TUBB2B

A range of cerebrocortical development malformations (MCD) ranging from simplified gyral patterns to the complete loss of gyri and sulci is associated with mutations in a cluster of highly homolog β-tublin genes, such as TUBB2A and TUBB2B.

CASE REPORT: The fetus had pachygyria, asymmetrical perisylvian polymicrogyria, dysplasia of the lateral sulcus and insula, agenesis of the splenium and partial agenesis of the body corpus callosum, cerebellar superior vermian hypoplasia with agenesis of the inferior vermis. Karyotype and microarray were normal. Trio Medical Exome Sequencing detected a de novo novel heterozygous mutation c.862G > A (p.E288K) in the tubulinpathy genes. Long-range PCR and Sanger sequencing specific for TUBB2A and TUBB2B gene detected a heterozygous variant c.862G > A specific to TUBB2B.

CONCLUSION: The combination of LR-PCR amplification and medical exome sequencing allows mutational assessment in tubulinopathy genes. Our study expands the spectrum of malformations associated with mutations in the β-tubulin gene TUBB2B [1)].

1)
Wang H, Li S, Li S, Jiang N, Guo J, Zhang W, Zhong M, Xie J. De Novo Mutated TUBB2B Associated Pachygyria Diagnosed by Medical Exome Sequencing and Long-Range PCR. Fetal Pediatr Pathol. 2018 Dec 26:1-9. doi: 10.1080/15513815.2018.1538273. [Epub ahead of print] PubMed PMID: 30585108.