Chitayat Syndrome (ERF Variant)

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Hand hyperphalangism leading to shortened index fingers with ulnar deviation, hallux valgus, mild facial dysmorphism and respiratory compromise requiring assisted ventilation are the key features of Chitayat syndrome. This condition results from the recurrent heterozygous missense variant NM_006494.2:c.266A>G; p.(Tyr89Cys) in ERF on chromosome 19q13.2, encoding the ETS2 repressor factor (ERF) protein. The pathomechanism of Chitayat syndrome is unknown. To date, seven individuals with Chitayat syndrome and the recurrent pathogenic ERF variant have been reported in the literature. Here, we describe six additional individuals, among them only one presenting with a history of assisted ventilation, and the remaining presenting with variable pulmonary phenotypes, including one individual without any obvious pulmonary manifestations. Our findings widen the phenotype spectrum caused by the recurrent pathogenic variant in ERF, underline Chitayat syndrome as a cause of isolated skeletal malformations and therefore contribute to the improvement of diagnostic strategies in individuals with hand hyperphalangism.