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MULTIPLE CONGENITAL MALFORMATIONS AS AN INDICATION FOR MEDICAL ABORTION

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Congenital malformations may be isolated or multiple and represent a relevant cause of spontaneous abortion and fetal death when they are incompatible with the normal embryo-fetal development.

We present a case of a 23-year-old primigravida, with a negative personal and family history of congenital anomalies. Due to an abnormal finding on the first trimester screening (PRISCA 1), a chorionic villus sampling was performed at 14 weeks of gestation, but chromosomopathy was not detected. An ultrasound examination at 17 weeks of gestation revealed hydrocephalus and megacolon, which indicated termination of pregnancy. Induction with oxytocin was performed and the fetus and placenta were sent for autopsy. A female fetus weighing 170 grams and 16 cm long. During inspection, facial dysmorphism was detected with low-set ears (below the level of the angulus oris), the neck was short and wide, and on the back of the neck there was a hygroma measuring 0.5x2 cm. An imperforate anus was detected. In the abdominal cavity, there was a slightly distended large intestine that was passable and opened into the urinary bladder. The described malformation was in favor of a persistent cloaca, which has been confirmed histologically. Renal agenesis was also detected. Histological analysis of the remaining organs showed extramedullary hematopoiesis and congestive changes. The placenta weighed 110 grams, and the funiculus umbilicalis had a paracentral insertion. On the maternal side, there was a retroplacental hematoma and microscopically there were areas with subacute chorioamnionitis, cystically degenerated areas of the amniotic membranes, intraplacental bleeding and areas of intra and intervillous fibrinoid deposition. Additional analyses (Chromosomal Microarray Analysis) were proposed, but the patient refused.

The aim of this case presentation is to emphasize that prenatal ultrasound examination allows the detection of major fetal defects and pregnancies can be terminated if the fetus is severely affected.

Keywords: congenital malformations, ultrasound, hygroma, imperforate anus, persistent cloaca.