

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

Kod ORPHA	Kod OMIM	Kod ICD10
79347	215105	Q77.3

Kod ICD11
LD24.04

*Źródło

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