

Familial Case of Pelizaeus-Merzbacher Disorder Detected by Oligoarray Comparative Genomic Hybridization: Genotype-to-Phenotype Diagnosis

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Abstract:

Introduction. Pelizaeus-Merzbacher disease (PMD) is an X-linked recessive hypomyelinating leukodystrophy characterized by nystagmus, spastic quadriplegia, ataxia, and developmental delay. It is caused by mutation in the PLP1 gene. **Case Description.** We report a 9-year-old boy referred for oligoarray comparative genomic hybridization (OA-CGH) because of intellectual delay, seizures, microcephaly, nystagmus, and spastic paraplegia. Similar clinical findings were reported in his older brother and maternal uncle. Both parents had normal phenotypes. OA-CGH was performed and a 436 Kb duplication was detected and the diagnosis of PMD was made. The mother was carrier of this 436 Kb duplication. **Conclusion.** Clinical presentation has been accepted as being the mainstay of diagnosis for most conditions. However, recent developments in genetic diagnosis have shown that, in many congenital and sporadic disorders lacking specific phenotypic manifestations, a genotype-to-phenotype approach can be conclusive. In this case, a diagnosis was reached by universal genomic testing, namely, whole genomic array.

Full text:

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