

## **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

Klasifikacja	Synonimy
Podtyp kliniczny	Xq22.3 microdeletion syndrome
	Zespół mikrodelecji Xq22.3
<b>Kod ORPHA</b>	<b>Kod OMIM</b>
1018	308940
<b>Kod ICD11</b>	<b>Kod ICD10</b>
LD2H.Y	Q87.8

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### \* Źródło

orphanet