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A case of VACTERL Association

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Objective

We present the case of a fetus with prenatal diagnosis of VACTERL sequence with Fallot heart disease.

Methods

This is a case report.

Results

Pregnant 32 years old, third pregnancy, the first son was healthy and the second had syringomyelia. Her past medical history included: factor V leiden mutation, thromobophlebitis managed with cardiovascular medicine and renal lithiasis. Screening during the first trimester of pregnancy showed a low risk result for T21 and T18. In the morphological ultrasound at week 16, bilateral equinovarial foot was evidenced, and therefore was referred to a fetal medicine unit. Week 17 transabdominal ultrasound was performed for morphological study: Fetus was cephalic with biometry according to dates. Suspected VACTERL sequence with Fallot heart disease with pulmonary hypoplasia, shortening of lower limbs with suspected absence of fibula and presence of bilateral equine feet. Unable to visualize left kidney, single umbilical artery. Anterior placenta under uterine segment. After informing the pregnant woman and her partner, both decide on voluntary termination of pregnancy (VTP) in week 18. Fetal postmortem was performed with the result of a female fetus of 17 weeks' gestation and 129 grams of weight. During the postmortem the following were confirmed: a Fallot cardiac disease with overlapping aorta and hypoplastic pulmonary artery (extreme Fallot), left renal agenesis and lower limbs with hyperflexion of feet and legs (arthrogryposis) and umbilical cord with 2 vessels. The Array-MLPA p245 analysis of fetal tissue was performed with normal result is performed.

Conclusion

VACTERL is an association of congenital malformations typically characterized by the presence of at least three of the following: vertebral defects, anal atresia, heart defects, tracheoesophageal fistulas, renal anomalies and abnormalities in the extremities. The annual incidence is 1 / 10.000 to 1 / 40.000 live births. No specific geographic distribution or predominance has been found in certain ethnic groups. Prenatal diagnosis can be challenging, since certain characteristics of the components can be difficult to determine before birth. The differential diagnosis includes Baller-Gerold syndrome, CHARGE syndrome, Currarino disease, 22q11.2 microdeletion syndrome, Fanconi anemia, Feingold syndrome, Fryns syndrome, MURCS association, speculum. oculo-auriculo-vertebral, Opitz G / BBB syndrome, Pallister-Hall syndrome, Townes-Brocks syndrome and VACTERL with hydrocephalus. We believe that performing a postmortem and studying Arrays in case of interruption of pregnancy is very important in these cases.