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NEK9-related lethal skeletal dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. [NEK9-related lethal skeletal dysplasia](#). ORPHA:464366

NEK9-related lethal skeletal dysplasia is a rare, lethal, primary bone dysplasia characterized by fetal akinesia, multiple contractures, shortening of all long bones, short, broad ribs, narrow chest and thorax, pulmonary hypoplasia and a protruding abdomen. Short bowed femurs may also be associated.