Management of Twin Pregnancy with Discordant Structural Fetal Anomalies
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Case Presentation

We report a case of dichorionic diamniotic twin pregnancy, discordant for fetal anomalies diagnosed prenatally at 16 weeks of gestation. The diagnosis of the anomaly was based on two and three-dimensional ultrasound. Twin two had normal fetal structures. Twin one was diagnosed with severe ventriculomegaly, caudal regression and absent right forearm and short right humerus (Figure 1). Parents opted for pregnancy termination, termination performed at 32 weeks without complications. Cytogenetic testing of the fetus showed normal karyotype therefore the recurrence risk remains low. In this case review we would like to share the ultrasound images, postnatal correlation, clinical management and the ethical aspects of this particular case.

Results

Cytogenetic Investigations:

Fetal amniotic fluid:

Microarray result-no significant abnormality detected. Genome wide array analysis indicated that there was no evidence of any copy number change of likely clinical significance for the diagnosed fetal anomalies. The array analysis result obtained was consistent with an XX (female) chromosome complement. This result is consistent with the preliminary rapid QF-PCR report.

Figure 1. Ultrasound images of the anomaly scan performed at 16 weeks and 3 days (1&2) and photographs taken in the post-natal period (3&4).

Discussion

It is well known that the risk of congenital anomalies in twins is higher than in singletons. One recent study has shown 1.5 times higher risk for fetal malformations in monochorionic (MC) pregnancies compared to dichorionic (DC) pregnancies. The excess risk was mainly attributed to the higher rate of complex anomalies. While the rate of cardiac abnormalities was higher in MC twins, the rate of CNS abnormalities was lower compared to DC twins [1].

The increase in the incidence of anomalies in DC twin pairs could occur from the contribution of the monzygotic dichorionic twin pairs and the pathology of embroy division. Woman has thus at least two fold risk of carrying a fetus with structural abnormality then if she were carrying a singleton. The vast majority of fetal abnormalities effect only one twin. In dichorionic twin pregnancies fetocide for the baby with anomalies can be as performed as it is performed in singleton pregnancies. Procedure involves ultrasound guided intracardiac injection of potassium chloride. Procedure when performed at 20 weeks has approximately 5-9% risk of loss of the health co-twin [2]. In the present case in order to reduce the risks to the healthy twin fetocide procedure delayed until 32 weeks gestation.

Incidence of absent forearm is rare less than 1:5000. Prognosis depends on the underlying pathology and the presence or absence of other structural abnormalities. It is essential to facilitate accurate prenatal diagnosis and parental counselling [3].

Caudal regression syndrome (CRS) is a rare congenital malformation with a degree of early gestational developmental failure. Although diabetes mellitus is the major risk factor for CRS, as seen in our case, sporadic presentations may occur. Clinicians should consider CRS when CRL is shorter than expected and incomplete vertebral ossification is observed in 3D ultrasound imaging [4].

The prenatal diagnosis and the management of discordant abnormalities in multiple pregnancies involve a multidisciplinary team of sonographer, fetal medicine specialist, clinical geneticist, neonatologist, and orthopedic surgeons to provide the parents with the information regarding etiology of the disorder, prognosis and options related to the pregnancy and recurrence risk for future pregnancies [5].

Early prenatal diagnosis of twins with discordant congenital defects is important. Supportive antenatal care is necessary in the management of twins with discordant congenital defects and multidisciplinary counseling services to parents are recommended for determination of options [6].

Ethics is essential for the responsible clinical management of multiple pregnancies and decision-making about the future of such pregnancies with pregnant women. The ethical concept of the fetus as a patient is presented as the basis for identifying a professionally responsible approach to selective termination for discordant twins when one or more fetuses are adversely affected by a complication of pregnancy or fetal anomaly.

The roles for directive counseling, such as making evidence based recommendations, and for non-directive counseling, such as offering evidence based alternatives but making no recommendations, are described methods of counseling. The professional responsibility model of perinatal ethics creates the informed process about the clinical management of multiple pregnancies and a practical framework to guide the clinical judgment of fetal medicine specialists [7].

References:

5. Collection 2011