Choroideremia- New findings could improve therapies development and efficacy.

New NEI study combined adaptive optics with indocyanine green dye to visualize the in vivo pathogenesis of choroideremia in the living human eye. This study viewed live cells in the retina layers of Choroideremia affected males and female carriers- light-sensing photoreceptors, retinal pigment epithelium (RPE), and choroidal blood vessels.

Findings: **RPE cells are dramatically enlarged** (as much as five-fold) in males and females with choroideremia.

greater disruption to the RPE was found in the fovea, the last area to be affected in choroideremia, than in either the photoreceptor or choriocapillaris layers. This finding suggests that **RPE disruption plays an important role in choroideremia**

The unexpected finding of patches of photoreceptors that were fluorescently labeled, but structurally and functionally normal, suggests that the **RPE blood barrier function** may be altered in choroideremia.

Enlarged RPE cells can be detected even when using only a **conventional ophthalmic imaging instrumentation.** This could prove valuable in identifying which patients would benefit the most from therapeutic interventions**.**

Link: https://www.nature.com/articles/s42003-022-03842-7

Aguilera, N., Liu, T., Bower, A.J. *et al.* Widespread subclinical cellular changes revealed across a neural-epithelial-vascular complex in choroideremia using adaptive optics. *Commun Biol* **5**, 893 (2022). https://doi.org/10.1038/s42003-022-03842-7

Enlarged retinal pigment epithelial (RPE) cells observed in choroideremia