

Opis choroby *

Definicja

Schinzel phocomelia syndrome, also called limb/pelvis hypoplasia/aplasia syndrome, is characterized by skeletal malformations affecting the ulnae, pelvic bones, fibulae and femora. As the phenotype is similar to that described in the malformation syndrome known as Al-Awadi/Raas-Rothschild syndrome, they are thought to be the same disorder.

Dane

Klasyfikacja

Zespół wad wrodzonych Al Awadi-Raas-Rothschild syndrome

Synonimy

Aplazja/hipoplazja kończyn i miednicy

Ciężki ubytek kończyn

Wrodzony brak kości łokciowej i strzałkowej

Zespół Al Awadi, Raasa i Rothschilda

Aplasia/hypoplasia of limbs and pelvis

Congenital absence of ulna and fibula

Severe limb deficit

Kod ORPHA

2879

Kod OMIM

276820

Kod ICD10

Q87.2

Kod ICD11

LD2F.1Y

*Źródło

orphanet