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Short Stature Homeobox Deficiency

National Cancer Institute

Source

National Cancer Institute. *Short Stature Homeobox Deficiency*. NCI Thesaurus. Code C120113.

A rare genetic deficiency characterized by mutations in the SHOX gene and reduced expression or function of the SHOX protein. It results in the disruption of normal bone development and growth starting before birth. It manifests with skeletal abnormalities and short stature.