

Generalized Arterial Calcification of Infancy (GACI): Diagnosis and Prognosis

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Received: April 13, 2017; **Published:** May 02, 2017

Abstract

GACI is a rare autosomal recessive disorder due to mutations of ENPP1 coding for ecto-nucleotide pyrophosphatase/phosphodiesterase 1. Most patients die within the first six months of life. In fetus, it manifests itself with anasarca resulting from heart failure leading to intrauterine fetal death.

Female fetus, mother aged 33, gravida 6, para 4, with a child born with an IVC spontaneously regressive, two infants with cardiac malformations, one miscarriage and one intrauterine fetal death. This pregnancy ended at 16 weeks of gestation in intrauterine fetal death. The fetal pathological analysis had been performed.

External examination showed fetal anasarca, cranio-facial dysmorphism, and excess of skin in the neck. Dissection revealed hypoplasia of lungs, cardiac calcifications situated in the left ventricle. The rest of the organs were normal. Histologic examination included calcium deposition in the left ventricle, intra-myocardial vessels, the glomeruli and renal tissue vessels. Fetal and parental karyotypes being normal, GACI diagnosis is retained.

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

Keywords: GACI; ENPP1

Introduction

GACI is a rare autosomal recessive disorder due to mutations of ENPP1 coding for ecto-nucleotide pyrophosphatase/phosphodiesterase 1. Most patients die within the first six months of life. In fetus, it manifests itself with anasarca resulting from heart failure leading to intrauterine fetal death.

Aim

We are studying the fetal form of this disorder and specifying the fetal pathological characteristics in order to improve the antenatal diagnosis. This case is reported in the fetal embryology pathology section of Maternity and neonatology center of Tunis.

Case Report

Female fetus, mother aged 33, gravida 6, para 4, with a child born with an IVC spontaneously regressive, two infants with cardiac malformations, one miscarriage and one intrauterine fetal death. This pregnancy ended at 16 weeks of gestation in intrauterine fetal death. The fetal pathological analysis had been performed.

External examination showed fetal anasarca, cranio-facial dysmorphism, and excess of skin in the neck (Figure 1 and 2). Dissection revealed hypoplasia of lungs, cardiac calcifications situated in the left ventricle (Figure 3). The rest of the organs were normal. Histologic examination include calcium deposition in the left ventricle, intra-myocardial vessels the glomeruli and renal tissue vessels. Fetal and parental karyotypes being normal, GACI diagnosis is retained.



Figure 1 Figure 2
Figure 1 and 2: Macroscopic examination of fetus.

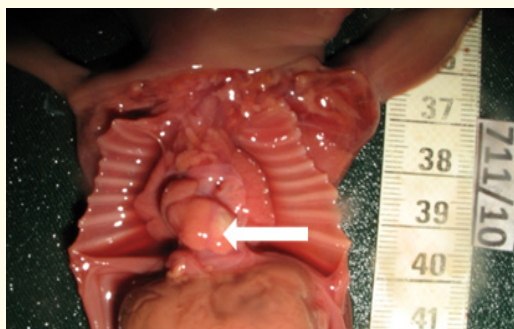


Figure 3: Hypoplasia of lungs, cardiac calcifications situated in the left ventricle.

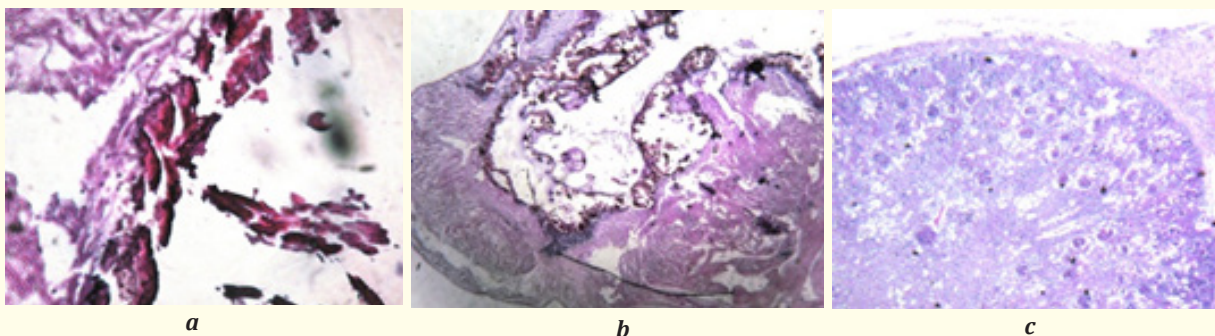


Figure 4: Calcium deposition in the left ventricle (a), intra-myocardial vessels (b), the glomeruli and renal tissue vessels (c).

Discussion

GACI is a lethal, congenital and its diagnosis is usually made at autopsy [1,2]. Fetal standard x-rays can detect calcifications of the aorta and its principal branches [3,4]. 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 [5,6]. Antenatally, the calcifications can be identified in ultrasounds examination After having eliminated some pathologies that have echographic signs in common like the oligoamnios [7].

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 terminal of pregnancy and the future pregnancies' prognosis by ultrasound and genetic searching for mutations.

Conflict of Interest

We declare that we have no conflict of interest.

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Volume 4 Issue 1 May 2017

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