Opis choroby *

Definicja

A rare renal disease characterized by the association of X-linked Alport syndrome (glomerular nephropathy, sensorineural deafness and ocular anomalies) and benign proliferation of visceral smooth muscle cells along the gastrointestinal, respiratory, and female genital tracts and clinically manifests with dysphagia, dyspnea, cough, stridor, postprandial vomiting, retrosternal or epigastric pain, recurrent pneumonia, and clitoral hypertrophy in females.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
Xq22.3 microdeletion syndrome
Zespół mikrodelecji Xq22.3

Kod ORPHA
1018

Kod OMIM
308940

Kod ICD10
Q87.8

Kod ICD11
LD2H.Y

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*Źródło

orphanet