

Case Report

Fibrodysplasia ossificans progressive: A case report

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Abstract

Fibrodysplasia ossificans progressiva (FOP) is an extremely rare autosomal dominant disorder having variable expressivity with complete penetrance. FOP incidence has been estimated to be 1 per 2 million. FOP caused by mutations in ACVR1 gene encoding bone morphogenetic protein type-1 receptor. To date, 15 types of mutations have been reported. The majority of cases were determined to be the result of a new mutation occurring sporadically. Here we report a 20 years old girl who's suffering FOP for 11 years.

Keywords: Fibrodysplasia ossificans progressiva, ACVR1, Autosomal dominant

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