

[Open Peer Review on Qeios](#)

Xq21 microdeletion syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. [Xq21 microdeletion syndrome](#). ORPHA:1435

Choroideremia-deafness-obesity syndrome is 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.