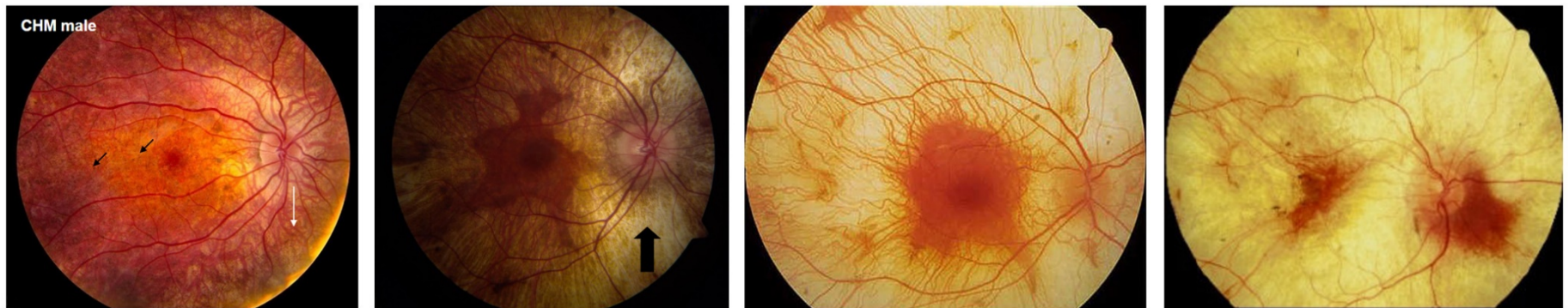


# Choroideremia (CHM)

- 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



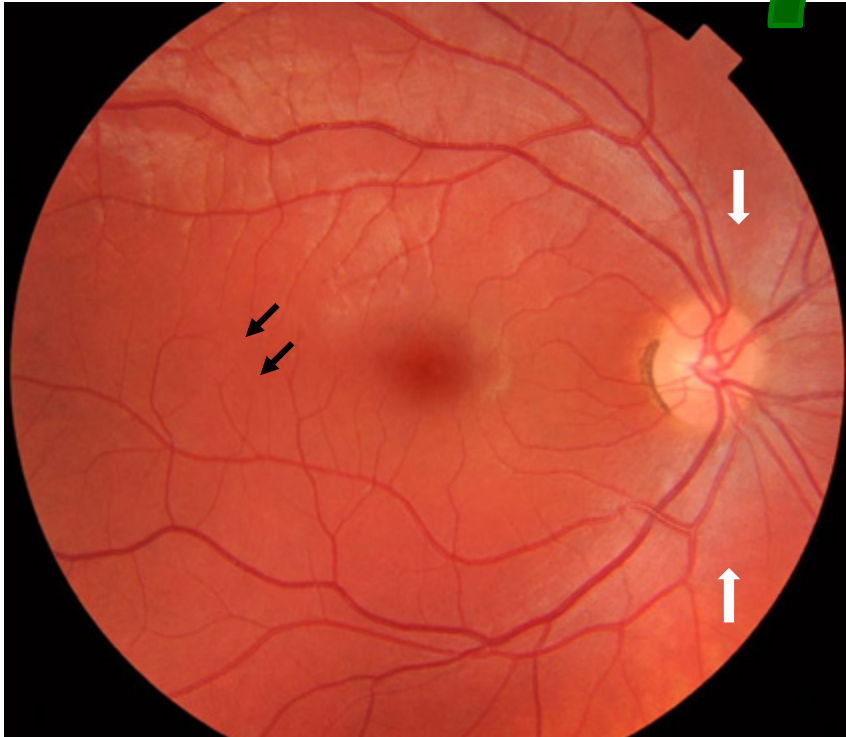
Night  
blindness in  
early  
childhood

A progressive  
narrowing of  
the visual field  
follows

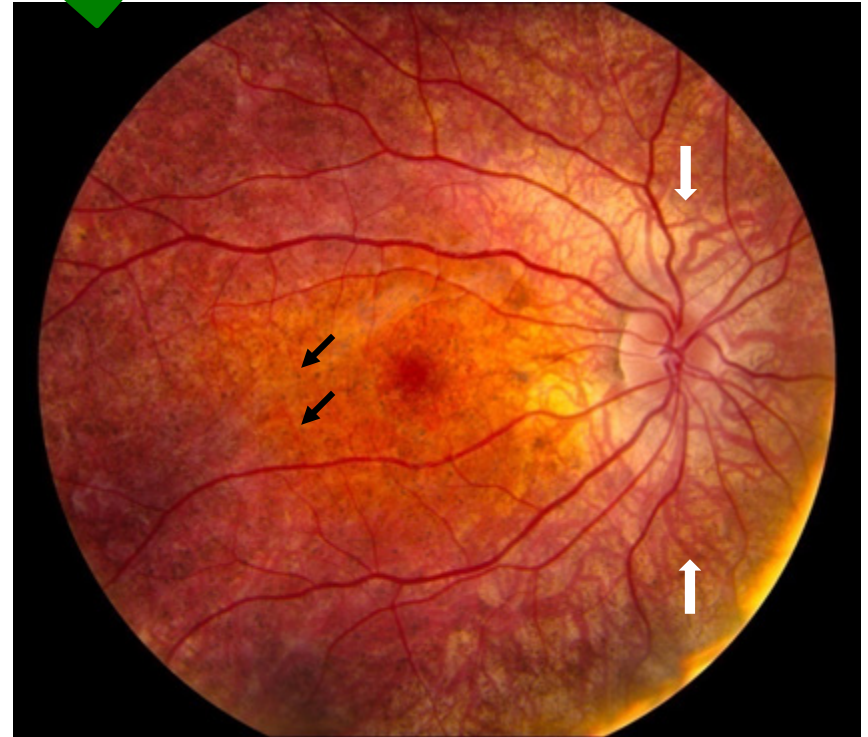
A huge  
decrease of  
visual acuity at  
the late stage

Blindness in  
late adulthood  
(60~70y)

# Early Stage



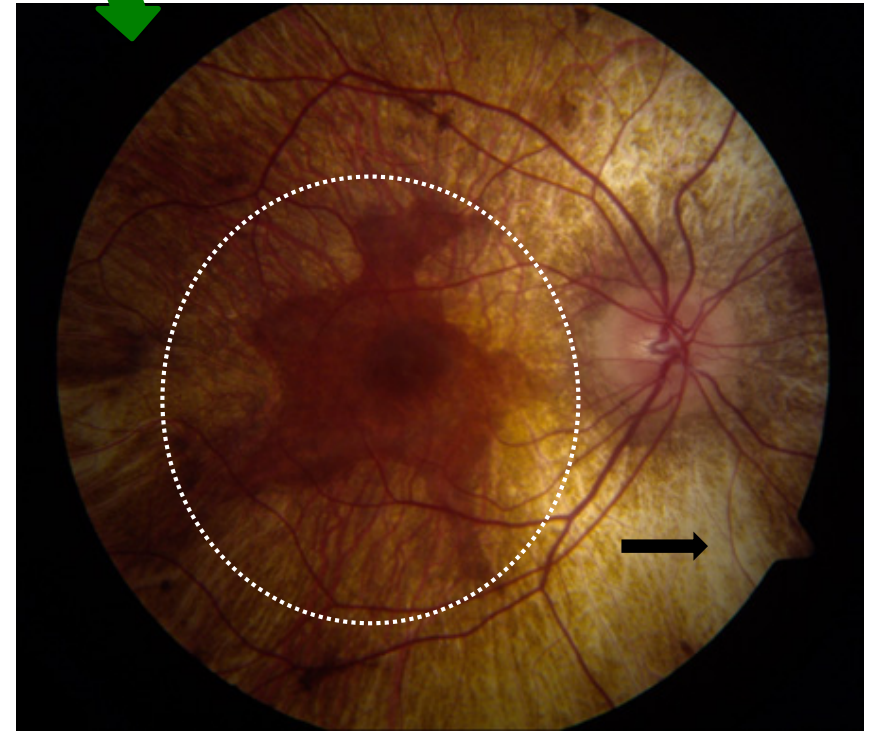
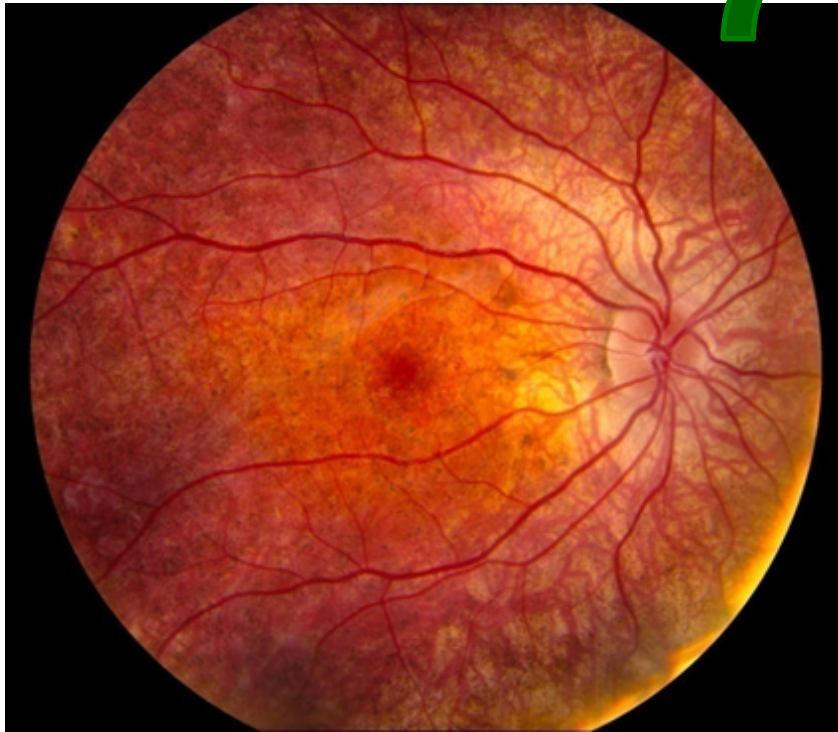
Normal Fundus



Fundus of an Early Stage CHM male

- Retinal pigment epithelial cell changes  $\Rightarrow$  Retinal hypo- or hyperpigmentation  $\Rightarrow$  "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)

# Mid Stage



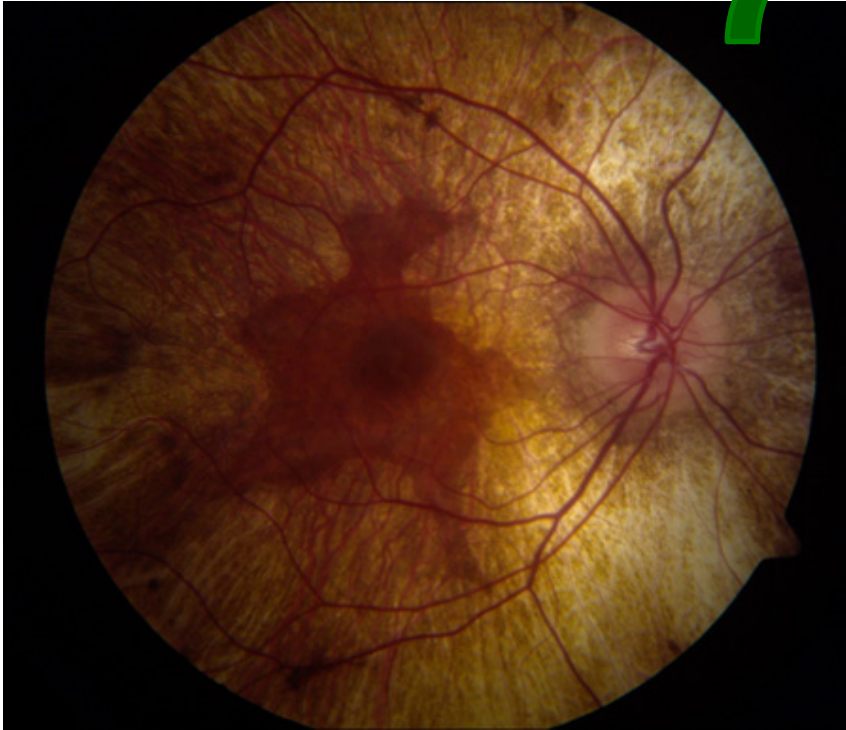
Fundus of an Early Stage CHM Male

Fundus of a Mid Stage CHM Male

- 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



Fundus of a Mid Stage CHM Male



Fundus of a Late Stage CHM Male

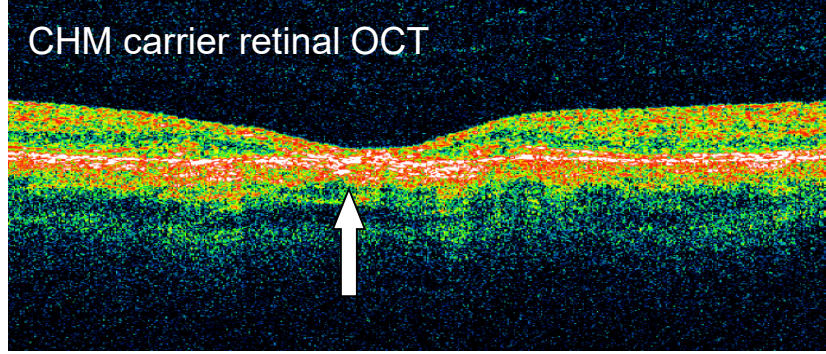
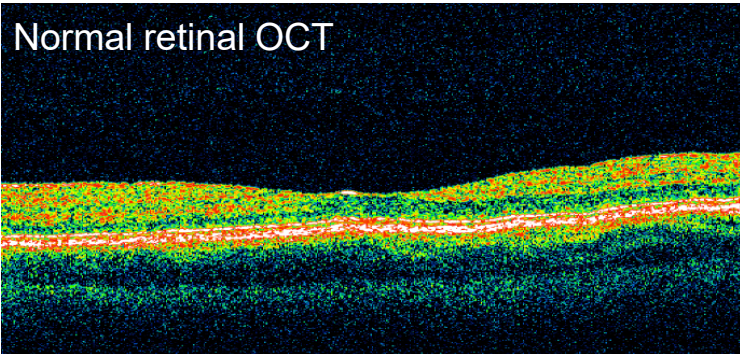
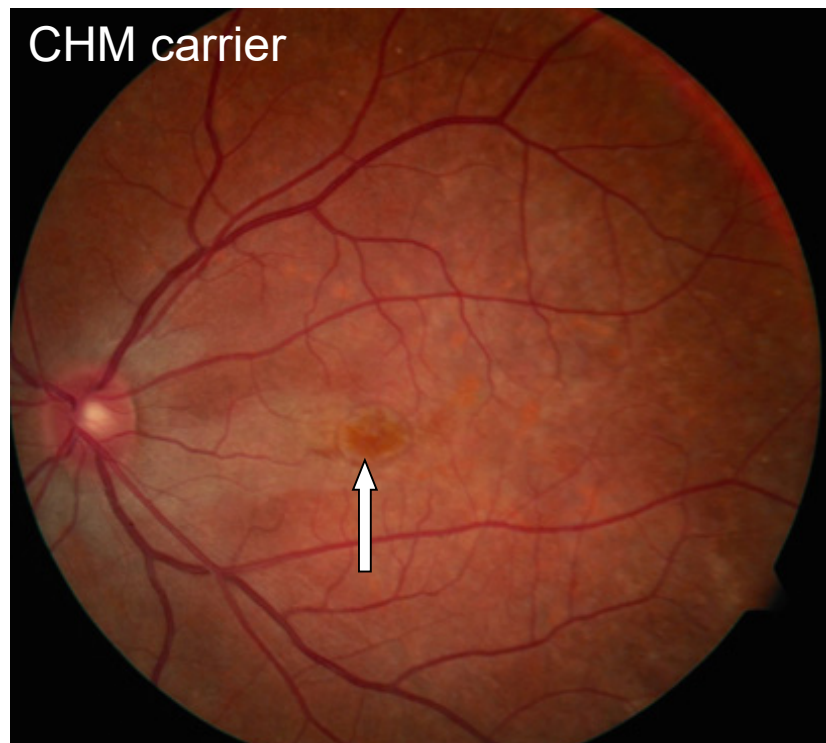
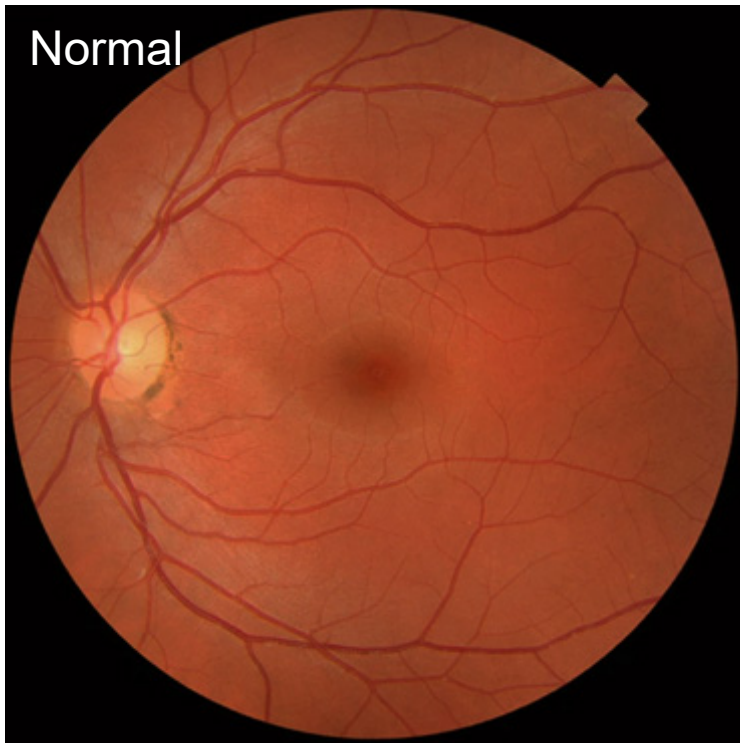
- Exposure of the white sclera, “white-out” appearance.
- Extensive choriocapillaris atrophy.

## **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

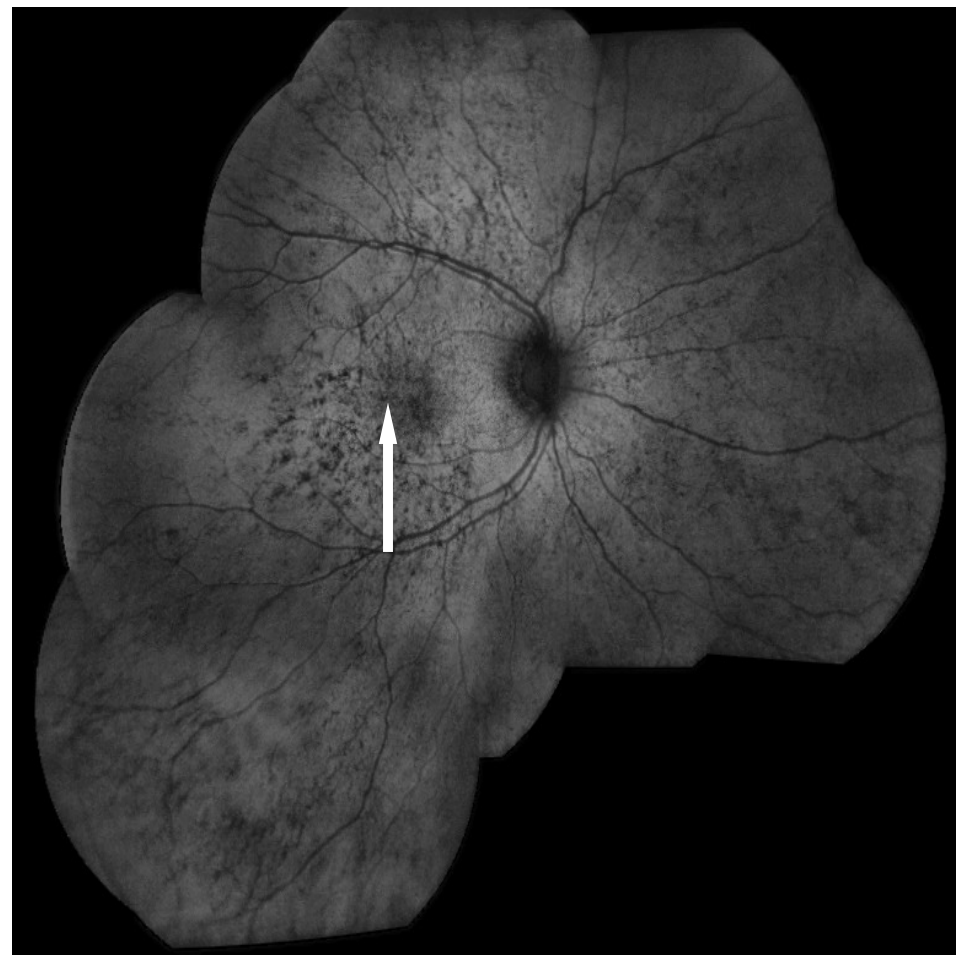


**OCT scan of the retina of a CHM female carrier showing central thinning of the retina (white arrow)**





**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

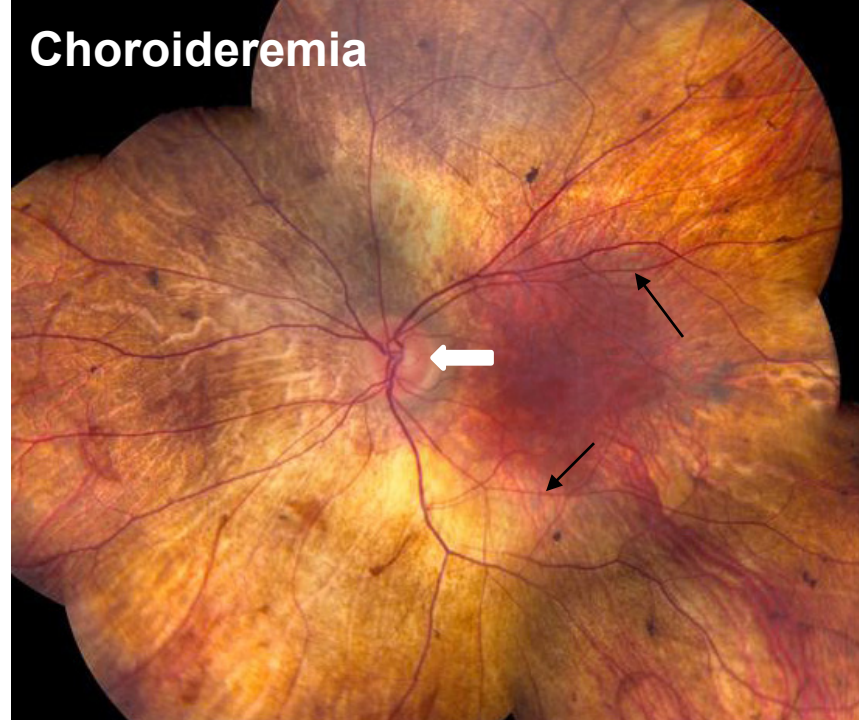
	<b>Choroideremia (CHM)</b>	<b>Retinitis Pigmentosa (RP)</b>
<b>Inheritance Pattern</b>	X-linked recessive	Autosomal dominant /Autosomal recessive/X-linked
<b>Cause</b>	CHM gene mutations	Mutations in more than 60 genes
<b>Gender</b>	Mainly affects males	Both males and females
<b>Symptoms</b>	Night blindness Visual field constriction (tunnel vision) Decreased visual acuity Photophobia	Night blindness Visual field constriction (tunnel vision) Decreased visual acuity Photophobia
<b>ERG finding</b>	Early stage: Decreased Scotopic response Late stage: Non-recordable	Early stage: Decreased Scotopic response Late stage: Non-recordable
<b>OCT finding</b>	Retinal thinning Loss of RPE and photoreceptors Preservation of the inner retinal layers Retinal tubulations	Retinal thinning Loss of RPE and photoreceptors Preservation of the inner retinal layers Retinal tubulations



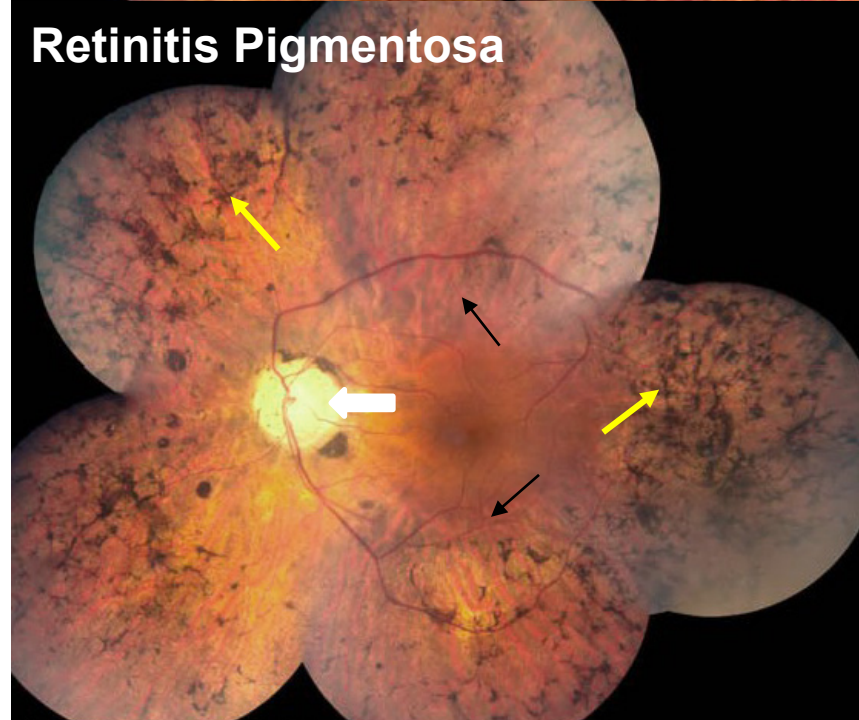
## Retinal findings in RP that differ from CHM (Later stage)

- 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.

Choroideremia



Retinitis Pigmentosa



## **Clinically diagnosed with CHM?**

Free genetic testing is available

[www.curechm.org/resources](http://www.curechm.org/resources) for details.

## **Other genetic testing resources:**

[www.eyewant2know.com](http://www.eyewant2know.com)

## Additional Resources

- **Choroideremia Research Foundation:**

[curechm.org](http://curechm.org)

- **Foundation Fighting Blindness:**

USA: [www.blindness.org](http://www.blindness.org)

Canada: [www.ffb.ca](http://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.