cases that are obvious in the prenatal or neonatal period [5, 6]. Defects in practically every organ have been described in patients with VACTERL and are considered additional criteria for this association: facial asymmetry, external ear malformation, lung lobation defects. Prenatal diagnosis is challenging considering many key features that can be suspected using ultrasound as the most specific diagnostic tool. The prognosis of VACTERL is determined by the number, severity, and nature of abnormalities present. Multiple surgeries are often required during childhood and various physical sequelae may follow, such as scoliosis, bowel dysfunction, gastro-esophageal reflux, dysphagia, airway morbidity, decreased cardiac, renal or limb function [4, 5]. Termination of pregnancy by medical reasons can be done in cases with prenatally diagnosed VACTERL association in order to avoid deliveries of polymalformative fetuses with poor postnatal prognosis and mental trauma of the parents.

CASE REPORT

A 18-year-old woman presented with spontaneous rupture of amniotic membranes and irregular uterine contractions 18 days before the estimated due date. The patient denied previous pregnancies, any accompanying diseases, usage of medications and complications during this pregnancy. Her history showed irregular monitoring of pregnancy and the fact that combined screening for chromosomal abnormalities and fetal morphology examination were not performed. Her physical examination did not show deviations from the normal ranges. A speculum examination showed leakage of yellowish amniotic fluid. Obstetrical examination established good pelvic score and cephalic presentation of the fetus. Abdominal ultrasound was performed and found live fetus in cephalic presentation with estimated fetal weight 1900 g, corresponding to 32 + 3 gestational weeks (g.w.) at term of amenorrhea 37 + 5 g.w., which made the diagnosis of fetal growth restriction.

Multiple anatomical defects were revealed during the ultrasound imaging such as chizencephaly (Fig. 1).

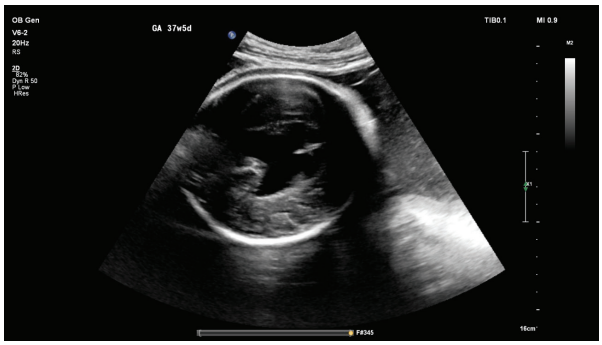

Fig. 1. Open-lip schizencephaly

Non-visible four-chamber view of the heart, hypoplastic left ventricle, atrial septal defect and ventricular septal defect (VSD) were revealed during the ultrasound imaging (Fig. 2).

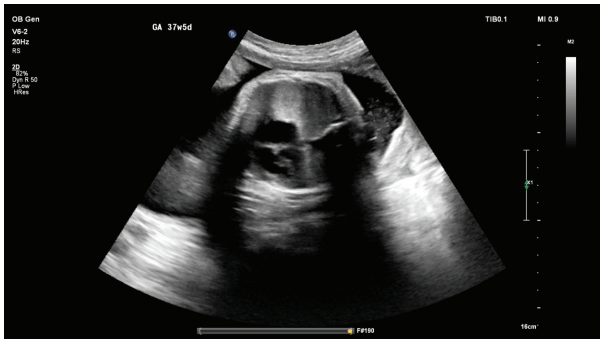

Fig. 2. AVSD

Presence of double-bubble sign, giving suspicious of duodenal atresia was also found during the ultrasound examination (Fig. 3).

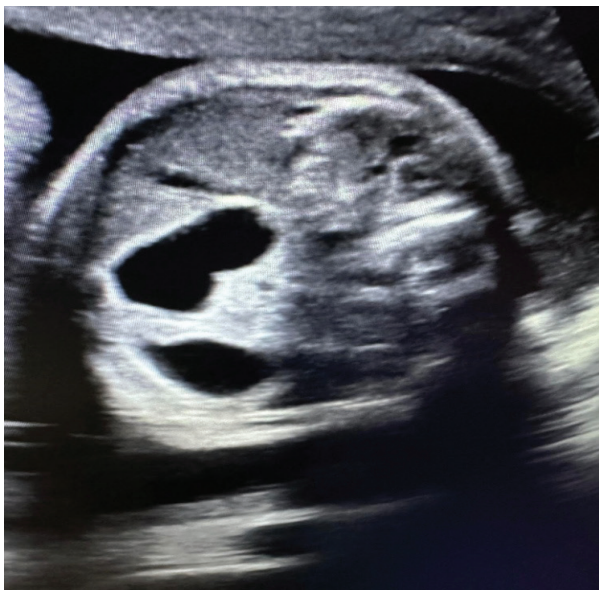

Fig. 3. Double-bubble sign seen in duodenal atresia

Skeletal disorders of upper limbs, including bilateral absence of radius and hand deformities were present (Fig. 4).

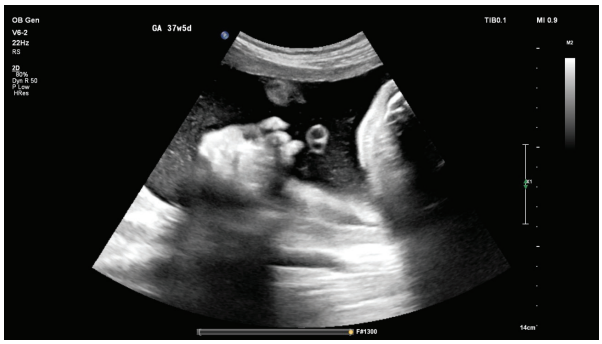

Fig. 4. Limb anomaly due to radial aplasia

The maximum vertical diameter of amniotic fluid was 9,9 cm, which demonstrated polyhydramnios despite of definite rupture of the amniotic sac (Fig. 5).

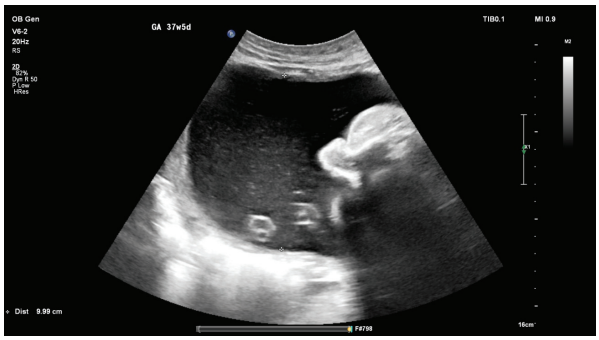

Fig. 5. Polyhydramnios

Placenta was localized on the posterior uterine wall with second degree of maturity. The presence of only two umbilical vessels was detected (Fig. 6).

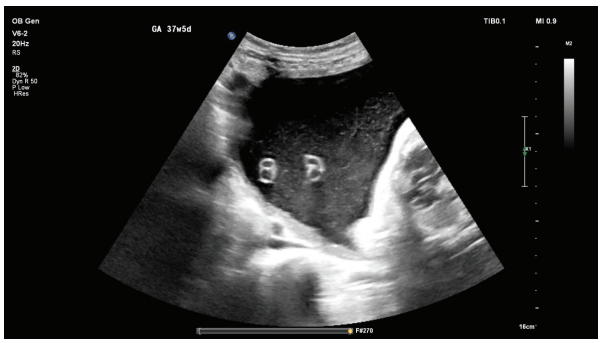

Fig. 6. Single umbilical artery of the umbilical cord

Doppler velocimetry showed increased resistance of single umbilical artery. The patient was informed about the polymalformative status of the fetus and uncertain prognosis of the newborn after delivery.

Monitoring data for fetal distress appeared in the late active phase of the first stage of labor. Regardless of the manifestation of fetal distress during the delivery, medical council decided that cesarean section was unjustified because of unsecure prognosis of the newborn and the labor should be completed vaginally in presence of neonatologist. A live male hypotrophic fetus was born weighing 1890 g with dull heart sounds and lack of spontaneous breathing. The newborn was intubated and placed on mechanical ventilation because of severe respiratory distress. Multiple malformations were detected such as malformative upper limbs, micrognathia, facial dysmorphism, low set ears, sandal gap. Unsuccessful attempt for esophageal probe was made. An X-ray revealed the absence of the phalanges of the fifth fingers, one bone of the forearm (ulna), missing body of the last thoracic vertebra, two air bubbles in the place of the stomach and duodenum, no gas was visualized in the lower parts of the gastrointestinal tract. A contrast study of the gastrointestinal tract demonstrated esophageal atresia, tracheoesophageal fistula and duodenal atresia. Echocardiography showed hypoplastic left ventricle, mitral atresia, ventricular septal defect, atrial septal defect, pulmonary hypertension. Consultation with geneticist with suspicious of VACTERL-association was made. The chromosomal study revealed normal karyotype, 46XY. Pediatric surgeon consulted the newborn who was referred for surgical correction of tracheoesophageal fistula. The parents were notified of the newborn's diagnosis and they denied the parental rights. Neonatal sepsis and multiple organ failure developed in the postoperative period. Despite intensive neonatal care and full-scale resuscitation, the child died 48 days after birth.

DISCUSSION

Defects of every organ have been reported in association with VACTERL [7, 8]. A single umbilical artery is a common ultrasound finding in fetuses with VACTERL, associated with intrauterine growth restriction [9]. The important ultrasonographic features include three or more typical defects of this disorder. Based on the ultrasound findings in our case VACTERL-association was the most likely diagnosis due to the combination of cardiac defects, tracheoesophageal fistula, esophageal and duodenal atresia, limb abnormalities, especially bilateral radial aplasia. Additional features like vertebral defects, facial dysmorphism were found postnatally. This clinical presentation demonstrates the importance and limitation of prenatal ultrasound diagnosis. Polyhydramnios, which is commonly seen in VACTERL-association, due to esophageal atresia improves the ultrasound image and makes detection of these abnormalities easier. The ultrasound can identify not only the defects but also specify the

prognosis of the fetus [10]. Therefore, the correct and regular monitoring of the pregnancy is mandatory in order to identify malformative fetuses and provide the opportunity for the parents to make choice regarding outcome of the pregnancy. The poor prognosis and lethal outcome in the presented case was as a result of combined severe cardiac defects, development of worsening respiratory failure, sepsis and multiple organ failure in the neonatal period.

CONCLUSION

Although prenatal diagnosis of VACTERL association is challenging, its suspicion is extremely important due to the generally poor prognosis of this association. When diagnosis before viability is possible, management options can be discussed with the couple, including medical termination of pregnancy. Even when the decision to continue the pregnancy is taken, prenatal diagnosis is very important to improve outcome of the newborn.

Conflict of Interest Statement: *The authors declare no conflicts of interest related to this work.*

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Informed Consent for a Clinical Case: *Written informed consent was obtained from the patient/parent for the publication of this case report, including any accompanying images.*

Ethical statement: *This study has been performed in accordance with the ethical standards as laid down in the Declaration of Helsinki.*

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