Opis choroby *

Definicja

A rare multiple congenital anomalies syndrome characterised by holoprosencephaly, predominantly radial limb deficiency (absent thumbs, phocomelia), heart defects, kidney malformations and absence of gallbladder. Variable manifestations include vertebral anomalies, cleft lip/palate, microphthalmia, absent nose, dysplastic ears, hearing loss, colobomas of the iris and retina and/or bifid uvula.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Steinfeld syndrome
Zespół Steinfelda

Kod ORPHA
3186

Kod OMIM
184705

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet