Outros

(21616) - PRENATAL DIAGNOSIS OF NOONAN SYNDROME IN A FETUS WITH ABNORMAL NUCHAL TRANSLUCENCY. THE IMPORTANCE OF NGS GENETIC TESTING.

Natalia Salgueiro[1]; Lisandra Castro[1]; Ariana Conceição[1]; Marcia Cardoso[1]; Elsa Garcia[1]; Marta Moreira[1]; Fabiana Ramos[2]; Margarida Reis-Lima[1]

1 - Unidade de Genética Molecular e Genómica-SynlabHealth Genética Médica, Porto; 2 - Serviço de Genética, Hospital Pediátrico de Coimbra

Introdução

Noonan syndrome (OMIM#163950) rasopathy, is characterised by postnatal short stature, distinctive facial features, congenital heart defects, variable degree of developmental delay and other structural abnormalities. Prenatal features of rasopathies can include increased nuchal translucency (NT) and/or cystic hygroma, distended jugular lymph sacs (JLS), hydrops fetalis, polyhydramnios, pleural effusion, ascites, cardiac defects and renal anomalies. It has been previously estimated that mutations in rasopathy genes are found in 6.7%–19% of fetuses with increased NT and additional anomalies on ultrasound.

Objectivos

We present a clinical case where fetal ultrasound, revealed increased nuchal translucency (NT) (NT>P99), cystic hygroma and pleural effusion

Metodologia

Chorionic villus biopsy was performed and Whole-Exome-Sequencing (WES) requested. The bioinformatic analysis was focused on our targeted panel of 35 genes, associated with rasopathies.

Resultados e Conclusões

A heterozygous variant in PTPN11 gene, c.206A>T (p. Glu69Val) was detected by WES. This variant causes an amino acid change from Glu to Val at position 69. According to ACMG guidelines we classified this variant as Pathogenic. The clinical information and the variant found is compatible with autosomal dominant Noonan Syndrome (OMIM# 163950).

The use of an extended NGS panel of known rasopathy genes should be used when a rasopathy is suspected and will be very helpful in the definitive diagnosis of this pathology for orientation and family counselling.

Palavras-chave : SYNLAB