Heterokaryotypic pregnancy: monzygotic monochorionic twins discordant for trisomy 13

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Objectives: To describe a rare case of heterokaryotypic monzygotic twins – monochorionic diamniotic – discordant for trisomy 13.

Methods: Case report

Results: A 23 years old primiparous women with spontaneous monochorionic twin pregnancy underwent first trimester screening in our department which was negative for trisomy 21, 18 and 13. One fetus had no structural abnormalities, while in the other one gastroschisis was detected. Due to this finding she underwent genetic consultation and amniocentesis was performed. The result of the karyotyping was 47XY + 13 for the fetus with gastroschisis (fetus A) and 46XY for the structurally normal fetus (fetus B). Monzygotic pregnancy was confirmed. During the 16th week scan another structural abnormality in fetus A was detected - right sided palate and lip cleft and in 20th week left sided hydronephrosis and megaureter was found. Due to the pathological finding the patient opted for selective reduction of the fetus A which was performed at 21st week of pregnancy in Fetal Medicine Center, Institute for the Care of Mother and Child in Prague, Czech Republic using bipolar cord occlusion. The intervention was performed without any complication. The pregnancy was then followed at our instiution until week 36 when the healthy fetus was delivered by cesarean section for intrauterine growth restriction, oligohydramnion and breech presentation. The measurements of the baby boy were 2400 grams and 44 centimeters, Apgar score 9-9-10. The boy had no structural abnormalities.

Conclusion: Heterokaryotypic monochorionic twins are very rare and karyotypic difference is caused by asymmetric X-chromosome inactivation and differential gene imprinting post-zygotic mitotic errors such as nondisjunction and anaphase lag. The most common cause of this is nondisjunction of homologous chromosomes. According to the phase of zygote formation in which the nondisjunction occurs, it is determined whether genetic abnormalities occur in all fetuses or in only one fetus. In other words, one fetus will have genetic abnormalities but another fetus will be normal if the nondisjunction occurs after zygote formation is completed.1,2,3 The incidence is not described in the literature. Our suspicion for this anomaly begun with discordant ultrasound findings during first trimester screening. Diagnosis was confirmed by amniocentesis. Discordant ultrasound findings in case of monzygotic twins should lead to suspicion for heterokaryotypic pregnancy.

References: