Genetic heterogeneity of retinitis pigmentosa

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Abstract	Genetic heterogeneity is a phenomenon in which a genetic disease can be transmitted by several modes of inheritance. The understanding of genetic heterogeneity is important in giving genetic counselling.The presence of genetic heterogeneity can be explained by the existence of:1.different mutant alleles at a single locus, and2.mutant alleles at different loci affecting the same enzyme or protein, or affecting different enzymes or proteins.To have an overall understanding of genetic heterogeneity, the heterogeneities of chronic hemolytic anemia caused by beta-globin abnormality, chronic hemolytic anemia due to thalassemias, and mucopolysaccharidoses have been chosen as examples.'The article is focused on the genetic heterogeneity of retinitis pigmentosa, a kind of retinal hereditary disease. The genetic heterogeneity of retinitis pigmentosa can be known by the facts thata.Retinitis pigmentosa can be either an isolated disease or a part of a certain syndrome.b.Retinitis pigmentosa as isolated disease can be genetic or non-genetic in nature.c. Retinitis pigmentosa as a genetic disease can be transmitted either by autosomal dominant, autosomal recessive, or X-linked recessive genes.The frequency of isolated hereditary retinitis pigmentosa varies between 30% to 50%. Clinically the dominant form is milder than the recessive form. Certain syndromes associated with retinitis pigmentosa which need to be mentioned are Usher syndrome, Laurence-Moon-Bardet-Biedl syndrome, and Kearn-Sayre syndrome.Key Words: genetic heterogeneity - retinitis pigmentosa - genetic disease - hemolytic anemia - Usher syndrome
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