

Title (en)
BEST'S MACULAR DYSTROPHY GENE

Title (de)
BEST'S MACULADYSTROPHIE-GEN

Title (fr)
GENE DE LA DYSTROPHIE CORNEENNE DE BEST

Publication
EP 1060184 A1 20001220 (EN)

Application
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Abstract (en)
[origin: WO9943695A1] Novel human and mouse DNA sequences that encode the gene CG1CE, which, when mutated, is responsible for Best's macular dystrophy, are provided. Provided are genomic CG1CE DNA as well as cDNA that encodes the CG1CE protein. Also provided is CG1CE protein encoded by the novel DNA sequences. Methods of expressing CG1CE protein in recombinant systems are provided. Also provided are diagnostic methods that detect patients having mutant CG1CE genes.

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C07H 21/02; **C07K 16/00**; **C07K 14/00**; **C12N 15/63**; **C12P 19/34**; **C12Q 1/00**

IPC 8 full level
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