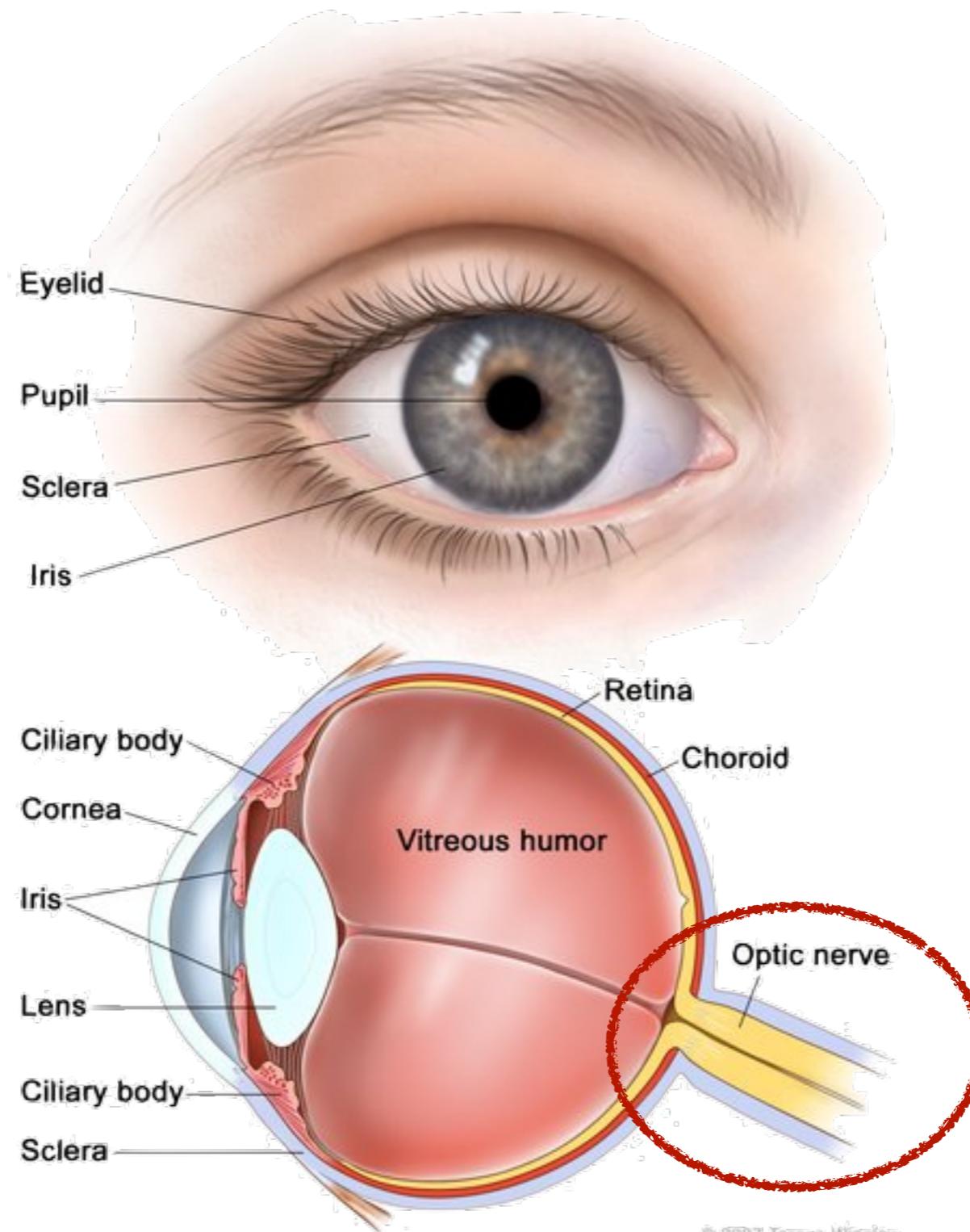
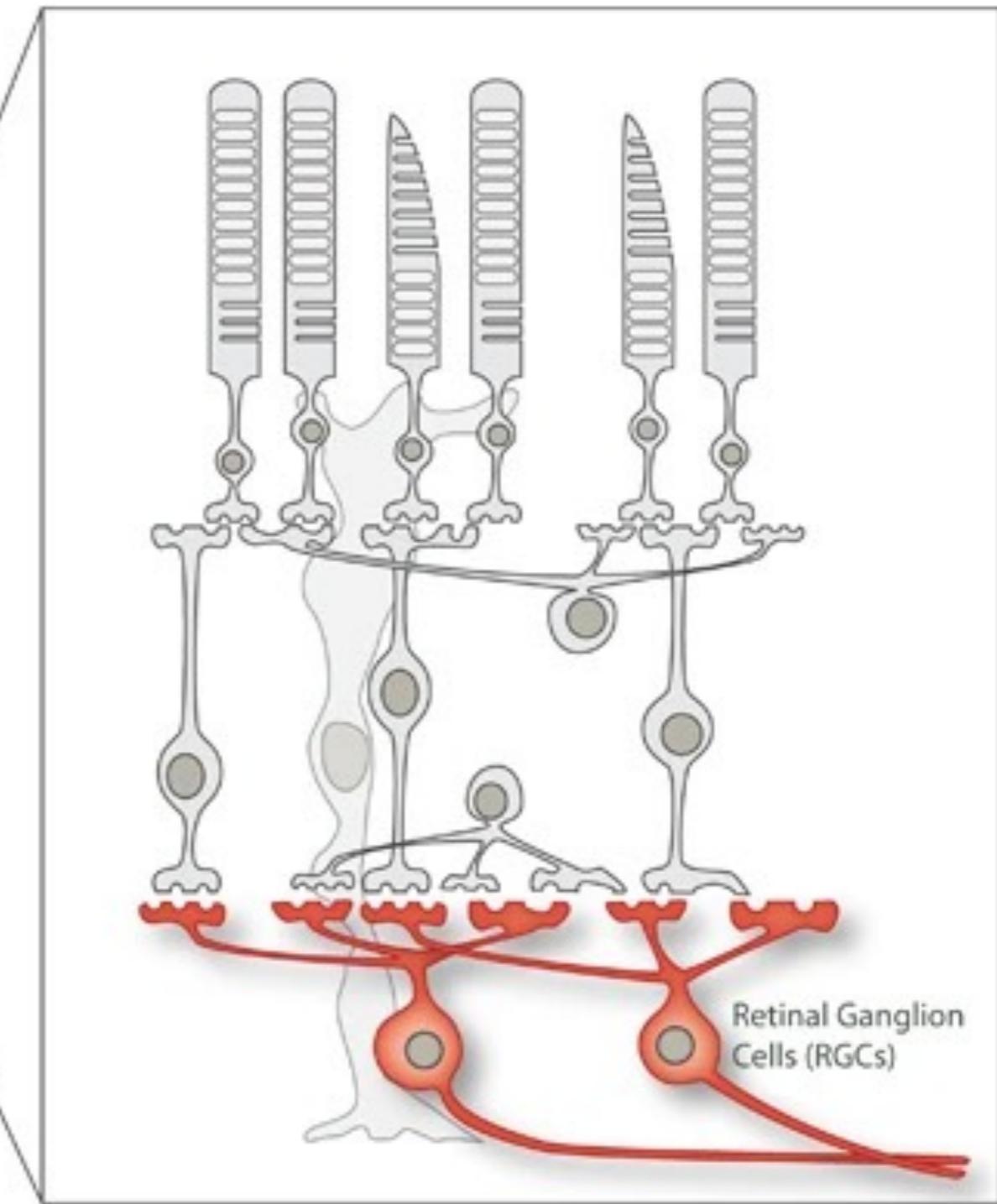
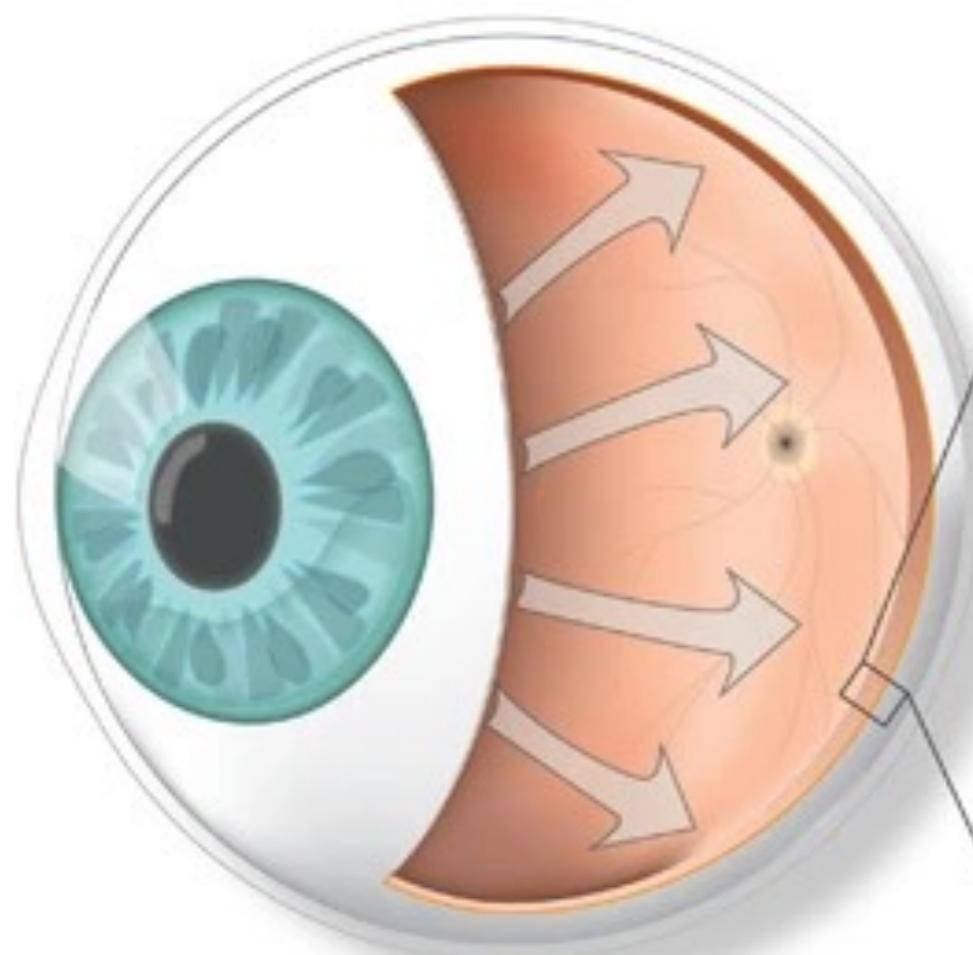


OPA 1
&
OPTIC ATROPHY TYPE 1

What is Optic Atrophy Type 1?



What cause Optic Atrophy Type 1?



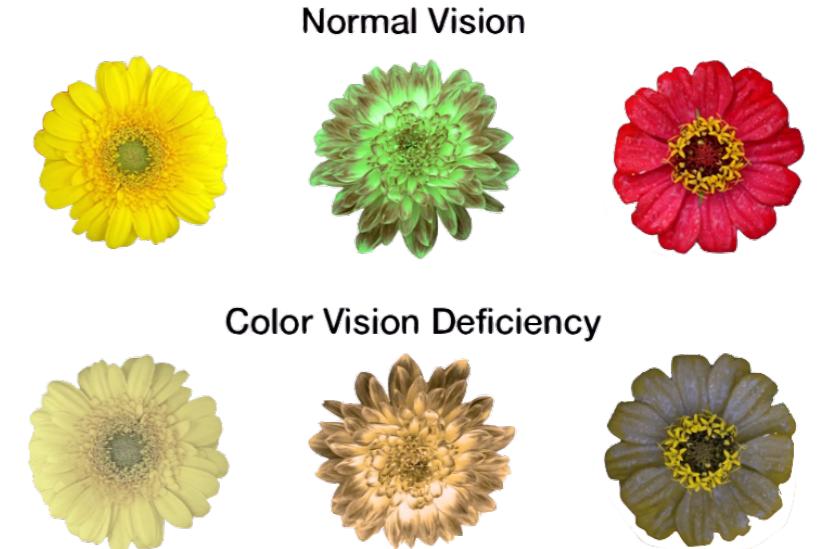
Visuals of Individuals with Optic Atrophy Type 1



Blurred



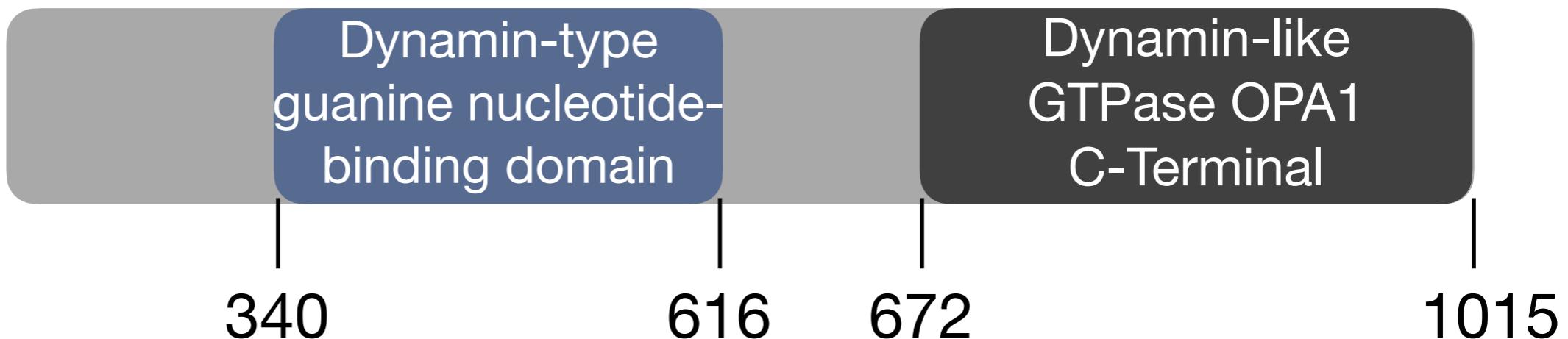
Blocked



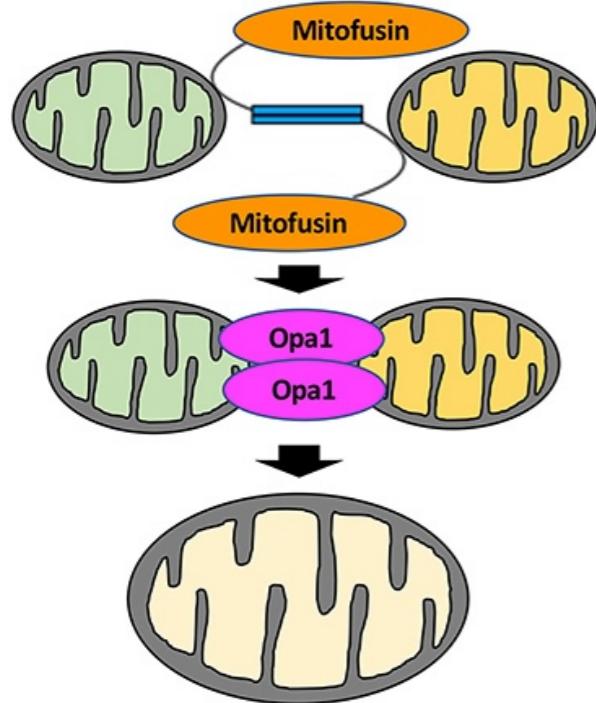
Color vision
deficiency

Domain & Gene Ontology

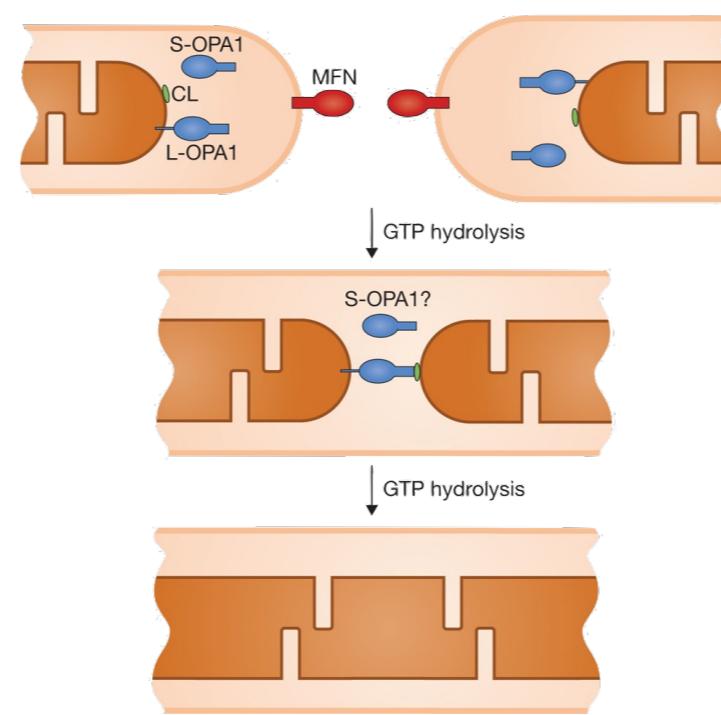
Human



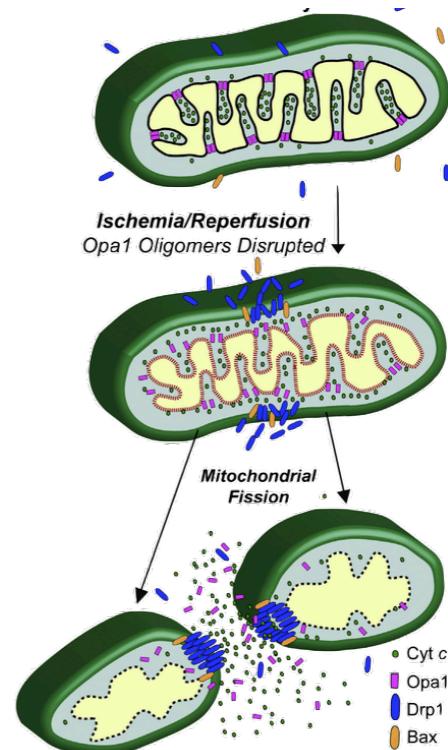
Biological process



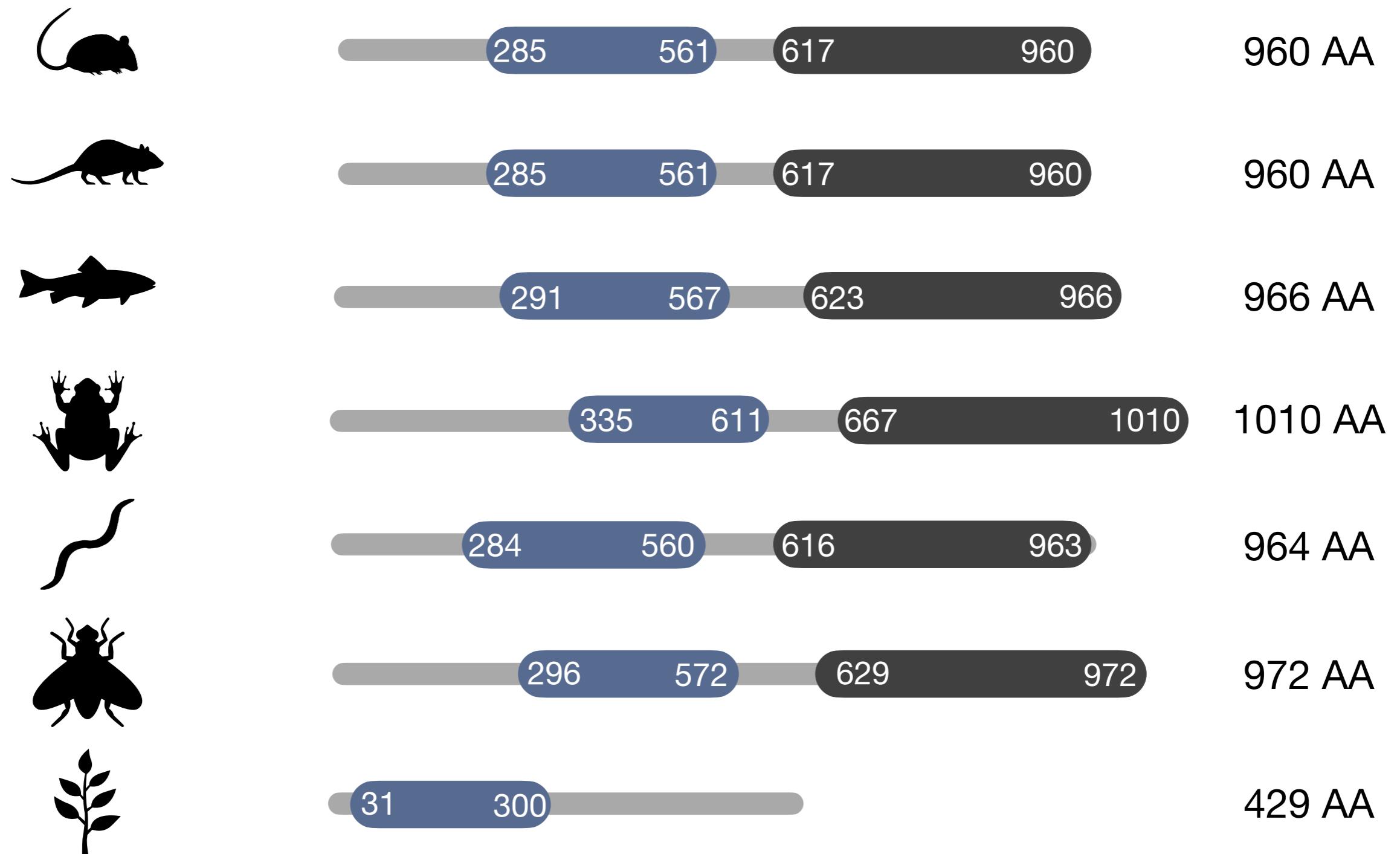
Molecular Function



Cellular Component



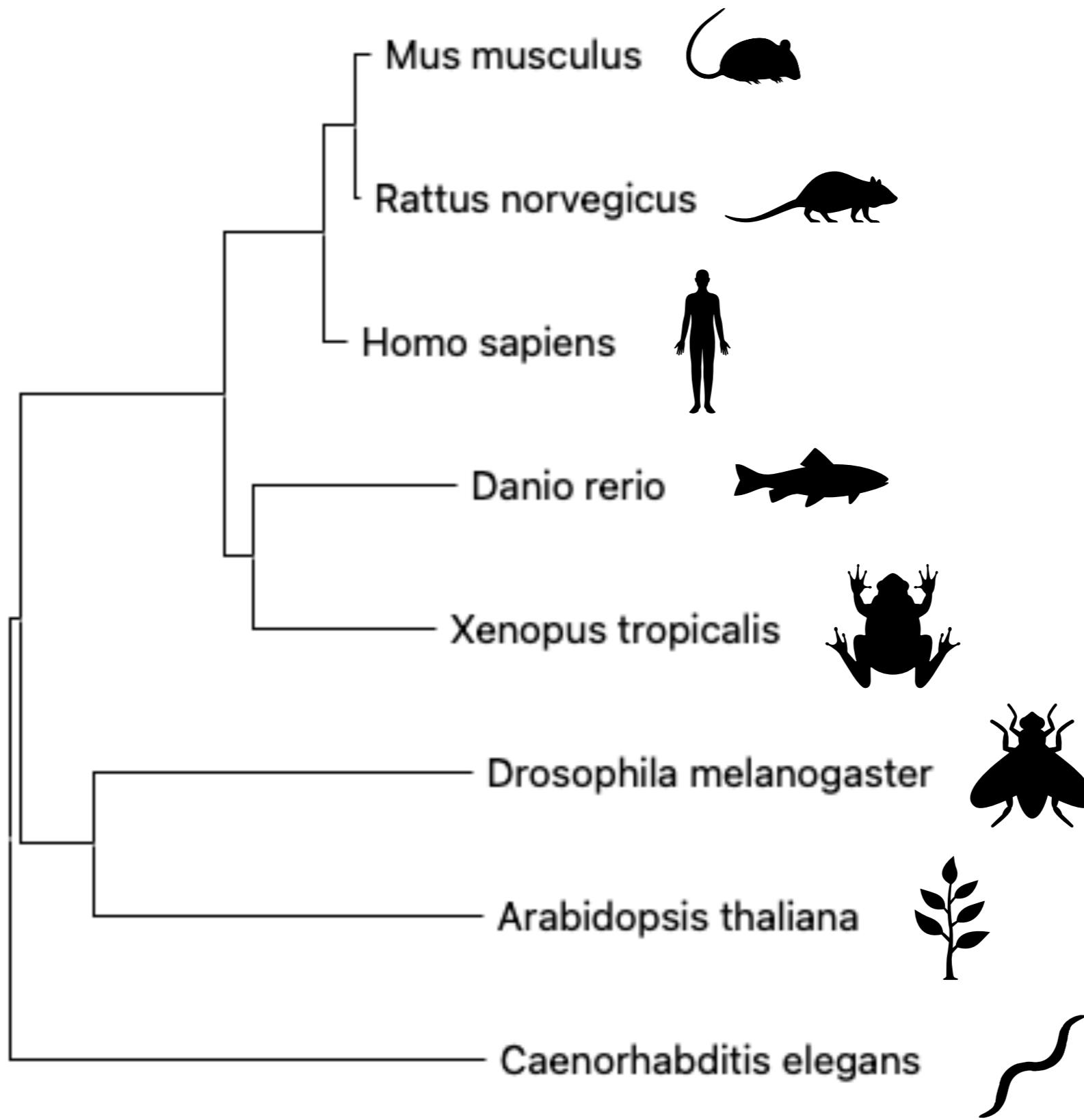
Gene in Model Organisms



Dynamin-type
guanine nucleotide-
binding domain

Dynamin-like
GTPase OPA1
C-Terminal

How are OPA1 Homologs Related?



Which Model Organisms to Use?

Knowledge Gap

Hypothesis

Primary Goal

Aim 1

Aim 2

Aim 3

Summary

References

- [1] Arruti, N., Rodríguez-Solana, P., Nieves-Moreno, et al . (2023). OPA1 Dominant Optic Atrophy: Diagnostic Approach in the Pediatric Population. *Current issues in molecular biology*, 45(1), 465-478.
- [2] Delettre-Cribaillet, C., Hamel, C. P., & Lenaers, G. (2007). Optic Atrophy Type 1. In M. P. Adam (Eds.) et. al., *GeneReviews®*. University of Washington, Seattle.
- [3] Ferré, M., Bonneau, D., Milea, D., et al. (2009). Molecular screening of 980 cases of suspected hereditary optic neuropathy with a report on 77 novel OPA1 mutations. *Human mutation*, 30(7), E692-E705. <https://doi.org/10.1002/humu.21025>
- [4] Formichi, P., Radi, E., Giorgi, E., et al. (2015). Analysis of opa1 isoforms expression and apoptosis regulation in autosomal dominant optic atrophy (ADOA) patients with mutations in the opa1 gene. *Journal of the neurological sciences*, 351(1-2), 99-108. <https://doi.org/10.1016/j.jns.2015.02.047>
- [5] Lenaers, G., Hamel, C., Delettre, C. et al. Dominant optic atrophy. *Orphanet J Rare Dis* 7, 46 (2012). <https://doi.org/10.1186/1750-1172-7-46>
- [6] Roubertie, A., Leboucq, N., Picot, M. C., et al. (2015). Neuroradiological findings expand the phenotype of OPA1-related mitochondrial dysfunction. *Journal of the neurological sciences*, 349(1-2), 154-160. <https://doi.org/10.1016/j.jns.2015.01.008>
- [7] Yu-Wai-Man, P., Griffiths, P. G., Burke, et al. (2010). The prevalence and natural history of dominant optic atrophy due to OPA1 mutations. *Ophthalmology*, 117(8), 1538-1546.e1. <https://doi.org/10.1016/j.ophtha.2009.12.038>
- [8] Zanna, C., Ghelli, A., Porcelli, A. M., et al. (2008). OPA1 mutations associated with dominant optic atrophy impair oxidative phosphorylation and mitochondrial fusion. *Brain : a journal of neurology*, 131(Pt 2), 352-367. <https://doi.org/10.1093/brain/awm335>