Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay and intellectual disability, progressive spondyloepimetaphyseal dysplasia, short stature, short fourth metatarsals, and dysmorphic craniofacial features (including microcephaly, hypertelorism, epicanthal folds, mild ptosis, strabismus, malar hypoplasia, short nose, depressed nasal bridge, full lips, small, low-set ears, and short neck). Craniosynostosis, generalized hypotonia, as well as asymmetry of the cerebral hemispheres and mild thinning of the corpus callosum on brain imaging have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
457395

Kod OMIM
616723

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet