WDR62

Autosomal recessive primary microcephaly (MCPH) is a group of rare neurodevelopmental diseases with severe microcephaly at birth. One type of the disorder, MCPH2, is caused by biallelic mutations in the WDR62 gene, which encodes the WD repeat-containing protein 62. Patients with WDR62 mutation may have a wide range of malformations of cortical development in addition to congenital microcephaly.

Zombor et al., describe two patients, a boy and a girl, with severe congenital microcephaly, global developmental delay, epilepsy, and failure to thrive. MRI showed hemispherical asymmetry, diffuse pachygyria, thick gray matter, indistinct gray-white matter junction, and corpus callosum and white matter hypoplasia. Whole exome sequencing revealed the same novel homozygous missense mutation, c.668T>C, p.Phe223Ser in exon 6 of the WDR62 gene. The healthy parents were heterozygous for this mutation. The mutation affects a highly conserved region in one of the WD repeats of the WDR62 protein. Haplotype analysis showed genetic relatedness between the families of the patients. Our findings expand the spectrum of mutations randomly distributed in the WDR62 gene. A review is also provided of the brain malformations described in WDR62 mutations in association with congenital microcephaly [1].

1)
Zombor M, Kalmár T, Nagy N, Berényi M, Telcs B, Maróti Z, Brandau O, Sztriha L. A novel WDR62 missense mutation in microcephaly with abnormal cortical architecture and review of the literature. J Appl Genet. 2019 Feb 1. doi: 10.1007/s13353-019-00486-y. [Epub ahead of print] PubMed PMID: 30706430.