Opis choroby *

Definicja

An extremely rare congenital limb malformation syndrome, described in only 3 patients to date,characterized by the association of hypoplasia or aplasia of the hand and foot phalanges, hemivertebrae and various urogenital and/or intestinal abnormalities (i.e. dysgenesis of the urogenital tract and rectum). There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Johnson-Munson syndrome
Zespół Johnsona i Munsona

Kod ORPHA
1112

Kod OMIM
207620

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet