Case Report

Thanatophoric Dysplasia Type 1 (TD1) without "Telephone Receivers"

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Abstract

Thanatophoric dysplasia (TD) is a relatively common neonatal lethal skeletal dysplasia. It is subdivided into type 1 (TD1) and type 2 (TD2). By definition, TD1 is differentiated from TD2 by bowed femori, the so called "telephone receivers". TD2 is distinguished by straight femori and cloverleaf skull deformity. We described a case of TD1 with atypical feature of almost straight femori, and was found to have characteristic TD1 missense mutation C746G in FGFR3 gene.

Keyword: Bowed femori; FGFR3 gene; TD1; Thanatophoric dysplasia