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X-linked lethal multiple pterygium syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked lethal multiple pterygium syndrome. ORPHA:79447

X-linked lethal multiple pterygium syndrome is a rare, genetic, developmental defect during embryogenesis characterized by the typical lethal multiple pterygium syndrome presentation (comprising of multiple pterygia, severe arthrogryposis, cleft palate, cystic hygromata and/or fetal hydrops, skeletal abnormalities and fetal death in the 2nd or 3rd trimester) with an X-linked pattern of inheritance.