# ORIGINAL ARTICLE

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# Catalog of 605 single-nucleotide polymorphisms (SNPs) among 13 genes encoding human ATP-binding cassette transporters: ABCA4, ABCA7, ABCA8, ABCD1, ABCD3, ABCD4, ABCE1, ABCF1, ABCG1, ABCG2, ABCG4, ABCG5, and ABCG8

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Abstract Single-nucleotide polymorphisms (SNPs) at some gene loci are useful as markers of individual risk for adverse drug reactions or susceptibility to complex diseases. We have been focusing on identifying SNPs in and around genes encoding drug-metabolizing enzymes and transporters, and have constructed several high-density SNP maps of such regions. Here we report SNPs at additional loci, specifically 13 genes belonging to the superfamily of ATPbinding cassette transporters (ABCA4, ABCA7, ABCA8, ABCD1, ABCD3, ABCD4, ABCE1, ABCF1, ABCG1, ABCG2, ABCG4, ABCG5, and ABCG8). Sequencing a total of 416kb of genomic DNA from 48 Japanese volunteers identified 605 SNPs among these 13 loci: 14 in 5' flanking regions, 5 in 5' untranslated regions, 37 within coding elements, 529 in introns, 8 in 3' untranslated regions, and 12 in 3' flanking regions. By comparing our data with SNPs deposited in the dbSNP database of the National Center for Biotechnology Information (US) and with published reports, we determined that 491 (81%) of the SNPs reported here were novel. We also detected 107 genetic variations of other types among the loci examined (insertion-deletions or mono- di-, or trinucleotide polymorphisms). The high-density SNP maps we constructed on the basis of these data should provide useful information for investigating associations between genetic variations and common diseases or responsiveness to drug therapy.

**Key words** Single-nucleotide polymorphisms (SNPs) · Insertion-deletion polymorphisms · High-density SNP

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maps · ATP-binding cassette transporter genes · Japanese population

## Introduction

Single-nucleotide polymorphisms (SNPs) at some gene loci can be useful markers of individual risk for adverse drug reactions or susceptibility to complex diseases, because an SNP itself may influence the quality and/or quantity of a gene product (see a review by McCarthy and Hilfiker 2000). With this in mind, investigators in many laboratories have undertaken projects to establish large collections of SNPs across the human genome (Sachidanandam et al. 2001). In the near future, the genomic approach, combining the large collection of SNPs and high-throughput genotyping procedures based on that information, is likely to revolutionize drug discovery and development as well as the practice of medicine (Gray et al. 2000; Meyer 2000; Roses 2000). Recently, we have focused on genetic loci corresponding to drug-metabolizing enzymes and transporters and have already described more than 1500 variations in those regions (Iida et al. 2001a-e, 2002a,b; Saito et al. 2001a,b, 2002a,b; Sekine et al. 2001).

The ATP-binding cassette (ABC) transporter superfamily is one of the largest gene families. These genes encode a functionally diverse group of membrane proteins involved in energy-dependent transport of a wide variety of substrates across membranes (see reviews by Allikmets et al. 1996; Broccardo et al. 1999; Klein et al. 1999; Dean et al. 2001). Eukaryotic ABC transporters are organized either as full transporters containing two transmembrane domains and two nucleotide-binding folds, or as half-transporters. Molecules of the latter type must form either homodimers or heterodimers to constitute functional transporters. The entire group is divided into seven distinct subfamilies on the basis of predicted protein structures and phylogenetic analysis. So far, a total of 48 human transporter genes belonging to one or other of the seven ABC subfamilies has been reported (http://nutrigene.4t.com/humanabc.htm).

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Some genetic variations present in genes that encode drug transporters and drug-metabolizing enzymes are likely to be associated with susceptibility to common diseases as well as with differences in therapeutic efficacy and/or side effects of drugs among individuals. Therefore, information concerning genetic variations in human transporter genes should be an important resource for understanding not only the etiology and risk of some diseases, but also the pharmacokinetics or pharmacodyamics of drugs. Regarding SNP analysis in the ABC transporter genes, we previously reported the construction of high-density SNP maps in the genomic segments corresponding to nine ABC transporter subfamily B genes, and eight ABC transporter subfamily C genes (Saito et al. 2002a,b). In this article, we provide highresolution maps of 13 ABC transporter (subfamilies A, D, E, F, and G) gene loci containing a total of 605 SNPs and 107 insertion-deletion polymorphisms that we detected in DNA from 48 Japanese volunteers.

# Subjects and methods

Blood samples were obtained with written informed consent from 48 healthy Japanese volunteers for this study, which was approved by the ethical committee of the RIKEN SNP Research Center. The detailed methods used to screen for SNPs are available from our website (http://snp.ims.u-tokyo.ac.jp/). In brief, we sequenced 96 chromosomes to screen SNPs using polymerase chain reaction (PCR) primers to amplify an entire gene, including about 2kb upstream of the first exon and downstream from the last exon, but excluding repetitive sequences. Each PCR was carried out using 20ng of pooled DNA for three individuals. All SNPs detected by the PolyPhred computer program (Nickerson et al. 1997) were confirmed by sequencing both strands of each PCR product.

Genbank accession numbers for the 13 *ABC* loci examined are as follows:

ABCA4: NT\_019258.1 ABCA7: NT\_025194.1 ABCA8: AC005922.1 and AC015844.5 ABCD1: U52111.2 ABCD3: NT\_019284.3 and X83467.1 ABCD4: AC005519.3 ABCE1: NT\_006296.2 ABCF1: NT\_007592.3 ABCG1: AP001746.1 ABCG2: NT\_022959.2 ABCG4: AP001315.3 and AP001182.2 ABCG5: AC084265.2 and AC11242.8 ABCG8: AC084265.2

# **Results and discussion**

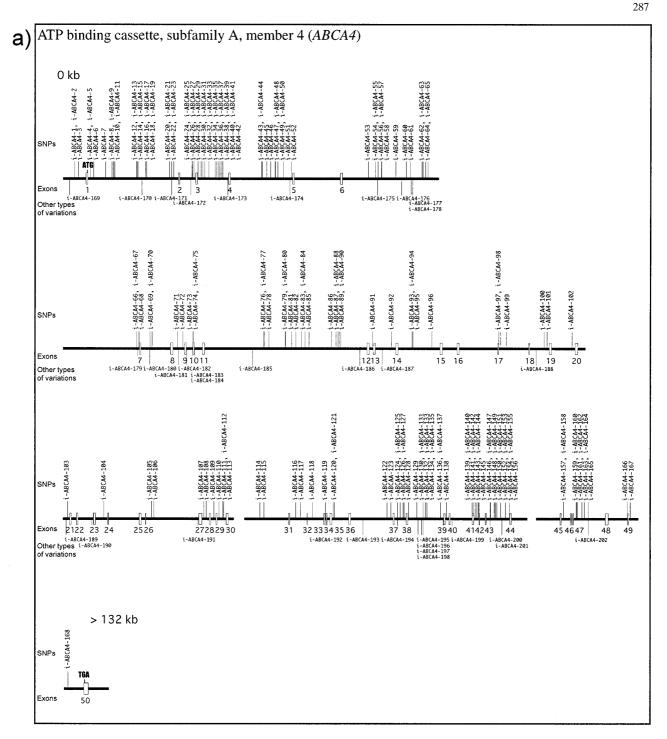
Sequencing of about 416kb of genomic DNA corresponding to 13 loci containing *ABC* transporter genes identified a total of 712 variations among 96 Japanese chromosomes, including 605 SNPs and 107 variations of other types. The exon-intron organization of each gene and locations of SNPs identified within each locus are illustrated schematically in Fig. 1; detailed information is given in Table 1. The classification of SNPs on the basis of their locations is also summarized in Table 2. Among the 605 SNPs, 14 were located in 5' flanking regions, 5 in 5' untranslated regions, 37 in coding regions, 529 in introns, 8 in 3' untranslated regions, and 12 in 3' flanking regions. The 17 nonsynonymous SNPs were among 37 coding SNPs we found. Although the frequency of nonsynonymous SNPs was a very small part of the total SNP counts (2.8%), the SNPs would likely influence protein function. By comparing our data with reports from elsewhere (Maugeri et al. 1999; Dvorakova et al. 2001; Hubacek et al. 2001; Lee et al. 2001; Lu et al. 2001) and the SNPs deposited in the dbSNP database at the National Center for Biotechnology Information, we were able to consider 491 of the 605 SNPs (81%) to be novel (Tables 1 and 2). In addition, the overall frequencies of nucleotide substitutions were counted as 40% for A/G, 33.9% for C/T, 8.6% for A/C, 8.1% for C/G, 6.1% for G/T, and 3.3% for T/A (Table 3).

#### ABCA genes

The ABC subfamily A comprises a group of full-size transporters characterized by the presence of a highly hydrophobic segment between two transmembrane domains. On the basis of database for "expressed sequence tags" (dbEST) information, current estimates suggest that at least 11 *ABCA* genes are present in the human genome (Broccardo et al. 1999).

ABCA4 locus. The ABCA4 (sometimes referred as ABCR) gene is expressed highly and exclusively in the retina, and encodes the outer-segment rim protein of rods (Allikmets et al. 1997). Mutations of ABCA4 have been implicated in a variety of retinal degenerative diseases associated with loss of vision (Klein et al. 1999; Dean et al. 2001). By screening approximately 84.5kb at the ABCA4 locus, we identified a total of 168 SNPs; 3 were in the 5' flanking region, 4 were in coding regions, and 161 were in introns. No SNPs were found in 5' untranslated, 3' untranslated, or 3' flanking regions. The average distribution of SNPs at this locus was 1 in every 503 nucleotides. The frequency of each type of substitution was 39.9% for A/G, 32.1% for C/T, 11.3% for A/C, 8.3% for C/G, 6.0% for G/T, and 2.4% for T/A. We also found 34 variations of other types at the ABCA4 locus.

*ABCA7* locus. The *ABCA7* gene encodes a 2146-aminoacid peptide (Kaminski et al. 2000). Northern blotting has revealed predominant expression in myelolymphatic tissues such as bone marrow, thymus, spleen, and peripheral blood. *ABCA7* is a sterol-sensitive gene that is inversely regulated by cholesterol import and export in macrophages; this regulatory response to cholesterol influx and efflux is similar to that of *ABCA1* and *ABCG1*. Hence, Kaminski et



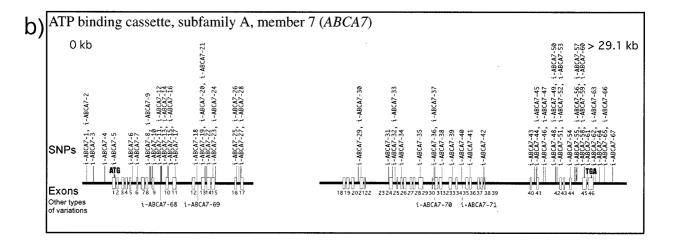
**Fig. 1a–I.** Genomic organizations and locations of single-nucleotide polymorphisms (SNPs) in 13 regions containing ATP-binding cassette transporter genes. Exons and introns are represented by *rectangles* and

*horizontal lines*, respectively. SNPs are indicated *above* the genes (designations correspond to those in the left-most column in Table 1). Other types of variations are indicated *below* the genes

al. (2000) suggested that ABCA7 might function as a cellular lipid exporter. By screening an approximately 21.2-kb region at the ABCA7 locus, we identified a total of 67 SNPs (4 in the 5' flanking region, 1 in the 5' untranslated region, 19 in coding regions, 37 in introns, 1 in the 3' untranslated region, and 5 in the 3' flanking region). The distribution of SNPs at this locus was 1 per 316bp on average. The frequency of each type of substitution was

44.8% for A/G, 29.9% for C/T, 7.5% for A/C, 13.4% for C/G, 3.0% for G/T, and 1.5% for T/A. We also found four other variations at the *ABCA7* locus.

*ABCA8* locus. *ABCA8*, located in the *ABCA*-transporter cluster of genes on chromosome 17q24, was isolated from human brain libraries during cDNA-sequencing projects (Nagase et al. 1998; Dean et al. 2001). ABCA8 may function



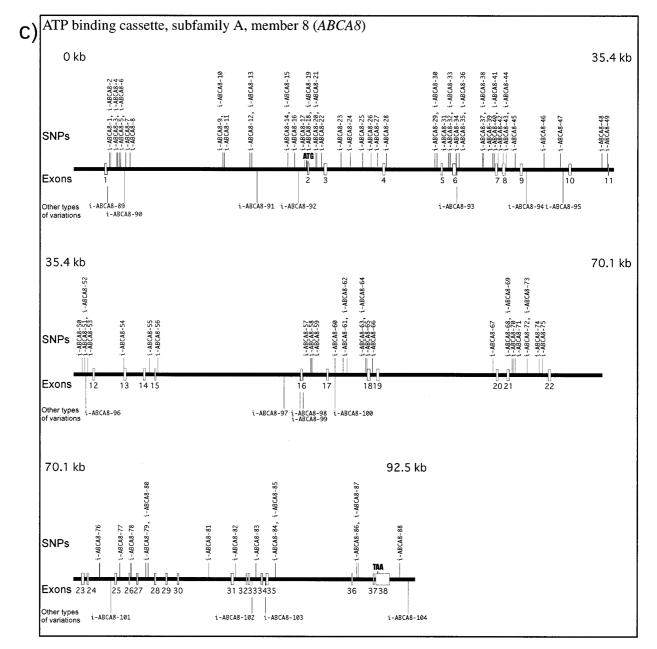
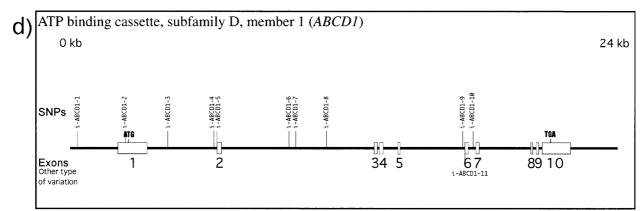
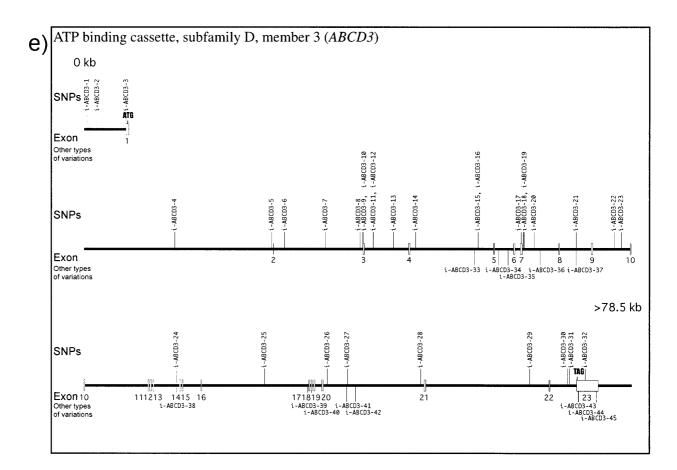
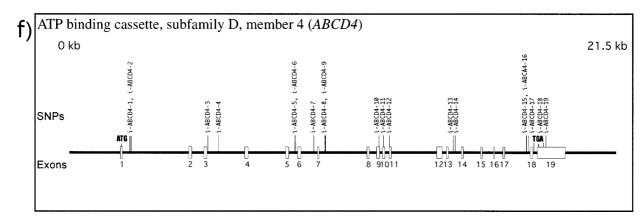


Fig. 1a–I. Continued







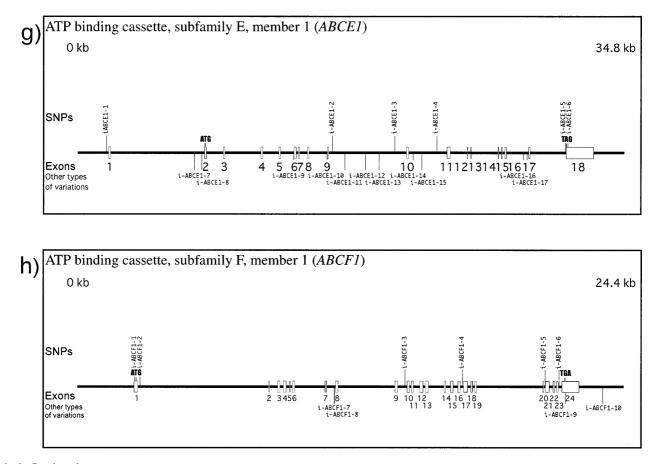


Fig. 1a-l. Continued

as a lipid transporter, but that has not been proven. Screening a region of approximately 57.7kb encompassing the *ABCA8* gene detected a total of 88 SNPs: 3 in coding regions, 84 in introns, and 1 in the 3' flanking region. The distribution of SNPs at this locus was 1 per 656bp on average. The frequency of each type of substitution was 40.9% for A/G, 33.0% for C/T, 5.7% for A/C, 10.2% for C/G, 5.7% for G/T, and 4.5% for T/A. We also found 16 variations of other types at this locus.

# ABCD genes

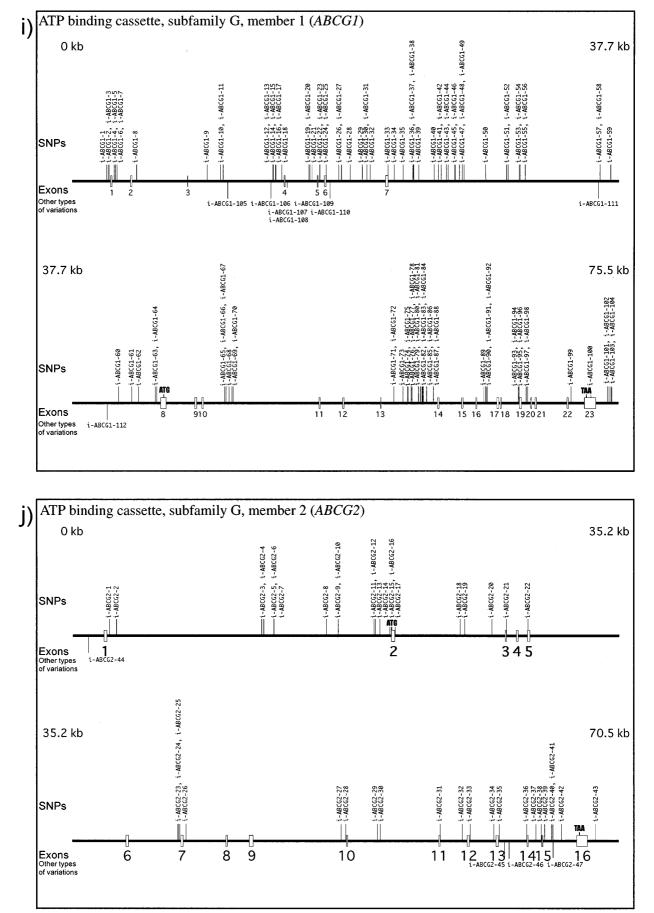
The ABCD subfamily at present consists of four genes, *ABCD1*, *ABCD2*, *ABCD3*, and *ABCD4*; all of them encode half-transporters located in the peroxisome, where their homo- and/or heterodimers regulate transport of very long chain fatty acids (Dean et al. 2001).

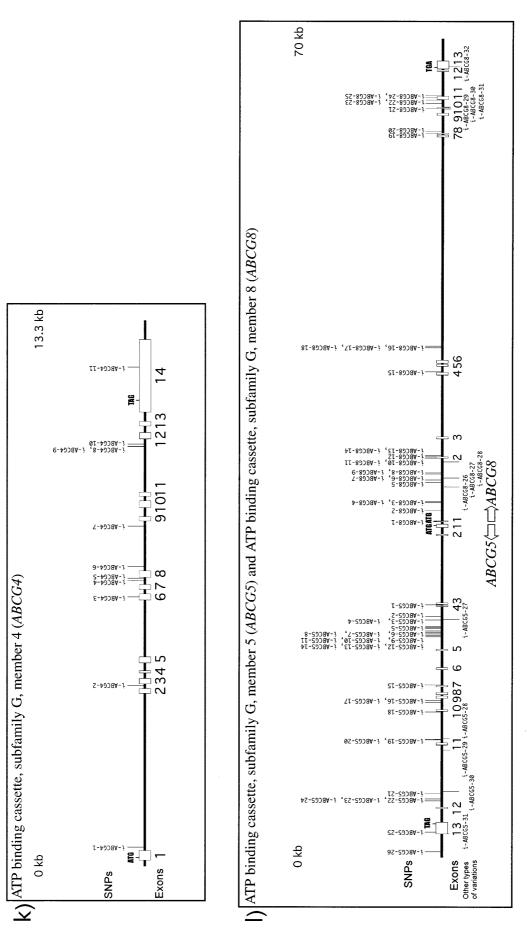
*ABCD1* locus. Positional cloning has characterized *ABCD1*, encoding a 745-amino-acid protein, as the "causative gene" for X-linked adrenoleukodystrophy (Mosser et al. 1993). ABCD1 may play a role in the import or anchoring of the peroxisome enzyme VLCF-CoA synthase (Dodd et al. 1997). Screening of an approximately 15-kb region containing the *ABCD1* gene identified a total of 10 SNPs: 1 in the 5' flanking region, 1 in the 5'

untranslated region, and 8 in introns. The distribution of SNPs at this locus was 1 per 1499 bp on average. The frequency of each type of substitution was 30% for A/G and 70% for C/T. We also found a one-base deletion polymorphism in intron 5.

*ABCD3* locus. *ABCD3* gene encodes a 70-kDa peroxisomal membrane protein (PXMP70) in the liver, and its mutations are responsible for Zellweger syndrome in some patients with this lethal inborn defect of peroxisome assembly (Gartner et al. 1992). Screening of approximately 37.1kb encompassing the *ABCD3* gene identified a total of 32 SNPs: 2 in the 5' flanking region, 1 in the 5' untranslated region, 2 in coding regions, 26 in introns, and 1 in the 3' untranslated region. The distribution of SNPs at this locus was 1 per 1158 bp on average. The frequency of each type of substitution was 53.1% for A/G, 28.1% for C/T, 9.4% for A/C, 3.1% for G/T, and 6.3% for T/A. We also found 13 variations of other types at this locus.

*ABCD4* locus. ABCD4, a 606-amino-acid protein located at the peroxisomal membrane, is a half-ABC transporter (Holzinger et al. 1997), but its amino acid sequence is not highly homologous to ABCD1 (24.5%), ABCD2 (24.9%), or ABCD3 (27.4%) (Shani et al. 1997). Screening approximately 13.4kb around the *ABCD4* gene detected a







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i-ABCA4-2	5' Flanking region	-819	of of a dark of the address	AT	0,00,000000000000000000000000000000000			
i-ABCA4-3	5' Flanking region	-680	accedanaccececeduagua agracracceattgraggg	CIA	toreatractoryser tradeg			
i-ABCA4-4	Intron 1	208	tqcccttcccaqqaaqatqt	GIA	tttctctctctcaccaca			
i-ABCA4-5	Intron 1	234	ctatcctcaaccacatgaaa	AG	tetttaectaecataecta			
i-ABCA4-6	Intron 1	510	ageteacgateaagteacag	1/C	ttaactggacacattatttt			
i-ABCA4-7	Intron 1	1527	gcttaacaaccagcataaaa	G/A	agagcagcatgggacacgct			
i-ABCA4-8	Intron 1	2077	caggactgtagctgctggcc	T/C	aaaatgagcccattcctgtg			
i-ABCA4-9	Intron 1	2174	ccctctcaatctggcctttc	G/C	ctggcatgggtgggcgactc			
i-ABCA4-10	Intron 1	2246	gctcccagggagatggagcc	A/G	ctcgggctgagggccttggc			
i-ABCA4-11	Intron 1	2364	ttctgtctggcacgcctccc	G/A	atggctccccacctgctacc			
i-ABCA4-12	Intron 1	4243	ctccctggggtatgcctgta	C/G	gcagttaagcgtcaaggaca			
i-ABCA4-13	Intron 1	4287	atgccgctctggggggggg	AC	gctgagcatgattttggaag			
i-ABCA4-14	Intron 1	4309	ctgagcatgattttggaagc	СЛ	ggcagaagggctattgtga			
i-ABCA4-15	Intron 1	4416	tgcagcaaccgcccccgccc	СЛ	ccgccaaaaacaaacacact			
i-ABCA4-16	Intron 1	4996	tttacccctggaacaggcag	G/A	ccaagctggcT/Cggtcccctc			
i-ABCA4-17	Intron 1	5007	aacaggcagG/Accaagctggc	1/C	ggtcccctccctgatacaca			
i-ABCA4-18	Intron 1	5080	gtgtgtggctggtttcttag	C/G	aagcaccatggttccaagtt			
i-ABCA4-19	Intron 1	5152	gggagatgaacgtaagtgga	G/A	ggcaggcctacaaggttgca			
i-ABCA4-20	Intron 1	7110	ccactggatctgcttttgga	AG	tcaagagtccttaagctcca			
i-ABCA4-21	Intron 1	7290	gattttgttggctttgcaa	T/A	ggatcacagtcatttattca			
i-ABCA4-22	Intron 1	7483	tctgagcctctttccttaac	T/C	gcagagtgagtggC/Ttacaga		+	
i-ABCA4-23	Intron 1	7497	cttaacT/Cgcagagtgagtgg	СЛ	tacagagaaatctttactac			
i-ABCA4-24	Intron 2	1067	tcaagcagcagcagcaactg	CIA	gtggagtcttcttgaactaa			
i-ABCA4-25	Intron 2	1106	aacactcctatgcccctctc	GIA	gcacaaaatgacG/Atgtcccc			dbSNP ID:rs1889404
i-ABCA4-26	Intron 2	1119	ccctctcG/Agcacaaaatgac	GIA	tgtcccccttgcttcccct			dbSNP ID:rs1889405
i-ABCA4-27	Intron 2	1243	cacccagcacagggactggc	ЪТ	cacatgagatgctcctgctt		+	
i-ABCA4-28	Intron 3	26	tgttgagatccctaccatgc	AG	ggggaggaagttgcacaccc			
i-ABCA4-29	Intron 3	101	agcatggagcactgagtgtt	СЛ	ttgtggctttgctgagcccc			
i-ABCA4-30	Intron 3	330	tgcttgggtggagtgaatca	T/C	tgtaggagaaaactcagtt			
i-ABCA4-31	Intron 3	470	tgaagtcaggtttacaaagt	C/G	aagtttacttcttgggagaa			
i-ABCA4-32	Intron 3	634	tgaaaaccaatgaccctct	T/C	ccaagaaaaatggccacata			
i-ABCA4-33	Intron 3	1016	ccttggggggggctcagtatg	AG	ttcttccaggagaagcctgc			
i-ABCA4-34	Intron 3	1554	gaaagttgggtttcatgttt	T/C	gcactcacattatgagtgaa			
i-ABCA4-35	Intron 3	1686	ctagacattctcacagagcc	AG	agggcagcaaggggggggtc			
i-ABCA4-36	Intron 3	1823	ttcacctctctccatggacc	A/G	gtctccctgctcctcaatg			
i-ABCA4-37	Intron 3	1938	caaattcctgggaacaaatc	GIA	ggttgacccagcT/Gttattct			
i-ABCA4-38	Intron 3	1951	acaaatcG/Aggttgacccagc	T/G	ttattctccctgtcccatca			
i-ABCA4-39	Intron 3	2063	ggctgtcagagcctacctgc	GЛ	tgaatgggtggaaggG/Acagg			
i-ABCA4-40	Intron 3	2079	ctgcG/Ttgaatgggtggaggg	G/A	caggtctcagagaattgggt			
i-ABCA4-41	Intron 3	2186	agacacacagagcatgggac	СЛ	gagaggcgagcagaccctgc			
i-ABCA4-42	Intron 3	2214	gagcagaccctgccaaaact	GIA	ggagactgaatagatcgctc			
I-ABCA4-43	Intron 4	2717	cgtgcttctgcacagccacc	1/C	gggaaggtatgccgatggtt			dbSNP ID:rs1211213
i-ABCA4-44	Intron 4	2802	attctcagcagggaggatta	AG	tggtaaagcccaggaatgg			dbSNP ID:rs1209515
i-ABCA4-45	Intron 4	3182	cccccagagccacagcagcc	C/G	tgtctcctgggtggtcttgt			
i-ABCA4-46	Intron 4	3515	agtataataaagcaggagc	CT	atagcccccaactctcaaga			
i-ABCA4-47	Intron 4	3907	aggggagtgacagtgggcac	CIA	actctcagggaacccA/Gttac			dbSNP ID:rs570878
I-ABCA4-48	Intron 4	3923	gcacC/Aact ct cagggaaccc	AG	ttactgtgagagaagccact			dbSNP ID:rs570926
I-ABCA4-49	Intron 4	3952	agagaagccactgtgccact	G/C	tgtggtcgaacttcaagacc			
I-ABCA4-50	Intron 4	4125	ggctgtccagcacacagggg	CIA	aggcctcttggccactgggg			dbSNP ID:rs481931
I-ABCA4-51	Intron 4	463/	aatcacttgccccaaggtca	5	cttaactgttaggtgttctt		+	

Table 1. Characterization of variations in the 13 ABC transporter gene loci

M6C/4423         Introni         5.11         Anccorrespondence company         M6           M6C/4433         Introni         2.857         concertic sequence company         M6           M6C/4435         Introni         2.857         concertic sequence company         M6           M6C/4435         Introni         2.875         concertic sequence company         M6      <	Arg/His423Arg/His Arg/His423Arg/His73747 Arg/His423Arg/His73747 Arg/His423Arg/His Arg/His423Arg/His Arg/His423Arg/His
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Inton 381561grattiticatgraaattarc tattiticatgraaattarcIntron 381562tattiticatgraaattarcIntron 381574ccaagtgaacacagggIntron 381574tgectgaacacagggIntron 381574tgectgaacacagggIntron 382867tgectgaacacagggIntron 382874tgectgaacacagggIntron 39123gagggaacttgragggs//cagtcaIntron 401904ggaacacggaggs//cagtcaIntron 41123ggaactaaacggacactIntron 41287ttggttcccagtttratgIntron 41287ttggttcccagtttratgIntron 41433gttctcggagacagtttatcIntron 43376tttggagcagtttgtcccIntron 43376acctcccccacacattttIntron 43376tttggagcaggtggggggggggggggggggggggggggg	Intron 38		1526	ccaacatttgctaagcaccc	GIA	ccttcaaaaacctggtattt			dbSNP ID:rs567370
Intron 381562tattttcatgraattatcc/AIntron 381674ccagctgacacacagtgcIntron 381674ccagctgacacacagtgcIntron 382867tgcctggacacacagtgcIntron 382867tgcctggacacacagtgcIntron 39123ggagggacctgtgtggggggIntron 40123ggagggacctgtgtggggggIntron 41123ggaaataactgacacacagtgcIntron 41122ggaaataactgacaccagtgggggggggggggggggggg	Intron 38		1561	gtattttcatgtaaattatc	C/A	G/Aatacacagctgctatggaa			
Intron 38         16/4         ccagetgaaccccogroc           Intron 38         2867         tggcctgggaaccccagroc           Intron 38         287         tggcctgggcaggggyCagg           Intron 38         2874         tggcctgggcaggcggg           Intron 40         123         gaggggaccttgttgggcc           Intron 40         123         gaggggaccttgttgggcc           Intron 41         5814         ggagggaccttgttgggcc           Intron 41         121         gaggggaccttgttgggcc           Intron 41         123         gaggggaccttgttgggcc           Intron 41         287         tctggcggggggcctggtttgf           Intron 41         287         tctgcgggggggggggggggggggggggggggggggg	Intron 38		1562	tattttcatgtaaattatcC/A	G/A	atacacagctgctatggaaa			
Intron 38286/ 10100 381287/ 2874tgcctggtcggtgacaaggggIntron 382874ctggacaaggggA/CagctccIntron 401904ggaaataaagtgactgttcatgIntron 41122gacactgtacagccagcccaIntron 41122gacactgtacagccagccaIntron 41122gacactgtacagccagccaIntron 41122atttggttcccagtctatgIntron 41122atttggttcccagtctagIntron 41287tcttggttcccagtctagIntron 41287tcttggttcccagtctagIntron 43328ttttgtggttcccggtttctcIntron 43376cttcttgggtccggtttctcIntron 43376tttgtggtggtggggIntron 43376tttgtgggggggggggggggggggggggggggg	Intron 38		1674	ccagetgaacaecaegtgee	G/A	ggtgtgtgctgatataaaca		÷	dbSNP ID:rs486479
Intron 38         28/4         ctagacaaagggAV.Gagctcc           Intron 39         123         gagggacctgtgggcccg           Intron 41         58/4         ctagaccaagggacctgtgggcccg           Intron 41         130         904         gagactaaaactggacct           Intron 41         122         atttrggttcccaagtcaggccagc           Intron 41         122         atttrggttcccaggaccagc           Intron 41         122         atttrggttcccaggaccagc           Intron 41         287         tctgcaggactgggtccg           Intron 41         411         cctcttcccagaccagcagg           Intron 41         411         cctcttcgcaggctagg           Intron 43         328         tctgtagccattgg           Intron 43         376         cttgtagccacttgftctc           Intron 43         376         ctccaccacattfttd           Intron 43         376         ctccaccacattfttd           Intron 43         376         ctccaccacattfttd           Intron 43         376         ctccaccacattfttd           Intron 43         822         ggagggatgggcggcgcg           Intron 43         915         ggagggatggcgcgcdccaca           Intron 43         154         ggagggatggcccccccccta           Intron 43	Intron 38		2867	tgcctggctagacaaagggg	AC	agctccC/Tgcccactagaaac			dbSNP ID:rs565155
Intron 40     100     gagaattaaactgagaccug       Intron 41     287     uttggttcccaggttttatg       Intron 41     287     uttggttcccaggtttatg       Intron 41     287     uttggttcccaggtttatg       Intron 41     287     uttggttcccaggtttatg       Intron 41     287     uttggttcccaggtttatg       Intron 41     411     ctcttcccctcttgctct       Intron 41     413     gtcctaggtccgggtgggggggggggggggggggggggg	Intron 38 Intron 30		28/4	ctagacaaagggggA/Cagctcc	5	gcccactagaaacttgcagg			
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Cooling regionBoldggaaaraaaacgaaactuIntron 41122atttggttcocagttttatgIntron 41287tctgagagacgggtcagcIntron 41287cctcttcocctcttgctctIntron 41284cG/AtatcttgggttcctcIntron 41284cG/AtatcttgggttcctcIntron 41284cG/AtatcttgggttcctcIntron 43328ttttgraggcargggttggttcIntron 43376cctcctcccacatttttIntron 43376ctcccacacatttttIntron 43376ctcccacacatttttIntron 43376ctcccacacatttttIntron 43376ctcccacacatttttIntron 43376ctcccacacatttttIntron 43376ctcccacacatttttIntron 43377ggaggatgggctcgcccaaaIntron 431071gggaggatgggctcccacacattIntron 431071gggaggatgggctccccacacattIntron 431071gggaggatggctcccacacattIntron 431671ttgggggatggctcccacacattIntron 451671ttgggggatggctcccacacattIntron 451671ttgggggatggctcccgtaaaIntron 451671ttgggggatggccccacacattIntron 45176ggggggggggcccggatcccgtaattIntron 45176gggggggggggcccggatcccgtaattIntron 45176ggggggggggcccggatcccgtaattIntron 45176ggggggggggccgggccggggggggccgggggggggccgggg			1904	gacactgtacagccagccca	AC	tcctgaccccttttcttcat			
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Coding region         exon 42         5844         CG/Atatcttgeagatttatcc           Intron 43         328         tttgeagccaattgette           Intron 43         376         acctecctcaacaattgette           Intron 43         370         acctecctcaacaacttttt           Intron 43         370         acctecctcaacaacttttt           Intron 43         376         ctccaacacattttt           Intron 43         376         ctccaacacattttt           Intron 43         370         acctggagagaggetggetcccctaa           Intron 43         870         ggagggagaggetggetcccctaa           Intron 43         822         gttagggetggetggetccccctaa           Intron 43         815         ggraggagaggetggetccccctaa           Intron 43         815         ggraggagaggetccccctaact           Intron 43         815         ggraggagaggetccccccaact           Intron 43         1242         actgagctgacatct           Intron 43         1242         actgaggaggetccccagaaa           Intron 43         1242         actgaggaggaggacccccagaaa           Intron 43         174         tgagggaggagccccccagaaa           Intron 45         176         ggaggaggagcccccagaaa           Intron 45         176         ggaggagagtccccccaaccacccaccaccacc	Intron 41		443	gttctcagtccggtttcttc	G/A	tatcttgcagatttatccA/Gg			
Intron 43         328         tttgtagcctattcctata           Intron 43         345         ttaak/Gaatgcaccattgcttc           Intron 43         370         acctccctccaccacattttt           Intron 43         376         ctccaccacattttt           Intron 43         376         ctccaccacattttt           Intron 43         376         ctccacacacatttt           Intron 43         670         ttaaacagattgctccccta           Intron 43         670         ttaaacagatggctgctgcccacat           Intron 43         822         gagaggatgggctggctgctcacat           Intron 43         815         ggcgggacgggggcgcgagaa           Intron 43         1242         actgggggggggggggggggggggggggggggggggggg		on 42	5844	cG/Atatcttgcagatttatcc	AG	ggcacctccagcccagcagt	Pro1948Pro		Maugeri et al. (1999)
Intron 43         345         taak/Gaatgaccattycttc           Intron 43         370         acctccctccacacatttt           Intron 43         376         ctccacacacttttt           Intron 43         376         ctccacacacatttt           Intron 43         376         ctccacacacatttt           Intron 43         670         ttcaacacatttt           Intron 43         670         ttcaacagagatgagctcccaca           Intron 43         822         gagaggatgagctcccacaca           Intron 43         822         gtcaggacgacgacgatcccagaa           Intron 43         1242         actgagctggacgctgagaa           Intron 43         1571         ttgaggacgacgacgatatt           Intron 43         1671         ttgaggacgtgacgtcacacatt           Intron 45         1671         ttgaggacgtgacgtcacacatt           Intron 45         1673         gtgtttggttcacacagaa           Intron 45         176         gtggttggttcacacagtcc           Intron 45         176         gtggttggttcacacagtcc           Intron 45         176         gtggttggttcacacagtcc	Intron 43		328	ttgtagcctattcctataa	AG	aatgcaccattgcttcC/Gcat			
Intron 43         370         acctocctcacacacattttt           Intron 43         376         ctccacacacattttt           Intron 43         670         ttaaacagactggcccctaa           Intron 43         701         gagagatggccccctaactc           Intron 43         701         gagagatggccccctaactc           Intron 43         701         gagagatggccccctaactc           Intron 43         822         gttaggtgccgccccaactc           Intron 43         915         ggcagacgggcccccaactc           Intron 43         157         gggagagggccccccaactc           Intron 43         1571         tagaggagactcgccagaaa           Intron 43         1671         ttggaggaggccccagaaatt           Intron 45         1671         ttggaggagccccagaaatt           Intron 45         1671         ttggaggaactcccagraatt           Intron 45         176         grgtgttggttcaccagraatt           Intron 45         176         grgtgttggttcaccagactca           Intron 45         176         grgtgttggttcaccagactca           Intron 45         176         grggggaggreggaggtcccagacact           Intron 45         176         grggggggggggggggggggggggggggggggggggg	Intron 43		345	taaA/Gaatgcaccattgcttc	C/G	cattacctccctccacacat			
Intron 43         3/6         ctccacacatttttA/Gcaaaa           Intron 43         670         ttcaaacagactggtccccta           Intron 43         701         gggggatggctcgtccactc           Intron 43         701         gggggatggctccccaca           Intron 43         822         gttaggtgctcgacgact           Intron 43         915         ggcgggagggctgggccccccaca           Intron 43         1242         actgggctgggggcgggacgaca           Intron 43         1671         tggaggatggctccccgtaatt           Intron 45         1671         tggaggatgtccccgtaatt           Intron 45         1671         ttggaggatgtccccgtaatt           Intron 45         176         ggggagggtccccccgtaatt           Intron 45         176         ggggaggtccccccgtaatt           Intron 45         176         ggggaggtccccccgtaatt           Intron 45         176         grggttgggtccccccgtcaatt           Intron 45         176         grggttgggtccccccgtcaatt	Intron 43		370	acctccctccacacattttt	AG	caaaaC/Tgtttcagggagttt			
Intron 43         0.0         tradacagateggetcocccta           Intron 43         701         gagaggatagctctcactc           Intron 43         822         gttaggtgstggctgacccccta           Intron 43         815         ggraggatggstggctgacactc           Intron 43         1242         ggraggatggstggstcstgaca           Intron 43         1242         actggstctggacgatactcgaca           Intron 43         1242         actgaggstggaggtggatgg           Intron 43         1671         tagagagtttacttccctcactc           Intron 45         176         ggaggatgttactcccgtaatt           Intron 45         176         gtgtttggttcaccacgactcactcactc           Intron 45         176         gtgtttggttcaccacgactcactcactcactcactcact	Intron 43		376	ctccacacatttttA/Gcaaaa	CT 202	gtttcagggagtttactgag			
Intron 43         701         gggggggggggggggggggggggggggggggggggg	intron 43		0/0	traaacagactggtccccta	2	gggcaggacagagggatga			
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Intron 43         1671         tegagagagittacttocate           Intron 43         1671         tegagagagttacttocate           Intron 45         176         tegagagagttactcocate           Intron 45         176         gtgtttggttactcocate           Intron 45         176         gtgtttggttactcocate           Intron 45         193         ctcc/1gggggaagacate	Intron 43		1242	uuuuauuauuuuuuuauua artaarrtaraarretaraaa	5	generatinggeneagacagg asantatannnttaananan			
Intron 43 2036 ttgaaggatactcagtaatt Intron 45 176 gtgtttggttcacacagctc Intron 45 193 ctcc//gggggaagaacagcca	Intron 43		1671	tagagaagtttacttccatc	G/A	addecacatdcatctttcta			
Intron 45 176 gtgtttggttaacaggtc Intron 45 193 ctcc/192ggaaaagcagtca	Intron 43		2036	ttgaaggatactcagtaatt	G/A	cttttttttgcagtattt			
Intron 45 193 ctcc/Tggggaaaacaagtca	Intron 45		176	gtgtttggttcacacagctc	СЛ	ggagaaaaacaagtcaC/Tggc			
Intron 47 230	Intron 45		193	ctcC/Tggagaaaacaagtca	СЛ	ggcacagccttgacttggga			
Intron 4/ 238	Intron 47		238	cccaagtctctggatggggc	AG	tctgatcaggatgcatgcag			

2	Region	Exon	Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
i-ABCA4-160	Intron 47		269	atgcatgcagagcctggctg	G/A	gatgagggggggggtgctacc			
i-ABCA4-161	Intron 47		326	accacttatctcaacagatc	C/G	gggacctgtggcctatttac			
i-ABCA4-162	Intron 47		715	aagtcactaagctggttggt	G/A	ggaggaacagcacataacC/Tc		÷	
i-ABCA4-163	Intron 47		734	tG/Aggaggaacagcacataac	СЛ	caccttatctatgctgaggt		+	
i-ABCA4-164	Intron 47		931	ggacactgcatagatatcta	T/C	agaaatagcagcatgtcagg			
i-ABCA4-165	Intron 47		1260	acactctctggtggaccatc	AC	ctcatccaagagagggtaac			
i-ABCA4-166	Intron 48		1663	tetegetettetettacete	T/C	aggtgtttgtaaattttgct			Maugeri et al. (1999)
i-ABCA4-167	Intron 49		127	agagagccccacccacca	СЛ	ggtccctaccaagtccccac			
i-ABCA4-168	Intron 49		-1545	gcagttaattccaaactttt	CIA	tcccttattggatgagatca			dbSNP ID:rs1932016
i-ABCA4-169	5' Flanking region		(-1441)–(-1400)	gtaaatctcagttgaatcag	(TCA)14~16	attttcaatctggttcctg			
i-ABCA4-170	Intron 1		4712-4720	gaggggggggggggggggggggg	(A)8~10	caqcctaattcaaqqatqaq			
i-ABCA4-171	Intron 1		7295-7304	ttqttqqctttqcaaT/Aqqat	CACAGTCAT/del	ttattcactcattcattcac			
i-ABCA4-172	Intron 2		951-952	cctqtccatcaqactcttct	TT/del	acctctccccqaqqaaqccca			
i-ABCA4-173	Intron 3		2642-2653	cctgggtgacagagcgagat	(A)10-12	tagcatgagatattattact		÷	
i-ABCA4-174	Intron 4		5202	cacaaagcatctgacacccc	C/del	atccadccctggctagctt			
i-ABCA4-175	Intron 6		3029-3044	cactaaaaacaaaaatttac	(A)16~18	cctgaaagaaattgcaggca		+	
i-ABCA4-176	Intron 6		5138-5139	ttcatgacagatcagatgtt	G/ins	ctttatggatttacaaaga			
i-ABCA4-177	Intron 6		5985	tttccttcttcaaacccccc	C/del	agactaggaaggtctgtc			
i-ABCA4-178	Intron 6		6094		T/del	agtttctgttgagccaaaga			
i-ABCA4-179	Intron 6		-161	tatttttcaattaaataaa	Avdel	gagtttttgtttctaaaag			
i-ABCA4-180	Intron 7		809-810	gggccgagtatgcacactga	TG/ins	tgtgggaagttagagaaG/Ca			
i-ABCA4-181	Intron 8		472-484	atcttccccacctttcacta	(T)10~13	ggtcttctatggggtaaagg			
I-ABCA4-182	Intron 9		48-71	gtaccctggacctcccagaa	(GT)11~13	gagagagatgtgccttcctg			
i-ABCA4-183	Intron 9		554	ataggggcagaaaagacaca	Avdel	ccaaagttctctctcactt			
I-ABCA4-184	Intron 10		11	catgatcagagtaagggggg	G/del	ttggaggatgggggggggg			
-ABCA4-185	Intron 11		4242		G/del	cctcctgtaaataggcccag			
i-ABCA4-186	Intron 11		13743-13753	tgctcttttgtgggtaatgg	(T)9~11	cctcttccaggagaagaaaa			
-ABCA4-18/	Intron 13		636-63/ 520 530	cggggtggagggttgggagg	G/ins	ctcatttgtcattatagatg			
-ABCA4-188	Intron 18		569-570	tgctgccctcatcttctctc	TT/del	aaactagttctgtatttctc			
I-ABCA4-189	Intron 20		(-304)-(-297)	tataacctgactttttttc	(A)7-9	ggattgctttttaaacata			
-ABCA4-190			1230-1240 4606 4605	gctgaattagttcccttggg	(1)9-11	agttaactcctgattttgc			
-ABCA4-191	Intron 23 Intron 33		4020-4033 115-116	garaarcaargcrgraaggg +	(A)9-10 CT/Abl	tggcattagagatccagacc			
	00 nortul		1070		0100				
I-ABCA4-193	Intron 30		290-291	raagcagctatcacttaaca	T/ins	tacaaaaccagagattatca			
-ABCA4-195	Intron 38		896 896	eccreadercadagecreggggg ataaaaanannnnnaaaaaa		caycoartececean/ Geoecere gaaggeagt goet grange			
-ABCA4-196	Intron 38		1209-1210	atagaararagagagagagagaga		4 + 1 1 2 2 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4 4			
i-ABCA4-197	Intron 38			gradatetadaadG/TC/Addadd	G/del	erceagarcergeraggger at aagaararaggcoorggga			
-ABCA4-198	Intron 38			agaccecatactactagagag	Adel	aaaaaaataaaaatttacccc			
I-ABCA4-199	Intron 40		152	tttctccaataatacaagt	Avdel	gaggatcgggttaaaatagg			
i-ABCA4-200	Intron 43		330	tqtaqcctattcctataaA/Ga	Adel	tgcaccattgcttcC/Gcatta			
i-ABCA4-201	intron 43		1354	tttaattggcccagccatgc	C/del	ttaataacttttatcatta			
-ABCA4-202	Intron 47		1305-1308		AAAG/del	caccaatggcccaagcccta			
P binding cas	ATP binding cassette. subfamily A. member 7 (ABCA7)	nber 7 (Ai	BCA7)						
i-ABCA7-1	5' Flanking region		-1596	agaatgttggccccctcccc	СЛ	tC/Tctgcatcctctgcagaag			
i-ABCA7-2	5' Flanking region		-1594	aatgttggccccctccccC/Tt	СЛ	ctgcatcctctgcagaagcc			
i-ABCA7-3	5' Flanking region		-1180	ggccagtgagtgacgggcag	G/A	tcgcccaaatagcagcgtgc			
i-ABCA7-4	5' Flanking region		-460	agagetgggggtegtectee	AG	gctgggcaactgcctgtctc			
i-ABCA7-5		exon 1	6	ctctgtcccgtcccctgccc	AG	gtctcaccatggccttctgg			
i-ABCA7-6	Intron 5		91	ccccgggccaaggacctccc	G/A	ttccaggcatccaggctgtc			
i-ABCA7-7	Coding region	exon 6	563	cagcttgttggaggccgctg	AG	ggacctggcccaggaggtac	Glu188Gly		
i-ABCA7-8	Intron 8		103	qccqqaqqqtcacqqaaact	AG	tttaaaaaataaaataa			

Comment         Comment <t< th=""><th>9</th><th>Region Ex</th><th>Exon P.</th><th>Position<sup>a</sup></th><th>Flanking sequence<sup>b</sup></th><th>Variation<sup>c</sup></th><th>Flanking sequence<sup>b</sup></th><th>Substitution</th><th>Repetitive sequence<sup>d</sup></th><th>dbSNP/Previous report</th></t<>	9	Region Ex	Exon P.	Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
Memory (memory	i-ABCA7-10		n 9	955	accggaccttcgaggagctc	AG	ccctgctgagggatgtccgg	Thr319Ala		
Material         Material         Material         Material         Material         Material         Material           Control Material         11         10         00000         00000         0000         0000         0000         0000         0000         0000         0000         00000         00000         0000         00000         00000         00000         00000         00000         000000         000000         000000         000000         00000000         00000000         0000000	i-ABCA7-11	Intron 9		421	tttttttttttttttttttttt	T/A	taagagatggagtctcactc		+	
Memory (monol)         60         Constructional (1990)         C/T         Experimentational (1990)         F	i-ABCA7-12	Intron 9		463	gttgcccaggctggactgca	G/A	tggC/Tgagatcttggctcact		+	
Columnol         64         Season         84         8	i-ABCA7-13	Intron 9		467	cccaggctggactgcaG/Atgg	СЛ	gagatcttggctcactgcaa		+	
Cubino monto         114         Constantinguistantes         No         Constantinguistantes         Medida           Cubino monto         10         90         времанстраницата         Medida         Medida           Cubino monto         10         0         времанстранистраницата         Medida <td< td=""><td>i-ABCA7-14</td><td>Intron 9</td><td></td><td>488</td><td>gagatcttggctcactgcaa</td><td>СЛ</td><td>ctccgcctcctggattcaag</td><td></td><td>+</td><td></td></td<>	i-ABCA7-14	Intron 9		488	gagatcttggctcactgcaa	СЛ	ctccgcctcctggattcaag		+	
Minute         10         станализация соста услугия соста услуги услуги услуги услуги услуги услуги услуги услуги услуги услугия	i-ABCA7-15		n 10	1184	cgcacacgctgatgtggggc	AG	cctggtgggcacgctggggcc	His395Arg		
Office         Office         Office         Application         Application           Office         Contrasticological         0.0         Construction         Application           Office         Construction         13.0         Construction         Application         Application           Construction         13.0         Construction         10.0         Construction         Application           Construction         13.0         Construction         10.0         Construction         Application           Construction         13.0         Construction         10.0         Construction         Application           Construction         10.0         Construction         10.0         Construction         Application           Construction         10.0         Construction         0.0         Construction         Application           Construction         10.0         Construction         0.0         Construction         Application           Construction         10.0         Construction         0.0         Construction         Application           Construction         20.0         Construction         0.0         Construction         Application           Construction         20.0         Construction <t< td=""><td>i-ABCA7-16</td><td></td><td></td><td>10</td><td>gagtgacggaggtgaggcc</td><td>1/C</td><td>gtccacctgcggggtctgtt</td><td></td><td></td><td></td></t<>	i-ABCA7-16			10	gagtgacggaggtgaggcc	1/C	gtccacctgcggggtctgtt			
Current I: Import I: Im	i-ABCA7-17		n 11	1388	cctgggccccggccacgtgc	GIA	catcaaaatccgcatggaca	Arg463His		
Minorial         124         controperior         Mode         constructions         Mode         constructions         Mode           minorial         0	i-ABCA7-18			115	caggctgcgaactttgcacc	1/G	ttacaccactccacgtgacc			
Minor II         35         99/99/97/02/93/93/94/64         AG         99/99/99/97/02/93/93/94/64           Colordy Mpor         wmi II         181         99/99/97/02/93/93/94/64         AG         99/99/99/94/94           Colordy Mpor         wmi II         181         99/99/97/02/94/94/94         AG         99/99/99/94/94           Colordy Mpor         wmi II         281         99/99/97/04/94         AM         AM           Minor II         181         99/97/97/04/94         AM         99/97/97/94         AM           Minor II         281         99/97/97/94         AM         99/97/97/94         AM           Minor II         281         99/97/97/94         AM         99/97/97/94         AM           Colordy Mpor         Wmi II         288         99/97/97/94         AM         AM           Colordy Mpor         Wmi II         288         99/97/97/94         AM         AM           Colordy Mpor         Wmi II         AM         AM         AM         AM           Colordy Mpor         Wmi II         AM         AM         AM         AM           Colordy Mpor         Wmi II         AM         AM         AM         AM           Colordy Mpor         Wmi II	i-ABCA7-19		n 13	1824	cccttcctgctcagcgccgc	AG	ctgctggttctggtgctcaa	Ala608Ala		
Colority 13         78         99300/groce/sector/gold         CA         99300/groce/sector/gold         Antifity           Mitori 13         73         91300/groce/sector/gold         CA         99300/groce/sector/gold         Antifity           Mitori 13         13         91300/groce/sector/gold         CA         99300/groce/sector/gold         Antifity           Mitori 13         13         91300/groce/sector/gold         CA         99300/groce/gold         Antifity           Color/groups         exist 1         263         05400/groups         Antifity         Antifity           Color/groups         exist 1         263         05400/groups         Antifity         Antifity           Color/groups         exist 1         263         05900/groups         Antifity         Antifity           Color/groups         exist 1         263         05900/groups         Antifity         Antifity           Color/groups         exist 1         263         05900/groups         Antifity         Antifity           Color/groups         exist 24000/groups         Antifity         Antifity         Antifity         Antifity           Color/groups         exist 24000/groups         Antifity         Antifity         Antifity         Antifity	i-ABCA7-20	Intron 13		55	ggtgcgctggagggtgacag	AG	cagggggggggccccacgtggg			
Control         181         Outspace conservation         06170y           Control         3         999393575039575303         00         04939336753030         06170y           Fiton 16         3         97939357530353         01         04939336753030         06170y           Fiton 16         181         010000000         181         0100000000         0100000000         010000000           Fiton 17         242         0000000000         000         0100000000         010000000         010000000           Fiton 17         242         000000000         000         000000000         010000000         01000000           Fiton 17         242         000000000         000         000000000         000         00000000           Fiton 17         243         000000000         000         000000000         000         00000000         00000000         000         00000000         000         00000000         000         000000000         00000000         00000000         00000000         00000000         00000000         00000000         00000000         00000000         00000000         00000000         00000000         000000000         000000000         000000000         000000000000000000000000000000000000	i-ABCA7-21	Intron 13		78	ggggcggccccacgtgggtg	CIA	gcgcccccaggccaatccag			
Ordingramment (s)         21(3)         0999909000000000000000000000000000000	i-ABCA7-22		n 14	1851	cgttgcctctcacagctggg	AG	gacatcctcccctacagcca	Gly617Gly		
Inten (5)         34         ggggggggreetergengeregenergener         C/C         geggggeretergenergenergenergenergenergen	i-ABCA7-23		n 15	2153	cgagggcgcgcagtggcaca	AC	cgtgggcacccggcctacgg	Asn718Thr		
Import (a)         (b)         (consistent and angle participations         (b)         (consistent and angle participations         (b)           Consist (a)         10         (consistent and angle participations         (b)         (consistent and angle participations         (b)           Consist (a)         100         (consistent and angle participations         (b)         (consistent and angle participations         (b)           Consist (a)         100         (consistent and angle consistent angle consi	i-ABCA7-24	Intron 15		8	qqcqqqqctccqqqqt	C/G	gcacctgctttgcgggaggc			
Munori         Bit         Construction construction         Cit         Construction construction         Munori           Curring monit         monit         2.2         Construction construction         Munori         Munori         Munori           Curring monit         monit         2.2         Construction construction         Munori	i-ABCA7-25	Intron 16		8	ctagacccagagggtgaggc	AIC	ctacaaqoottaataqotoq			
Configuration         Configur	-ABCA7-26	Intron 16		161	<pre>ccondcocccccccccccccccccccccccccccccccc</pre>	E C				
Contropy (0)         Contropy (0)<			17	101				1 0117061 011		
Miletion         Main         Main         Main         Main         Main         Main           Cumprision         Excit         302         Graphergenergenergener         Main         Main         Main           Cumprision         Excit         302         Graphergenergenergener         Main         Main           Cumprision         Excit         Graphergenergenergener         Main         Main         Main           Cumprision         Excit         Scit         Graphergenergenergener         Main         Main           Cumprision         Excit         Scit         Graphergenergenergener         Main         Main           Cumprision         Excit         Scit         Graphergenergener         Main         Main         Main           Cumprision         Excit         Sc				2002	caccccaeccccagcagcg		gragaggggggggggggggg	renvaoren renvaoren		
Control         Dis         Dis <thdis< th=""> <thdis< th="">         Dis</thdis<></thdis<>	-ABCA/-28		/L U	1242	cccggcctgagtcctggcgt	C/A	tccgttcgcagcctggagaa	Vali8U/Val		
Control         Control <t< td=""><td>-ABCA/-29</td><td></td><td></td><td>166</td><td>cgagacagtaagagttgggg</td><td>AG</td><td>tagacagaggttcccctgga</td><td></td><td></td><td></td></t<>	-ABCA/-29			166	cgagacagtaagagttgggg	AG	tagacagaggttcccctgga			
Intron         138         993549970 concept concept         147         993549970 concept concept         147           Confing region         8005         317         99340000         801         9934990000         801         147         993400000         801         147         993400000         801         147         9934000000         801         9934000000         801         9934000000         801         9934000000         801         99340000000         801         993400000000         801         993400000000         801         99340000000         801         147         14300000         8000000000000000000000000000000000000	-ABCA7-30		n 21	3027	ctgctgggagaccgtgtggc	CT	gtggtggcaggtggccgctt	Ala1009Ala		
Control region         Art1         gggattergegreetened         Displayergegreetened         Lend 136, Lend           Control region         exons         3.83         gggattergegreetened         Displayergegreetened         Lend 136, Lend           Control region         exons         3.83         gggattergegreetened         Displayergegreetened         Lend 1346, Lend           Control region         exons         3.83         gggattergegreetened         Displayergegreetened         Ant1346, Lend           Control region         exons         3.84         consergedergegreetened         Displayergegreetene         Ant1346, Lend           Control region         2.33         gggattergegreetene         Displayergegreetene         Ant1346, Lend           Control region         2.34         gggattergegreetene         Displayergegreetene         Ant134, Lend           Control region         2.34         gggattergegreetene         Displayergegreetene         Ant136, Lend	-ABCA7-31			1386	gggtggggggggggggggggggggggggggggggggggg	СЛ	tccctgaagcacccctttgt			
Intron 23 (onling region         147 (onling region         support comparison         147 (onling region         concrete spaganet sector         6/A         tot comparison         147 (onling region         concrete spaganet sector         6/A         tot comparison         147 (onling region         concrete spaganet sector         6/A         tot comparison         147 (onling region         concrete spaganet comparison         0/A         concrete spaganet comparison         0/A         0/	-ABCA7-32		n 23	3417	gggatctccgacaccagcct	C/G	gaggaggtgtgaggcctggg	Leu1139Leu		
Config region         Biolity records         Biolity region         Biolity region <td>-ABCA7-33</td> <td></td> <td></td> <td>147</td> <td>ggagctctggtggctcagat</td> <td>GIA</td> <td>tcccttgggaaggcctgggg</td> <td></td> <td></td> <td></td>	-ABCA7-33			147	ggagctctggtggctcagat	GIA	tcccttgggaaggcctgggg			
Odding region         excercing address of the concorregation consist and the concorrect concorrect consist and the concorrect concorrect concorrect consist and the concorrect conco	-ABCA7-34		n 25	3528	gctggcctagacgtaaccct	AG	cggctcaagatgccgccaca	Leu1176Leu		
Nime         Bit         Conceptings activity         Control         Contro	-ABCA7-35		n 29	4046	cccagcctgccagtgtagcc	GIA	gcccggtgcccggcgcctgc	Arg1349GIn		
Coding region exon 31         238         ctgeocretegear geoccaseg         AG         terrageocretegear geoccaseg         AG           Intron 32         2         1         castact tragegeorect         GA         tragecocret grage         AG           Intron 32         2         45         castact tragegeorect grage         GA         tragecocret grage         AG           Intron 38         305         999         997         GA         tragecocret grage         AG           Intron 40         154         cotact extremescent trageacy         AG         997         tragecocret grage         AG           Intron 40         154         cotact extremescence         AG         997         tragecocregoer         AG           Intron 41         258         cotact extremescence         TC         994         tradecocreacy         AG           Intron 41         269         cotact extremescence         TC         994         castact extremescence         AG           Intron 41         261         cotact extremescence         TC         995         cotact extremescence         AG           Intron 41         261         cotact extremescence         TC         995         cotact extremescence         AG           Intron 41	-ABCA7-36			81	cccctgggagctctcccgg	CIA	cccccggccctcagctccc			
Intron (2)         1         catagragacycycic sol (3)         Control (2)         1         catagragacycic (1)         (3)         (4)         cycarcycycycycycy (1)         (4) </td <td>-ABCA7-37</td> <td></td> <td>n 31</td> <td>4239</td> <td>ctgcctgcatggcccccacag</td> <td>AG</td> <td>tacggaggcttctcgctggg</td> <td>Arg1413Arg</td> <td></td> <td>dbSNP ID:rs881768</td>	-ABCA7-37		n 31	4239	ctgcctgcatggcccccacag	AG	tacggaggcttctcgctggg	Arg1413Arg		dbSNP ID:rs881768
Minorial         34         acadestructore conformation         6A         transpondent control         transpondent control <thtranspont< th="">         transpont         <thtransponde< td=""><td>-ABCA7-38</td><td>Intron 32</td><td></td><td>- ;</td><td>caaggagcagctgtctgagg</td><td>G/C</td><td>tgcactgtgagtccctccac</td><td></td><td></td><td></td></thtransponde<></thtranspont<>	-ABCA7-38	Intron 32		- ;	caaggagcagctgtctgagg	G/C	tgcactgtgagtccctccac			
Coding region         246         catactutoggageccaga         GA         cagadgactotitigace         Coding region	-ABCA7-39	Intron 33		Z :	ccactgcttgccactgccct	GIA	tctggccccttgtaggcagg			
Coding regionexon 3650579915499099154154GalageogradiateIntron 40377ctetaactacactergaaAG9915415499154154Intron 40277ctetaactacacageggaaC/G6925451593454Ala186AAlaIntron 415892gtegageoccegggaaccactergactergaaC/G99160575475454Intron 41286ctetaageoccegggaaccagegaaC/G99160575475454Intron 41286ctetageoccegggaaccagegaaC/G99160575475454Intron 41389tegageoccegocceaIntron 41991actategeoccegocceaC/G9954564649545464Intron 41991actategeoccegocceaIntron 41991actategoccegocceaC/G995456464954664Intron 41991actategoccegocceaC/G995456464954664Intron 41991actategoccegocceaC/G9954564769763664Intron 411001ccepcceac/rac//rac//racC/G9954564769763664Intron 411001ccepcceac/rac//racC/G995456476976364Intron 411001ccepcceac/rac//racC/G95456476946646464Intron 411001ccepcceacy/racC/G95456476946646464Intron 411001ccepcceacy/racC/G95456476966464646464Intron 411001ccepcceacy/racC/G95456476966464646464Intron 4110111011ccepcceacy/racC/GIntron 412039956949946469469469464A/G	-ABCA7-40			245	cagtactttgggaggccgag	GIA	caggaggactgcttgtggcc		+	
Intron 30         65         gecceracettic gaa         AG         gecceracettic conserved           Intron 40         271         ctgagecccoggeacccagter         TC         cgageccctic conserved           Intron 40         271         ctgageccccggeacccagter         TC         cgageccccggagaacccagter           Intron 41         2582         tgageccgtccccagter         TC         geccacteggagaacccagter         Ala1864Ala           Intron 41         2582         tgageccgtccccagttgag         TC         geccacteggagaacccagter         Ala1864Ala           Intron 41         258         tgageccgtccccagttgag         TC         geccactegcccagttgag         Ala1864Ala           Intron 41         269         tgagecgtcccagttgag         TC         geccactcccactccagttg         Ala1864Ala           Intron 41         269         tggeccgtcccagttgag         TC         geccactccattccattcat         Ala1864Ala           Intron 41         269         tggeccgtcccact/ract         TC         geccactcattcattcat         Ala1864Ala           Intron 41         1001         cccatagcctgcccast         TC         geccactcattcat         Ala1864Ala           Intron 41         1010         cccatagcctgcccast         TC         geccactgccttcattcat         Ala18644la           Intron 41	-ABCA7-41		n 36	5057	ggtgagccggatcttgaaac	AG	ggtcttccttatcttccccc	GIn1686Arg		
	-ABCA7-42	Intron 38		65	ggcccactcacctttctgaa	AG	gacctgcactctccccaggta			
Inton 40277ttpagecceggegecceart thron 41700 $100$ miton 412662 $100$ miton 412662 $100$ miton 41 $200$ $200$ miton 41 $200$ $200$ miton 41 $200$ $200$ miton 41 $1001$ $1001$ $1001$ miton 41 $1001$ $10$	-ABCA7-43	Intron 40		154	ctctacctcccacacgcgga	C/G	caggccctgagacacccctg			
Coding regionexon 415582gtggccggggaccagtgggtaAla1864AlaIntron 41286ctccttgactttgcctttggccgtgcccagtggctAla1864AlaIntron 41389tggcggrctgacttgcgctttggccgtgccagtgggtc+Intron 41389tggcggrctgactggcctgcccaccaCTggccgtcccaccca+Intron 41389tggcggrctgactggcctgcccaccaCTggccgtccacccacca+Intron 41389tggcggrctggcctggccggcctgcccaccaCTggccgtccaccca+Intron 41389tggcggcctggcccggcccgcccacd/racCTact/racc/ract+Intron 411001ccaccacacca/racCTact/ract/ract+Intron 411001ccagcccgcccacgcccacd/racCTact/racgccccaccacaccaccaccacca+Intron 411131tgccgcccgcccaccaccaccaccaccaccaccaccaccac	-ABCA7-44	Intron 40		277	ctgagcccccggcgccccca	1/C	ccccagcgtggcccgggaac			
Intron 41         286         ctecttgactcgccacttgcrag         T/C         ggecctgcccacttgact         +           Intron 41         89         tggecctgcccacttgcrag         C/T         cgtttactcgcccact         +           Intron 41         94         actatggccctgcrccaa         C/T         actC/GacA/ggctccaaccaa         +           Intron 41         994         actatggccctgccccaa         C/T         actC/GacA/ggctccaaccaaccaaccaac         +           Intron 41         1001         acctaggcccgaccaac/Tract/Gac         C/T         acct/GacA/ggctccaaccaac         +           Intron 41         1031         tggcccggcccaac/Tract/Gac         C/T         actC/GacA/ggctccaaccaaccaaccaaccaaccaaccaaccaaccaa	-ABCA7-45		n 41	5592	gtggcccgggaacccagtgc	T/C	gcgcacctcagcatgggata	Ala1864Ala		
Intron 41         389         tggccgttcccagttgcag         C/T         cgtttcactgccttccat         +           Intron 41         991         ccaactaggccctgcccac/Tac         C/T         cgtttcactgctcccaccac         +           Intron 41         994         accactaggccctgcccac/Tac         C/T         cacc/TeatC/GccA/Ggctccaccaccac         +           Intron 41         998         tggccctgcccac/Tac/Tcat         C/T         catC/Gac/Ggctccaccaccac         +           Intron 41         1001         ccctgccccac/Tac/Tcat         C/G         cac/Ggttccaccaccaccac         +           Intron 41         1051         actcatggcctgccac/Tac         C/G         gctccaccaccaccaccac         +           Intron 41         1031         ccctgccccac/Tac         C/G         gctccaccaccaccaccac         +           Coding region         exon 45         585         gggcgcggcccaggcccact         A/G         gccctcgccccaccaccac         +           Intron 44         203         ggcgcggggggggcccacgggggggggggggggggggg	-ABCA7-46	Intron 41		286	ctccttgactctgccttctg	T/C	ggccctgcccacttgctcct			
Intron 41         901         ccacactatgocccgocccc         C/T         acc/Tract/GccA/Ggttccacccac         +           Intron 41         994         actatggcctggccccgcccac/Tac         C/T         act/TgatC/GccA/Ggttccacccac         +           Intron 41         998         actatggcctggcccgcccac/Tac         C/T         actC/GccA/Ggttccacccac         +           Intron 41         1001         cctggcccgcccac/Tac/Tac         C/G         cct/Ggttccacccaccac         +           Intron 41         1051         actcatgcggcgcccaccac/Tac         C/G         cct/Ggttccacccaccac         +           Intron 41         1051         actcatgcgggggcccaccaccaccaccaccaccaccaccaccacc	-ABCA7-47	Intron 41		389	tggccgttccccagtttgcag	СЛ	cgtttcactgcctcttccat			
Intron 41         994         actatggccctgcccac/Tac         C/T         catC/GccA/Ggtccaccaca         +           Intron 41         1001         ccctgccccac/Tac/Tac/Tac         C/G         ccat/Ggtccaccacaca         +           Intron 41         1051         ccctgccccac/Tac/Tac/Tac         C/G         ccat/Ggtccaccacacaca         +           Intron 41         1051         ccctgccccac/Tac/Tac         C/G         gctcaccacacacacacacacacacacacacacacacaca	-ABCA7-48	Intron 41		991	cacactatggccctgcccca	СЛ	acC/TcatC/GccA/Ggctccaccca		+	
Intron 41         908         tggccctgcccac/Tac/Trat         C/G         cccA/Ggctccaccaccatg         +           Intron 41         1001         ccctgcccac/Tac/Trat/Ggc         A/G         gctccaccaccaccatg         +           Intron 41         1001         ccctgcrcaccaccatg         A/G         gctccaccaccaccaccatg         +           Intron 41         1131         tgccctgcrcactgrcgcccatt         A/G         gctccaccaccaccatcac         +           Intron 41         1131         tgccctgcrcactgrcgcccatt         A/G         gctccaccaccaccaccatcac         +           Intron 41         1131         tgccctgcrcactgrccatt         A/G         gcccctgcrcactaccatcac         +           Intron 41         201         ggcgggggggggggggggggggggggggggggggggg	-ABCA7-49	Intron 41		994	actatggccctgccccaC/Tac	СЛ	catC/GccA/Ggctccaccacac		+	
Intron 41         1001         ccctgccccaC/TacC/TcatC/dcc         NG         gctccaccacacacggcc         +           Intron 41         1051         actatggcccgcccatt         AG         gctccaccacacacacacacacacacacacacacacacac	I-ABCA7-50	Intron 41		<b>866</b>	tggccctgccccaC/TacC/Tcat	C/G	ccA/Ggctccacccacacatg		+	
Intron 41         1051         actcatgctggtcaccca         CT         accatggcccgcccatac         +           Intron 41         1131         tgccctgccccatgcccatt         AG         tgcccctgccccatac         +           Coding region         exon 44         5085         gggcggggccccggccc         GA         gcccctgccccatac         +           Coding region         exon 44         5085         gggcggggccccggccc         GA         gcccctggcccgggggg         Leu1995Leu         +           Intron 44         203         gggcggggggggggggggggggggggggggggggggg	-ABCA7-51	Intron 41		1001	ccctgccccaC/TacC/TcatC/Gcc	AG	getecacceacacatggee		+	
Intron 41         1131         tgccctgcccatt         MG         tgccctgctcaactcaa         +           Coding region         exon 44         5985         gaaggetctgggggg         Leu1995Leu         +           Intron 44         201         ggegggggggggggggggggggggggggggggggggg	-ABCA7-52	Intron 41		1051	actcatgctggctccaccca	СЛ	accatggccccgccccatac		+	
Coding region         Exon 44         5085         gaagcgctctgctcgccg         Leu1995Leu           Intron 44         201         gggcaggagccggggggggggggggggggggggggggg	-ABCA7-53	Intron 41		1131	tgccctgccccatgcccatt	AG	tgccctgctccacactcaa		+	
Intron 44         201         ggcgcaggagcaggaggggggggggggggggggggggg	-ABCA7-54		n 44	5985	gaagegetetgetegegeet	G/A	gccatcatggtgaatgggcg	Leu1995Leu		
Intron 44         233         ctgggtggatttagaagaca         C/T         aatcaggtggggggggggggggggggggggggggggggg	-ABCA7-55	Intron 44		201	ggcgcaggaccaggaggcgt	G/C	agccgggggctctgggfgga			
Intron 44         313         agttagggagggggggggggggggggggggggggggg	I-ABCA7-56	Intron 44		233	ctgggtggatttagaagaca	L'	aatcaggtgtgcgttggagt			
Intron 44         33/         99509990asteritating region           Coding region         \$501         99509990asteritating region           Coding region         \$501         99509990asteritating region           Coding region         \$501         \$50299590astericectggg         \$67           Coding region         \$501         \$5029959950astericectggg         \$67           Coding region         \$501         \$502999950astericectggg         \$67           Coding region         \$501         \$5029999950astericectggg         \$67           Coding region         \$500         \$5029999950astericectggg         \$67           Vintanside region         \$500         \$5029999990astericectggg         \$67           Vintanside region         \$500         \$5029999990astericectggggggggggggggggggggggggggggggggggg	I-ABCA7-57	Intron 44		313	agttaggggagggcctggtt	AG	gtgggcgggccataggaaa			
Coding region         exon 45         613         tggcggccggggtccgggtcccggg         G/I         cgggggcgcgggggcgcgggggcgggggggggggggg	i-ABCA7-58		ļ	337	ggcggggccataggaaagtg	0/C	ggcgggggtatttattgtgt			dbSNP ID:rs1609436
Cooling region         Exon 45         b159         ctgcgcgagggggg         U/I         cgcctgcgcttccagctg         Given 37           Intron 45         27         acgccgcgggggcggggggggggggggggggggggggg	I-ABCA/-59		n 45	6133	tggcggccgagttccctggg	6/1	cggagctgcgcgaggcacat	Ala2045Ser		
inuoli 40 z/ acgocogogocoggocoggocoggocoggocoggocog v/v gggaggocaggocoggocoa 3'Untanslated region exon 46 6580 aaggocggaggagaggoggaggaggogggaggaggogggggaggogggggg	-ABCA/-60		0 45	6159 77	ctgcgcgaggcacatggagg	5	cgcctgcgcttccagctgcc	GIYZU53GIY		
				21	acggcgccgggggtcggggctg	200	gggaggcaggcrggggggcca			
			n 46	0869	aaggctggagagaagccgtg	G/C	tggtgaaaccgtgtgcatgt			dbSNP ID:rs1134007

Q	Region Exon	Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution Repetitive seguence <sup>d</sup> dbSNP/Previous report
i-ABCA7-64	3' Flanking region	376	cttacaqqaqccqqtqtcc	СЛ	gaaacacaaaccaaaaccaa	
i-ABCA7-65	3' Flanking region	687	cagcagggagacttggggag	GIA	G/Aqqqaqaqttcacactqc	
i-ABCA7-66	3' Flanking region	688	agcagggagacttgggggagg/A	GIA	gggagagattcacactqcq	
i-ABCA7-67	3' Flanking region	1169	cctcgacctgacccacttca	СЛ	ggggctgcagggcqgqtgat	
i-ABCA7-68	Intron 9	398-422	cgtgaactaccacgtcctgc	(T)22~26	aagagatggagtctcactct	+
i-ABCA7-69	Intron 12	175184	ggggactctgagggtctggt	(G)8~10	actctgagggtctggggggcc	
i-ABCA7-70	Intron 30	81-87	cccctgggagctctcccgg	(C)6-7	ggccctcagctccccttccc	
i-ABCA7-71	Intron 34	349-361	agaaagagaaagagagaaag	(A)12-14	cagaaatgtgctttgggtga	+
ATP binding casse	ATP binding cassette, subfamily A, member 8 (ABCA8)	(ABCA8)				
i-ABCA8-1	Intron 1	204	ctggtaattaatattagata	A/G	ataaaacattgagttagaa	
i-ABCA8-2	Intron 1	266	aacattatgttgttttaaac	AG	taactgagtgtagaaataag	
i-ABCA8-3	Intron 1	674	gttctgaattttacaacagt	AG	taagaggataaaaatgagtc	dbSNP ID:rs2041321
i-ABCA8-4	Intron 1	733	ttgccatatgtataataaag	T/A	attcatgtttttgctagcct	
i-ABCA8-5	Intron 1	861	agactggagtttgcatgcta	СЛ	ctaagactgtagctgattcc	
i-ABCA8-6	Intron 1	205	gaggagatcatcctcttggc	СЛ	aatgtctattaacttcgcca	
i-ABCA8-7	Intron 1	1262	cagaaacttttgccctctct	G/A	taggctagctcactgtgaaa	
i-ABCA8-8	Intron 1	1537	agetetettaaaagtateea	T/C	gctgaattttctgcacctta	
i-ABCA8-9	Intron 1	7622	tcgttaacagcaatgataat	1/C	tagcccatccttatccC/Taga	
i-ABCA8-10	Intron 1	7639	aatT/Ctagcccatccttatcc	СЛ	agaaacaacaggctcataag	
i-ABCA8-11	Intron 1	7720	tccatgtgttacaaactgcc	СЛ	tggagaacagaaaagagaa	
i-ABCA8-12	Intron 1	9397	cataatatatatacatatgc	G/A	cacacacacatatacaca	
i-ABCA8-13	Intron 1	9519	agtagttcatgttggaacaa	T/C	atgcttgagaaatgcagaaa	
i-ABCA8-14	Intron 1	11880	acacggagatcatatatatg	G/C	aaataC/Tgtgtgtgtgtttttt	dbSNP ID:rs1860445
i-ABCA8-15	Intron 1	11886	agatcatatatatgG/Caaata	СЛ	gtgtgtgtatttttcctgat	dbSNP ID:rs1860446
i-ABCA8-16	Intron 1	12341	ctgggtgcaatctggggaca	GЛ	aaactacatagtaattaaat	dbSNP ID:rs887380
i-ABCA8-17	Intron 1	12973	ttgataacaggcacagggca	T/C	cacaaataaatgatggaaca	+
i-ABCA8-18	Intron 1	13100	cattggagtattaggctacg	1/C	tttttgttgttgcaggat	
i-ABCA8-19	Intron 1	13128	ttgtttgcaggatatttctt	1/C	ttcttaagaacttcatatta	
i-ABCA8-20	Intron 2	420	caattagttttcttcaaaaa	AG	gtagaaagttggaattgta	
i-ABCA8-21	Intron 2	505	catataaaaaatcttgatta	AT	actttggtatattttaaaaa	
i-ABCA8-22	Intron 2	819	gcaatgccttggaactatct	СЛ	ttaaaacacattgactttca	
i-ABCA8-23	Intron 3	915	ttgtgttcgatagatcagta	G/A	ggtgactagttaacaatgat	+
i-ABCA8-24	Intron 3	1539	aaagggaaatctgtggtgat	СЛ	gccctgtcattcattcatag	
i-ABCA8-25	Intron 3	2341	tteetttetttgteaaette	СЛ	gtccaaattccactcaagct	
i-ABCA8-26	Intron 3	2882	tattctatttctgtactct	AG	ttaatattctataataataa	
i-ABCA8-27	Intron 3	3314	atttaaatatctatctctct	AG	tatttaccatttcaaattta	
i-ABCA8-28	Intron 4	88	gaggttagtatgccaaatta	GIA	agcatcactatctgtcataa	
i-ABCA8-29	Intron 4	3264	ttccattggcctattatgcc	СЛ	gtgttatatccagtgttaga	
i-ABCA8-30	Intron 4	3403	aagagaccaacaaaattctt	C/G	atcagcagaaagcacagga	
i-ABCA8-31	Intron 5	389	gcttactgaatatataaatt	G/C	agaaaagccatgccaagcaa	
i-ABCA8-32	Intron 5	479	tgagagtggtgagtaactca	AG	aatgcctggactccG/Aaggtc	
i-ABCA8-33			actcaA/Gaatgcctggactcc	G/A	aggtcccagcaggtcaatga	
i-ABCA8-34	Coding region exon 6		atgggtcttcgggattcagc	GIA	ttctggtgagtcaaacgcag	Ala264Ala
i-ABCA8-35	Intron 6	200	cctcccaagtagctgggact	GIA	caggtgccgA/Gccaccatgcc	+
i-ABCA8-36	Intron 6	210	agctgggactG/Acaggtgccg	AG	ccaccatgcctggataattt	+
I-ABCA8-37	Intron 6	1751	gtgagttattattgtgttgg	СT	tttgcagctgttttgttttt	
i-ABCA8-38	Intron 6	1808		GU	aatattgtaaaacaaagaa	
i-ABCA8-39	Intron 6	2412	tattcctaattctaaagaat	1/C	ctgcccaaacttttacctt	
i-ABCA8-40	Intron 6	2506		G/A	agttatcttagaA/Gtccattt	
I-ABCA8-41	Intron 6	2519	gaatgaaG/Aagttatcttaga	AG	tccatttcaggtcttccttt	

Table 1. Continued

1	Region Exon	Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
i-ABCA8-42	Intron 7	28	agtgaattaaatatctttcc	AG	tccacctatagcctaaaaat			
i-ABCA8-43	Coding region exon 8	991	taaagaaatctttcctcacc	GIA	acctantcatattectecte	GIV331Ser		
i-ABCA8-44	Intron 8	74	tggaatccataggctgtaat	СЛ	atttacaaactcagcattgt			
i-ABCA8-45	Intron 8	635	ccagaaattctccctcatgc	СЛ	gccaatttacatggatcacc			dbSNP ID:rs767690
i-ABCA8-46	Intron 9	1417	acacatacttaaatatattt	1/C	ctctqttctacttttqtttt			
i-ABCA8-47	Intron 9	2504	agaggaaaattatggtttgg	G/A	aatgaaataaagcagaaata			
i-ABCA8-48	Intron 10	2013	tggccaaagatctttccaac	СЛ	tgtgccagtggttcacagga			
i-ABCA8-49	Intron 10	2378	ctgaagaaattgtcacttt	GIA	aagtatctttttttttc			
i-ABCA8-50	Intron 11	-697	aaaaaaaaaaaaaaagagagag	AG	gagaaagaaaatatttgtta			
i-ABCA8-51	Intron 11	-528	tataaagttagaaaaaat	GЛ	aA/Gtatgttttagaaatagat			
-ABCA8-52	Intron 11	-526	taaaagttagaaaaaatG/Ta	AG	tatgttttagaaatagatgt			
i-ABCA8-53	Intron 11	-342	ctcaaaggagttttagccat	G/A	taataacttactattaatct			
i-ABCA8-54	Coding region exon 13	1632	ggttcagtcaccatctataa	СЛ	aataagctttcagaaatggc	Asn544Asn		
I-ABCA8-55	Intron 14	252	cttattgcaaaataagtgaa	GIA	ttgagtttctaagagatcaa			
i-ABCA8-56	Intron 15	130	ttttgtttttgagacggagt	AC	tcgatcatctcggctcactg		+	
I-ABCA8-57	Intron 16	534	acatatacattcattcaaat	AG	cacattttatggtgacaaca			
i-ABCA8-58	Intron 16	588	gaatcatcaggaaagtgtta	СЛ	gcaaattctgattagtactt			
i-ABCA8-59	Intron 16	645	atttaaagaaaatttgtaga	СŢ	gttttaggtggaatgaagaa			
i-ABCA8-60	Intron 17	431	tgtcaggtttttctttttt	T/A	ttctttatgttagaaattgg			
i-ABCA8-61	Intron 17	970	ttttgcacaggttttattag	AC	ttcagtccactcccagcttc			dbSNP ID:rs1373068
i-ABCA8-62	Intron 17	1390	getgtaaactegttttgtga	CIA	ttaggtaccccatgattcta			
i-ABCA8-63	Intron 17	2452	cacgttatacctatagtaac	G/A	cggaagaG/Ctctaatcatgag			
i-ABCA8-64	Intron 17	2460	acctatagtaacG/Acggaaga	G/C	tctaatcatgagatG/Ccttag			
-ABCA8-65	Intron 17	2475	gaagaG/Ctctaatcatgagat	G/C	cttagcagagccaatctcta			
i-ABCA8-66	Intron 18	152	gaagaagcacaggagagg	СЛ	agaatcttgacatccaaagg			
I-ABCA8-67	Intron 19	7477	aaaatctattttgaaagaca	СЛ	ttggaactaaaaaaatcttt			
-ABCA8-68	Intron 21	196	ttgtttaagtaaaataaaa	T/C	G/Caacaaaacatttttcaaag			
i-ABCA8-69	Intron 21	197	tgtttaaagtaaaataaaaT/C	G/C	aacaaaacatttttcaaaga			
-ABCA8-70	Intron 21	287	actgtggtgggggggggg	GЛ	ggggggggggggggggggggggggggggggggggggggg		+	
-ABCA8-71	Intron 21	403	cctgcacaatgtgcacatgt	AG	ccctaaaacctaaagtataa		+	
i-ABCA8-72	Intron 21	1194	totogototgocaccoageo	GIA	gagtgcagtggcA/Gggatcat		÷	dbSNP ID:rs1156340
-ABCA8-73	Intron 21	1207	cccagccG/Agagtgcagtggc	AG	ggatcatagctcactgtaac		+	
		1968	taatcaaagaagcaactatg	AC	gaatcatgaaaataacatgg			dbSNP ID:rs991210
-ABCA8-75	Intron 21	2215	atgttggatattttttgagt	T/C	aagctgtcacatttgagata			dbSNP ID:rs991209
-ABCA8-76	Intron 24	263	ctcctagatatagacaaaaa	AC	caaggtgcacaatggccatg			
-ABCA8-77	97 uonu	212	cctgattaatatatgggaag	GIA	aagggtaaggggtagtggga			
-ABCA8-78		67	aataattttcagttctgtac	AG	cactgtgaaacttctttat			
	17 UDUU	515 664	gtgtctcccaaaccacatca	6/1	tttcatcttttgctattaca			
	Intron 20	1061	cctggatattatcagactta	GIA	aatggagaggaaaagtcaat			
HABCA8-82	Intron 31	112	caaaaarrayaracaagggg ototaaataotaasoooaaat		rgaaarrgacrttaartgta			
-ABCA8-83	Intron 33	401	etteteactagettotgaga	20	acactgyytayatttaaat actatattattaaattttatat			
i-ABCA8-84	Intron 35	480	ctqqtaacaqcatcatcctq	AT	socsocsscatttttcatadaca			dhSNP ID:rs1373065
-ABCA8-85	Intron 35	484	taacagcatcatcctgA/Ttgt	AG	tttattttcatagacagaaa			
-ABCA8-86	Intron 36	258	tttgcatgtatgttggtaaa	AG	cctaagtcaaaactcagtta			
i-ABCA8-87	Intron 36	375	atattatttactgtcttag	C/G	ctgtatataagaaactgac			
HABCA8-88	3' Flanking region	674	gcggtggacatagaaagccc	G/A	gaagcttcttgatgtgctta			
-ABCA8-89	Intron 1	56-57	ttttgcttttgtgtgtgagt	TT/del	gtttcagaggttttgtcttt			
-ABCA8-90	Intron 1	1180-1191	taaagtataataataaacg	(A)9~11	gaaattcctcctgtacagag			
-ABCA8-91	Intron 1	9877-9885	ctcctgcaaataggtatgac	(A)8-12	tcaactgagtacaaaagget			

i-ABCA8-92 i-ABCA8-93 i-ABCA8-94 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-100 i-ABCA8-96 i-ABCA8-100 i-AB	Intron 1 Intron 6 Intron 9 Intron 1 Intron 15 Intron 15 Intron 15		Position <sup>®</sup>	Flanking sequence	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence	dbSNP/Previous report
i-ABCA8-93 i-ABCA8-94 i-ABCA8-95 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-99 i-ABCA8-100 i-ABCA8-96 i-ABCA8-100 i-AB	Intron 6 Intron 9 Intron 1 Intron 15 Intron 15 Intron 15 Intron 16		12588	gtactagagtgcactccttt	T/del	gcaacaggacggccaaagga			
i-ABCA8-94 i-ABCA8-95 i-ABCA8-95 i-ABCA8-97 i-ABCA8-97 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-97 i-ABCA8-97 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-100 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-97 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-96 i-ABCA8-100 i-ABCA8-	Intron 9 Intron 9 Intron 11 Intron 15 Intron 15 Intron 16		78	tcaatgcatctttttttt	T/del	gaaatggagtctcgctctgt			
i-ABCA8-95 i-ABCA8-95 i-ABCA8-96 i-ABCA8-98 i-ABCA8-98 i-ABCA8-100 i-ABCA8-100 i-ABCA8-101 i-ABCA8-102 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-95 i-ABCA8-98 i-ABCA8-98 i-ABCA8-98 i-ABCA8-98 i-ABCA8-98 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-101 i-ABCA8-100 i-ABCA	Intron 9 Intron 11 Intron 15 Intron 15 Intron 16		265	gtatatggtattttttttt	T/del	agacctcttagaaagctagt			
i-ABCA8-96 i-ABCA8-97 i-ABCA8-98 i-ABCA8-99 i-ABCA8-101 i-ABCA8-101 i-ABCA8-102 i-ABCA8-102 i-ABCA8-102 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-104 i-ABCA8-103 i-ABCA8-10	Intron 11 Intron 15 Intron 15 Intron 16		2666	attttttaaaggtatcca	A/del	tagtcattctcaatttcttc			
i-ABCA8-97 i-ABCA8-98 i-ABCA8-99 i-ABCA8-100 i-ABCA8-101 i-ABCA8-101 i-ABCA8-102 i-ABCA8-103 i-ABCA8-1	Intron 15 Intron 15 Intron 16 Intron 17		-447	ggatattctgggtttttttt	T/del	ctacaaactcaagttttttg			
cassett	Intron 15 Intron 16 Intron 17		8407	gtggaataatttttgactta	T/del	gcatttggtcaaataaaatt			
i-ABCA8-99 i-ABCA8-100 i-ABCA8-101 i-ABCA8-101 i-ABCA8-102 i-ABCA8-102 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-103 i-ABCA8-100 i-ABCA8	Intron 16 Intron 17		9458-9470	tatgtcgagtaacatatgtc	(T)11~15	ctgaatgccagcttgcagtt			
i-ABCA8-100 i-ABCA8-101 i-ABCA8-102 i-ABCA8-103 i-ABCA8-104 ATP binding casset	Intron 17		54-56	tgaataatagtcatcatcat	CAT/del	aattattatcattacaacta			
i-ABCA8-101 i-ABCA8-102 i-ABCA8-103 i-ABCA8-104 ATP binding casset			433	tcaggtttttcttttttT/At	T/del	ctttatatagaaattagac			
i-ABCA8-102 i-ABCA8-103 i-ABCA8-104 ATP binding casset	Intron 24		1462	actccatctcaaaaaaaaa	A/del	gagagaaaaaaattetgeat			
i-ABCA8-103 i-ABCA8-104 ATP binding casset	Intron 33		155	Caatartttdraaaaaaa	A/del	aatottootaatatt			
i-ABCA8-104 ATP binding casset	Intron 34		184	0441400-00004444444 ###CFC####AGF+++++++	T/del	45 C C C C C C C C C C G C G C C C C C C			
ATP binding casset(	3' Flanking region		1240	atecttggaecaaaaaaaa	Adel	ctttatctgtgctttgcgtg			
ATP binding cassett									
	te, subfamily D, me	mber 1 (A	BCD1)						
	5 Fidmking region	1 0000	2//1-	agteceagggetagggeaca	A FO	gcaccctcctgcctaactcg			
			80- 900	acaatcottocagocacotg	5 5	crcaacrgcrgccccaggca			UVOIAKOVA ET AI. (2001)
	Intron 1		DOE VCOC	gggcacaarggcarccarcc	5				
	Intron 1		3066	gagaceregeceedaat	5 5	graacctctggctctcggcc			
i-ABCD1-5	Intron 2		2020						606602281
	Intron 2		2758 3758	agaaguuruccuuguuucc					
i-ABCD1-8	Intron 2		J£30 4612	gegagacageaceregeagee		ctrcgctccatggctgccat			
i-ABCD1-9	Intron 5		2748		5 C				021120233
i-ABCD1-10	Intron 6		212	aauggeeuguguguguggeeu atotatataaaaatatataca	50	gggcarryggagceretaa gggcggggaf af gagcaf af			071110761
i-ABCD1-11	Intron 5		2835	ggcgtcagcggctgttgccc	C/del	tgcaggtggaggaaggcatg			
:	:								
I P binding cassett	ATP binding cassette, subtamily D, member 3 (ABCD3)	mber 3 (A	BCD3)		0.0				
-ABCD3-1	5' Flanking region		-2034		200	gautugaacuctuugaguca			
		1 1000	0117-			ccrradgccrrrarugaaag			
		-	-6763	geageegeegeegeege atactttgccatttgagata	1/C	seesevereeeeeeeeeeeeeeeeeeeeeeeeeeeeeee			
i-ABCD3-5	Intron 1		-117	atatttctatctcactatq	AG	tataaaaaaaaaaaaaaaaacact			dbSNP:rs2147794
i-ABCD3-6	Intron 2		731	ctttggacctatactagttt	СЛ	cttaggcattgtgcttagaa			
i-ABCD3-7	Intron 2		3551	accacagtggtcttttttt	AG	tatttaaaaaaattattggg			
i-ABCD3-8	Intron 2		5936	cagaactcacttccttattc	AG	gtttttagataacattgttt		+	
i-ABCD3-9			6083	tggttctttaattttatgat	A/G	tgtttgttatagctatctta			
-ABCD3-10		exon 3	162	atgcagaaagggggaaaaa	G/A	gagcgagctgtggtggacaa	Lys54Lys		dbSNP:rs16946
-ABCD3-11	Intron 3		614	tetettgtttetgaagtatt	AT	tttcattttattttatgtga			
I-ABCD3-12	Intron 3		651 2002	gtgaaatgctagggtactgc	5.5	atacagctaccctaaatggt			
-ABCU3-13			306	ggctaatttggccagcttcc	5	accetetgeaggeettette			005NP:151041282
			097	aaaycartrocaaagaarcac		rrgagcargrrratragaag			
FABCD3-15	Intron 4		4708	tttroccadatttatocatot tttroccasatttatocatot	9/6 8/6	greecercacagaaaagaaat at aaat aaaaaaat			4CZOCI I SI' JNICOD
1.4BCD3-17		<u>avon 7</u>	555		A/G	ureecaedaaaaaaaaaaaaaaaaaaaaaaaaaaaaaaaaa	Dro185Dro		
-ABCD3-18			8	atttatadaattgattt	G/A	padacataacagcagcaga taaacataacagcagcatat			dbSNP:rs681187
i-ABCD3-19	Intron 7		124	aaatatttaatgcttttata	AG	gaaaattagagttgttgtaa			
i-ABCD3-20	Intron 7		838	ggtcacagttgacctagata	1/C	acaqttttqaqacaaaaqaa			
i-ABCD3-21	Intron 8		1150	aatcttgaatacttactagc	AC	catattgtgctagatagt			
i-ABCD3-22	Intron 9		1493	tcatcttcttccataggctt	A/G	ggtgtggagaggagatagaa			
i-ABCD3-23	intron 9		1971	ctcttcatctataaaatacc	T/C	ttttttttgtgtcgtgatcc			dbSNP:rs2147795
i-ABCD3-24	intron 13		1534	tctgttgagttgggggatcct	AG	tggaaacctcttccttcatc			
i-ABCD3-25	Intron 16		4310	gaaaagtgaatgctgagtag	GЛ	ttagccaggcttgatttaga			

agtrosgesaac NG CI certaactedgecocata restaacacagecocataa restaacacagecocataa CI certaactedgegaage cateratecocataa CI certaactedgegaage Cateratecocataa CI certaactedgecocata CI certaactedgecocat CI certaacted	₽	Region Exon	Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation <sup>c</sup>	Flanking sequence	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
664         Consangianananana         AC         pagarancentgrant consant           77         Extragraggerancensgrant         AG         Extragraggerancensgrant           76         Extragraggerancensgrant         AG         Extragraggerancensgrant           76         Extragraggerancensgrant         AG         Extragraggerancensgrant           76         Extragraggerancensgrant         AG         Extragraggerancensgrant           76         Extragraggerant         AG         Extragraggerant           76         Extraction operation         AG         Extragrangerant           76         Extraction operation         AG         Extraction operation           76         Extraction operation         AG         Extraction operation           76         Extraction operation         AG         AG           77         Signet operation         AG         AG           76         Signet operation         AG         AG           77         Signet operation         AG         AG           76         Signet operation         AG           77         Signet operation         AG           76         Signet operation         AG           77         Signet operacing signet	i-ABCD3-26	Intron 20	273	ttctaaaagttcagagaaac	T/A	ctgtagctcattattcctgg			
883         CT         Cgacast end state of comparations         CT <thcc< th="">         Cgacast end state comparations         <t< td=""><td>i-ABCD3-27</td><td>Intron 20</td><td>1664</td><td>ctcaaaagaaaaaaaaaaa</td><td>AC</td><td>aaaaacacatgatccataa</td><td></td><td>+</td><td></td></t<></thcc<>	i-ABCD3-27	Intron 20	1664	ctcaaaagaaaaaaaaaaa	AC	aaaaacacatgatccataa		+	
17.1         tccetaseoggeneration         AG         tctcaseoggeneration         AG         tctcaseoggeneration         AG         tctcaseogcottate           868         grappresatatation         GG         castagrapprescoan         GG         castagrapprescoan           868         grapprescuttation         GG         castagrapprescoan         GG         castagrapprescoan           873         graat castagrap         GG         castagrapprescoan         GG         castagrapprescoan           873         graat castagrap         GG         GG         graat castagrap         GG           874         graat castagrap         GG         GG         graat castagrap         GG           875         graat castagrap         GG         GG         GG         GG         GG         <	i-ABCD3-28	Intron 20	6693	cttaaggtttgtgttttact	СЛ	tgagcaattagtatttccca			
220         createradecretical         AG         anticacconstruction         AG           200         cognitication and off         and         anticacconstruction         AG           201         cognitication and off         and         anticacconstruction         AG           202         cognitication and off         and         anticacconstruction         AG           203         cognitication and off         and         anticacconstruction         AG           213         cognitication and off         The         and         and         and           213         cognitication and         The         and         and         and         and           213         cognitication and         The         and	i-ABCD3-29	Intron 21	7171	atcataaacagagaaataat	AG	tcttaaatqaqctctqaaaa			
S8         agradoaataaiccicac         GA         coorganication           68         agraditage         The         agraditage           68         agraditage         The         agraditation           68         agrituitageantitutti         The         agrituitageantitutti           1227         gggaatggggggraditation         The         agrituitageantitutti           1286         gggaatggggggraditage         The         agrituitageantitutti           1286         gggaatgggggggraditage         The         agrituitageantitutti           1596         trantactacaaaag         The         agrituitageantitutti           1596         trantactacaaaag         The         agrituitageantitutti           1696         trantactacaaaag         The         agrituitageantitutti           1708         ggaatcaagtuggggggraditage         The         agrituitageacaaa           171         trantactacaaaag         The         agrituitageacaa           173         trantactactacaaagtug         The         agrituitageacaa           174         graagaacaagtug         The         agrituitageacaa           175         graagaacaagtug         The         agrituitageacaa           176         graggattagtactaa         The </td <td>i-ABCD3-30</td> <td>Intron 22</td> <td>1220</td> <td>ctagaaatcaaaggcattta</td> <td>AG</td> <td>aatataqccaaqcctttatq</td> <td></td> <td></td> <td></td>	i-ABCD3-30	Intron 22	1220	ctagaaatcaaaggcattta	AG	aatataqccaaqcctttatq			
46         addititation         TC         addititation           202         Testificitatianen         Thei         adoititation           202         Testificitatianen         Thei         adoititation           203         Testificitatianen         Thei         adoition           2037         Testificitatianen         Thei         adoition           2037         Testificitatianen         Thei         adoition           2037         Testificianen         Thei         adoition           2037         Testificianen         Thei         adoition           2037         Testificianen         The         adoition           2037         Testificianen         The         adoition           2037         Testificicanen         The	i-ABCD3-31	Intron 22	1358	agtagcaaaataatcatcac	G/A	ccagtgatcatgtgaaggag			
4461tractfctfctfgagttagg0.0instrttinggafttaggertittett0.1ingtgageraacterstegt1.12.7gggattgggeraacterstegt1.12.7gggattggggggggggggggggggggggggggggggg	i-ABCD3-32		2499	cagcgtttattatacagtgg	T/C	agatttctttagctgccaca			dbSNP:rs698951
66         gutttinggattlittitt         70el         accast gast arggiggerent           1727         gggatggerenttittitt         70el         accast gast arggiggerent           1736         gggatggerent         70el         accast gast arggiggerent           1736         gggatggerent         70el         accast gast arggiggerent           1736         gggatggerent         70el         accast gast arggiggerent           1737         gggatggerent         70el         accast gast arggiggerent           1670         gast grotter arg         70el         accast gast arggiggerent           1670         gast grotter arggiggerent         70el         accast gast arggiggerent           1670         gast arggiggerent         70el         accast gast arggiggerent           1701         gast arggiggerent         70el         accast gast arggiggerent           1702         gast arggiggerent         70el         accast gast arggiggerent           1703         gggatt arggiggerent         70el         accast gast arggiggerent           1704         gast arggiggerent         70el         gast arggiggerent           1705         ggggatt arggiggerent         70el         gast arggiggerent           1706         gggggggerent         70el         gast argg	i-ABCD3-33	Intron 4	4448-4461	taactttcttgtagttagcg	(T)11~14	aactgttttactttttaggg			
<ul> <li>Turger generation of the standard of the standard</li></ul>	i-ABCD3-34	Intron 5	268	gtttttggcatttttttt	T/del	aaccttcagtccaggttttc			
<ul> <li>17.27 ggaatgggggtgtatta</li> <li>17.8 ggaatggggggtgtata</li> <li>17.8 ggaatgggggggtata</li> <li>17.8 ggaatgggggggaata</li> <li>17.8 cagattattutttt</li> <li>17.8 tattattattatt</li> <li>17.8 ggaatggggaggaa</li> <li>17.8 tattattattattat</li> <li>17.8 ggaatggggaggaa</li> <li>17.8 tattattattattatt</li> <li>17.8 tattattattattatt</li> <li>17.8 tattattattattatt</li> <li>17.8 tattattattattatt</li> <li>17.8 tattattattattatt</li> <li>17.8 tattattattattattattattattattattattattat</li></ul>	i-ABCD3-35	Intron 5	891–902	ttggtgtaaaacctgtagtg	(T)10~13	aacaaatgcaaatatagtgt			
<ul> <li>12.8 capatitactititit 10el actigatacticage</li> <li>15.8 capatitactititi 10el actigatacticage</li> <li>15.6 tgatactataaagacea (0):</li> <li>15.7 gtastcastergatega</li> <li>15.8 gtastcastergaged</li> <li>15.9 gtastcastergaged</li> <li>15.0 gtastcastergaged</li> <li>15.1 gtastcasterged</li> <li>15.2 gtastcasterged</li> <li>15.1 gtastcasterged</li> <li>16.1 gtastcasterge</li> <li>17.1 gtastcasterged</li> <li>17.2 gtastcasterged</li> <li>17.1 gtastcasterged</li> <li>18.1 gtastcasterged</li> <li>19.1 gtastcasterge</li> <li>19.1 gtastcasterge</li> <li>10.1 gtastcasterge</li> <li>11.1 gtaged</li> <li>11.1 gtaged</li> <li>12.1 gtastcasterge</li> <li>13.1 gtastcasterge</li> <li>14.1 gtaged</li> <li>15.2 gtastgagegeggeged</li> <li>16.1 gtastgaged</li> <li>17.2 gtastgagegeggeggeggegged</li> <li>17.2 gtastgagegeggeggegged</li> <li>18.1 gtastgaged</li> <li>19.1 gtastcasterge</li> <li>10.1 gtastcasterge</li> <li>10.2 gtastgagegegggeggegged</li> <li>10.2 gtastgagegegggeggegged</li> <li>10.3 gtastgagegeggggegged</li> <li>10.4 gtastgageggggggeged</li> <li>11.4 gtastgageggggggggggggggggggggggggggggggg</li></ul>	i-ABCD3-36	Intron 7	1226-1227	gggaatgggggggtgtatcta	T/ins	tacaactttccatgtaattt			
<ul> <li>1566 tgaaccaasagcee TAins gritectalatoritieg</li> <li>1566 tgaaccaasagcee TAins gritectalatoritieg</li> <li>1567 graagactergeresaaag (N)</li></ul>	i-ABCD3-37	Intron 8	1129	cagatttacttttttttt	T/del	aatcttgaatacttactagc			
<ul> <li>1-334 traitacticasgaciga ().1-13</li> <li>1-334 traitacticasgaciga ().1-13</li> <li>1-573 traitacticasgaciga ().1-13</li> <li>1-573 gradgacasaccia ().1-13</li> <li>1-273 traitacticaticas ().1-14</li> <li>1-273 traitacatticyclasat ().1-14</li> <li>1-273 traitacatagacaca</li> <li>1-273 traitacatagacaca</li> <li>1-274 traitacticyclasat ().1-14</li> <li>1-274 traitacticyclasat ().1-14</li> <li>1-274 traitacatticyclasat ().1-14</li> <li>1-275 traitacatagacaca</li> <li>1-275 traitacatagacaca</li> <li>1-276 traitacatagacaca</li> <li>1-276 traitacatagacaca</li> <li>2-277 traitacatagacaca</li> <li>2-278 traitacatagacaca</li> <li>2-278 traitacatagacaca</li> <li>2-278 traitacatagacaca</li> <li>2-279 traitacatagacaca</li> <li>2-270 traitagacattacticatacaca</li> <li>2-270 traitagacattacticatacaca</li> <li>2-270 traitagacattacticatacaca</li> <li>2-270 traitagacatagacticat</li> <li>2-270 traitagacacacaca</li> <li>2-270 traitagacacacacaca</li> <li>2-270 traitagacacacacacaca</li> <li>2-270 traitagacacacacacacacacacaca</li> <li>2-270 traitagacacacacacacacacacacacacacacacacacaca</li></ul>	i-ABCD3-38	Intron 13	1595-1596	tgaaacataataaagcacac	TA/ins	gttatcattaatactttatg			
12     gttcctcaggtaagictag     Cdel     tragettaftttgjfcta       12001     gttagtctratttttt     (M)+-a     caastaagiggactaa       12001     gtagattttftttt     (M)+-a     caastaaggactcaatag       12001     gtagattttfttttt     (M)+-a     caastaaggactcaatag       12001     gtagattttfttttt     (M)+-a     caastaaggactcaatag       12001     gtuggctttgttttttt     (M)+-a     caastaaggactcaatag       1201     gtuggctttggtactaa     (M)+-a     caastaadcaaggacaa       1201     gtuggctttggacaata     (M)+-a     caastaggactcaatag       1201     gtuggctttggacaata     (M)+-a     caastaggactcaata       1201     gtuggctttggacaata     (M)+-a     caastaggactagaa       1201     gtuggctttggacaata     (M)+-a     caastagacaatag       1201     gttggggggggagggacaata     (M)+-a     caastagacaatag       1201     gttggggggggggggggggggggggggggggggggg	i-ABCD3-39	Intron 16	7337-7351	ttaattacttcacagactga	(T)13~15	caqqttcqatctqqqqctaa			
<ul> <li>1670 geagettergetteaate</li> <li>1710 geagettergetteaate</li> <li>1711 traatcanttingtaate</li> <li>1712 traatcanttingtaate</li> <li>1713 traatgagetagegegedeate</li> <li>1714 traatcanttingtaate</li> <li>1715 traatgagetergegedeate</li> <li>1716 traatgagetergegedeate</li> <li>1716 traatgagetergegee</li> <li>1716 geogegetergetergeaate</li> <li>1716 geogegetergetergeaate</li> <li>1716 geogegetergetergeaate</li> <li>1716 geogegetergetergeaate</li> <li>1716 geogegetergegegee</li> <li>1716 geogegetergee</li> <li>1716 geogegetergegee</li> <li>1716 geogegetergee</li> <li>1716 geogegetergee</li> <li>1716 geogegetergee</li> <li>1717 geogegetergee</li> <li>1718 geogegegegee</li> <li>1718 geogegegegee</li> <li>1718 geogegegegee</li> <li>1718 geogegegegee</li> <li>1718 geogegegegee</li> <li>1718 geogegegegee</li> <li>1728 geogegegegee</li> <li>1739 geogegegegee</li> <li>1740 geogegegegee</li> <li>1740 geogegegegee</li> <li>1740 geogegegegee</li> <li>175 geogegegegegee</li> <li>176 geogegegegegee</li> <li>176 geog</li></ul>	i-ABCD3-40	Intron 18	12	gttcctcaggtaagacctag	C/del	ttgagttatctttgatctaa			
<ul> <li>-2273 tratccattitegtaate ()</li> <li>-2273 tratccattitegtaate ()</li> <li>-2089 stragtagettag</li> <li>-2079 traatcaadtgagettag</li> <li>-2079 stragtstuttittegaaa</li> <li>-2079 stragtstuttittegaaa</li> <li>-2079 stragtstuttittegaaa</li> <li>-2079 stragtstuttitte ()</li> <li>-208 stragtstaacaageaacaa</li> <li>-208 stragtstaace</li> <li>-208 stragtstaace</li> <li>-209 stragtstaate</li> <li>-209 stragtstaageacaa</li> <li>-209 stragtstaageacaa</li> <li>-200 stragtstaageacaa</li> <li>-200 stragtstaate</li> <li>-200 stragtstaate</li> <li>-200 stragtstaate</li> <li>-200 stragtstaadea</li> <li>-200 stragtstaadea</li> <li>-200 stragtstaate</li> <li>-200 strattstaate</li> <li< td=""><td>i-ABCD3-41</td><td>Intron 20</td><td>1652-1670</td><td>gcaagactctgtctcaaaag</td><td>(A)17~20</td><td>cacatgatccataatagggg</td><td></td><td>÷</td><td></td></li<></ul>	i-ABCD3-41	Intron 20	1652-1670	gcaagactctgtctcaaaag	(A)17~20	cacatgatccataatagggg		÷	
<ul> <li>-2079 taaataaagttgggttag</li> <li>-2079 taaataaagtgggttag</li> <li>-2079 taaataaagttgggttag</li> <li>-2079 taaataagttgggttagtt</li> <li>-2081 gttggctagttutttt</li> <li>-2081 gttggctagttutttt</li> <li>-2081 gttggctutttttttgaaa</li> <li>-2081 gttggctuttttttgaaa</li> <li>-2081 gttggctutttgaaa</li> <li>-2081 gttggctutttgaaa</li> <li>-2081 gttggctutttgaaa</li> <li>-2081 gttggctutttgaaa</li> <li>-2081 gttggctuttgaaa</li> <li>-2081 gttggctuttgaaa</li> <li>-2081 gttggctuttgaaa</li> <li>-2081 gttgttuttgaaa</li> <li>-2082 gttgggggtutttgaaa</li> <li>-2082 gttgttuttgacccaaa</li> <li>-2082 gttgtttgggctagggggggggggggggggggggggggg</li></ul>	i-ABCD3-42	Intron 20	2262-2273	ttaatccatttttqtaaatc	(T)11-13	accttaaattaaccaactatc			
<ul> <li>P-2001 gtigagettagttittit (A)</li></ul>	i-ABCD3-43		2072-2079	taaaataaaattaaacttaa	(T)8~9				
<ul> <li>1-336 actatitue gata</li> <li>1-336 tigget attitue gata</li> <li>17 titage actor agtitue gata</li> <li>17 titage actor agtitue gata actor agtitue actor</li> <li>18 tigge actor agtitue gata actor agtitue actor</li> <li>19 titage actor agtitue gata actor agtitue actor</li> <li>11 titage actor agtitue gata</li> <li>11 titage actor agtitue actor</li> <li>12 agtitue actor</li> <li>13 agtitue actor</li> <li>14 agtitue actor</li> <li>15 agtitue actor</li> <li>16 agtitue actor</li> <li>17 agtitue actor</li> <li>10 agtitue actor</li> <li>11 titage actor agtitue actor</li> <li>11 titage actor actor</li> <li>12 actor</li> <li>12 actor</li> <li>13 actor</li> <li>14 actor</li> <li>15 actor</li> <li>16 agtitue actor</li> <li>17 agtitue actor</li> <li>10 agtitue actor</li> <li>10 agtitue actor</li> <li>11 agtitue actor</li> <li>12 actor</li> <li>12 actor</li> <li>13 actor</li> <li>14 actor</li> <li>14 actor</li> <li>15 acta actor</li> <li>16 agtitue actor</li> <li>17 agtitue actor</li> <li>16 agtitue actor</li> <li>17 agtitue actor</li> <li>18 actor</li> <li>10 agtitue actor</li> <li>10 agtitue actor</li> <li>11 actor</li> <li>12 acta actor</li> <li>13 actor</li> <li>14 actor</li> <li>15 acta actor</li> <li>16 agtitue actor</li> <li>17 acta actor</li> <li>18 actor</li> <li>19 actor</li> <li>10 acta actor<td>i-ABCD3-44</td><td></td><td>2080-2091</td><td>attaaacttaattttttt</td><td>(A)10-11</td><td>caaaqcaaacaaattaactaq</td><td></td><td></td><td></td></li></ul>	i-ABCD3-44		2080-2091	attaaacttaattttttt	(A)10-11	caaaqcaaacaaattaactaq			
76EggeattettttttgaaaG/Aaggaeeteegggggeaeg71tttaggoesteetttttttgaaaG/Aaggaeeteegggggggggggggggggggggggggggggg	i-ABCD3-45	3' Untranslated region exon 23	3349-3368	acttattttctgttcagatt	(A)16-19	ctcagatatcctatacaacc			
76tggcattettttttgaaaa6/Aaggaacteggtgereaa71tttagecacgttggtettggt7/Gttgggccaatggagtgereag73tttagecacgttggtettgg6/Aggggtaccacteggtggagtgereag73ggggttettagegereag6/Aggggereacteggttetgaceaa73ggggttettagegereag6/Aggggereacteggttegaat73ggggttettagegereag6/Aggggagggereag74tagtectagggereag6/Aggggagggereag75ggggttettagegereag6/Agggereag76tagtectagggereag6/Agggereag77tagtectaggagtereag6/Agggereag78ggggttettaggereag6/Agggereag79tagtettaggaceg6/Agggereggattggaceg70ttdgttettagecectag6/Agggereggattggaceg71ttdgttettagecectggaggereag6/Agggereggattggaceg79ttdgttettagecectggaggereag6/Agggereggattggaceg70ggggattggggereag6/Agggggeregggeregggereggereggereggeregge									
<ul> <li>regearcontitution</li> <li>regearcontitution</li> <li>regearcontitutities</li> <li>regearcontitutities</li> <li>regearcontitutities</li> <li>regearcontitutities</li> <li>regearcontitutities</li> <li>regearcontitutities</li> <li>regearcontitutities</li> <li>regegarcontitities</li> <li>regegarcontities</li> <li>regegarcontities</li></ul>		Issette, subramily D, member 4 (	ABCD4)						
<ul> <li>criticiagiticitigacaci</li> <li>criticiagiticitigaci</li> <li>criticiagiticitigaci</li> <li>critageconstruction</li> <li>gragocastgragacicati</li> <li>gragocastgragacicati</li> <li>gragocastgragacicati</li> <li>gradoggraditigacaci</li> <li>gradoggraditigagacaci</li> <li>gradoggraditigagacaci</li> <li>gradoggraditigagaci</li> <li>gradoggraditigagacaci</li> <li>gradoggraditigagacaci</li> <li>gradoggraditigad</li> <li>gradoggraditigat</li> <li>gradoggraditigad</li> <li>gradoggraditigad</li> <li>gradoggraditigad</li> <li>gradoggraditigad</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>gradoggraditigat</li> <li>graditigadacacacacitigat</li> <li>graditigadacacacitigat</li> <li>gradi</li></ul>	-ABCD4-1		2/6	tggcattcttttttgaaaa	G/A	aagaacctcaggtgcacaaa			
<ul> <li>rtagcacgrigatottot</li> <li>tragcacgrigatottot</li> <li>crtasocactogagg</li> <li>ggggrtaccactogagggggggggggggggggggggggggggggg</li></ul>			925	cttctcagttcttgacaccc	1/C	gtgggccaatgcaaggctcc			
<ul> <li>accaccoccuatedage</li> <li>accaccoccuatedage</li> <li>accaggacagggacage</li> <li>ragrettageggeccate</li> <li>ragrettageggecage</li> <li>ragrettageggecage</li> <li>ragrettageggeggggacage</li> <li>ragrettageggeggggggggggggggggggggggggggg</li></ul>				traagcacgttgatcttgct	AVG AVG	ttggcccacgtgggactgat			
<ul> <li>according and adjust and a second adjust a</li></ul>				ccracccccarrcagragg	614	gggcraccacctgcrcactc			
<ul> <li>gadagggterectoggagg</li> <li>gadatigtereccogagg</li> <li>gaggettereccogagg</li> <li>gaggettereccogagg</li> <li>gaggettereccoggg</li> <li>gaggettereccoggg</li> <li>gaggettereccoggg</li> <li>gaggettereccoggg</li> <li>gaggettereccoggg</li> <li>gaggetteregggettereccoggge</li> <li>getteregggetteregggettere</li> <li>getteregggegggggegggegggegggegggegggeggg</li></ul>			230	ataggagcagagggcccatt	G/A	ggccactcagattggacagg			dbSNP ID:rs2074945
III9999tttttttagtggcreagat679999tttttttagtggcreagat689995tttggaaattgreagat699945tttggaastggreagat619945tttggaastggreagat639945ttggaastggreagat649945ttggaastggreagat679945ttggaastggreagat689145ttggaastgereagat699145ggreettgaastgreagat699145ggreettggaastgereagat61915ggreettggaastgereagat62915ggreettggaastgereagat63915ggreettggaastgereagat64915ggreettggaastgereagat65915ggreettggaastgereagat66916ggreettggaastgereagat67916ggreettggaastgereagat68915ggreettggaastgereagat69916ggreettggereagat60916ggreettggaastgereagat61916ggreettggaastgereagat62916ggreettggereagetgggereaget63916ggreettggaastgereagetgggereaget64916ggreettggereagetgggereaget65916ggreettggaastgereagetgggereaget66916ggreettaggaggereaget67916ggreettaggaggereaget68916ggreettaggaggereaget69916ggreettaggaggereaget60916ggreettaggagereaget61916ggreettaggaggereaget62916ggreettaggaggereaget63916ggreettaggaggereaget64916ggreettaggaggereaget65916ggreettaggaggereaget66916ggreettaggaggereaget67916ggreettaggagtgggreettag68916ggreettaggagt			612 113	gacaggggctacctgagagg	6/	aacaggagtcagggctgagg			
67gastretragreated67gastretragreated68reastretreaccedet69reastretreaccedet61atcagtretreaccedet61atcagtretreaccedet61atcagtretreaccedet61atcagtretreaccedet61atcagtretreaccedet61atcagtretreaccedet61atcagtretreaccedet62atcagtretreaccedet63acgetretregegeacce64atcagtretregegeaccedet65acgetretregegeaccedet66agtregegetregger67acgagtreggecected68accedetreggeacce69acgetretreggeacced69acgetretreggeacced61acgetreggeagegegegege62acgetreggeacced63acgetreggeagegegegegegege64gegagegeggeggeggeage65acteccetreggeacced66acteccetreggeacced67gegagegeggeggeage68acceacgeagegeggeage69acceacgeageage69acceacgeageageacce61gegagactreggeage63trettreacgaageage64gegactretreacce65acceacgeageageage66acceacgeageageageageagea67gegagagageageageageageageageageageageage				ggggutttctaccccataac	S S S	grccaggaaagggrcagact			dDSNP ID:rs20/4946
00     yaccustorsysted     No     yaccustorsysted       01     treatgygacstysted     G/A     agtgyggtragogsgygg       02     gcgsgatcstystedscoccg     G/A     agtgyggtragogsgygg       03     toggtystystedscoccg     G/A     agtgyggtragogsgygg       04     gtgyggggggggggggggggggggggggggggggggg		intron 7	767	tagtettagtggeetagegt	6/P	gggcctgaaattgtcaaatg			
00     rccargogaaccrgaaccor     0,M     cagagettageaccrggrc       01     rccargogaaccrgaaccor     0,M     agrgggettagaaccrggrc       02     gggagtagettggrcargaacgrc     0,M     agrgggettgggacgrggacgr       03     rggattgggccargaccor     0,M     agrgggettgggccarggrcarg       04     agrgggettgggcrgggrcarggrcargggrcarg     0,M     agrgggettgggcrggggcggggcggggcggggcggggcgg			207	yaaar Lyrcaaaryaaryaa		decreatecereteded			
0.0     acceptoreconstruction       0.1     accenterconstruction       0.1     accenterconstructon       0.1     accenterconstruct	i-ABCD4-10		910 981		4/5 4/0	cagagettageaccetggte	Ala304 I hr		
0.0     greggadactord     0.0     acgogadactord       0.1     triggattoggadactord     0.0     acgogattoggadactord       4.8     crotroprocentance     0.0     greggattoggadactord       4.8     crotroprocentance     0.0     greggattoggadactord       4.8     crotroprocentance     0.0     greggattoggadactord       4.8     greggadactord     0.0     greggattoggadactord       4.8     greggadactord     0.0     greggattoggadactord       4.1     greggadactord     0.0     greggattoggadactord       0.01     grectoggadactord     0.0     greggattoggadactord       0.01     grectoggadactord     0.0     greggadactogattogattogattogat       0.01     grectoggattoggadactord     0.0     greggattoggadactoccod       0.01     grectoggattogadactord     0.0     greggattogcadactoccod       0.02     catatgettaggaagtoccod     0.0     greggattogcadactoccod       0.03     catatgettaggaagtoccod     0.0     greggattogcadactoccod       0.03     catatgetaggaagtoccod     0.0     greggattogcadactoccod       0.03     tutgettggagaagtoccod     0.0     greggattogcadactoccod       0.03     tutgettggagaactoccod     0.0     greggattogcadagtoccod       0.04     greggattogcadgtogcadactoccod     0.0			100	arcagergerceaeccager		arcdaccrgrccacgacgcr	Da 2021		
<ul> <li>46 greggreggreggreggreggreggreggreggregger</li></ul>	i-ABCD4-12		107	gcgagatcctgggcgagagc	6/A	agtggggcttggacacgtga	Glu367Lys		
<ul> <li>44 cutodiadacacca</li> <li>45 cutodiadacacca</li> <li>46 gtgggaggagaagaggagaacaata</li> <li>46 gtgggaggagagagagagagagagagagagagagagaga</li></ul>	i-ABCD4-14	Intron 13	161			agcagcreergaggcaggra			
<ul> <li>46 guggggggggggggggggggggggggggggggggggg</li></ul>		Intern 17	202						
<ul> <li>9505939393939393939393939393939393939393</li></ul>	i-ARCD4-16	Introp 17	946	concentrations and	200	crecectgaggeagger			
<ul> <li>9001 9900099904000 and 9000</li> <li>9001 9900099904000 and 9000</li> <li>604</li> <li>618</li> <li>618</li> <li>618</li> <li>619</li> <li>619</li> <li>614</li> <li>614</li> <li>614</li> <li>615</li> <li>614</li> <li>615</li> <li>614</li> <li>615</li> <li>614</li> <li>614</li></ul>	i-ARCD4-17	Intron 18	41						
<ul> <li>B3 generagetorege</li> <li>B3 generagetorege</li> <li>B6 aactcagattctcggaactgat</li> <li>B7 ctgaaattattgcaattc</li> <li>C7 ctgaaattattgcaattc</li> <li>C7 ctgaaattattgcaattc</li> <li>C7 ctgaaattattgcaattc</li> <li>C7 crgaaattattgcaattc</li> <li>C7 crgaaattattatgcaattc</li> <li>C7 crgaaattattgcaattc</li> <li>C7 crgaaattattgcaattc</li> <li>C7 crgaaattattgcaattc</li> <li>C7 crgaaattgaacggatggtgggg</li> <li>C7 crgaaattcaattc</li> <li>C7 crgaaattcaattct</li> <li>C7 crgaattcaattcgc</li> <li>C7 crgaattcaattcaattcgc</li> <li>C7 crgaattcaattcaattcaattcaattcaattcaattcaa</li></ul>	-ABCD4-18		2001	4466649444444444444444 4000944544544444444444	G/A				
<ul> <li>aactcagattctcggcacct</li> <li>ctgaaattatatgcaaattc</li> <li>ctgaaattatatgcaaattc</li> <li>ttgtgtaggaagctgataca</li> <li>ttgtgtaggaagctgataca</li> <li>ttgtgtaggaagctgataca</li> <li>attcgtcagaggta</li> <li>ttggtgtaggtggtgggggg</li> <li>ttggtgtggtggggggg</li> <li>ttggtcaaaatttata</li> <li>ttggtcaaaatttata</li> <li>ttgtctcaaaatttata</li> <li>ttaattcacattattgc</li> <li>ttaattcacattatttgc</li> </ul>	i-ABCD4-19		2083	catatgettgaagtgetgat	1/G	acctacaaatgatttcagat			dbSNP ID:rs2466
<ul> <li>actcagattctcggcacct</li> <li>ctgaaattatatgcaaattc</li> <li>ttgtgtaggaagctgataca</li> <li>ttgtgtaggaagctgataca</li> <li>ttgtgtaggaagctgataca</li> <li>attgtccagaaattc</li> <li>attgtccagaacgatggta</li> <li>ttg</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgattga</li> <li>ttggttgattgactgaggg</li> <li>ttggttgattgacttga</li> <li>ttggttgattgacttga</li> <li>ttgattcacattgattga</li> <li>ttgattcacattgat</li> </ul>	ATP binding ca	ssette, subfamily F, member 1 (,	ARCE1)						
Intron 9         237         ctgaaattatatgaaatta         Cit           Intron 9         4203         ttgtgtaggaagtgaatta         Cit           Intron 10         1811         ccaagaaacttaggaagtgataca         T/G           Intron 10         1811         ccaagaaacttaggaagtgataca         T/G           Intron 10         1811         ccaagaaacttaggtaggaagtgataca         T/G           Intron 17         2301         atatccagaaacagatggta         T/G           Intron 18         1810         tggttgatgatgatggtagttggt         G/G           Intron 1         5349-5363         ttggttggtagatgatggtggttggtggtggtggtggt         G/G           Intron 1         5349-5363         ttggttggtagatggtggttggttggtggt         (1):-16           Intron 1         5345-5854         taaattcacaaatttata         (1):-16           Intron 1         5345-5854         ttaattcacattgtcaaaatttata         (1):-16           Intron 1         5345-5854         ttaattcacattgtcaaaatttata         (1):-16           Intron 8         1155-1169         ttaattcacattgtcaaaatttata         (1):-16           Intron 9         1025-1169         ttaattcacattgtcaaaatttgtc         (1):-16	i-ABCE1-1	5' Flanking region	-158	aactcadattctcdddct	СЛ	radradrt ddrt trocraar			
Intron 9         4203         ttgtgtaggaagctgataca         T/G           Intron 10         1811         ccaagaaacttcagctttct         C/T           Intron 17         1811         ccaagaaacttcagctttct         C/T           Intron 17         2301         atatccagaaacaggtggt         T/C           Intron 17         2301         atatccagaaacaggtggt         T/C           3'Untranslated region         xon 18         180         tggstgattagactgatcg         T/C           Ntron 1         5349–5363         tttgstcgggstgattagactg         T/1-6         T/1-16           Intron 1         5345–5854         tacatttgstcaaaatttata         (T)1+16         T/1-16           Intron 8         1155–1169         ttaattcacatgsttgsttgstfgst         (T)1+16         T/1-16           Intron 9         1052–1024         ttaattcacatgatttgst         (T)1+16         T/1-16	i-ABCE1-2	Intron 9	237	ctgaaattatatgcaaattc	СЛ	gtagctttataggaagcaga			
Intron 10         1811         ccaagaaacttcagcttct         C/T           Intron 17         2301         atatcoagaaacgatgta         7/C           3'Untranslated region         exon 18         1810         tgatgatgatggctggtagt         7/C           3'Untranslated region         exon 18         1810         tggatgattgggctggtggtggtggtgg         7/C           3'Untranslated region         exon 18         1810         tggatgattgggcggcggtggggg         7/C           3'Untranslated region         8xon 18         5845-5853         ttggttggtggggggggggggggggggggggggggg	i-ABCE1-3	Intron 9	4203	ttqtqtaqqaaqctqataca	1/G	taatttqacatatqaqatqt			
Intron 17         2301         atatccagaaacagatggta         T/C           3'Untranslated region         exon 18         1810         tggatgattagactgactct         G/C           3'Untranslated region         exon 18         1810         tggatgattagactgactct         G/C           Intron 1         5349–5563         tttgtttgggttggggg         (1):1-16           Intron 1         5345–5854         tacatttgtcaaaatttata         (1):1-16           Intron 8         1153–1169         tcttttcaaatttgc         (1):1-16           Intron 9         1023–1264         ttctttcaatttgc         (1):1-16	i-ABCE1-4	Intron 10	1811	ccaagaaacttcagctttct	СЛ	ttcacttaaatataqqaaac			
3'Untranslated region         1810         tggatgattagactgactct         G/C           Intron 1         5349-5363         tttgtctgggttgggg         (1):13-16           Intron 1         5349-5363         tttgtctgggttggggg         (1):1-16           Intron 1         5349-5363         tttgtctgggttggggggggggggggggggggggggg	i-ABCE1-5	Intron 17	2301	atatccagaaacagatggta	T/C	gtgcagaacaggttgtacag			
Intron 1         5349–5363         ttggtctggggtgggg         (T):13-16           Intron 1         5845–5864         tacattgccaaaatttata         (T):3-10           Intron 5         836–851         tacattgccaaaatttata         (T):3-10           Intron 8         1153–1169         tccttgcaactgattggc         (T):3-17           Intron 9         1023–1264         ttgctctgttgggggggg         (T):3-17	i-ABCE1-6		1810	tggatgattagactgactct	G/C	agaatattgataagccattt			
Intron 1         5845-5854         tacattgtcaaaatttata         (T)9-10           Intron 5         836-851         taaattcacatgattcgta         (T)19-16           Intron 8         1155-1169         tctttcaaacttattttgc         (T)19-17           Intron 9         1023-124         ttactctgtttcatttgc         (T)13-17	i-ABCE1-7	intron 1	5349-5363	tttgtctgggttggttgggg	(T)13-16	gagactgggtctgactctca			
htton 5 836-861 taaattcacatgattctgta (1):4-16 htton 8 1153-1169 tcttcacaatcattattgc (1):3-17 htton 9 1023-1024 ttoctctatttcaattctc (1):3-17 htton 9	i-ABCE1-8	Intron 1	5845-5854	tacatttgtcaaaatttata	(T)9~10	gcagataatcatttcatctc			
Infrom 8 1153-1169 tetttecaaacttatttge (T):3-17 Infrom 9 1023-1024 ttggetetgettegaatgeetet T/ins	i-ABCE1-9	Intron 5	836-851	taaattcacatgattctgta	(T)14-16	aggatcctcctgactggcag			
Intron 9 1023-1024 ttactctatttcaaatctct T/ins	i-ABCE1-10	Intron 8	1153-1169	tctttcaaacttatatttgc	(T)13~17	catagtttcatgtttgatga			
	i-ABCE1-11	Intron 9	1023-1024	ttgctctgtttcaaatctct	T/ins	attcatgggccagcagctcg			

2	Region Ex	Exon	Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
i-ABCE1-12	Intron 9		2338-2346	agtgtagatggacctcgggg	(A)8~9	ctagttaaggaaaagtaata			
i-ABCE1-13	Intron 9		3213-3221	ttccaattttccattgttac	(T)8~9	cttgccagattactcctgaa			
i-ABCE1-14	Intron 10		284-299	tcctctgcattttggcttct	gcagtattactgtagt/del	atttgtcattttcaaattaa			
i-ABCE1-15	Intron 10		840-853	ttttttggtttctttctttc	(T)13~14	aatcttggaggaatctttt			
i-ABCE1-16	Intron 16		1163-1172	gattagaaatccaggttaaa	(T)9~10	gtttgcacaaaaatattac			
i-ABCE1-17	Intron 16		1372–1382	taaaatttaatcaaaattga	(T)10~11	ctcttagtcctcaaaccctt			
ndino casset	ATP binding cassette_subfamily F_member 1 (48//54)	or 1 (ARI	CF4)						
	5 I Intranslated region acon 1		50		ť				
		-		guession		cgccgccggaagcggaaata			
I-ABCF 1-2	Intron 1		101	gcacgagactgaccgggccc	C/G	tgcgggagttactgcgcatg			
i-ABCF1-3	Intron 9		321	tggaggtagcggtttgtcag	AG	ggetteeetgeagggagaaaa			dbSNP ID:rs1264440
i-ABCF1-4	Intron 16		103	ctacatttcaaqqactaccq	СЛ	acadaactcaaatttatat			dhSNP ID re1264437
LARCE1-5	Intron 20		80						104407 ISI'DI JNOON
			8	rgacercaaccacec		crcrtrcrcggggagaaa			
<u>0</u>	Intron 23		ŝ	agtgtgccctcatccctgct	C/A	catggggaccaagctgtagt			
i-ABCF1-7	Intron 7		342-354	acagagcgagactccgtctc	(A)10~14	gaaaaaaaaaaaaacattt			
i-ABCF1-8	Intron 7		356-369	cqtctcaaaaaaaaaaaaa	(A)13-15	catttcatcagacctgtctt			
i-ABCF1-9 3' L	3' Untranslated region exon 24	n 24	2425	tcaaccaaccccaacaataa	A/del	2014 t + 704 t 000 000 000 000 000 000 000 000 000			
~			1067-1068	attaatttgatcaattgtct	T/ins	guerececcedagageee aatatgtcgtactctagatt			
ding case of	ATD hinding cassatta subfamily G mombord / ADCC4/								
IUIIIY CASSEL		er I (AD	(LO)		,				
	o Flanking region		-380	gcaataatcattggctagag	GIA	tattgtgatatgatgtcatt			
I-ABCG1-2	5' Flanking region		-206	aatctgaccaaaagtgagag	AC	tgggtagattttcctacttg			dbSNP ID:rs1893590
FABCG1-3	5' Flanking region		-136	gctttacgcccagtgacttg	T/G	gagggaacagaactgccctt			dbSNP ID:rs1378577
i-ABCG1-4	Intron 1		199	caccaaatattggtgagctg	СЛ	ctggatttgggagatgcagt			
i-ABCG1-5	Intron 1		291	acttgggggtccggtgtgagg	AC	tectacaeteaatttetata			
I-ABCG1-6	Intron 1		318	actcggtttctgtgatggtg	T/A	atacaggggggggggtcacagggtt			
I-ABCG1-7	Intron 1		468	gateccaacggatttetaga	T/C	ccetecaaaaaaceetttaa			
i-ABCG1-8	Intron 2		434	ctgggtacaggttttgttcc	G/A	attaatctactattaaatat			
i-ABCG1-9	Intron 3		1839	ttaaaatgagttgtttttct	C/G	ctaaagcctttaqqqqaqttq			
-ABCG1-10	Intron 3		3076	tttgtcacttccttcgtctc	СЛ	ggetetaetteeetggggggt			
i-ABCG1-11	Intron 3		3352	gttccttggaggaaacgtgg	GIA	gtacacagtggttccagtta			
-ABCG1-12	Intron 3		8030	acagtgaagcacaaggcagc	CT	gaagacacagcaggcaggtc			
-ABCG1-13	Intron 3		8066	aggtcaggtctgtqtgcaca	T/C	tqqcaqqctqcA/Gtqcaqacc			
i-ABCG1-14	Intron 3		8078	tqtqcacaT/Ctqqcaqqctqc	AG	tgcagaccagcctC/Tggccca			dhSNP ID:rs692383
-ABCG1-15	Intron 3		8092	ggctgcA/Gtgcagaccagcct	C/T	gacccagatagagagagaga			
-ABCG1-16	Intron 3		8285	ctggacatgtgactcccctg	СЛ	accoaccetcaccetcaca			
-ABCG1-17	Intron 3		8342	gecagaggeceaagtecteag	1/G	at accort t t t c c c c t t c a			dhSND ID:rsq15845
I-ABCG1-18	Intron 3		8860	caddtaataatacaatccaa	T/C	10000000000000000000000000000000000000			
-ABCG1-19	Intron 4		2246	00000000000000000000000000000000000000	0/L	00000000000000000000000000000000000000			dhSND ID:re664010
-ABCG1-20	Intron 4		2319		0.: U	22274222244444444444444444444444444444			
-ABCG1-21	Intron 4		2557		5/0				
i-ABCG1-22	Intron 5		139	tancradacaecaecaecaecaecaecaecaecaecaecaecaecae	021 1/A				
-ABCG1-23	Intron 5		171	00400004000040000000000000000000000000	0/0				
-ARCG1-24	Intron 6		ţ						
-ABCG1-25	Intron 6		21	cay craccy caay cy cry co		ccagggguggucae/Agaatot anntatonatttananttti			
-ARCG1-26	Intron 6		1101						
ABCG1-27	Intron 6		1011			geragreeregaagaga		+	
-ARCG1-28	Intron 6		2282			grgggcaracaaggggrggc			
	e contra		2022	credearcaegeagereea	- L 2 (	garccratraatrgggggggg			
67-1				araggcacacatggggaaaa	6/1	gcacccaccaagagtcctca			dbSNP ID:rs2032260
-ABCG1-30			3853	cctgggcttcagcaggggcc	1/C	cacacctgcaatgggtgC/Tct			*
-ABCG1-31	Intron 6		3871	ccT/Ccacacctgcaatgggtg	CU	ctggggagagggtgcagatg			
-ABUG1-32			C/14	tccaaagcccagatttggtg	1/C	ttttggggctcttttggaat			
-ABCG1-33			4	ctggtggaggaagaaaggta	G/A	ggagggcggctgctttgtgt			
	Intron 7				1				

Intron / Intron 7		Flanking sequence	Variation <sup>c</sup>	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup> dbSNP/Previous report
	1426	aatteteetteteaaettaa	AG	gaaatattttatagaaaaat		
	2342	agagcctgcaatgggccgcc	G/A	agggacctgcccatgactca		
	2399	gaggggttgacagacaggat	AG	tgtctgC/Gtgtgtttccagctg		
	2406	tgacagacaggatA/Gtgtctg	C/G	tgtgttccagctgctggttt		
Intron 7	1167	ccctctctgtgcccactgtt	5/5 •	tcccaacaccagcctgttct		
Intron 7	4752	raraarayarrooraya aatttaaaaaaaaa	2 E	aacataattgrgagaggaac		
Intron 7	5026	occadatetatagaseecaeecaa ceagatetatagasttteag	- D			
Intron 7	5532	aaatattaaatattooooooo	EC C	רכמממממטטמטיטיסטרמטכממט מררפפמוד רפתפוד או זיד הד		
Intron 7	5681	got anadt graf ggaaggees	- 0/1	uctaurcauaccause netaeteeetiittttii		
Intron 7	6290	tcacagcagattcatgagag	T/A	toaatotttagooococototot toaatotttagooocoatot		dhSND ID:re881304
Intron 7	6386	agatgeteceetecageeag	СЛ	acattttotocototgago		TO DO
Intron 7	6758	acctgcatggtgggtgcccc	C/G	ctdccttcctctactdcctt		dbSNP ID:rs915846
Intron 7	7029	tgggtcagattaaatatc	СЛ	tqaaqqactaaaacqtaaaaa		dbSNP ID:rs225440
Intron 7	7176	ttgctcacattgtgaaaaaa	C/G	gcaaaaagatgggttttcag		dbSNP ID:rs225441
Intron 7	9243	gcctgagagcgctggcagta	GIA	gaaggqtcgccaqtqtqqac		
Intron 7	11224	tctggtttagaggaaaat	GIA	ggcagcatcatttgtcacc		dbSNP ID:rs915847
Intron 7	11371	gggetetettggageeettt	1/6	teteteceaaceetacatet		
Intron 7	12420	gggatttcgaatctcaacac	T/C	ctqaqctctqtqctttcccc		
Intron 7	12484	gagttgtcctccaagagaat	СЛ	tttgtatggttccttttctg		dbSNP ID:rs225446
Intron 7	12955	ctggggttggtgggagccac	AG	gtctcacacctattqqcaqq		dbSNP ID:rs225447
Intron 7	12985	ctattggcaggtcgtgaaca	1/C	tgttcttggatttgcaaata		
Intron 7	20041	acatggccggcttcccttct	T/C	cctcG/Agaatggcctggaatt		
Intron 7	20046	gccggcttcccttctT/Ccctc	G/A	gaatggcctggaattcgatc		
Intron 7	21058	acaagacttagaatttgacc	G/A	tgattttaaaactattctaa		
Intron 7	26189	ttcttggatgtggccatgca	СЛ	gggggcaagggtttgatgag		
Intron 7	27453	atcatgtggtttgggggaaa	G/C	ctgggaccccacttggtaca		
Intron 7	28098	caggaaggagacagctgctg	G/C	tgctgcttagagttaggcgc		dbSNP ID:rs225376
Intron 7	29670	ccttcagttgtaataggcag	AG	aggagcgcacgaggaggctg		dbSNP ID:rs225378
Intron 7	29810	attgtttctcctggttttgt	T/C	tgtgttgactttccctttaa		
ntron 10	1995	cagatcccttggttgctggg	СЛ	aggtagtaggagaggttttt		dbSNP ID:rs25383
Intron 10	2116	aaacagggcttgagtcctcc	GIA	taagggacaggagaccttcc		
Intron 10	2145	aggagaccttcccacatcct	G/A	gcaagaattcttctttttc		dbSNP ID:rs225384
Intron 10	2437	cagactaaatgcacaattct	G/A	gattgagctgactgtattga		dbSNP ID:rs25385
ntron 10	2689	tgtaaagatggagaagaac	AG	cagtagtcgcttgctgtgag		+ dbSNP ID:rs225386
ntron 10	2804	tgtgactcatggcctctgcc	AG	ggggactgggctggccctgc		dbSNP ID:rs225387
Intron 13	1196	tgaaaagaaaatggatgagt	G/A	gaaA/Cccaaaagagagaaaat		
	1200	aagaaaatggatgagtG/Agaa	AC	ccaaaagagagaaaatgtgg		
Intron 13	2400	aagcagaggcttttccaccc	6/A	gagactcaagaagctgctcc		
Internation	2430	grggrgaagragagcrgagc	A .	cacgggggggggccctccatcc		
Intron 13	2007		4/5 1	ctgtgccccgagccgagctt		dbSNP ID:rs170441
Intron 13	2022 2850	cagcaggcrccgrgcrgaag		cacagcaagccaggcccttg		
Intron 13	2010			gagerggaagacccagaaca		
ntron 13	3506	geerereedgageageagea ggradretggggagaga	- 27	9998400094499046990 201277757737777777777777777777777777777777		
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Intron 13	3721					
Intron 13	3852					
Intron 13	3025	caccagagccactcagtcgg				dDSNP ID:rs225410
Intron 13	3070	90000000000000000000000000000000000000				
Intron 13	<b>4</b> 2915	acceaceageererereaga nanonnotnoant annation	-/C	agcottocagaagotgttto		
Intron 13	1020					
	7764	gaaaccaccagaaarrgrgc	A P	tcctctcatgtgtccattca		dbSNP ID:rs492338

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	Flanking sequence <sup>b</sup> Variation	ion <sup>c</sup> Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
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Internal         Circle         Circl	₽	Region Exon	n Position <sup>a</sup>	Flanking sequence <sup>b</sup>	Variation	Flanking sequence <sup>b</sup>	Substitution	Repetitive sequence <sup>d</sup>	dbSNP/Previous report
07.0     construction     0.0     construction     0.0       08.0     opport/construction     0.0     construction     0.0       08.0     opport/construction     0.0     opport/construction	i-ARCG5-22	Intron 11	4623		Ц				
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000         despect consequence         Old         space comparation         Space comparation         Old         Space comparation         Space comparation <t< th=""><th>i-ABCG5-25</th><th>3' Untranslated region exon</th><th></th><td>tgaggattaaaataaaaac</td><td>СЛ</td><td>gtaggaatgggctcaacagt</td><td></td><td></td><td></td></t<>	i-ABCG5-25	3' Untranslated region exon		tgaggattaaaataaaaac	СЛ	gtaggaatgggctcaacagt			
<ul> <li>Monto grand and a concentration of the second and a concent of the</li></ul>	i-ABCG5-26	3' Flanking region	1560	catagcactcagcaagaaac	G/C	tqtqctaaaqactqaqqttc			
<ul> <li>2.2.2.2.1 aggregreggreggregence in Advantage</li> <li>2.2.2.2.2.2.2.2.2.2.2.2.2.2.2.2.2.2.2.</li></ul>	i-ABCG5-27	Intron 4	1078-1080	gggcacagctccctgggagc	AGG/del	agaactcccgatagcagagt			
<ul> <li>39 gaartaagtorogoto (M)n-r</li> <li>cutturturgrageto (M)n-r</li> <li>cutturturgrageto (M)n-r</li> <li>cutturturgrageto (M)n-r</li> <li>cutturturgrageto (M)n-r</li> <li>cutturgragetorogoto (M)n-r</li> &lt;</ul>	i-ABCG5-28	Intron 10	2321-2327	agcgggttggggtgagccctt	TAACATT/del	aqqtaqqtqtqqtqttqqct			
<ul> <li>-0004 cittitugiergenergener</li> <li>-2731 tacoctaaaacttaarget (h)10</li> <li>cetterergenergenergenergenergenergenergen</li></ul>	i-ABCG5-29	Intron 11	422-433	ggaattaagactagtcagac	(A)10~12	geetgeagaataaaagaetg			
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<ul> <li>asgagaricagar (agreed and a grant agreed and a grant agreed and a grant agreed agreed and a grant agreed agr</li></ul>	i-ABCG5-31	3' Untranslated region exon		taccctaaaacttaaagtat	(A)11~13	cctaccgaaaaaaaaaaaaa			
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Hubble         Bit         Controportion         Acr         Consequences         Acr         Acr <th>i-ABCG8-1</th> <th>5 Untranslated region exon</th> <th>1 -19</th> <td>a a dadadat dradadad</td> <td>GЛ</td> <td>racaratet at acaret</td> <td></td> <td></td> <td>1 of al (2001)</td>	i-ABCG8-1	5 Untranslated region exon	1 -19	a a dadadat dradadad	GЛ	racaratet at acaret			1 of al (2001)
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	i-ABCG8-32	3' Untranslated region exon 1		tcctggggacagtgaggaca	A/del	tgaccctacagatgctcagc			

This, insertion polymorphism, det, detending polymorphism, and t, single-indecodue polymorphism <sup>a</sup> Nucleotide numbering is according to the mutation nomenclature (Dunnen and Antonarakis 2000) <sup>b</sup>S' and 3' sequences to each variation are denoted by small letters <sup>c</sup> Variation is shown by capital letter <sup>d</sup> + indicates the variation located with a repetitive sequence

Table 1. Continued

Table 2. Summary of genetic variations identified in 13 ABC transporter genes

Gene	5' flanking region	5' UTR <sup>a</sup>	Coding region	Intron	3' UTR <sup>b</sup>	3' flanking region	Other types of variations	SNP	Identity to dbSNP/previous report	New SNP
ABCA4	3	0	4	161	0	0	34	168	43	125
ABCA7	4	1	19	37	1	5	4	67	3	64
ABCA8	0	0	3	84	0	1	16	88	10	78
ABCD1	1	1	0	8	0	0	1	10	3	7
ABCD3	2	1	2	26	1	0	13	32	7	25
ABCD4	0	0	3	14	2	0	0	19	3	16
ABCE1	1	0	0	4	1	0	11	6	0	6
ABCF1	0	1	0	5	0	0	4	6	2	4
ABCG1	3	0	0	96	1	4	8	104	29	75
ABCG2	0	0	2	40	0	1	4	43	5	38
ABCG4	0	0	1	9	1	0	0	11	2	9
ABCG5	0	0	0	24	1	1	5	26	0	26
ABCG8	0	1	3	21	0	0	7	25	7	18
Total	14	5	37	529	8	12	107	605	114	491

SNP, single-nucleotide polymorphism

<sup>a</sup>5' Untranslated region

<sup>b</sup>3' Untranslated region

**Table 3.** Distribution of SNPs by types of nucleotide substitutions

Gene	A/G	C/T	A/C	C/G	G/T	T/A	Total
ABCA4	67	54	19	14	10	4	168
ABCA7	30	20	5	9	2	1	67
ABCA8	36	29	5	9	5	4	88
ABCD1	3	7	0	0	0	0	10
ABCD3	17	9	3	0	1	2	32
ABCD4	10	4	2	1	2	0	19
ABCE1	0	4	0	1	1	0	6
ABCF1	1	3	1	1	0	0	6
ABCG1	41	38	6	10	5	4	104
ABCG2	18	13	3	0	6	3	43
ABCG4	6	3	1	1	0	0	11
ABCG5	8	10	2	2	3	1	26
ABCG8	5	11	5	1	2	1	25
Total	242 (40%)	205 (33.9%)	52 (8.6%)	49 (8.1%)	37 (6.1%)	20 (3.3%)	605 (100%)

SNP, Single-nucleotide polymorphism

total of 19 SNPs: 3 in coding regions, 14 in introns, and 2 in the 3' untranslated region. The distribution of SNPs at this locus was 1 per 703 bp on average. The frequency of each type of substitution was 52.6% for A/G, 21.1% for C/T, 10.5% for A/C, 5.3% for C/G, and 10.5% for G/T.

# ABCE and ABCF families

The *ABCE* and *ABCF* subfamilies contain genes encoding peptides that include ATP-binding domains likely to be derived from ABC transporters but that have no transmembrane domains. Their functions in the membrane transport system are not well understood (Dean et al. 2001).

*ABCE1* locus. ABCE1, a 599-amino-acid protein, contains two ATP/GTP-binding motifs; it was originally identified as protein inhibitor RLI, which can regulate RNase L activity in the 2-5A pathway (Bisbal et al. 1995). Interferons induce transcription of numerous genes involved in defense against viral infections, in control of cell proliferation, and in differentiation, as well as in modulation of immune responses (Williams 1991). The 2-5A/RNase L system, one of the major pathways induced by interferons, might play a more general role in regulating RNA turnover and stability in cells (Hassel et al. 1993). RNase L is an endoribonuclease that, on activation by 2-5A, leads to inhibition of protein synthesis by cleaving mRNAs at the 3' side of UpNp sequences (Floyd-Smith et al. 1981; Zhou et al. 1993).

Screening an approximately 26.2-kb region containing the *ABCE1* gene identified a total of six SNPs, 1 in the 5' flanking region, 4 in introns, and 1 in the 3' untranslated region. The distribution of SNPs at this locus was 1 per 4374bp on average. The frequency of each type of substitution was 66.7% for C/T, 16.7% for C/G, and 16.7% for G/T. We also found 11 variations of other types at the *ABCE1* locus.

ABCF1 locus. ABCF1 was first isolated as a protein of unknown function that was modulated by tumor necrosis

factor alpha in cultured synoviocytes (Richard et al. 1998). The *ABCF1* cDNA encodes an 807-amino-acid peptide that contains two signature motifs of the ABC transporter family, a 2ATP/GTP-binding site A motif, and three nuclear localization signals. ABCF1 is likely to play a key role in the initiation of mRNA translation, through interaction with eukaryotic initiation factor 2 and association with ribosomes (Tyzack et al. 2000). Screening approximately 10.8kb at the *ABCF1* locus identified a total of six SNPs, 1 in the 5' untranslated region and 5 in introns; their distribution was 1 in 1795 bp, on average. The frequency of each type of substitution was 16.7% for A/G, 50% for C/T, 16.7% for A/C, and 16.7% for C/G. We also found four

variations of other types at this locus.

# ABCG genes

The ABCG subfamily is comprised of six 'reverse' halftransporters that have a nucleotide-binding fold at the amino terminus and a transmembrane domain at the carboxyl terminus.

ABCG1 locus. One of the members in the ABCG subfamily, ABCG1, was originally named human White on the basis of 33% identity in amino-acid sequence to the product of the White gene of Drosophila melanogaster, which transports precursors of eye pigments, (guanine and tryptophan) along with brown and scarlet, in the eye cells of the fly (Chen et al. 1996; Croop et al. 1997; Klein et al. 1999; Dean et al. 2001). Klucken et al. (2000) showed that ABCG1 was regulated by cholesterol uptake or HDL3-mediated lipid release in human macrophages. Regulation of ABCG1 mRNA levels by the liver-specific X receptor/retinoid X receptor pathway, which is involved in lipid metabolism (Repa et al. 2000; Venkateswaran et al. 2000), suggests a function for ABCG1 in homeostasis of cholesterol in macrophages. The ABCG1 gene contains multiple promoters, spans more than 98kb of genomic DNA, and consists of 23 exons that encode proteins with different amino-terminal sequences generated by alter-native splicing (Kennedy et al. 2001).

Screening an approximately 66.2-kb region around the *ABCG1* gene identified a total of 104 SNPs: 3 in the 5' flanking region, 96 in introns, 1 in the 3' untranslated region, and 4 in the 3' flanking region. The distribution of SNPs at this locus was 1 per 637bp on average. The frequency of each type of substitution was 39.4% for A/G, 36.5% for C/T, 5.8% for A/C, 9.6% for C/G, 4.8% for G/T, and 3.8% for T/A. We also found eight variations of other types at the *ABCG1* locus.

*ABCG2* locus. *ABCG2*, the second member of the ABC transporter subfamily G, is also known as the placental ABC transporter, breast-cancer resistance protein, or mitoxantrone resistance-associated protein (Allikmets et al. 1998; Doyle et al. 1998; Miyake et al. 1999). *ABCG2* is often amplified and overexpressed in human cancer cells and is capable of mediating drug resistance even in the absence of

multidrug resistance proteins MDR1 and MRP1 (Litman et al. 2000; Robey et al. 2001). Screening 29.1kb at the *ABCG2* gene locus identified a total of 43 SNPs, 2 of them in coding regions, 40 in introns, and 1 in the 3' flanking region. The distribution of SNPs at this locus was 1 per 676 bp on average. The frequency of each type of substitution was 41.9% for A/G, 30.2% for C/T, 7.0% for A/C, 14.0% for G/T, and 7.0% for T/A. We also found four variations of other types at the *ABCG2* locus.

*ABCG4* locus. The *ABCG4* gene on chromosome 11q23.3 consists of 14 exons spanning 12.6kb of genomic DNA; the 3.5-kb transcript encodes a 646-amino-acid peptide (Engel et al. 2001). This half-size ABC transporter possesses a nucleotide-binding fold followed by a cluster of six transmembrane-spanning domains, and is thought to function in cholesterol transport. Screening an approximately 8.2-kb region at the *ABCG4* gene locus identified a total of 11 SNPs, 1 in a coding region, 9 in introns, and 1 in the 3' untranslated region. The distribution of SNPs at this locus was 1 per 748 bp on average. The frequency of each type of substitution was 54.6 for A/G, 27.2 for C/T, 9.1% for A/C, and 9.1% for C/G.

ABCG5 and ABCG8 loci. The ABCG5 and ABCG8 genes encode proteins of 651 and 673 amino acids, respectively, which share 28% identity. Both proteins contain an Nterminal ATP-binding motif (Walker A and B motifs), an ABC transporter signature motif, and six transmembrane segments in the terminus. Several mutations in the ABCG8 gene and one mutation in the ABCG5 gene have been found in patients with sitosterolemia (Berge et al. 2000). The two genes are located in close proximity, in a headto-head configuration on chromosome 2p21. Screening an approximately 15.7-kb region at the ABCG5 gene locus identified a total of 26 SNPs, 24 of them intronic, 1 in the 3' untranslated region, and 1 in the 3' flanking region. The distribution of SNPs at this locus was 1 per 603 bp on average. The frequency of each type of substitution at the ABCG5 locus was 30.8% for A/G, 38.5% for C/T, 7.7% for A/C, 7.7% for C/G, 11.5% for G/T, and 3.8% for T/A. We also found five variations of other types at this locus.

Screening an approximately 31.2-kb region around the *ABCG8* gene identified a total of 25 SNPs: 1 in the 5' untranslated region, 3 in coding regions, and 21 in introns. The distribution of SNPs at this locus was 1 per 1249bp on average, and their frequencies were 20.0% for A/G, 44.0% for C/T, 20.0% for A/C, 4.0% for C/G, 8.0% for G/T, and 4.0% for T/A. We also found seven variations of other types at the *ABCG8* locus.

Altogether we have collected a total of 712 variations, including 605 SNPs and 107 variations of other types, among 13 genes encoding ABC transporters in a 96-chromosome sample from the Japanese population. The SNP collection reported here provides genetic data that should be helpful for personalized medical services and also for identifying alleles involved in drug efficacy, side effects, and/or complex genetic diseases.

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