

Opis choroby *

Definicja

A rare, genetic multiple congenital anomalies syndrome characterized by atrioventricular septal defects and blepharophimosis, in addition to radial (e.g. aplastic radius, shortened ulna, fifth finger clinodactyly, absent first metacarpal and thumb) and anal (e.g. imperforate or anteriorly place anus, rectovaginal fistula) defects.

Dane

Klasyfikacja

Zespół wad wrodzonych Houlston-Ironton-Temple syndrome
Zespół Houlstona, Irontona i Temple'a

Kod ORPHA

1352

Kod OMIM

600123

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet