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

Synonimy
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