Retinitis Pigmentosa

WHAT IS RETINITIS PIGMENTOSA?

Retinitis pigmentosa (RP) is a group of diseases characterized by gradual vision loss. The vision loss usually affects the peripheral vision (side vision) and dark/night vision first, but usually worsens to involve the central vision as well. This is caused by changes in the retina (pigment and neural cells that line the back of the eye).

WHAT CAUSES RP?

RP is usually caused by an inherited genetic abnormality. The inheritance pattern can be dominant (passed from one generation to the next), recessive (an abnormal gene from 2 normal vision parents), or X-linked (unaffected mother passes abnormal gene to affected son). RP can also occur sporadically (by a new mutation).

WHAT ARE THE SYMPTOMS/FINDINGS ASSOCIATED WITH RP?

Decreased peripheral vision (side vision) and poor vision in the dark or in dim lighting are the most common early symptoms of RP. Visual function is typically better with good lighting. Progressive loss of peripheral vision may eventually involve the central vision. Symptoms range from mild to severe, depending upon the stage and course of the disease. An ophthalmologist (eye M.D) may detect changes and abnormalities in the retina before visual symptoms are noticed. The changes are most commonly pigment clumping in the retina, thinning of the blood vessels and/or paleness of the optic nerve [figure 1].
Occasionally cataracts or retinal swelling/inflammation may occur as well.

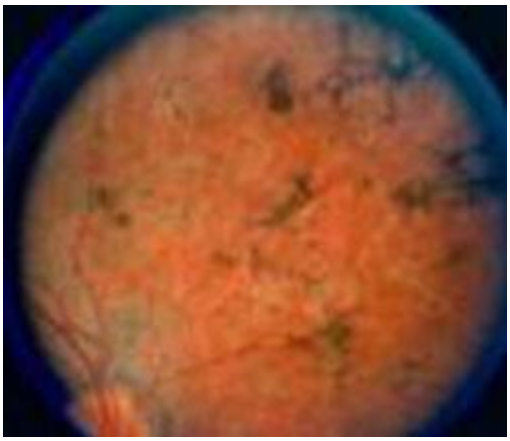

Fig. 1: Retinal appearance in retinitis pigmentosa.

WHAT TESTS CAN BE PERFORMED TO CONFIRM THE DIAGNOSIS?

The Eye MD may perform an electroretinogram (ERG) in the office, lab or operating room outfitted with special equipment. Electrodes are placed on the skin and cornea to measure the electrical response of the retinal cells to a flash of light. A visual field test is often performed at least once per year to assess the peripheral vision. Dark adaptation and molecular analysis (blood sample) for genetic abnormalities are less frequently administered tests. However, genetic testing is becoming more widely available.

ARE THERE OTHER ABNORMALITIES IN CHILDREN WITH RP?

RP is usually an isolated finding. However, RP can be associated with other systemic abnormalities including hearing loss (Usher Syndrome).

HOW DOES THE DISEASE PROGRESS?

The age of onset and progression of RP is extremely variable. Some affected people maintain good vision into the 5th or 6th decade, while others have profound vision loss early in life. It is important to have serial exams with an ophthalmologist to maximize vision and provide resources for help and support.

ARE THERE ANY TREATMENTS FOR RP?

Although there is no cure, serial examinations and treatment (by an Eye MD) of refractive error by glasses, removal of visually significant cataracts, and treatment of any retinal swelling of the retina will help maintain the best possible vision. There are also many low vision aids that can be useful. Vitamin

supplementation may slow vision loss, but this treatment is controversial and requires direct physician supervision. There are many ongoing investigations to help people with RP. In the future, gene therapy, stem cell therapy, and/or development of an artificial retina may provide significant benefit. Parents and physicians typically communicate with teachers and vision personnel (including low vision specialists) to maximize the education experience.

More technical information can be found on the EyeWiki Site.

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