Choroideremia (CHM)

- A slowly progressive inherited retinal dystrophy.
- Rare, prevalence is estimated to 1/50,000 in the general population.
- Caused by CHM gene loss-of-function mutations.
- Inherited in an X-linked recessive pattern, thus mainly affects males.
- Account for approximately 4 percent of all blindness.

**A Classic Course**

Night blindness in early childhood

A progressive narrowing of the visual field follows

A huge decrease of visual acuity at the late stage

Blindness in late adulthood (60~70y)
Fundus of an Early Stage CHM male

- Retinal pigment epithelial cell changes ➔ Retinal hypo- or hyperpigmentation ➔ “salt and pepper” appearance (black arrows)
- The pigment layer is more transparent, allowing one to see the deep blood vessels of the choroid (white arrow)
The pigment layer is possibly absent in the peripheral (black arrow).
The pigment layer is only preserved in the central area called the macula (within the white circle).
Late Stage

- Exposure of the white sclera, “white-out” appearance.
- Extensive choriocapillaris atrophy.
Female carriers of choroideremia

- Female carriers can experience a milder form of Choroideremia.
- Decline of visual function with difficulty seeing at night generally occurs after age of 50.
- In rare cases, female carriers may exhibit severe symptoms similar to those of affected males.
Female carrier with signs in the eye

OCT scan of the retina of a CHM female carrier showing central thinning of the retina (white arrow)
Fundus of female carrier (left) and autofluorescent image of the same eye (right). Note areas of speckling of the pigment layer representing its deterioration (arrow)
# The Differential Diagnosis of Choroideremia

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<th>Retinitis Pigmentosa (RP)</th>
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Retinal findings in RP that differ from CHM (Later stage)

- The optic nerve can be much paler, termed as waxy appearance. (White arrows)
- The blood vessels, especially the arterioles, can be thinner. (Black arrows)
- Areas of pigment, termed as bone spicules, are more commonly seen. (Yellow arrows)
- Cystoid Macular Edema (CME) is more common.
Clinically diagnosed with CHM?
Free genetic testing is available
www.curechm.org/resources for details.

Other genetic testing resources:
www.eyewant2know.com
Additional Resources

• **Choroideremia Research Foundation:**
curechm.org

• **Foundation Fighting Blindness:**
USA: www.blindness.org
Canada: www.ffb.ca

• **Choroideremia patient survey:**
English: http://choroideremiasurvey.questionpro.com/
French: http://choroideremieenquete.questionpro.com/

• **Join the Choroideremia Patient Registry:**
https://www.curechm.org/research/join-patient-registry
Contact Us

For questions or comments about this presentation please contact us at:

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For information regarding care options for you and your family, please contact your ophthalmologist.