Atelosteogenesis type II

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Atelosteogenesis type II. ORPHA:56304

Atelosteogenesis II is a lethal perinatal bone dysplasia characterized by limb shortening, normal sized skull with cleft palate, hitchhiker thumbs, distinctive facial dysmorphism and radiographic skeletal features, caused by mutations in the diastrophic dysplasia sulfate transporter gene.