Condition: Retinitis pigmentosum

Inheritance:

Genetically heterogeneous; in some instances, heterozygosity for two distinct genes has been found to cause RP, referred to as digenic inheritance

Genetic etiology:

Multiple distinct genes responsible for disorder.

Frequency:

Approximately 1/5,000.

Clinical features:

Retinitis pigmentosum is a term for a group of disorders characterized by dysfunction of photoreceptors in the eye, loss of vision, and accumulation of pigment in the retina. Loss of rod function usually occurs first, resulting in night blindness. Retinitis pigmentosum is a component of many genetic syndromes, and may occur in isolation.

Management:

Supportive management, including optical aids; treatment with vitamin A may be helpful.

Genetic counseling:

Depends on mode of genetic transmission; genetic testing for many of the genes responsible for RP is possible.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.