

Opis choroby *

Definicja

Cono-spondylar dysplasia is a rare genetic primary bone dysplasia disorder characterized by early-onset severe lumbar kyphosis, marked brachydactyly and irregular, pronounced cone epiphyses of the metacarpals and phalanges. Additional reported features include developmental delay, intellectual disability, hypotonia, epileptic seizures and mild facial dysmorphism (incl. long and thin or square-shaped face, slight mid-face hypoplasia, hypertelorism, epicanthic folds, low-set ears, anteverted nostrils). Radiographic findings also reveal hypoplasia of iliac wings and anterior defect of vertebral bodies.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Short stature-kyphosis-hypoplasia of basal ilia-cone epiphyses-facial dysmorphism syndrome
Zespół niskiego wzrostu, kifozy, hipoplazji nasady kości biodrowej, stożkowatych nasad i dysmorfii twarzy

Kod ORPHA

420794

Kod OMIM

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Kod ICD10

Q77.7

Kod ICD11

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[*Źródło](#)

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