Case Report

Thanatophoric Dysplasia Variant, San Diego Type in a Chinese Fetus, Caused by C746G Missense Mutation in \textit{FGFR3} Gene

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Abstract

Thanatophoric dysplasia variant - San Diego type (TD-SD) is a variant of thanatophoric dysplasia (TD), sharing with TD subtype TD1, common mutations in the \textit{FGFR3} gene. We describe the first local case of TD-SD with missense mutation C746G in \textit{FGFR3} gene. Radiologically, major features consist of macrocephaly, wafer thin vertebral bodies, and metaphyseal flaring of tubular bones that differentiate from other types of thanatophoric dysplasia.

\textbf{Keyword :} \textit{FGFR3} gene; Platyspondyly lethal skeletal dysplasia; San Diego type; Thanatophoric dysplasia