X-linked dominant chondrodysplasia punctata

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. X-linked dominant chondrodysplasia punctata. ORPHA:35173

X-linked dominant chondrodysplasia punctata (CDPX2) is a rare genodermatosis with great phenotypic variation and characterized most commonly by ichthyosis, chondrodysplasia punctata (CDP), asymmetric shortening of the limbs, cataracts and short stature.