

Open Peer Review on Qeios

## Camurati-Engelmann Syndrome

National Cancer Institute

## Source

National Cancer Institute. <u>Camurati-Engelmann Syndrome</u>. NCI Thesaurus. Code C84610.

An autosomal dominant skeletal disorder caused by mutations in the TGFB1 gene. It is characterized by thickening of the bones, particularly the long bones of the extremities. It is associated with muscle weakness and tiredness.

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