Pyle disease

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pyle disease. ORPHA:3005

Pyle disease is a bone dysplasia characterised by genu valgum, metaphyseal anomalies with broadening of the long bones extending into the diaphyses and giving the femora and tibiae an 'Erlenmeyer flask'' appearance, widening of the ribs and clavicles, platyspondyly and cortical thinning.