Dysplazja typu Silvermana i Handmakera
Kod Orpha: 1865 Kod OMIM: 224410

Opis choroby *

Definicja
Dyssegmental dysplasia, Silverman-Handmaker type is a rare, genetic, primary bone dysplasia disorder, and lethal form of neonatal short-limbed dwarfism, characterized by anisospondyly, severe short stature and limb shortening, metaphyseal flaring and distinct dysmorphic features (i.e. flat facial appearance, abnormal ears, short neck, narrow thorax). Additional features may include other skeletal findings (e.g. joint contractures, bowed limbs, talipes equinovarus) and urogenital and cardiovascular abnormalities.
Dane

Klasyfikacja
Choroba

Kod ORPHA
1865

Kod OMIM
224410

Kod ICD10
Q77.7

Kod ICD11
LD24.3

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.