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(71) Applicants: **PXE INTERNATIONAL, INC.** [US/US]; 23 Mountain Street, Sharon, MA 02067 (US). **UNIVERSITY OF HAWAII** [US/US]; 2800 Woodlawn Drive, Suite 280, Honolulu, HI 96822 (US).

(72) Inventors: **BOYD, Charles, D.**; 3330 Paty Drive, Honolulu, HI 96822 (US). **CSISZAR, Katalin**; 3330 Paty Drive, Honolulu, HI 96822 (US). **LESAUX, Olivier**; 2745 Terrace Drive, Apartment A, Honolulu, HI 96822 (US). **URBAN, Zsolt**; 2630 Dole Street, Apartement A1, Honolulu, HI 96822 (US). **TERRY, Sharon**; 23 Mountain Street, Sharon, MA 02067 (US).

(74) Agent: **WALLER, Patrick, R., H.**; Testa, Hurwitz & Thibeault, LLP, High Street Tower, 125 High Street, Boston, MA 02110 (US).

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(54) Title: METHODS AND COMPOSITION FOR DIAGNOSING AND TREATING *PSEUDOXANTHOMA ELASTICUM* AND RELATED CONDITIONS

(57) Abstract: Methods and compositions are provided for diagnosing and treating *Pseudoxanthoma elasticum* (PXE) patinet and PXE carriers. Methods and compositions are based on the discovery that PXE mutations are located in the MRP6 (ABCC6) gene.

INTERNATIONAL SEARCH REPORT

Inte onal Application No
PCT/US 01/05741

A. CLASSIFICATION OF SUBJECT MATTER
IPC 7 C12Q1/68 G01N33/53

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 G01N C12N C07K

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)
EPO-Internal, BIOSIS

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category °	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
A	LE SAUX O ET AL: "Pseudoxanthoma Elasiticum maps to an 820-kb region of the p13.1 region of chromosome 16" GENOMICS, vol. 61, no. 1, 15 November 1999 (1999-11-15), pages 1-10, XP002201011 the whole document ---	1
A	COTTON R G H: "CURRENT METHODS OF MUTATION DETECTION" MUTATION RESEARCH, AMSTERDAM, NL, vol. 285, 1993, pages 125-144, XP000443992 ISSN: 0027-5107 the whole document --- -/--	1-33

[X] Further documents are listed in the continuation of box C.

[X] Patent family members are listed in annex.

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"A" document defining the general state of the art which is not considered to be of particular relevance

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Date of the actual completion of the international search	Date of mailing of the international search report
4 June 2002	18/07/2002

Name and mailing address of the ISA	Authorized officer
European Patent Office, P.B. 5818 Patentlaan 2 NL - 2280 HV Rijswijk Tel. (+31-70) 340-2040, Tx. 31 651 epo nl, Fax: (+31-70) 340-3016	Osborne, H

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C.(Continuation) DOCUMENTS CONSIDERED TO BE RELEVANT

Category °	Citation of document, with indication,where appropriate, of the relevant passages	Relevant to claim No.
A	WO 98 37764 A (BAYLOR COLLEGE MEDICINE ;UNIV UTAH (US); US HEALTH (US); UNIV JOHN) 3 September 1998 (1998-09-03)	
T	LE SAUX ET AL: "Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum" NATURE GENETICS, vol. 25, - June 2000 (2000-06) XP002201012 page 2	1-33
T	RINGPFEIL F ET AL: "Pseudoxanthoma elasticum: Mutations in the MRP6 gene encoding a transmembrane ATP-binding cassette (ABC) transporter" PROC. NATL. ACAD. SCI. USA, vol. 97, May 2000 (2000-05), pages 6001-6, XP002201013 the whole document	1-33

INTERNATIONAL SEARCH REPORT

Information on patent family members

Inte onal Application No
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Patent document cited in search report	Publication date	Patent family member(s)	Publication date
WO 9837764 A	03-09-1998	AU 742094 B2	20-12-2001
		AU 6538698 A	18-09-1998
		EP 0989805 A1	05-04-2000
		JP 2001516209 T	25-09-2001
		WO 9837764 A1	03-09-1998