

Generalised arterial calcification of infancy

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Objective

We studied the fetal form of generalised arterial calcification of infancy and specified the fetal pathological characteristics in order to improve the antenatal diagnosis. This case was reported in the fetal embryology pathology unit of maternity and neonatology centre of Tunis.

Methods

A 33 year-old woman, gravida 6, para 4, with a child born with an IVC which spontaneously regressed, two infants with cardiac malformations, one miscarriage and one intrauterine fetal death. This pregnancy ended up at 16 weeks of gestation in intrauterine fetal death. Fetal pathological analysis was performed on a female fetus. External examination showed fetal anasarca, cranio-facial dysmorphism, and excess of skin in the neck. Dissection revealed hypoplasia of lungs and cardiac calcifications situated in the left ventricle. The rest of the organs appeared normal. Histologic examination revealed calcium depositions in the left ventricle, intramyocardial vessels, the glomeruli and renal tissue vessels. Fetal and parental karyotypes were normal, and a diagnosis of GACI was made.

Results

GACI is a lethal, congenital abnormality and its diagnosis is usually made at autopsy. Fetal standard x-rays can detect calcifications of the aorta and its principal branches. Histologic examination reveals calcifications of pulmonary and systemic arteries. Calcium deposits are found in the lamina of large and medium-sized arteries and fibrointimal hyperplasia causing luminal stenosis. Damaged arteries are turned into rigid "pipestems". Sometimes, glomerular tufts and periarticular calcifications are found. Antenatally, the calcifications can be identified in ultrasounds examination. This disorder, that used to be considered idiopathic, is actually due to an enzymatic deficit causing a lack of inorganic pyrophosphate inhibitor of hydroxyapatite deposits. The diagnosis is confirmed by searching for mutations of ENPP1 gene.

Conclusion

Antenatal diagnosis of GACI requires multidisciplinary care in order to decide the termination of pregnancy and the future pregnancies' prognosis by ultrasound and genetic searching for mutations.