

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

Zespół wad wrodzonych

Synonimy

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