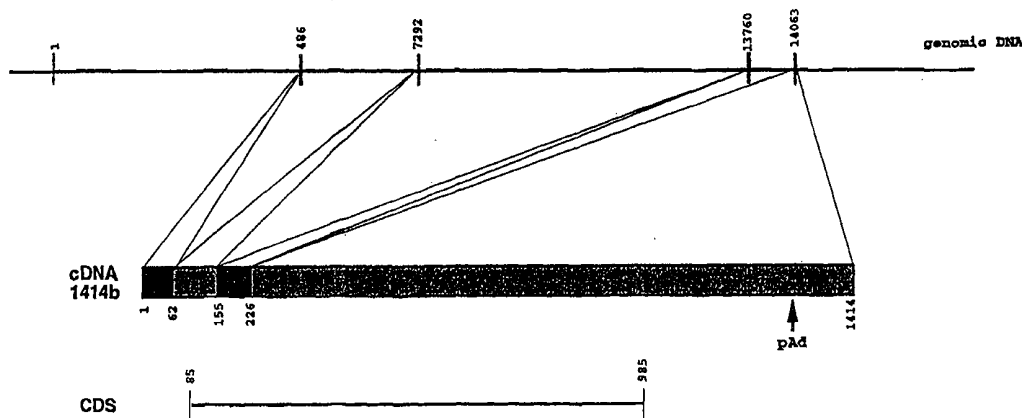




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<p>(51) International Patent Classification ⁷ : C12N 15/54, 9/10, C12Q 1/68, C07K 16/40, G01N 33/50</p>	A3	<p>(11) International Publication Number: WO 00/05382</p> <p>(43) International Publication Date: 3 February 2000 (03.02.00)</p>
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(54) Title: A NUCLEIC ACID ENCODING A GERANYL-GERANYL PYROPHOSPHATE SYNTHETASE (GGPPS) AND POLYMORPHIC MARKERS ASSOCIATED WITH SAID NUCLEIC ACID



(57) Abstract

The present invention relates to a purified or isolated polynucleotide encoding human geranylgeranyl pyrophosphate synthetase, the regulatory nucleic acids contained therein, a polymorphic marker thereof and the resulting encoded protein, as well as to methods and kits for detecting this polynucleotide and this protein. The present invention also pertains to a polynucleotide carrying the natural regulatory regions of the *hGGPS* gene which is useful, for example, to express a heterologous nucleic acid in host cells or host organisms as well as functionally active regulatory polynucleotides derived from said regulatory region. The invention also consists in genetic markers, namely biallelic markers, which may be useful for the diagnosis of diseases related to an alteration in the regulatory or coding regions of *hGGPS*, such as pathologies related to a defect in the mevalonic biosynthetic pathway.

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INTERNATIONAL SEARCH REPORT

International Application No

PCT/IB 99/01353

A. CLASSIFICATION OF SUBJECT MATTER

IPC 7 C12N15/54 C12N9/10 C12Q1/68 C07K16/40 G01N33/50

According to International Patent Classification (IPC) or to both national classification and IPC

B. FIELDS SEARCHED

Minimum documentation searched (classification system followed by classification symbols)

IPC 7 C12Q C12N

Documentation searched other than minimum documentation to the extent that such documents are included in the fields searched

Electronic data base consulted during the international search (name of data base and, where practical, search terms used)

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Y	abstract; example 1	25,26
X	WO 97 45535 A (FILIPOWICZ WITOLD ;CIBA GEIGY AG (CH); GENSCHIK PASCAL (FR)) 4 December 1997 (1997-12-04) abstract * see especially SEQ ID NO:8, positions 268-273 * page 51 -page 53	15,38
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Patent family members are listed in annex.

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Date of the actual completion of the international search

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INTERNATIONAL SEARCH REPORT

International Application No

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C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT		
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A	OHNUMA S -I ET AL: "ARCHAEBACTERIAL ETHER-LINKED LIPID BIOSYNTHETIC GENE. EXPRESSION CLONING, SEQUENCING, AND CHARACTERIZATION OF GERANYLGERANYL-DIPHOSPHATE SYNTHASE" JOURNAL OF BIOLOGICAL CHEMISTRY,US,AMERICAN SOCIETY OF BIOLOGICAL CHEMISTS, BALTIMORE, MD, vol. 269, no. 20, 20 May 1994 (1994-05-20), pages 14792-14797, XP002030964 ISSN: 0021-9258 the whole document	
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