Published on *My46* (https://www.my46.org) 01/20/2022

My46 Trait Profile

Retinitis Pigmentosa

Retinitis Pigmentosa is a group of conditions that causes progressive vision loss beginning with decreased night vision and loss of peripheral vision. It can be caused by mutations in any one of many different genes.

Characteristics of Retinitis Pigmentosa

Retinitis Pigmentosa (RP) is a group of conditions that causes progressive vision loss. Initial symptoms are often night vision loss with eventual loss of side (peripheral) vision. The clarity or sharpness of vision (i.e., visual acuity) may be lost in later stages of disease. RP may have an onset from childhood to adulthood. There can be variation (even within the same family) as to how mildly or severely an individual is affected. In most cases, the disease affects both eyes similarly. The vision loss is caused by photoreceptor dysfunction. Photoreceptors are the light sensing cells that make up the retina (the back of the eye). In most affected individuals, RP is found as an isolated condition. However, RP may be a feature of a genetic syndrome, such as Usher syndrome (RP and hearing loss) or Bardet-Biedl syndrome (RP, polydactyly, obesity, kidney problems, etc). Approximately 1 in 3,500 to 1 in 5,000 individuals in the United States and Europe are affected with RP.

Diagnosis/Testing

The diagnosis of RP is made based on the results of clinical examinations and ophthalmic tests. An electroretinogram (ERG) test is done to determine how well the photoreceptors are functioning. With this test, eye drops are placed on the eye to dilate the pupils, after which content lens-like electrodes are put on the surface of the eye. Additional tests used to diagnosis and follow people affected with RP include the visual field test, visual acuity, ocular coherence tomography (OCT), and a dilated eye exam. The visual field test is used to determine the extent of visual field loss. The OCT image provides a cross section view of the retina. In some cases, especially early in the disease, the appearance of the retina on dilated eye exam may appear normal or near normal. However, most affected individuals will have some changes to their retina including blood vessel narrowing (attenuation), pigment clumping (bone spicules), and retinal atrophy or degeneration.

Nearly 60 different genes have been identified in which mutations are known to cause RP. Genetic testing is available for these genes to determine the genetic basis for disease. Approximately 40-50% of individuals affected with RP are the only person in their family known to be affected with the condition.

Management/Surveillance

There is no treatment for RP. After the initial diagnosis of RP is made, individuals should continue to be evaluated by an ophthalmologist with tests such as visual field tests to monitor the progression of the disease. It is also important to monitor for the development of any complications of the disease. For example, individuals with RP tend to develop cataracts at a younger age, which may be removed. Individuals with RP may develop cystoid macular edema (CME; an eye condition that causes swelling of the retina). CME can be detected through the OCT and may be treated with medication.

It has been suggested that vitamins may be helpful in slowing the progression of RP. Eyes should also be protected from bright sunlight. Individuals with low vision may benefit from low vision adaptive devices, such as computer programs and better lighting, to help them use the remaining vision that they have. Affected individuals may also benefit

from occupational therapy, job rehabilitation, or mobility training to help them remain independent.

Several clinical treatment trials are currently underway in the United States and other countries for individuals affected with RP including oral medication, gene therapy, and stem cell therapy. A retinal implant was recently approved for the improvement of vision in individuals who have advanced RP.

Mode of inheritance

RP may be inherited in one of three patterns of inheritance: autosomal dominant, autosomal recessive, and X-linked recessive. There have been a very small number of RP families reported with digenic inheritance, which means that more than one gene is responsible for the RP in a single family.

Autosomal dominant inheritance:

Autosomal dominant inheritance accounts for approximately 15-25% of RP. This inheritance pattern means inheriting one mutation is enough for an individual to be affected with RP. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Autosomal recessive inheritance:

Autosomal recessive inheritance accounts for approximately 5-20% of RP. This inheritance pattern means that an individual has to inherit two mutations (i.e., one from each parent) to be affected. If both parents are carriers of a mutation they have a 1 in 4 (25%) chance with each pregnancy of having a child with the condition.

X-linked recessive inheritance:

Approximately 5-15% of RP can be inherited in an X-linked recessive pattern of inheritance. The gene mutations causing this type of inheritance are found on the X chromosome. An X-linked recessive pattern means that in females, both copies of a gene (i.e., one on each X chromosome) must have a change or mutation, whereas in males, only one copy of a gene must have a mutation to be affected. A female with a mutation in one copy of a gene on the X chromosome is said to be a “carrier” for an X-linked condition, and is typically not affected.

Risk to family members

The risk to family members depends on the pattern of inheritance.

Autosomal dominant inheritance:

The risk to family members depends on whether or not the individual with RP has a parent affected with RP. If a parent also has the condition, the risk of having a child with RP is 50% with each pregnancy. If a parent does not have RP, the risk of other siblings being affected is very low.

Autosomal recessive inheritance:

Parents of a child with RP are carriers of RP. If a sibling of a child with RP is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of RP.

X-linked recessive inheritance:

If a father is affected with RP, his daughters will be carriers of RP and his sons will be unaffected. If a mother is a carrier of RP, each daughter has a 1 in 2 (50%) chance of being a carrier and each son has a 1 in 2 (50%) chance of being affected with RP.

A wide range of changes can be found in female carriers. Some female carriers may remain relatively asymptomatic, but have abnormalities on the ERG test. Other females will report that they have difficulty with night vision and/or abnormalities with their field of vision, particularly as they get older. There have been reports of females who are as affected as males, but typically at a later age of life.

Special considerations

None

Resources

Foundation Fighting Blindness
http://www.blindness.org
Genetics Home Reference: Retinitis pigmentosa
http://ghr.nlm.nih.gov/condition/retinitis-pigmentosa
National Eye Institute

http://www.nei.nih.gov
eyeSmart®: Retinitis Pigmentosa
http://www.geteyesmart.org/eyesmart/diseases/retinitis-pigmentosa.cfm
eyeGENE® Research Project
http://www.nei.nih.gov/eyegene
Kellogg Eye Center
http://www.kellogg.umich.edu/patientcare/downloads/Understand-Retinitis-Pigmentosa.pdf

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