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Chondrodysplasia punctata, Toriello type

INSERM

Source

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

Chondrodysplasia punctata, Toriello type. ORPHA:79347

Chondrodysplasia punctata, Toriello type is a rare, non-rhizomelic, primary bone dysplasia syndrome characterized by calcific stippling of epiphyses in association with minor facial abnormalities, short stature and ocular colobomata. In addition, patients present chondrodysplasia punctata, brachycephaly, flat facial profile with small nose, flat lower eyelids and low-set ears, developmental delay, brachytelephalangy and deep palmar creases. Complex congenital cardiac disease and central nervous system anomalies (including partial absence of corpus callosum, small vermis, enlargement of the cisterna magna and/or of the anterior horns of the lateral ventricles) have been reported.