

Title (en)

GENE RESPONSIBLE FOR STARGARDT-LIKE DOMINANT MACULAR DYSTROPHY

Title (de)

FÜR STARGARDT-ÄHNLICHE, DOMINANTE, MAKULA-DYSTROPHY VERANTWORTLICHES GEN

Title (fr)

GENE RESPONSABLE DE LA DYSTROPHIE MACULAIRE DOMINANTE DE STARGARDT

Publication

**EP 1283897 A4 20040804 (EN)**

Application

**EP 01935445 A 20010511**

Priority

- US 0115464 W 20010511
- US 20499000 P 20000516

Abstract (en)

[origin: WO0187921A2] The gene responsible for Stargardt-like macular dystrophy has been identified, along with its normal allelic form. The mutant gene encodes a mutant protein containing a frameshift mutation, resulting in abnormal fatty acid synthesis and transport in the retina. Also disclosed are assays for Stargardt-like macular dystrophy and therapies.

IPC 1-7

**C12P 21/06**; **C12N 1/20**; **C07K 14/47**; **A61P 27/00**; **G01N 33/53**; **C12Q 1/68**

IPC 8 full level

**C07K 14/47** (2006.01); **C12Q 1/68** (2006.01); **C12Q 1/6883** (2018.01)

CPC (source: EP)

**A61P 27/00** (2017.12); **C07K 14/47** (2013.01); **C12Q 1/6883** (2013.01); **C12Q 2600/156** (2013.01)

Citation (search report)

- [AD] EDWARDS A O ET AL: "Autosomal dominant Stargardt-like macular dystrophy: I. Clinical characterization, longitudinal follow-up, and evidence for a common ancestry in families linked to chromosome 6q14.", AMERICAN JOURNAL OF OPHTHALMOLOGY. UNITED STATES APR 1999, vol. 127, no. 4, April 1999 (1999-04-01), pages 426 - 435, XP002283260, ISSN: 0002-9394
- [A] DATABASE EMBL [online] 7 March 2000 (2000-03-07), SIMS S, XP002283261, retrieved from EMBL Database accession no. AL159162
- See references of WO 0187921A2

Designated contracting state (EPC)

AT BE CH CY DE DK ES FI FR GB GR IE IT LI LU MC NL PT SE TR

DOCDB simple family (publication)

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DOCDB simple family (application)

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