Prenatal diagnosis of DiGeorge syndrome associated with a CNS malformation followed by a virtual autopsy of the aborted fetus – a case report

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Objective: DiGeorge syndrome is one of the most common microdeletion syndromes. It is caused by the deletion of a part on the long arm of the chromosome 22 and its incidence is 1/4000. Many organ systems can be affected. The syndrome may include congenital heart defects (VCC), abnormalities of the orofacial region, congenital renal anomalies, mental retardation, immune disorders due to thymic hypoplasia or aplasia with consequent T-cell deficit, and hypocalcemia. The aim of this study was to assess whether an MRI virtual autopsy of aborted fetus is an alternative to the conventional autopsy in cases of CNS malformations, multiple organ anomalies or eventually in cases where the parents do not agree with a classic autopsy.


Results: First examination at 12 weeks of pregnancy: NT 2.9 mm, PAPP-A 1.29 MoM, free-β hCG 0.79 MoM, risk for trisomy 21 1/1750, risk for trisomy 13 and 18 1/27400, abnormal fetal profile, and high suspicion of orofacial cleft. Second examination at 16 weeks showed adequate fetal growth, median cleft lip and palate, high suspicion of VCC (ventricular septal defect and abnormal crossing of the great vessels). Amniocentesis was performed. Fetal echocardiography at 17 weeks revealed persistent truncus arteriosus (PTA). Third examination at 20 weeks found symmetric fetal growth, median cleft lip and palate, VCC – PTA, lobar holoprosencephaly, and grade III hydronephrosis of the right kidney. The conventional karyotype was 46,XX. However, SNP array revealed 22q11.21 microdeletion. The patient decided to terminate the pregnancy. Because of the CNS malformation, an MRI virtual autopsy was performed.

Conclusion: In some cases, a classic autopsy cannot confirm the prenatal diagnosis of a CNS malformation due to advanced maceration/autolysis of the brain. MRI dominates in the imaging of the brain and soft tissues. The diagnosis and management of congenital malformations should be a team work including not only gynecologists and geneticists but also relevant specialists (cardiologists, surgeons, neurologists), radiologists and pathologists. It is the only way how to provide the family with sufficient and valid information about the malformation of the fetus and the prognosis for future pregnancies.