

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

Synonimy

Al Awadi-Raas-Rothschild syndrome
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