Outros

(21524) - MITOCHONDRIAL HSD10 DISEASE: REPORT OF PRENATAL PRESENTATION

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Introdução

Few cases are known of HSD10 disease, an X-linked disorder caused by deleterious variants in *HSD17B10*. This disease arises from 2-methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD) dysfunction in isoleucine metabolism presenting with impaired mitochondrial RNase P complex activity. Increased excretion of 3-hydroxy-2-methylbutyrate and tiglylglycine without elevation of 2-methylacetoacetate is a hallmark of this disease. The neonatal form presents with immediately postnatal onset of severe metabolic acidosis with death in the first months of life.

Objectivos

Describing the first prenatal presentation HSD10 disease, of our knowledge

Metodologia

We report a boy with neonatal HSD10 disease and unusual prenatal presentation.

Resultados e Conclusões

Our patient was the second son of a non-consanguineous couple. Prenatal ultrasound showed mild subcutaneous edema in the cephalic pole and feet, increased aortic valve echogenicity, ascending aorta dilatation and hyperechogenic fetal bowel. He was born prematurely at 34 weeks of gestation with generalized edema and hypotonia, developing metabolic acidosis in the first day of life with hyperlactacidemia refractory to bicarbonate. Urinary organic acids analysis showed massive levels of lactic acid and non-specific increase of multiple organic acids. Muscle biopsy studies excluded mtDNA depletion and described increased mitochondrial proliferation with complex I and IV deficiency. Our patient developed multiorgan failure, dying on the second day of life. Autopsy described glomerulocystic kidney disease. Whole exome sequencing detectied a hemizygous variant of unknown significance in *HSD17B10*, maternally inherited and not present in his younger healthy brother or his healthy maternal uncle. Fibroblasts enzymatic studies showed MHBD decreased activity, supporting the diagnosis of HSD10 disease.

With this case, we describe the first prenatal presentation of HSD10 disease and a potential association of this disease with glomerulocystic kidney disease. This case highlights the relevance of considering the diagnosis of mitochondrial disorders in the prenatal period, and preserving samples for further functional/biochemical studies.

Palavras-chave : HSD10 disease, mitochondrial disorder, Prenatal