

Opis choroby *

Definicja

A spectrum of congenital malformative disorders characterized by the co-occurrence of distal limb anomalies (usually bilateral cleft feet and/or hands) and renal defects (e.g. unilateral or bilateral agenesis), that can be associated with a variety of other anomalies such as those of genitourinary tract (genital anomalies, ureteral hypoplasias, vesicoureteral reflux), abdominal wall defects, intestinal atresias, and lung malformations. Familial cases have been reported in which an autosomal recessive inheritance was suspected.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

971

Kod OMIM

201310

Kod ICD10

Q87.2

Kod ICD11

LD2F.1Y

*Źródło

orphanet