

Opis choroby *

Definicja

Lowe-Kohn-Cohen syndrome is an extremely rare anorectal malformation syndrome characterized by imperforate anus, closed ano-perineal fistula, preauricular skin tag and absent renal abnormalities and pre-axial limb deformities. There have been no further descriptions in the literature since 1983.

Dane

Klasyfikacja Synonimy

Zespół wad wrodzonych Deafness-nephritis-ano-rectal malformation syndrome
Głuchota - zapalenie nerek - malformacja odbytu i odbytnicy
Hearing loss-nephritis-ano-rectal malformation syndrome

Kod ORPHA
2408

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
LD2H.Y

*Źródło

orphanet