Case Report

Desbuquois Syndrome Atypical Hands Subtype with No Mutation in FLNB Gene

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Abstract

Desbuquois syndrome is characterised by craniofacial dysmorphism, carpotarsal, vertebral and joint abnormalities; with associated developmental delay. It is genetically heterogeneous and has been subdivided into two groups, the "typical hands" and "atypical hands" groups. We hypothesised that the "atypical hands" subgroup may be due to Filamin B (FLNB) mutations, and the objective of this study is to evaluate if mutation in FLNB gene is associated with this clinical subgroup. After sequencing the whole FLNB gene, we report that no mutation was found in our case of Desbuquois syndrome.

Keyword: Atypical hands; Desbuquois syndrome; FLNB gene; Skeletal dysplasia