Abstract

Full text links

Genet Mol Res. 2014 Aug 15;13(3):6287-92. doi: 10.4238/2014.August.15.11.

Modulation of expressivity in PDGFRB-related infantile myofibromatosis: a role for PTPRG?

Linhares ND[1], Freire MC[2], Cardenas RG[1], Bahia M[3], Puzenat E[4], Aubin F[4], Pena SD[5].

Author information

Abstract

Infantile myofibromatosis is a rare genetic disorder characterized by the development of benign tumors in the skin, muscle, bone, and viscera. The molecular pathogenesis is still incompletely known. An autosomal dominant form had been reported as causally related with mutations in the gene for platelet-derived growth factor receptor beta (PDGFRB). We report here two siblings with infantile myofibromatosis and with a PDGFRB mutation identified by exome sequence analysis. However, the unaffected mother also had the same PDGFRB mutation. We showed that both children had also inherited from their healthy father a heterozygous mutation in the gene for receptor protein tyrosine phosphatase gamma (PTPRG), an enzyme known to dephosphorylate PDGFRB. We suggest that in this family, the additional mutation in PTPRG may explain the full phenotypic penetrance in the siblings affected, in comparison with the unaffected mother.

PMID: 25158255 [PubMed - indexed for MEDLINE] Free full text

Publication Types, MeSH Terms, Substances, Supplementary Concepts

LinkOut - more resources

PubMed Commons

PubMed Commons home

0 comments

How to join PubMed Commons