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CASES OF PRENATAL DIAGNOSIS OF REVERSE ARTERIAL PERFUSION SYNDROME IN YAKUTIA

ABSTRACT

There are six cases of Twin Reversed Arterial Perfusion Syndrome (or Acardia) in ultrasound prenatal diagnostics in the period from 2009 – July, 2013; one of the cases is presented in the triplet pregnancy.

Keywords: pregnancy, ultrasound prenatal diagnostics, complications of monochorionic twins, Twin Reversed Arterial Perfusion (TRAP) Syndrome, Acardia.

INTRODUCTION

TRAP syndrome, or Acardia syndrome –a rare complication of monochorionic twins (frequency is 1:35000) characterized with the arterioarterial and arteriovenous anastomoses of twins cord vessels [3, 4].

A fetus with Acardia syndrome (recipient) is nonviable, and a single vascular supply source is the circulatory system of a donor twin.Term "reversed arterial perfusion" is used to characterize the bloodstream toarecipient twin: oxygenated bloodcirculates not with the umbilical vein, as it should be, but the umbilical artery, and deoxygenated blood circulates to placenta with the umbilical vein [1, 5].

With the increasing of the gestational age of the recipient twin the need of oxygenated blood is gaining too, and this factor leads to considerable increase of a heart stress of the donor twin, resulting in heart failure [2].

Accordingly, without prenatal adjustment conductionthis is largely responsible for the loss of donor fetus in 50-75% cases, especially if a tentative mass of the recipient twin is about more than 50% from a mass of the donor twin [6].

Six various cases of Acardia syndrome from November, 2009 to July, 2013, one of which was observed at triplet pregnancy, are presented.

Pregnant women were referred on a complex prenatal research from central district hospitals and health centers of Yakutsk to the genetic consultation of the National Hospital No. 1. Ultrasonography analyses were conducted on the ultrasonic device Voluson E8, Toshiba-Xario in the genetic consultation of NH No. 1. There also prenatal karyotyping was carried out.

Considering that departures of the families in the Federal Centers of Prenatal Diagnostics for laser coagulation of an umbilical artery of a fetus recipient were complicated during the described period, tactics of active non-interference was chosen.

In two described cases pregnancy came to the end with the birth of children. In one of them – neonatal loss in the 10th days of life, and in other case – the birth of the child with a cerebral palsy.

High frequency of occurrence of this pathology in Yakutia is interesting, considering the fact, that the period from 2009 to 2012 the number of birth was 64833 (2009 – 15848, 2010 – 15868, 2011 – 16195, 2012 – 16922).

CLINICAL SURVEILLANCE

Case No. 1

Pregnant woman, 35 years, the inhabitant of the suburb of Yakutsk, is referred from LLC Victory Clinic to the genetic consultation of the National medical center with the clinical conclusion of ultrasonography: Pregnancy of 13,4 weeks. Monochorionicmonoamniotic twins. The Acardia syndrome, for the decision of further actions in pregnancy care. The patient is somatically healthy. According to the gynecologic history, the patient has one births in time, the child is healthy; three induced abortions in early gestational age, without complications. This pregnancy is the fifth. The patient has no social habits. The partner is healthy.

Accordingly to the ultrasonography research the diagnosis was completely confirmed. During the prenatal karyotyping – a karyotype of fetuses 46, XY {11} - normal. The prenatal genetic consultation was carried out. The couple was ordered to conduct the pregnancy in one of the Federal Centers of Prenatal Diagnostics, considering rare type of defect and high risk of antenatal mortality, but at the gestational age of 17,5 weeks with the dynamic ultrasonography an antenatal death of fetuses was diagnosed.

Results

An autopsy of fetuses confirmed the diagnosis. The first fetus (donor): male, weight is 120 g, height is 21 cm, without anatomical defects.

The second fetus (recipient: 10 g, 8 cm. The following defects are found: absence of the calvarial bones in a facial part, orbits, nose, auricles, and acoustic meatus. In the oral cavity a small-sized tongue is detected. Fetus body is extremely edematous. Humeruses are "immured" in a skinfold of the body; forearms and hands are easily detected. The top and lower extremities are without bony skeleton, easily are bended along the axis.

Placenta, umbilical cord: one placenta, weight is 50 g, the size is 10x8, 5x1 cm, with defect of a tissue, and covers are absent. The place of an attachment of the umbilical cords isn't detected because of the serious defects of the placenta. The second fetus (recipient) has an umbilical artery with hypoplasia.

Case No. 2

Pregnant woman, 32 years, referred from Zhigansky District to the female consutive department of National Medical Center of Yakutsk with the diagnosed anomaly of the twins fetal development and a positive gynecological anamnesis. From Female consultative department of NMC the pregnant was sent to ultrasonography research in the radiology department of NMC, there were established: Pregnancy of 27/5 weeks. Monochorionicdiamniotic twins. Frank polyhydramnios of the first

fetus. Abnormal development of the twins fetus: the TRAP (acardia) syndrome. A thickened placenta. The umbilical edema of gelatin of Wharton of the first fetus.

From the anamnesis: the fifth pregnancy. The first two pregnancies completed with birth in time, children are healthy. The third pregnancy is terminated in the gestational age of 17-18 weeks for medical reasons - congenital defect of central nervous system (an acrania, spina-bifida). The forth pregnancy completed with a late spontaneous miscarriage on 20th week, without congenital defects.

With this pregnancy the patient was registered since 10 weeks. With the ultrasonic analysis in a district healthcare center at the gestational age of 11,5 weeks congenital defects was not established. The patient has the third degree of obesity. She is married at the second time, the partner (from hearsay) is healthy. Social habits: smoking to the 5 weeks of the current pregnancy. Has no professional harmfulness.

In the genetic consultation of NMC a geological monitoring was conducted. Considering the duration of gestation, the prenatal karyotyping was not conducted.

The patient was hospitalized in the pathologic pregnancy department of the municipal clinical hospital in Yakutsk, where in the 30 gestational weeks of pregnancy with a pericardial effusion and an edema of gelatin of Wharton of the first fetus (donor) the elective operative delivery is carried out.

Results

Feature of this defect is a defective fetus does not have some parts of a body and internals, including heart. Its delivery occurs at the expense of a healthy fetus. Using special formulas it is possible to know the weight of an acardiac fetus, and that is a determining factor for further pregnancy prolongation. Upon reaching a certain weight, the CVS stress of a healthy fetus considerably increases.

At the gestational age of 27,5 weeks, according to ultrasonic analysis conclusion, the recipient fetus is described as: "… the fetus with a bizarre shape is detected close to a fetal surface - it has no head, but partially has a top part of a body, thoracic organs and an abdominal cavity. It was succeeded to visualize one top extremity, fingers aren't differentiated. Lower extremities are two. In the abdominal cavity a kidney is detected. Weight - 980 g. An umbilical cord of the second fetus is short (about 20 mm). The place of an attachment of the umbilical cord of the second fetus is in a close proximity with the place of an attachment of an umbilical cord of the first fetus. In the CFM the wide network of the anastomoses is detected. The relation of mass of the acardiac fetus to mass of the anatomically healthy fetus makes 64%".

At the gestational age of 30 weeks the pregnant woman was sent to an operative delivery.

The mass of the acardiac monster (in the birth moment) was 1190 g. From the protocol of autopsy of the second fetus (recipient): female fetus, weight is 1140 g, height is 23 cm. The head is absent, the body is deformed, shortened. Procurvation of a backbone, the upper part ends blindly. The right upper extremity is edematous, a hand is with hypoplasia and has 5 vestigial fingers. The left upper extremity is absent, there is a cartilaginous apophysis d - 0,5 cm and 0,8 cm long. The lower extremities with vestigial feet, which have two fingers on the left and three fingers on the right foot.

There is one placenta, weight is 520 g, size is 24x20x2 cm. Lobularity is accurate, maternal surface is rough. The umbilical cords: the first fetus (donor) - with a diameter about 3 cm, gelatin of Wharton is extremely edematous; the second fetus (recipient) - in the diameter of 1,3 cm. Three (!) vessels are detected.

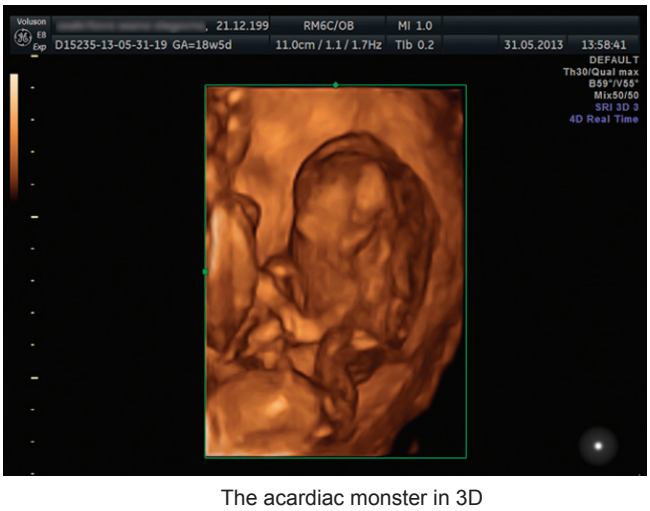
Attachment of the umbilical cords: a cord of the first fetus (donor) - paracentral, the second fetus (recipient) - velamentous.

The first fetus is female (donor), weight is 1760 g, height is 43 cm. At the birth an assessment on a scale Apgar score was 6/6 at birth. The patient was artificially ventilated in ICU. On the 10th day with stable condition the newborn girl was transferred to the neonatal pathology unit of the Maternal and Child Health Center of national hospital, NMC No1. At the present moment the child has a disability status because of cerebral palsy.

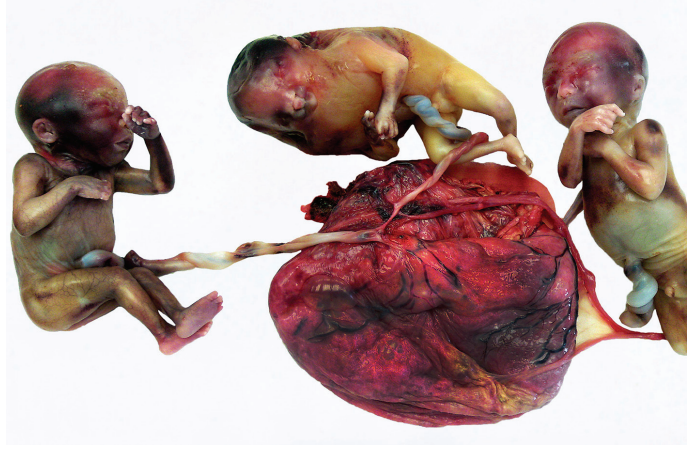
Case No. 3

The pregnant woman, 25 years, referred from Namsky District to the genetic consultation of NMC in the gestational age of 24 weeks with the clinic ultrasonic conclusion: Pregnancy 20, 5 weeks. Monochorionicmonoamnionic twins. The second fetus has an acardia syndrome. Polyhydramnios.

From the anamnesis: the first pregnancy completed with births in time and at terms, the child is healthy. Further, there were two cases of a spontaneous miscarriage in gestational age of 12 weeks. The fourth pregnancy completed with an operational delivery concerning the placental abruption in the gestational age of 29 weeks. Current pregnancy is the fifth. Registered on 20 weeks. Somatically healthy. Denies social habits. Civil marriage is the second. The partner is healthy (from hearsay).



The acardiac monster in 3D



The triplet pregnancy in the combination with the TRAP syndrome

Results

According to the ultrasonic research the gestational age was 24,2 weeks when the monochorionic twins was revealed. The second fetus has only the lower extremities, a part of an intestine and haunch bones. It has no head, the upper part of a body and the upper extremities. A subcutaneous tissue is frank edematous. Cord of the second fetus is short, and it is detected in the fixed position of a fetal surface of a placenta. The umbilical fetal cords are attached to the placenta at a close proximity to each other which gives impression that loops are bound. The first fetus is without anatomical defects, and it has a reasonable gestational age of 23,3 weeks.

The amniotic band was failed to visualize. There is one thickened placenta with signs of a premature maturity.

Conclusion of ultrasonography research: Pregnancy 24,2 weeks. Monochorionicmonoamniotic twins. Abnormal development: the TRAP syndrome of the second fetus (acardiaacephalus). The intrauterine growth retardation of the first fetus is not excluded. Moderate asymmetry of lateral ventricles of cerebrum of the first fetus. Polyhydramios. Thickening and premature maturing of a small-sized placenta.

In default of a possibility of the pregnant woman to departure to the Federal Centers of Prenatal diagnostics, with consent of the family it was procured an operative pregnancy interruption.

Results of an autopsy of fetuses confirmed the diagnosis of the ultrasonic research.

The first fetus is female, weight is 104 g, height is 37 cm, without anatomical defects.

Weight of the second fetus (donor) is 80 g, and looked like a shapeless mass with the lower extremities and a pelvis to a lumbar spine. There is no other part of the body. The sex cannot be detected – there are no external genitals. The fundament is presented. The frank edemas of extremities. From a pelvic part to the lower third of a cnemis the musculocutaneous folds are detected, which are interfering an extension of knee joints (pterygiums).

Case No. 4

A pregnant woman, 37 years, referred from the Phthisiology scientific production center to the genetic consultation of the National medical center No. 1 in the gestational age of 28,5 weeks suspected a twins congenital defects. She was hospitalized from the Eveno-Bytantaysky District with the diagnosis: Pregnancy of 28 weeks. Twins. Infiltrative tuberculosis of the right lung. Chronic hepatitis B. It is the second pregnancy, wanted. Labors in 1994, a healthy boy (given on adoption). Registered on D since 16 weeks.

The clinical ultrasonography conclusion: Pregnancy of 25,1 weeks. Monochorionicdiamniotic twins. Abdominal dropsy and antenatal death of the second fetus. Hyperechoic focal spots of the first fetus. Shorting of long bones of the first fetus. Edema of a gelatin of Wharton of the first fetus. Frank hypamnions of the first fetus. Oligoamnios of the II fetus. It is impossible to exclude a twin-to-twin transfusion syndrome.

According to the clinical conclusion the pregnant woman is hospitalized in the pathologic pregnancy department of the municipal clinical hospital in Yakutsk where the operative delivery in the gestational age of 28 weeks was carried out. The first fetus (donor) is female, weight is 1144 g, height is 38 cm, Apgar score is 6/6. She has been living for 10 days. The second fetus had a bizarre shape and weight is 2400 g (a fetus – the donor).

Results

In this case there is a wrong conclusion, though a specialist tried to describe an acardiac fetus: "… during examination of the second fetus the cardiac rate and the motion activity – aren't detected. There is a frank edema of soft tissues of a bandwidth at the head level which is about 60 mm, and at the level of an anterior abdominal wall is about 50 mm.

In edematous tissues multiple cystophorous formations (the overall is 90x72 mm) are visualized. The fetus sizes without edema of soft tissues are reasonable to 23 weeks of pregnancy.

The attachment place of an umbilical cord of the second fetus cannot be visualized. An umbilical cord of the first fetus with the signs of an edema of a gelatin of Wharton.

The autopsy of the second fetus: MCA - Acardia (Hemiacardius), corpus callosum aplasia, frontal cerebral hernia, hydrocephalus, anophthalmia, agenesia of a nose, auricles and acoustical meatuses. Hypoplasia of an upper and lower jaw bone with fragmentation. Agenesia of a trachea, extrapulmonary bronchus and lungs. Sinistral diaphragmatic pseudohernia with an eventration of loops in a thoracic cavity. Agenesia of a esophagus, liver and lien. Atresia of a fundament and rectum and genitals. Agenesia of the upper extremities, webbed toes of IV and V fingers of a left foot, agenesia of the IV finger of a right foot. Cystic lymphangioma of the soft tissues of a posterior surface of a neck and back. The umbilical cord of the second fetus has two vessels and a battledore placenta.

Case No. 5

A pregnant woman, 20 years, referred from a healthcare center of the high level to the genetic consultation of NMC with a triplet pregnancy and an ectopic first fetus, with the gestational age of 12,3 weeks. The current pregnancy is the third, wanted. The patient became pregnant in the next menstrual cycle after interruption of the second non-developing pregnancy. The husband is 21 years old. Partners are somatically healthy, have no occupational hazards and social habits. The examination was conducted on the ultrasonograph devices Toshiba Xario and Voluson E8.

Results

According to the first ultrasonic research conclusion in the gestational age of 12,4 weeks: Pregnancy of 12,4 weeks. Dichorionicdiamnionic triplets. Antenatal death, edema of the third fetus. Augmentation of the second fetus. The prenatal karyotyping of fetuses is carried out. A karyotype of fetuses 46, XY [11] and 46, XY [20] - normal. Chromosomal fetal pathology is excluded. In the gestational age of 14-15 weeks the woman is hospitalized to the gynecology department of NMC, for the decision about further prenatal care actions.

According to the ultrasonic research in the term of 15 weeks it is determined: in an uterine cavity there are three fetuses. The third fetus with parietal-coccygeal length of 74 mm (13,4 weeks) … "in energy regime" vessels are registered. Heart of the fetus isn't visualized, but the fetal movement becomes perceptible. On examination of brain structures - a cerebellum has a banana form. The bladder isn't detected. The frank edema of soft tissues, a hydrothorax are detected. Facial structures aren't visualized accurately because of the edema. The amniotic band isn't visualized accurately. During examination of the umbilical cord of the second and third fetuses it are revealed that cord vessels are in a close proximity to each other, at distance of 12-15 mm from a anterior abdominal wall. Pulse frequency of vessels of the acardiac fetus is 167 beats per minute; the donor fetus - 160 beats per minute.

At a dymanic research in the

gestational age of 22,5 weeks there are signs of a cardiovascular collapse of the second fetus (donor) – free fluid in thoracic cavity and abdominal cavity, therefore, with the consent of a family, the pregnancy was interrupted.

On autopsy the diagnosis about acardia syndrome of the third fetus was confirmed. From the protocol: a fetus weight is 360 g, height is 21 cm. The head is detected. There are no palpebral fissures. Bones of a nose aren't detected. There is a bilateral cleft of an upper lip and hard palate. In a neck, a breast to an epigastrium and a back to a loin there is a non-pitting edema of tissues. Hypoplasia of auricles, low located. Heart, an esophagus, a stomach, a liver, a pancreas are absent. Lungs without share division. In kidneys there are three spherical formations of 1,3 cm long. Adrenals in the typical place aren't detected.

There is one battledore placenta for the second and third fetuses, and velamentous placenta of the first fetus with an anastomosis of one vessel to a nearby umbilical cord with a distortion around it.

Case No. 6

Pregnant woman, 29 years, referred from LLC Victory Clinic to the genetic consultation of NMC with twins pregnancy with the acardia syndrome of the first fetus in the gestational age of 13,5 weeks. The current pregnancy is the fourth, unplanned, wanted. In the anamnesis there are one physiological labors, two abortions. The pregnancy was complicated by early toxicosis, an ARVI with temperature of about 102 degrees, an urogenital infection. The partners are somatically healthy, have no occupational hazards and social habits. The examination was conducted on ultrasonic devices Voluson Pro and Voluson E8.

Results

The conclusion of the first ultrasonography in Victory Clinic at the gestational age of 13 weeks: Pregnancy 13 weeks 5 days. Monochorionicdiamniotic twins. The Acardia syndrome: a nanocephalia, a hypodermic edema, a cervical hygroma, a hydrothorax, an omphalocele of the first fetus. A velamentous placenta with the abnormal number of vessels.

The ultrasonic research conclusion in the genetic consultation was confirmed the diagnosis. At the gestational age of 14,3 and 19 weeks it was additionally revealed: an aplasia of radial bones, oligodactyly and clinodactyly of the acardiac fetus. The prenatal karyotyping of fetuses is carried out. Chromosomal pathology is excluded.

In the gestational age of 19 weeks with the consent of the family the pregnancy was interrupted on medical authority. During the autopsy the ultrasonic diagnosis of the acardia of the second fetus was confirmed.

From the protocol: a fetus' weight is 250 g, height is 18 cm. A tissues surrounding heads, shoulders, body, the upper extremities to hands are presented by a vascular tumor (lymphangioma). Fetus head: the right parietal bone isn't formed, the brain is covered with a pachymeninx. A right orbit is covered with soft tissues, extruded. Nasal bones are absent, a rhegma of median soft tissues of an upper lip is detected. Heart and lungs aren't detected. In a thoracic cage a "slugged" yellowish tissue. Lungs, a liver, a lien are absent. There are kidneys on both sides (histologically primitive). On About the locomotion system: the right upper extremity is shortened at the expense of a shorting of forearm bones, a hand has 4 fingers and rotated inside with an acampsia of a radiocarpal joint. The left extremity is shortened at the expense of forearm bones, on a hand there are 4 distorted fingers. Lower extremities: the left foot has 3 fingers, right - 4 fingers.

CONCLUSIONS:

1. For the described period, a frequency of TRAP syndrome in Yakutia made 1: 10 805 labors. It is three times more than in other regions.
2. One of the reasons of high detection frequency of TRAP syndrome is rising of quality level of the prenatal diagnostics in the Republic of Sakha (Yakutia).
3. With the aim of frequency reduction of TRAP syndrome and reduce the endocrine pathology of women of childbearing age the extensive use of periconceptional care is highly recommended.
4. With detection of TRAP syndrome a laser coagulation of an umbilical artery of a fetus recipient is highly recommended before the progression of volemic complications.

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