DISEASE:
Xq21 microdeletion syndrome

NAME:	Xq21 microdeletion syndrome
DESCRIPTION:	An X-linked retinal dystrophy characterized by choroideremia, causing in affected males progressive nyctalopia and eventual central blindness. Obesity, moderate intellectual disability and congenital mixed (sensorineural and conductive) deafness are also observed. Female carriers show typical retinal changes indicative of the choroideremia carrier state.
ORPHACODE:	1435
SYNONYMS:	Ayazi syndrome Del(X)(q21) Monosomy Xq21
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	POU3F4
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Deafness, X-linked

Related Laboratories

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

Related Analytes

- POU class 3 homeobox 4