Ghosal Type Hematodiaphyseal Dysplasia- A rare and unusual cause of cytopenias

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Abstract

Ghosal Type Hematodiaphyseal Dysplasia (GHDD) is an autosomal recessive disorder characterized by metadiaphyseal dysplasia, defective hematopoiesis and steroid sensitive anemia. The diagnosis of this rare syndrome is important as early diagnosis helps in initiation of treatment leading to improvement in anemia and bony changes. Herein, we report two cases where the diagnosis was confirmed by mutation analysis. Treatment typically involves steroid therapy. Both our cases were started on steroids after which haemoglobin & other counts have normalized.They need very low dose steroids to maintain normal hemoglobin and counts.

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