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(71) Applicant (for all designated States except US): **DECODE GENETICS EHF** [IS/IS]; Sturlugata 8, IS-101 Reykjavik (IS).

(72) Inventors; and

(75) Inventors/Applicants (for US only): **SULEM, Patrick** [FR/IS]; Eskihlid 22, IS-105 Reykjavik (IS). **GUDB-JARTSSON, Daniel** [IS/IS]; Sogavegur 38, IS-108 Reykjavik (IS).

(74) Agent: **JONSSON, Thorlakur**; deCODE Genetics ehf., Sturlugata 8, IS-101 Reykjavik (IS).

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Declarations under Rule 4.17:

— of inventorship (Rule 4.17(iv))

Published:

— with international search report (Art. 21(3))

— before the expiration of the time limit for amending the claims and to be republished in the event of receipt of amendments (Rule 48.2(h))

— with sequence listing part of description (Rule 5.2(a))

(88) Date of publication of the international search report:
13 August 2009

(54) Title: SEQUENCE VARIANTS FOR INFERRING HUMAN PIGMENTATION PATTERNS

(57) Abstract: The present invention discloses variants that are predictive of human pigmentation patterns. The invention further relates to variants that are useful for determining risk of skin cancer, including melanoma and basal cell carcinoma. The disclosed variants can be utilized for the determination of the natural pigmentation patterns of a human individual, and for determining a susceptibility to melanoma and basal cell carcinoma, from a sample of genetic material. Methods and kits including the variants described are useful in e.g. forensic testing and diagnostic applications.



INTERNATIONAL SEARCH REPORT

International application No
PCT/IS2008/000017

A. CLASSIFICATION OF SUBJECT MATTER

INV. C12Q1/68

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)

C12Q

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, EMBASE, WPI Data

C. DOCUMENTS CONSIDERED TO BE RELEVANT

Category*	Citation of document, with indication, where appropriate, of the relevant passages	Relevant to claim No.
X	LANDI MARIA TERESA ET AL: "MC1R, ASIP, and DNA repair in sporadic and familial melanoma in a Mediterranean population" JOURNAL OF THE NATIONAL CANCER INSTITUTE (CARY), vol. 97, no. 13, July 2005 (2005-07), pages 998-1007, XP002518138 ISSN: 0027-8874 table 1 page 1000, column 1, paragraph 2 page 1001, column 2, paragraph 2 page 1003, column 2, paragraph 1 abstract the whole document ----- -/--	1-3, 7-31, 35-44, 49-52, 57-68, 87-89

☒ Further documents are listed in the continuation of Box C.

☒ See patent family annex.

* Special categories of cited documents:

- *A* document defining the general state of the art which is not considered to be of particular relevance
- *E* earlier document but published on or after the international filing date
- *L* document which may throw doubts on priority claim(s) or which is cited to establish the publication date of another citation or other special reason (as specified)
- *O* document referring to an oral disclosure, use, exhibition or other means
- *P* document published prior to the international filing date but later than the priority date claimed

- *T* later document published after the international filing date or priority date and not in conflict with the application but cited to understand the principle or theory underlying the invention
- *X* document of particular relevance; the claimed invention cannot be considered novel or cannot be considered to involve an inventive step when the document is taken alone
- *Y* document of particular relevance; the claimed invention cannot be considered to involve an inventive step when the document is combined with one or more other such documents, such combination being obvious to a person skilled in the art.
- *G* document member of the same patent family

Date of the actual completion of the international search

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Name and mailing address of the ISA/

European Patent Office, P.B. 5818 Patentlaan 2
NL - 2280 HV Rijswijk
Tel. (+31-70) 340-2040,
Fax: (+31-70) 340-3016

Authorized officer

Helliot, Bertrand

INTERNATIONAL SEARCH REPORT

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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	FARGNOLI MARIA CONCETTA ET AL: "Re: MC1R, ASIP, and DNA repair in sporadic and familial melanoma in a Mediterranean population" JOURNAL OF THE NATIONAL CANCER INSTITUTE (CARY), vol. 98, no. 2, January 2006 (2006-01), pages 144-145, XP002518139 ISSN: 0027-8874 table 1 abstract the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
A	WO 01/62964 A (ISIS INNOVATION [GB]; WINSEY SAMANTHA [GB]; HALDAR NEIL [GB]; WOJNAROW) 30 August 2001 (2001-08-30) table 3 the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
A	US 2003/175721 A1 (BOX NEIL F [AU] ET AL) 18 September 2003 (2003-09-18) claims 4,5 the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
A	US 2005/064440 A1 (ROTH RICHARD B [US] ET AL) 24 March 2005 (2005-03-24) figure 1 the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
A	US 2006/051768 A1 (HOON DAVE S [US] ET AL) 9 March 2006 (2006-03-09) paragraphs [0011], [0012] the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89

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INTERNATIONAL SEARCH REPORT

International application No
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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	US 7 166 427 B1 (LANG JAS C [US]) 23 January 2007 (2007-01-23) column 3, lines 54-64 the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
A	EP 1 835 036 A (EXONHIT THERAPEUTICS SA [FR]; HOPITAUX UNIVERSITAIRES DE GEN [CH]; UNI) 19 September 2007 (2007-09-19) paragraphs [0036], [0050] the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
P,X	SULEM PATRICK ET AL: "Genetic determinants of hair, eye and skin pigmentation in Europeans" NATURE GENETICS, vol. 39, no. 12, December 2007 (2007-12), pages 1443-1452, XP002518141 ISSN: 1061-4036 the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89
P,X	GUDBJARTSSON DANIEL F ET AL: "ASIP and TYR pigmentation variants associate with cutaneous melanoma and basal cell carcinoma" NATURE GENETICS, vol. 40, no. 7, July 2008 (2008-07), pages 886-891, XP002518142 ISSN: 1061-4036 the whole document	1-3, 7-31, 35-44, 49-52, 57-68, 87-89

INTERNATIONAL SEARCH REPORT

International application No.
PCT/IS2008/000017

Box No. II Observations where certain claims were found unsearchable (Continuation of item 2 of first sheet)

This international search report has not been established in respect of certain claims under Article 17(2)(a) for the following reasons:

1. ☐ Claims Nos.:
because they relate to subject matter not required to be searched by this Authority, namely:

2. ☐ Claims Nos.:
because they relate to parts of the international application that do not comply with the prescribed requirements to such an extent that no meaningful international search can be carried out, specifically:

3. ☐ Claims Nos.:
because they are dependent claims and are not drafted in accordance with the second and third sentences of Rule 6.4(a).

Box No. III Observations where unity of invention is lacking (Continuation of item 3 of first sheet)

This International Searching Authority found multiple inventions in this international application, as follows:

see additional sheet

1. ☐ As all required additional search fees were timely paid by the applicant, this international search report covers allsearchable claims.

2. ☐ As all searchable claims could be searched without effort justifying an additional fees, this Authority did not invite payment of additional fees.

3. ☐ As only some of the required additional search fees were timely paid by the applicant, this international search report covers only those claims for which fees were paid, specifically claims Nos.:

4. ☒ No required additional search fees were timely paid by the applicant. Consequently, this international search report is restricted to the invention first mentioned in the claims; it is covered by claims Nos.:

1 (part), 2, 3 (part), 7-16, 17-18 (part), 19-31, 35-36, 37 (part), 38-39
40-41 (part), 42, 43 (part), 44, 49-51, 52 (part), 57-60, 61 (part), 62
63-64 (part), 65-67, 68 (part), 87-89

Remark on Protest

- ☐ The additional search fees were accompanied by the applicant's protest and, where applicable, the payment of a protest fee.
- ☐ The additional search fees were accompanied by the applicant's protest but the applicable protest fee was not paid within the time limit specified in the invitation.
- ☐ No protest accompanied the payment of additional search fees.

FURTHER INFORMATION CONTINUED FROM PCT/ISA/ 210

This International Searching Authority found multiple (groups of) inventions in this international application, as follows:

1. claims: 1 (part), 2, 3 (part), 7-16, 17-18 (part), 19-31, 35-36, 37 (part), 38-39, 40-41 (part), 42, 43 (part), 44, 49-51, 52 (part), 57-60, 61 (part), 62, 63-64 (part), 65-67, 68 (part), 87-89.

- Methods of determining a susceptibility to a skin cancer in a human individual;
 - A method of screening a candidate marker for assessing susceptibility to at least one skin cancer selected from the group consisting of melanoma, basal cell carcinoma and squamous cell carcinoma;
 - A method of identification of a marker for use in assessing susceptibility to at least one skin cancer selected from melanoma, basal cell carcinoma and squamous cell carcinoma;
 - A method of genotyping a nucleic acid sample obtained from a human individual;
 - A kit for assessing susceptibility to at least one skin cancer selected from melanoma, basal cell carcinoma and squamous cell carcinoma in a human individual;
 - A computer-readable medium having computer executable instructions for determining susceptibility to a skin cancer selected from melanoma, basal cell carcinoma and squamous cell carcinoma, the computer readable medium;
 - An apparatus for determining a genetic indicator for a skin cancer selected from melanoma, basal cell carcinoma and squamous cell carcinoma in a human individual;
- wherein at least one polymorphic marker associated with the ASIP gene is used and the marker is rs1015362, rs4911414 or rs6060043.

2. claims: 1 (part), 2, 3-4 (part), 5, 7-11, 15-16, 17-18 (part), 19-36, 37 and 40-41 (part), 42, 43 (part), 44-46, 52 (part), 53-54, 60, 61 (part), 62, 63-64 (part), 65-67, 68 (part), 87-89.

- The same methods, kit, computer-readable medium and apparatus as hereinabove wherein at least one polymorphic marker associated with the TYR gene is used and the marker is rs1126809 or rs1393350.

3. claims: 1 (part), 2, 3-4 (part), 6-11, 15-16, 17-18 (part), 19-31, 35-36, 37 and 40-41 (part), 42, 43 (part), 44, 47-48, 52 (part), 55-56 and 60, 61 (part), 62, 63-64 (part), 65-67, 68 (part), 87-89.

FURTHER INFORMATION CONTINUED FROM PCT/ISA/ 210

- The same methods, kit, computer-readable medium and apparatus as hereinabove wherein the at least one polymorphic marker associated with the TYRP1 gene is rs1408799.

4. claims: 69-89

- A method of inferring at least one pigmentation trait of a human individual;
- A kit.

INTERNATIONAL SEARCH REPORT

Information on patent family members

International application No

PCT/IS2008/000017

Patent document cited in search report		Publication date	Patent family member(s)	Publication date
WO 0162964	A	30-08-2001	AU 3393301 A	03-09-2001
US 2003175721	A1	18-09-2003	NONE	
US 2005064440	A1	24-03-2005	NONE	
US 2006051768	A1	09-03-2006	NONE	
US 7166427	B1	23-01-2007	NONE	
EP 1835036	A	19-09-2007	AU 2007226234 A1	20-09-2007
			CA 2646269 A1	20-09-2007
			EP 1996727 A2	03-12-2008
			WO 2007105114 A2	20-09-2007