

Title (en)

VARIANTS IN COMPLEMENT REGULATORY GENES PREDICT AGE-RELATED MACULAR DEGENERATION

Title (de)

VARIANTEN KOMPLEMENTÄRER REGULATORGENE ALS PROGNOSÉ FÜR ALTERSBEDINGTE MAKULADEGENERATION

Title (fr)

VARIANTS DE GENES DE REGULATION DU COMPLEMENT PERMETTANT DE PREDIRE LA DEGENERESCENCE MACULAIRE LIEE A L'AGE

Publication

EP 1989319 A4 20091111 (EN)

Application

EP 07750527 A 20070213

Priority

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Abstract (en)

[origin: WO2007095185A2] Methods for identifying a subject at risk for developing AMD are disclosed, as are kits which can be used to practice the methods. The methods include identifying specific protective or risk polymorphisms or genotypes from the subject's genetic material, including polymorphisms in the BF, C2 and/or CFH genes. Microarrays and kits for use in these methods are also provided.

IPC 8 full level

C12Q 1/68 (2006.01)

CPC (source: EP)

C12Q 1/6883 (2013.01); C12Q 2600/112 (2013.01); C12Q 2600/118 (2013.01); C12Q 2600/156 (2013.01); C12Q 2600/158 (2013.01); C12Q 2600/172 (2013.01)

Citation (search report)

- [AP] WO 2006062716 A2 20060615 - UNIV YALE [US], et al
- [X] DATABASE PROBE [online] 16 September 2005 (2005-09-16), XP002452757, retrieved from NCBI Database accession no. Pr503071 & DATABASE SNP [online] 3 December 2002 (2002-12-03), XP002550606, retrieved from NCBI Database accession no. rs641153
- [X] DATABASE PROBE [online] 16 September 2005 (2005-09-16), XP002452758, retrieved from NCBI Database accession no. Pr501784 & DATABASE SNP [online] 3 December 2002 (2002-12-03), XP002550607, retrieved from NCBI Database accession no. rs4151667
- [X] DATABASE PROBE [online] 25 January 2006 (2006-01-25), XP002452759, retrieved from NCBI Database accession no. Pr003131349 & DATABASE SNP [online] 3 September 2003 (2003-09-03), XP002550608, retrieved from NCBI Database accession no. rs547154
- [X] DATABASE PROBE [online] 5 October 2005 (2005-10-05), XP002452760, retrieved from NCBI Database accession no. Pr001347746 & DATABASE SNP [online] 3 September 2003 (2003-09-03), XP002550605, retrieved from NCBI Database accession no. rs9332739
- [X] GUNDERSON KEVIN L ET AL: "A genome-wide scalable SNP genotyping assay using microarray technology", NATURE GENETICS, vol. 37, no. 5, May 2005 (2005-05-01), pages 549 - 554, XP002452745, ISSN: 1061-4036
- [PXY] GOLD BERT ET AL: "Variation in factor B (BF) and complement component 2 (C2) genes is associated with age-related macular degeneration", NATURE GENETICS, vol. 38, no. 4, April 2006 (2006-04-01), pages 458 - 462, XP002452747, ISSN: 1061-4036
- [PXY] MALLER JULIAN ET AL: "Common variation in three genes, including a noncoding variant in CFH, strongly influences risk of age-related macular degeneration", NATURE GENETICS, vol. 38, no. 9, September 2006 (2006-09-01), pages 1055 - 1059, XP002452748, ISSN: 1061-4036
- [Y] ZIPFEL P F ET AL: "Complement and diseases: Defective alternative pathway control results in kidney and eye diseases", MOLECULAR IMMUNOLOGY, PERGAMON, GB, vol. 43, no. 1-2, 1 January 2006 (2006-01-01), pages 97 - 106, XP025037099, ISSN: 0161-5890, [retrieved on 20060101]
- [A] DATABASE PROBE [online] 24 January 2006 (2006-01-24), XP002452761, retrieved from NCBI Database accession no. Pr003249825 & DATABASE SNP [online] 3 December 2002 (2002-12-03), XP002550609, retrieved from NCBI Database accession no. rs1048709
- [AD] HAGEMAN GREGORY S ET AL: "A common haplotype in the complement regulatory gene factor H (HF1/CFH) predisposes individuals to age-related macular degeneration", PROCEEDINGS OF THE NATIONAL ACADEMY OF SCIENCES OF THE UNITED STATES OF AMERICA, vol. 102, no. 20, May 2005 (2005-05-01), pages 7227 - 7232, XP002391909, ISSN: 0027-8424
- [A] DATABASE HAPMAP PROJECT HOMEPAGE [online] XP002452762, retrieved from [HTTP://WWW.HAPMAP.ORG/DOWNLOADS/ASSAY-DESIGN_PROTOCOLS.HTML](http://WWW.HAPMAP.ORG/DOWNLOADS/ASSAY-DESIGN_PROTOCOLS.HTML)
- [A] FAN BAO JIAN ET AL: "Molecular diagnostics of genetic eye diseases", CLINICAL BIOCHEMISTRY, vol. 39, no. 3, January 2006 (2006-01-01), pages 231 - 239, XP002452749, ISSN: 0009-9120
- [A] CHARBEL ISSA P ET AL: "The complement system and its possible role in the pathogenesis of age-related macular degeneration (AMD)", OPHTHALMOLOGE 2005 GERMANY, vol. 102, no. 11, 2005, pages 1036 - 1042, XP002452750, ISSN: 0941-293X
- See references of WO 2007095185A2

Citation (examination)

MATSUZAKI HAJIME ET AL: "GENOTYPING OVER 100,000 SNPS ON A PAIR OF OLIGONUCLEOTIDE ARRAYS", NATURE METHODS, vol. 1, no. 2, 1 November 2004 (2004-11-01), NATURE PUBLISHING GROUP, GB, pages 109 - 111, XP009082656, ISSN: 1548-7091, DOI: 10.1038/NMETH718 & "Homo sapiens SSO probe urn:lsid:perlegen.hapmap.org:Assay:25175.4056446:1 for variation rs547154", Retrieved from the Internet <URL:<http://www.ncbi.nlm.nih.gov/probe/3131349>> & "Homo sapiens bead probe urn: LSID:sanger.hapmap.org:Assay:1730462:1 for variation rs4151667", Retrieved from the Internet <URL:<http://www.ncbi.nlm.nih.gov/probe/?term=pr501784&report=fullreport>> & "Homo sapiens bead probe urn:LSID:sanger.hapmap.org:Assay:1727808:1 for variation rs641153", Retrieved from the Internet <URL:<http://www.ncbi.nlm.nih.gov/probe/503071?report=fullreport>>

Designated contracting state (EPC)

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AL BA HR MK RS

DOCDB simple family (publication)

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