Outros

(21609) - CAKUT SPECTRUM: RENAL HYPODYSPLASIA /APLASIA TYPE 3 PRENATAL DIAGNOSIS ACHIEVED BY WES. CASE REPORT

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Introdução

Renal hypodysplasia/aplasia tipo 3 (RHDA3) is an autosomal dominant disorder characterized by abnormal kidney development beginning in utero. The phenotype is highly variable, even within families, and there is evidence for incomplete penetrance. Some affected individuals have bilateral renal agenesis, which is usually fatal in utero or in the perinatal period, whereas others may have unilateral agenesis that is compatible with life, or milder manifestations, such as vesicoureteral reflux. Renal aplasia falls at the most severe end of the spectrum of Congenital Aomalies of the Kidney and Urinary Tract (CAKUT acronym).

Objectivos

We present a case which fetal ultrasound at 15 weeks, revealed kidney anomalies with oligoamnios

Metodologia

Chorionic villus biopsy was performed at 15 weeks, as well as Aneuploidy screening and Array-CGH analyses. Whole-exome-sequencing (WES) was performed and the bioinformatic analysis was focused on our targeted panel with 113 genes, related with renal anomalies – CAKUT Spectrum

Resultados e Conclusões

Aneuploidy screening and Array-CGH analysis revealed a normal result. WES identified a heterozygous variant in GREB1L gene: 556T>C (p.Cys186Arg), classified as a variant of uncertain significance, according to ACMG recommendations.

Since pathogenic variants in GREB1L gene are associated with dominant RHDA3 disorder, clinical reassessement of the fetus, as well as the parents study of the referred variant were performed.

The finding that the variant was been inherited "*the novo*", along with the clinical reassessement of the fetus, allowed us to reclassify the variant as probably pathogenic and to conclude it is compatible with RHDA3 disorder (OMIM# 617805).

Once again, WES technology revealed to be a highly useful tool to prenatal diagnostic.