PRENATAL SONOGRAPHIC DIAGNOSIS OF PIERRE-ROBIN SEQUENCE - A CASE REPORT

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Abstract

The rare anomalies diagnosed in prenatal ultrasonographic examination include Pierre-Robin sequence. The aim of the report was to demonstrate a well-documented case of Pierre-Robin sequence detected by ultrasonography at the 24th week of gestation, present the differentiation of Pierre–Robin sequence in the prenatal ultrasonography and to emphasize the importance of early sonographic diagnosis of the anomaly.

The prenatal diagnosis of Pierre-Robin sequence in prenatal ultrasonographic assessment is of great importance as it permits planning further course of pregnancy as well as EXIT procedure and postnatal treatment.

Key words: *fetus, micrognathia, retrognathia, Pierre-Robin sequence, ultrasonography*

INTRODUCTION

Fetal ultrasonography allows the evaluation of the fetal face from the first weeks of gestation, however craniofacial anomalies and difficulties in the assessment of two- and three-dimensional (2D and 3D) images result in their scarce detection[1]. The aim of the report was to demonstrate a well-documented case of Pierre-Robin sequence detected by ultrasonography at 24 weeks of gestation, present the differentiation of Pierre–Robin sequence in prenatal ultrasonography and to emphasize the importance of early sonographic diagnosis of this anomaly.

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CLINICAL REPORT

25-year-old prima gravida, pregnant with unrelated partner. Family, medical and gynecological histories were negative. Results of biochemical tests were normal. Ultrasound examination performed at 14 weeks of gestation by primary care obstetrician showed normal fetal development. During the next ultrasound examination carried out at the 24th week of gestation, the same obstetrician registered abnormalities of the fetal head suspecting hydrocephalus. The gravida was referred to our tertiary center for further diagnostic assessment. The ultrasound examination performed during the same week of gestation revealed a round outline of the head of the single male fetus. An improper image of the prosencephalon was present. Within the fetal head, only temporal lobes, regular thalami and cerebellum were visualized. The cisterna magna was widened up to 13 mm.

The facial skeleton outline was abnormal, micrognathia, retrognathia and symmetrical eyeballs with transparent

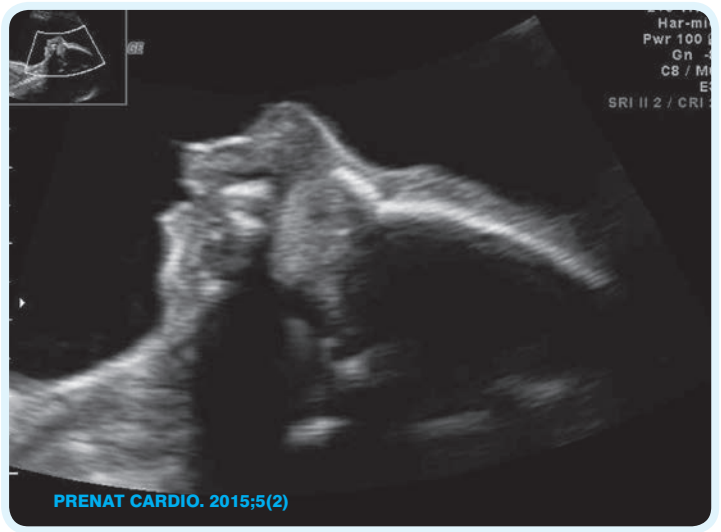

Figure 1. Fetus with sonographic features of Pierre-Robin sequence, 2D ultrasound examination of the fetal head, sagittal plane, 24 weeks of gestation.

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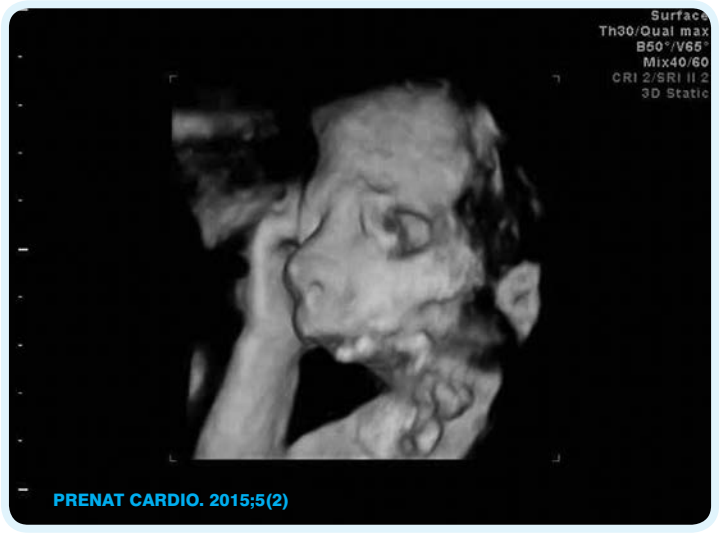

Figure 2. Fetus with sonographic features of Pierre-Robin sequence, 3D ultrasound examination of the fetal head, a left side view, 24 weeks of gestation.

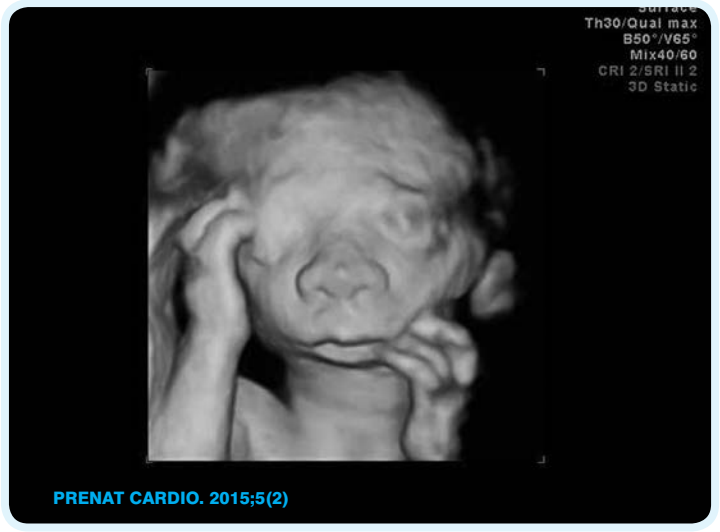

Figure 3. Fetus with sonographic features of Pierre-Robin sequence, 3D ultrasound examination of the fetal head, an en-face view, 24 weeks of gestation.

lenses, Outside Ocular Diameter of 37.5 mm, which corresponded to the 23rd week of gestation. The nasal bone was present and lip continuity was maintained. A long labial groove and small low-set auricles were observed.

Spinal column was symmetrical with maintained continuity. The outline of upper and lower limb long bones, as well as of hands and feet, was normal. The structure of the thoracic cavity was described as proper. Regular outlines of abdominal organs were stated. The umbilical cord was trivascular. The echocardiographic examination revealed no anomalies. The results of the examination were as follows: a single male fetus, at 24 weeks of gestation according to date of the last menstrual period and 24 weeks according to biometry, with dysmorphia of the craniofacial skeleton with mandibular retrognathia, micrognathia and alobar holoprosencephaly and hypoplasia of the external ears, with no other apparent abnormalities, Pierre-Robin sequence.

The mother made the decision to terminate pregnancy without amniocentesis.

The pregnancy was terminated at 24 weeks in the regional hospital. The findings of the prenatal sonographic diagnosis were confirmed upon pathological examination of the fetus and genetic examination performed after termination of pregnancy. Two years later the woman gave birth to a healthy male infant.

DISCUSSION

Pierre-Robin sequence (PRS) may be an isolated combination of defects or part of other congenital anomalies and syndromes, which include mandibular micrognathia, retrognathia as well as retroglossoptosis and hypoplasia of the external ear[2].

It is difficult to precisely analyse the fetal head in the ultrasonographic examination prior to the 23rd-24th week of gestation. The predominance of three-dimensional (3D) over two-dimensional (2D) ultrasonography is being discussed nowadays. However, 3D ultrasonography is of unquestionable importance in visualising the facial appearance and presenting the possible problem connected with facial defect to the parents and physicians. Moreover, it allows the diagnostician to more precisely evaluate some structures of the craniofacial skeleton, lips and external ear defects or the position of the fetal mandible allowing prenatal diagnosis of the anomaly or sequence of anomalies[3].

Micrognathia has been reported in over 300 syndromes and in association with retrognathia has been described in about 50 syndromes of congenital defects[4]. Pierre-Robin sequence is a non-chromosomal syndrome and should be differentiated from other non-chromosomal anomalies and more rarely from trisomies 18 and 13, in which micrognathia occurs in approximately 70%[5].

Systemic anomalies as well as local disorders including craniofacial anomalies in the fetus seriously worsen prognosis and may favor decision to terminate the pregnancy upon mother's request, which may be carried out before the end of the 24th week of gestation according to Polish law[6]. If the mother decides against termination of pregnancy or the diagnosis of Pierre Robin sequence by prenatal examination is made after the 24th week of gestation, sonographic examination allows planning of further treatment in both perinatal and postnatal periods.

Fetal ultrasonography is an important imaging modality in the assessment of fetuses with potential airway obstruction as planning and simulation for perinatal treatment is essential prior to delivery. In these cases EXIT (ex-utero intrapartum treatment) procedure in the perinatal period, which involve perinatal anesthetic and otolaryngological management, should be deliberated[7, 8]. EXIT procedure

Syndrome, anomaly, sequence	Characteristic features in the ultrasonographic examination
Pierre-Robin sequence	Micrognathia, retrognathia, macroglossia, external ear defects
Trisomy 13	Micrognathia, the central nervous system defects (holoprosencephaly), numerous anomalies of the craniofacial skeleton (anophthtalmia/micophthalmia, hypotelorism, proboscis, nasal defects, clefts), oligohydramnios or proper amount of amniotic fluid, polydactyly, the urinary system defects, heart defects
Trisomy 18	Micrognathia, the central nervous system defects (not holoprosencephaly), heart defects, osseous and skeletal defects
FADS (Fetal Akinesia Deformation Sequence)	Micrognathia, joint contractures, chest hypoplasia, talipes equinovarus, ulnar position of the hands
Multiple Pterygium Syndrome	Micrognathia, joint contractures, protuberances on knees and elbows
Neu-Lexova syndrome	Micrognathia, hypoplasia of corpus callosum and cerebellum, hypotelorism, short limbs, joint contractures, small fetus, hydramnios
Nager syndrome	Micrognathia, lack of fingers and toes (ecrodactyly), mesomelic dysplasia, exernal ear defects
Treacher-Collins syndrome	Micrognathia, external ear defects
Apert syndrome	Craniosynostosis, numerous craniofacial defects, symmetrical finger and toe syndactyly
Achondrogenesis	Micrognathia, micromelia, cranial vault hypermineralisation, generalised fetal edema
Campomelic dysplasia	Micrognathia, bowed and short femoral and tibial bones
Diastrophic dysplasia	Micrognathia, micromelia, hitch-hiker thumb, kyphoscoliosis
Femoral hypoplasia with numerous craniofacial defects	Micrognathia, limited hypoplasia of femoral bones

Table 1. The most common differentiation of Pierre–Robin sequence in the prenatal ultrasonographic examination (32 syndromes with micrognathia are listed by Benacerraf[11]).

should be also considered in cases of severe micrognathia and rethrognathia affecting function of upper respiratory system diagnosed upon prenatal sonographic examination, which are specific features of Pierre-Robin sequence [9]. In the postnatal period few surgical interventions have been proposed to manage airway obstruction in children with Pierre Robin sequence, which can be categorized into tongue repositioning procedures including subperiosteal release of the floor of the mouth musculature (SRFM), mandibular distraction procedures as well as bypass procedures including tracheostomy[10].

Early prenatal diagnosis of Pierre-Robin sequence upon prenatal ultrasonographic assessment allows for the counseling of parents as well as specialists involved in both perinatal and postnatal treatment before delivery.

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