Cloning and characterization of the human choroideremia gene.

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Abstract

Positional cloning has previously resulted in the identification of a gene which is disrupted by deletions in patients with the classic choroideremia (CHM) phenotype. More subtle mutations had been identified in 4 exons of the 3' portion but not elsewhere in the CHM gene. We have now isolated and characterized the complete open reading frame of the CHM gene and determined its exon-intron structure. The CHM gene encodes a protein of 653 amino acids, which is highly homologous to the mouse and rat CHM proteins, and, to a slightly lesser extent, to the human CHM-like (CHML) protein. The open reading frame (ORF) of the human CHM gene consists of 15 exons, spanning at least 150 kb of Xq21.2, and it is possible that there is an additional exon corresponding to the 5' non-coding region of the gene. Cloning of the 5' end of the CHM gene and the elucidation of its intron-exon structure enabled us to localize the X-chromosomal breakpoint in a CHM female with an X;7 translocation between exons 3 and 4.

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activating a cryptic exon.

Molecular basis of choroideremia (CHM): mutations involving the Rab escort protein-1 (REP-1) gene.

Detection and characterization of point mutations in the choroideremia candidate gene by PCR-SSCP analysis and direct DNA sequencing.

Mutation analysis in Canadian families with choroideremia.