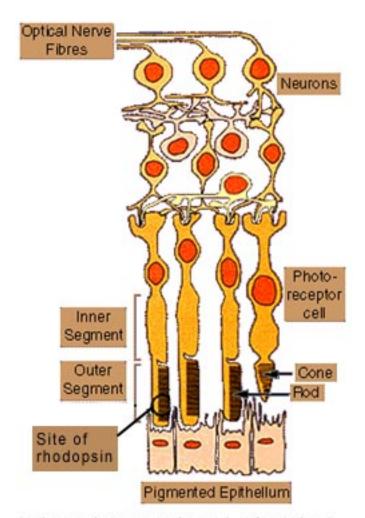


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## Diseases of the Eye



In the eye, light enters the pupil, is focused and inverted by the cornea and lens, and then is projected onto the retina at the back of the eye. The retina consists of several layers of cells, shown above. The only light-sensitive step during vision takes place in the outer segment of photoreceptor cells, and is catalysed by the molecule rhodopsin. Light causes rhodopsin to change shape, which then triggers a signal to be sent through the layers of cells that make up the retina, resulting in a neural signal to the brain. (Adapted from Gebhard Schertler's web page, MRC-LMB, Cambridge, UK, with permission.)

2 Genes and Disease

The function of our eyes is to allow us to see the objects in our surroundings at variable distances and under various conditions of lights. This function is achieved by a very complex arrangement of layers and structures found in the eye. In addition, two pockets of transparent fluid — the aqueous and vitreous humors — nourish eye tissues and help maintain constant eye shape.

The eye is comprised of three layers: an outer protective white coating called the sclera; a middle layer (choroid) containing blood vessels which nourish the eye; and an inner layer (retina) which contains the nerves that bear information to the brain for processing.

The cornea is the clear portion found at the front of the eye and serves to bend light rays. The iris, an extension of the choroid, is the colored portion of the eye and is made up of a spongy tissue. The pupil (black) is an opening in the iris that allows light into the eye. The lens then helps focus the light rays onto photoreceptors, which absorb and convert the light into electrical signals that carry information. The optic nerve contains fibers that transmit these signals to the brain for interpretation of the objects seen.

With the recent advances in molecular genetic techniques, new genes that cause eye disease are rapidly being identified, such as for those diseases discussed here. In many instances, these findings allow researchers to develop innovative strategies for preventing or slowing the progress of genetic eye diseases.

## **Diseases**

Best disease

Glaucoma

Gyrate atrophy of the choroid and retina

Retinoblastoma