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Down syndrome and transposition of great arteries

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Objective

Transposition of great arteries (TGA) is one of the most common and severe congenital heart diseases. TGA is very rarely associated with genetic syndromes, such as Turner, Noonan, Williams or Marfan syndromes, and in Down syndrome, it is virtually absent. The only genetic syndrome with a strong relation with TGA is Heterotaxy. It is also associated with fewer extracardiac anomalies (<10 percent) compared to other congenital heart defects.

Methods

We present one case of transposition of great arteries and ventricular septal defect as only findings on ultrasound examination.

Results

A 36-year-old patient G2P1 had a low risk at first trimester screening test, 1/ 2531 for T21 and 1/10. 000 for T18 and a normal first trimester scan. The patient had unremarkable medical history and prenatal exposure. After genetic counselling, the patient opted for an amniocentesis which gave a positive result for trisomy 21. There were not other fetal anomalies detected at the 20 weeks anomaly scan. After the amniocentesis results, the couple decided to have a termination of pregnancy.

Conclusion

The specific developmental aspects that result in ventriculoarterial discordance in D-transposition of the great arteries are not fully delineated. In addition to ventriculoarterial discordance, there are other cardiac anomalies or functional defects that are often seen in patients with D-TGA. Although transposition of the great arteries is one of the most difficult congenital heart defects to detect by fetal ultrasound, some cases may be diagnosed, especially if sonographic screening includes views of the outflow tracts.