FACTA UNIVERSITATIS
Series: **Medicine and Biology** Vol. 17, No 1, 2015, pp. 42–44 UC 611.012:575.22

Case Report

A LIVEBORN INFANT WITH TRIPLOIDY 69,XXX: CASE REPORT

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Abstract. *Since liveborn babies with triploidy are quite rare, we report here a new case of a liveborn female baby, with a karyotype 69,XXX. Ultrasound examination of the fetus, in the 37th week (9th month) of uncontrolled pregnancy, discovered severe intrauterine growth restriction and olygoamnion. A sample of fetal blood was taken by cordocentesis, in order to analyze fetal chromosomes. Two days later, the liveborn child was delivered by Cesarean section and died the same day, 7 hours and 55 minutes later, due to respiratory insufficiency. Autopsy revealed deformity of the joints, ectrodactyly of feet, deformity of the face (large, low positioned ears, hypotelorism and hypoplastic mandibula), with irregular position of both hands, hypoplastic lungs, kidneys, suprarenal glands, gallbladder and thymus agenesis. Chromosome analysis performed from fetal blood lymphocytes taken by cordocentesis showed karyotype 69,XXX. We would like to emphasize the significance of a health education of pregnant women, in order to establish regular examinations, and thus improve diagnostic and disease management possibilities.*

Key words: *Triploidy, multiple anomalies, cordocentesis*

Introduction

Triploidy presents a chromosome abnormality which is characterized by an extra-haploid set of chromosomes that could be inherited either from the father or from the mother. Data that have been obtained so far, point to the fact that triploidy could be present even in 2 per cent of the human fertilization and such pregnancies are usually ended with spontaneous abortion in the period from the seventh to the seventeenth week of pregnancy [1, 2]. Liveborn babies with triploidy are very rare, therefore, in this report we present a case of a female born baby, with karyotype 69,XXX, that lived for 7 hours and 55 minutes.

Case Report

A thirty-eight-year-old patient in the 37th (9th month) week of gestation, in her fourth pregnancy, was referred to our clinic for advice about delivery. She had three healthy children, so she decided not to control her pregnancy regularly. She had one ultrasound examination in the 8th week of pregnancy and had never been biochemically screened prenatally. Maternal-fetal physicians performed ultrasound examination of the fetus, and discovered severe intrauterine growth restriction and olygoamnion. With a suspicion on trisomy 18, they immediately took a sample of fetal blood by cordocentesis, in order to analyze fetal chromosomes.

The patient was held in the clinic, and two days later, she gave birth to a liveborn child by Cesarean section. The newborn had symmetric restriction of growth, it was 900g heavy and 35 cm long (under the third percentile), hypothrophic, with deformities of face and limbs. The baby died that same day, 7 hours and 55 minutes later, due to respiratory insufficiency.

Autopsy revealed deformity of the joints, ectrodactyly of feet, deformity of the face (large, low positioned ears, hypotelorism and hypoplastic mandibula), with irregular position of both hands, hypoplastic lungs, kidneys, suprarenal glands, gallbladder and thymus agenesis (Figs 1–3).

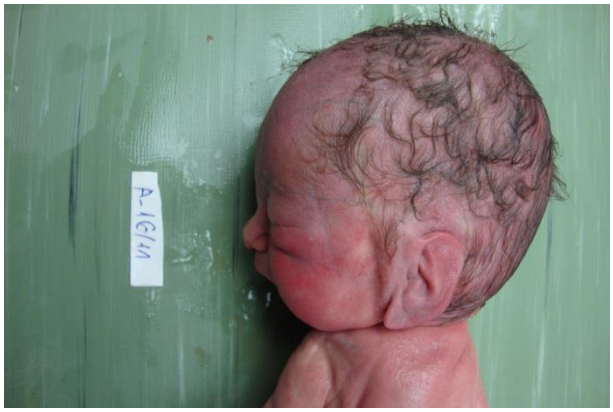

Fig. 1. Deformities of the face and head

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Received April 29th, 2015 / Accepted August 26th, 2015

Fig. 2. Ectrodactyly of feet

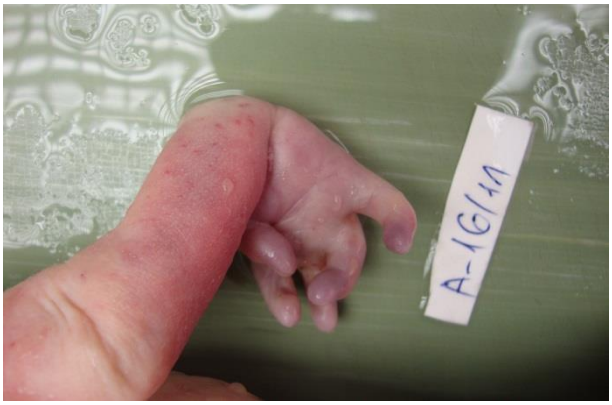

Fig. 3. Hand deformities

Chromosome analysis performed from fetal blood lymphocytes taken by cordocentesis showed karyotype 69,XXX**.** Triploidy was detected in 50 metaphase cells after G-banding with trypsin-Giemsa (GTG).

Discussion

Triploidy is the third most frequent case of chromosome abnormality and could be the cause of 15 to 18 per cent of all spontaneous miscarriages**.** It appears in 1:10 000 of liveborn children (out of that number 51 to 69 percent are male babies) [2, 3]. The frequency of triploidy in fetuses is 1:2000 [4].

Three possible mechanisms of triploidy appearance are described hereby:

1) nondisjunction in meiosis I or meiosis II of spermatogenesis (sperm formation), resulting in an extra set of paternal chromosomes (diandry)

2) nondisjunction in meiosis I or meiosis II of oogenesis (egg formation), resulting in an extra set of maternal chromosomes (digyny)

3) double fertilization of a normal egg, resulting in an extra set of paternal chromosomes (dispermy)

It has been reported that mechanism 1 accounted for 23.6% of triploidy cases, mechanism 2 for 10%, and mechanism 3 for 66.4% [5].

Most cases of triploidy result from dispermy [6–8]. Several studies have reported that the majority (62–77%) of cases of triploidy of maternal origin result from nondisjunction in meiosis II, although another investigation found nondisjunction to be evenly distributed between meiosis I and meiosis II [6, 7, 9].

Fetal nuchal translucency thickness (the sonographic appearance of a subcutaneous collection of fluid behind the fetal neck) in the first trimester is frequently increased for fetuses with triploidy [10–12]. Triploidy associates minor facial anomalies (facial asymmetry, low set ears), mild ventriculomegaly, multiple major structural defects of the internal organs, intrauterine growth retardation (asymmetric most frequently) and sindactily (of third and forth fingers most frequently). Other possible ultrasound findings in triploidy are: hypertelorism and microphtalmus, micrognatia, major facial anomalies (cleft), agenesis of corpus callosum, cardiac malformations, septal defects, single umbilical artery, omphalocele, renal anomalies, holoprosencephaly [4].

Unusual findings for triploidy in the presented case were hypotelorism, ectrodactyly and thymus agenesis.

It has been mentioned in the literature that the fetuses, whose haploid set is maternal in origin have a better chance to be born. Further, infants with triploid/diploid mosaicism will have longer survival than those with true triploidy [13].

Conclusion

In the presented case, the baby with triploidy was born due to uncontrolled pregnancy. In most cases, fetal chromosomal aberrations are presented with ultrasonographically detectable anomalies. That is the reason why thorough anatomical survey is a necessary part of fetal screening.

We would like to emphasize the significance of health education of pregnant women, in order to establish regular examinations, so we would have better diagnostic and disease management possibilities.

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