

Open Peer Review on Qeios

## Thanatophoric Dysplasia

National Cancer Institute

## Source

National Cancer Institute. <u>Thanatophoric Dysplasia</u>. NCI Thesaurus. Code C85187.

A severe autosomal dominant inherited disorder caused by mutations in the FGFR3 gene. It is characterized by multiple skeletal abnormalities, including extremely short limbs. It results in the death of the neonate.

Qeios ID: 4UWR71 · https://doi.org/10.32388/4UWR71