

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  
**EP 99908345 A 19990222**

Priority  
• US 9903790 W 19990222  
• US 7594198 P 19980225  
• US 11292698 P 19981218

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.

IPC 1-7  
**C07H 21/02**; **C07K 16/00**; **C07K 14/00**; **C12N 15/63**; **C12P 19/34**; **C12Q 1/00**

IPC 8 full level  
**C12N 15/09** (2006.01); **C07K 14/47** (2006.01); **C07K 16/18** (2006.01); **C12N 5/10** (2006.01); **C12P 19/34** (2006.01); **C12P 21/08** (2006.01); **C12Q 1/68** (2006.01)

CPC (source: EP)  
**C07K 14/47** (2013.01)

Designated contracting state (EPC)  
AT BE CH DE DK ES FI FR GB GR IE IT LI LU NL PT SE

DOCDB simple family (publication)  
**WO 9943695 A1 19990902**; CA 2321129 A1 19990902; EP 1060184 A1 20001220; EP 1060184 A4 20020925; JP 2002504559 A 20020212

DOCDB simple family (application)  
**US 9903790 W 19990222**; CA 2321129 A 19990222; EP 99908345 A 19990222; JP 2000533447 A 19990222