DISEASE:
X-linked Alport syndrome-diffuse leiomyomatosis

NAME:	X-linked Alport syndrome-diffuse leiomyomatosis
DESCRIPTION:	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.
ORPHACODE:	1018
SYNONYMS:	Xq22.3 microdeletion syndrome
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	COL4A6 COL4A5
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RELATED CONTENT

Related Genetic Tests

- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- collagen type IV alpha 5 chain
- collagen type IV alpha 6 chain

Related Gene Panels

- Alport (X-linked and recessive) (3 genes) - UZA
- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG