

Opis choroby *

Definicja

Pelvic dysplasia-arthrogryposis of lower limbs syndrome is a rare, genetic, dysostosis syndrome characterized by intrauterine growth restriction, short stature (with short lower segment), lower limb joint contractures and muscular hypotrophy, narrow, small pelvis, lumbar hyperlordosis with scoliosis, and foot deformity (short, overlapping toes). Imaging reveals ovoid/wedge-shaped vertebral bodies, pelvic and skeletal hypoplasia with metatarsal fusion in the lower limbs, and normal skull and upper limbs.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Ray-Peterson-Scott syndrome
Zespół Ray'a, Petersona i Scotta

Kod ORPHA

2840

Kod OMIM

602484

Kod ICD10

Q68.8

Kod ICD11

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*Źródło

orphanet