Choroideremia

Choroideremia (CHM) is a slowly progressive, X-linked (heritable) retinal disorder. The disorder is rare, occurring in 1 in 50,000 individuals in the general population. Affected males develop night blindness in their early years and gradually lose peripheral vision which results in legal blindness by age 40.

9 year old male: the pigment layer has a “salt and pepper” appearance and is more transparent (thinned) allowing one to see the deep blood vessels of the choroid.
13 year old male: the retinal pigment epithelial layer is patchy, probably thinned, and lacking pigment (arrow)
18 year old male: The pigment layer is now very thin and possibly absent (black arrow) except in the central area called the macula (white arrow), which remains healthy or intact. The overlying retina remains but is transparent.
26 year old male: the sclera (whitish) is visible with deep choroidal blood vessels (white arrow) in the areas outside the central macula (black arrow). The visual field is compromised with shaded areas representing parts where the patient is not seeing well.
Retinal findings in Retinitis pigmentosa (RP) differ from CHM

- Optic nerve is much paler than CHM.
- Blood vessels are thinner than in CHM.
- Areas of pigment (also termed bone spicules) are seen in the retina in RP more commonly.
- Atrophy (loss of structure) of the retinal pigment epithelium under the macula is seen in this case of RP.
Female carriers of choroideremia

- In general female carriers experience a milder form of choroideremia.
- Some carriers experience a decline in visual function with difficulty seeing at night after age 50.
- In rare cases, female carriers may have symptoms similar to an affected male.
Female carrier with signs in the eye

2-D scan of the retina of a choroideremia carrier showing “remodeling” of the retina and central thinning of the retina (Right image, left eye, arrow)
Fundus of female carrier (left) and autofluorescent image of the same eye (right). Note speckling of the pigment layer (arrow).

These two images have been created by combining multiple photos of the eye at slightly different positions.