

Chitayat Syndrome (ERF Variant)

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Chitayat syndrome (CHYTS, MIM #617180) is a rare autosomal dominant clinical condition caused by a single missense pathogenic variant in the ERF gene (19q13.2, MIM*611888), which encodes the ETS2 Repressor Factor (ERF) protein. The characteristic features reported to date for this condition are facial dysmorphism, hyperphalangism and respiratory complications during the newborn period. Herein, we report the sixth patient worldwide with a confirmed molecular diagnosis of CHYTS. Our documentation of pectus carinatum, hypoplastic phalanges (as in two previously described patients), and lack of hyperphalangism broadens the phenotypic spectrum of CHYTS. Moreover, our identification of a heterozygous mutation [c.266A>G or p.(Tyr89Cys)] [rs886041001] in this patient provides further evidence that this condition is caused by a recurrent pathogenic variant in ERF.