Retinitis Pigmentosa

Overview:
Retinitis Pigmentosa is a group of genetic eye disorders that progressively causes atrophy of the retina and typically affects both eyes. It is one of the most common forms of inherited retinal degenerative conditions, and affects 1 in 3,700 people within the United States. Typically the disease begins in childhood or early adulthood and the rate of progression varies between cases. It affects the peripheral retina first (the outer retina) and progressively moves toward the central retina. In some instances, retinitis pigmentosa progresses slowly and goes relatively unnoticed (until in later stages of the disease), with atrophying of the retina occurring over a life-span. In other cases, retinitis pigmentosa progresses quickly and can cause severe vision problems early in life.

Mutations in over 50 genes have been shown to cause retinitis pigmentosa and there are three main categories of the disease; autosomal dominant, autosomal recessive and x-linked. The main difference between recessive genes and dominant genes is that in order to develop the disease, you need two copies (from both parents) of recessive genes, whereas you only need one copy of a dominant gene (from either parent). X-linked genes are responsible for determining gender (XX for females and XY for males). Most genes causing retinitis pigmentosa are autosomal recessive. Autosomal dominant Retinitis Pigmentosa is the least pervasive of the three, whereas X-linked Retinitis Pigmentosa is the most pervasive.

Typically the first symptom of retinitis pigmentosa is night blindness. Patients with night blindness have problems seeing or are clumsy in dimly lit rooms. This occurs because the photoreceptor responsible for seeing in low-light settings (retinal rods) become damaged in retinitis pigmentosa. Loss of peripheral vision occurs as the disease progresses and can cause tunnel vision (a visual field of less than 10 degrees as measured by a Humphrey visual field test). In the late stage of the disease, central vision can start to decrease, typically to 20/200 or below.

How can the doctor determine the extent of Retinitis Pigmentosa?
The doctor will perform a dilated exam using a slit lamp to determine the extent of the disease and the affect it is having on the macula. Since Retinitis Pigmentosa primarily affects the outer retina, the doctor will use an indirect ophthalmoscope to check the periphery. Several tests are performed to help determine the extent of the disease.

What tests are performed?
Testing is important because it helps the doctor to precisely document the extent of the retinitis pigmentosa and how much atrophy is present, and measure changes that occur. The three types of tests described below are performed in our clinic. The four tests described below are done in our clinic:

**Optical Coherence Tomography (OCT)** is a high definition image of the retina taken by a scanning ophthalmoscope with a resolution of 5 microns. The doctor can use these images to objectively determine the amount of atrophy within the retina and to detect changes that occur between visits.

**Fundus Photography** is an image taken by a digital fundus camera to visually document the retinitis pigmentosa.

**Fluorescein Angiography** is a test that documents blood circulation in the retina using fluorescein dye which luminesces under blue light. Fluorescein is injected into a vein in your arm and digital fundus pictures are taken afterwards for 10 minutes. Fluorescein Angiography is used to document the extent of atrophy within the retina and how blood circulation has been affected by the disease progression.

**Humphry Visual Field** is a test that documents the amount of peripheral vision a person has, and is performed individually for each eye. The eye not being tested is covered so that you are unable to see with it. With the eye that is being tested, the technician will ask you to look into the machine and fixate on a point in the center. During the test, you will push a button each time you see a flashing light. It is important that the eye being tested remains fixated on the point straight ahead through-out the test. If you move your eye around during the test, the results will be inaccurate and the test will have to be repeated.

What are the risk factors?
Retinitis pigmentosa is a genetically inherited disease caused by either autosomal dominant, autosomal recessive, or x-linked genes. Due to the lower number of autosomal dominant and x-linked genes that cause retinitis pigmentosa, most cases are autosomal recessive. Typically neither parent has retinitis pigmentosa, and it is common that there are no cases of the disease within several generations or in extended family members. However, it has been estimated that around 1% of the population within the United States are carriers of retinitis pigmentosa causing genes.

What are the symptoms?
If the disease progress is slow, some patients may not notice any symptoms or may not realize that there have been any changes in their vision. Common symptoms of retinitis pigmentosa include:

1. Night Blindness
2. Loss of peripheral vision (tunnel vision)
3. Loss of central vision (in later stages)

What treatments are available for wet macular degeneration?
Currently there are no treatments for Retinitis Pigmentosa. The National Eye Institute recommends that high doses of vitamin A (15,000 IU) be taken daily to help slow the progression of Retinitis Pigmentosa. This recommendation is based on a large-scale trial that was performed in 1993. A recent study published in the Archives of Ophthalmology reported that the addition of Omega-3 (1,200mg per day) when just starting Vitamin A therapy helps to further slow the progression of Retinitis Pigmentosa. The study also reported that in patients who had been taking high doses of vitamin A for over 2 years, the addition of omega-3 only slowed the decrease in visual field sensitivity. Another recent study, also published in the Archives of Ophthalmology, reported that the addition of lutein (12 mg per day) to vitamin A therapy helped slow the progression of visual loss within the mid-periphery (beyond 30 degrees). More tests are needed to verify the results of these two studies. Genetic testing and counseling can help determine which type of retinitis pigmentosa you have and implications it may have on your family.

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