Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Toriello-Higgins-Miller syndrome
Zespół Toriello, Higginsa i Millera

Kod ORPHA
79347

Kod OMIM
215105

Kod ICD10
Q77.3

Kod ICD11
LD24.04

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*Źródło

orphanet