Retinitis Pigmentosa

WHAT IS RETINITIS PIGMENTOSA?

Retinitis pigmentosa (RP) is an inherited disease which causes gradual vision loss in both eyes due to damage to the retina.

The retina contains cells that respond to light called photoreceptors. The cells that work in dim light are called rods while other cells called cones work in bright lighting and provide detailed vision. Retinitis pigmentosa damages the rods first and eventually the cones too. The vision loss usually affects the peripheral vision (side vision) and dark/night vision first, but then worsens to involve the central vision as well.

WHAT CAUSES RP?

RP is usually caused by defect in a gene that is needed for photoreceptors (see above) to work well. Over 100 separate genes have been identified as causes of RP. Retinitis pigmentosa GTPase regulator (RPGR) is one of the most common genes to cause RP.

Each person usually has 2 copies of a gene. If the problem gene is dominant (15-25% of RP cases), one abnormal copy can result in the disease and many family members are usually affected. If the gene is recessive (5-20% of RP cases), both copies have to be defective and sometimes no one else in the family has the disease. Retinitis pigmentosa can also be X-linked (10-15% of cases). Those defined as girls at birth have two X chromosomes while boys have only one X chromosome, leading to differences in how the different sexes are affected. RP can also occur spontaneously (by a new noninherited mutation).

WHAT ARE THE SYMPTOMS/FINDINGS ASSOCIATED WITH RP?

Most people begin to have problems with vision in their teenage years, but some people don't have trouble until their 40's. Both eyes are usually equally affected. Decreased peripheral vision (side vision) and poor vision in dim lighting are the most common early symptoms of RP. Vision can be very good in bright lighting, but 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 pick up changes 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.

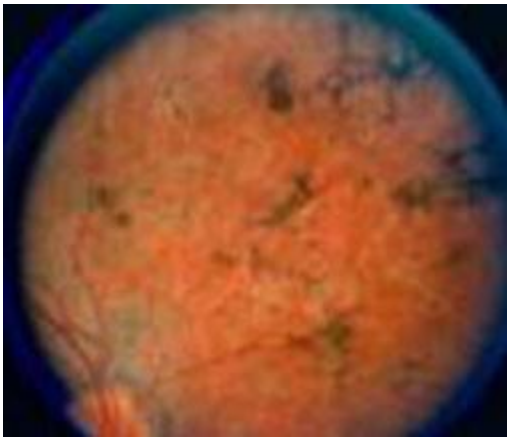

Fig. 1: Retinal appearance in retinitis pigmentosa.

WHAT TESTS CAN BE PERFORMED TO CONFIRM THE DIAGNOSIS?

The ophthalmologist may perform an electroretinogram (ERG) to check on the function of the rods and cones in the retina. Electrodes are placed on the skin or cornea (with contact lens) to measure the electrical response of the retinal cells to a flash of light.

A visual field test is often performed to assess the peripheral vision.

A specialized picture of the retina called an OCT (optical coherence tomography) can also show fluid in the retina and assess the health of the retina.

Genetics evaluation with testing and detailed history of affected family members may help with diagnosis and guide possible future treatments.

ARE THERE OTHER PROBLEMS IN CHILDREN WITH RP?

RP is usually a problem that only affects the eyes. However, RP can be seen in those with other disorders including hearing loss as seen in Usher Syndrome.

HOW DOES THE DISEASE PROGRESS?

The age when RP starts and the course it takes is extremely variable. Some affected people maintain good vision into their adult years, while others have severe vision loss early in life. It is important to have regular eye exams with an ophthalmologist to maximize vision and provide resources for support.

ARE THERE ANY TREATMENTS FOR RP?

There is currently no cure for RP. However, there are some supportive treatments that might be helpful. For example, sometimes glasses can improve the vision slightly. Cataracts (cloudy lens) if present can be removed, and treatment of any swelling in the retina (macular edema) with medications or injections will help keep the best possible vision.

It can be important for parents and physicians to communicate with teachers and vision personnel (including low vision specialists) to maximize the education experience. There are also many low vision aids that can be helpful.

Experimental treatments to slow the effects of RP include vitamin A supplementation (ß-carotene) or lutein, but this treatment is controversial and requires direct physician supervision.

There are many ongoing studies to help people with RP see better. Artificial retinal arrays, an implant in the eye to help stimulate the retina, may help those with severe visual loss. Exciting gene therapy trials attempt to replace defective genes which may slow the disease. Other ideas being investigated but not ready for patients include: protective medicines, injections of light sensitive proteins into the retina (optogenetics), injecting new proteins adapted to react to light (photochemical switch), and injecting stem cells. Studies of these new treatments have been encouraging but many challenges remain.

More technical information can be found on the EyeWiki Site.

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