Mutation analysis in Canadian families with choroideremia.

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Comment in
Ophthalmic Genet. 1996 Jun;17(2):43-6.

Abstract
Choroideremia (CHM) is an X-linked heritable progressive dystrophy of the choroid and retina. The condition predominantly affects males beginning in early childhood and eventually results in blindness after a period of 30-40 years. The CHM gene was localized to Xq21 and cloned in the past few years. The gene encodes for Rab escort protein-I, a protein involved in the isoprenylation of intracellular proteins. With the isolation of the gene, a number of mutations have been identified in patients affected by CHM using molecular techniques. Our group reports the characterization of mutations in four Canadian families affected by CHM. In addition, an intragenic polymorphism was identified in exon 5. Finding the mutations in these families will result in accurate predictive testing for carriers, avoid unnecessary repeated examination of at-risk individuals, and add to our understanding of the cause of this disorder.

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