

Title (en)  
HUMAN GALACTOKINASE GENE

Title (de)  
MENSCHLICHES GALACTOKINASE GEN

Title (fr)  
GENE DE LA GALACTOKINASE HUMAINE

Publication  
**EP 0783567 A4 19990915 (EN)**

Application  
**EP 95921460 A 19950526**

Priority  
• US 9506743 W 19950526  
• US 9410825 W 19940923

Abstract (en)  
[origin: WO9609374A1] This invention relates to human galactokinase and the identification of galactokinase mutations, a missense and nonsense, as well as isolated nucleic acids encoding same, recombinant host cell transformed with DNA encoding such proteins and to uses of the expressed proteins and nucleic acid sequences in therapeutic and diagnostic applications.

IPC 1-7  
**C12N 5/10**; **C12N 9/12**; **C12N 15/54**; **C12N 15/63**; **C12N 15/85**; **C12Q 1/00**; **C12Q 1/68**; **C07K 16/40**

IPC 8 full level  
**A01K 67/027** (2006.01); **A61K 35/12** (2006.01); **A61K 35/407** (2006.01); **A61K 35/44** (2006.01); **A61K 38/45** (2006.01); **A61K 48/00** (2006.01); **A61P 25/00** (2006.01); **A61P 43/00** (2006.01); **C12N 5/10** (2006.01); **C12N 7/00** (2006.01); **C12N 9/12** (2006.01); **C12N 15/09** (2006.01); **C12N 15/10** (2006.01); **C12N 15/54** (2006.01); **C12P 21/02** (2006.01); **C12P 21/08** (2006.01); **C12Q 1/68** (2006.01); **G01N 33/50** (2006.01); **G01N 33/573** (2006.01); **G01N 33/577** (2006.01)

CPC (source: EP)  
**A61P 25/00** (2017.12); **A61P 43/00** (2017.12); **C12N 9/1205** (2013.01); **A01K 2217/05** (2013.01)

Citation (search report)  
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• [A] STAMBOLIAN: "Galactose and Cataract", SURVEY OF OPHTHALMOLOGY, vol. 32, no. 5, 1988, pages 333 - 349, XP002108634  
• [A] ELSAS ET AL: "Galactosemia A Molecular Approach to the Enigma", INTERNATIONAL PEDIATRICS, vol. 8, no. 1, 1993, pages 101 - 109, XP002108635  
• [PX] STAMBOLIAN ET AL: "Expression and Mutation Analysis of the Gene for Human Galactokinase, a Cause of Congenital and Presenile Cataracts", INVESTIGATIVE OPHTHALMOLOGY AND VISUAL SCIENCE, vol. 36, no. 4, 15 March 1995 (1995-03-15), pages S843, XP002109090  
• See references of WO 9609374A1

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

DOCDB simple family (publication)  
**WO 9609374 A1 19960328**; AU 2653695 A 19960409; AU 7958194 A 19960409; BR 9509211 A 19980127; EP 0783567 A1 19970716; EP 0783567 A4 19990915; JP H10505757 A 19980609; JP H10506529 A 19980630; MX PA94008258 A 20050826; NO 971359 D0 19970321; NO 971359 L 19970521; NO 971363 D0 19970321; PL 319344 A1 19970804; WO 9609408 A1 19960328; ZA 947865 B 19951009

DOCDB simple family (application)  
**US 9506743 W 19950526**; AU 2653695 A 19950526; AU 7958194 A 19940923; BR 9509211 A 19950526; EP 95921460 A 19950526; JP 51084496 A 19940923; JP 51085896 A 19950526; MX 9408258 A 19940923; NO 971359 A 19970321; NO 971363 A 19970321; PL 31934495 A 19950526; US 9410825 W 19940923; ZA 947865 A 19941007