Leber's Congenital Amaurosis (LCA-2)

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January 21, 2008

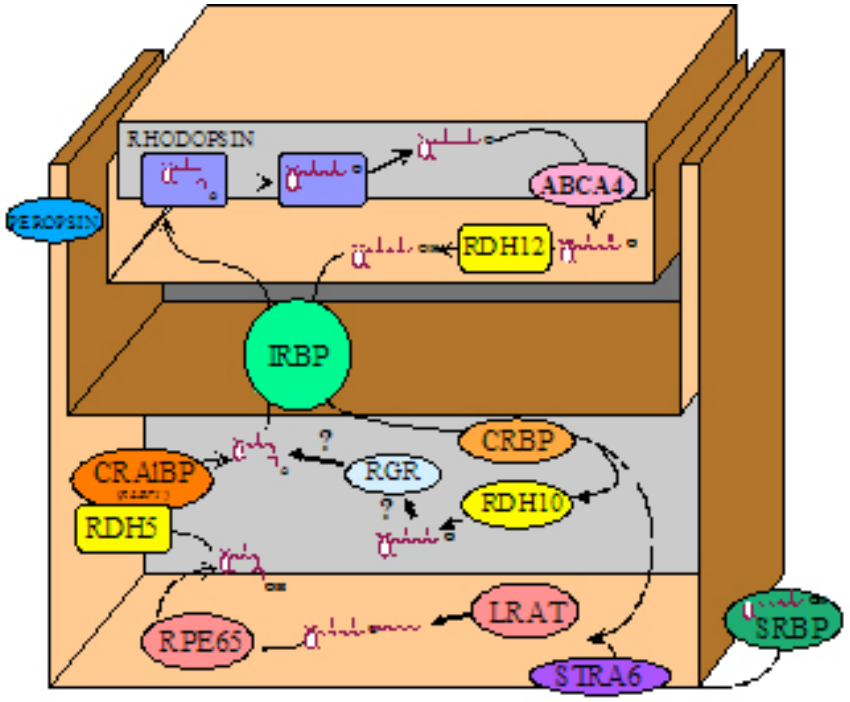
BioChem118

What is LCA?

- Eye disease
- Most common cause of inherited blindness
- Autosomal recessive
- 11 different types of LCA (1-11)
- LCA-2 causes a defect the gene RPE65, which prevents the retina from detecting light

Diagram of LCA

- Rhodopsin is a photoreceptor
- Lots of enzymes involved
- RPE65 is mutated (4bp deletion)
- Rhodopsin does not function properly



Classical Diagnosis

- Check for blindness or severe visual impairment
- Reduced photopic electroretinogram (ERG)
- Retina pigment migration
- Oculo-digital sign
- Family history

Classical Treatment

- No cure
- Prevention of secondary complications
- Surveillance

Genetic Diagnosis

- Sequence analysis
- Carrier testing
- Prenatal diagnosis

Genetic Treatment

- RPE65 injected into retina
- University of Pennsylvania and University College London had successful clinical trials