Outros

(21618) - KABUKI SYNDROME-1. WHOLE EXOME SEQUENCING (WES) - THE KEY IN A PRENATAL CASE DIAGNOSTIC.

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Introdução

Kabuki Syndrome is a rare, multisystem disorder and can vary greatly from one patient to another. It is characterized by distinctive facial features including, long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, and cleft or high-arched palate. Affected individuals have mild to severe developmental delay and intelectual disability. Other characteristics features include scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. We describe a case of an ongoing pregnancy where the ultrasound revealed abnormality of the face and semilobar. holoprosencephaly.

Objectivos

We present a clinical case of a 35-year-old pregnant women whose ultrasound revealed semilobar holoprosencephaly with severe facial anomaly.

Metodologia

At 13 weeks microrray-CGH and WES analysis were performed. Bioinformatic analysis was focused on our targeted 23 genes panel, related with holoprosencephaly and differential diagnoses.

Resultados e Conclusões

The array-CGH analysis revealed a negative result which led to WES analysis. A heterozygous variant in KMT2D gene, c.7066del (p.Gln2356fs) was detected by WES. This variant causes an abnormal/truncated KMT2D protein. According to ACMG guidelines we classified this variant as Pathogenic. The clinical information provided allowed us to conclude that this variant was compatible with autosomal dominant Kabuki syndrome, 1 (OMIM#147920). This result was given in time to, together with the ultrasound information propose TOP.

With this technology, necessary and of great value, we were able to solve prenatal diagnosis cases and affirm that WES is indeed a very powerful tool.

Palavras-chave : SYNLAB