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A case of complex cardiac anomaly in a case of 22q11.2 deletion syndrome

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Objective

The purpose of our paper is to present a case of a rare cardiac anomaly (interrupted aortic arch type B, large malalignment- type ventricular septal defect and pulmonary valve dysplasia) in a fetus diagnosed with 22q11.2 deletion whom we followed throughout the pregnancy until term; we will also review approach to diagnosis and newest data regarding prognosis and outcome.

Methods

The patient is a 34-years old IG IP who obtained a spontaneous pregnancy while starting diagnostic work-up for infertility and she was referred to our department for the second trimester anomaly scan. The first trimester anomaly scan was reported as normal and she had a low-risk NIPT result for trisomies 21, 18 and 13. At 22 weeks the differential diagnosis was made between severe aortic coarctation and tubular hypoplasia with VSD and IAA. The fetal thymus appeared as small and also we noticed a dilated CSP and a characteristic appearance of the fetal profile.

Results

The fetal echocardiography performed by a pediatric cardiologist established the final diagnosis as IAA type B with VSD and pulmonary valve dysplasia. Invasive genetic testing revealed 22q11.2 deletion and genetic counseling was offered to the patient who decided to continue the pregnancy. The patient had regular follow-up with ultrasound scans every 2-3 weeks to monitor fetal growth and condition and amniotic fluid volume. She opted to deliver in Germany so that neonatal cardiac surgery can be performed in an experienced Center for Cardiac Malformations.

Conclusion

We discuss a case of 22q11.2 deletion syndrome with a complex cardiac malformation emphasizing on ultrasound aspects in a continuous manner until delivery.