

Prenatal diagnosis of craniosynostosis associated with previously undescribed heterozygous variants genes

Tamara Yarygina, Inna Klimova, Roman Shmakov

CASE PRESENTATION

39 y.o. patient G2P2 with breast cancer T4N0M0 first diagnosed in pregnancy was referred to a tertiary center at gestational age of 33+4weeks. The pregnancy course was uneventful until the time of referral. Fetal abnormalities did not reported at 1 and 2 trimester. US revealed AGA-fetus with abnormal skull shape suspected for scaphocephaly.

In 3D-skeleton mode in a coronary view revealed premature closer of sagittal suture with bony groove in the area of the synostotic sagittal suture.

The figure shows abnormal images of the skull of this fetus and normal scans for comparison. At 34+2 weeks, the patient developed spontaneous labor.

A live girl with birthweight 2690 g, Apgar score of 7/8, had wide nasal bridge, short nose, long filter, and an elongated head in the anteroposterior direction. CT scan confirmed craniosynostosis. The child underwent successful surgery at the age of three months

Based on the totality of the information, these results are assessed as a variant of uncertain clinical significance and require careful comparison with the clinical picture and analysis of the origin of the variant.

EXOME SEQUENCING did not reveal pathogenic or likely pathogenic variants related to the patient's phenotype; however, previously undescribed heterozygous variants were found in the CDC42BPB and RPS6KA3 genes, which are common in patients with Chilton-Okura-Chang syndrome, Coffin-Lowry syndrome, and in patients with X-linked intellectual disability

