

## **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

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