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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 ATP-binding cassette transporters (*ABCA4*, *ABCA7*, *ABCA8*, *ABCD1*, *ABCD3*, *ABCD4*, *ABCE1*, *ABCF1*, *ABCG1*, *ABCG2*, *ABCG4*, *ABCG5*, and *ABCG8*). Sequencing a total of 416 kb 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 maps · ATP-binding cassette transporter genes · Japanese population

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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).

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 high-resolution 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 2 kb upstream of the first exon and downstream from the last exon, but excluding repetitive sequences. Each PCR was carried out using 20 ng 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 416 kb 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 84.5 kb 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-amino-acid 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

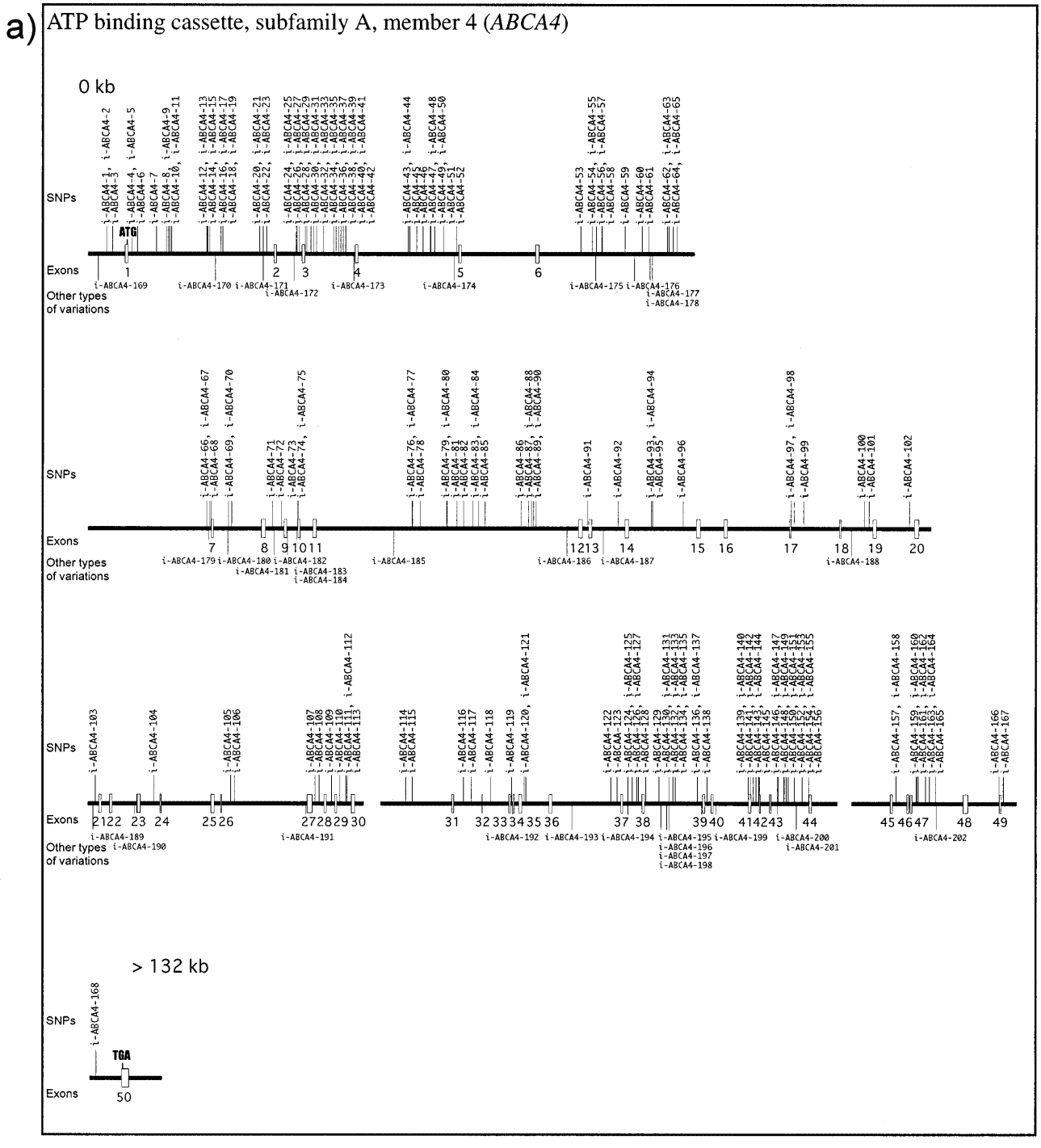
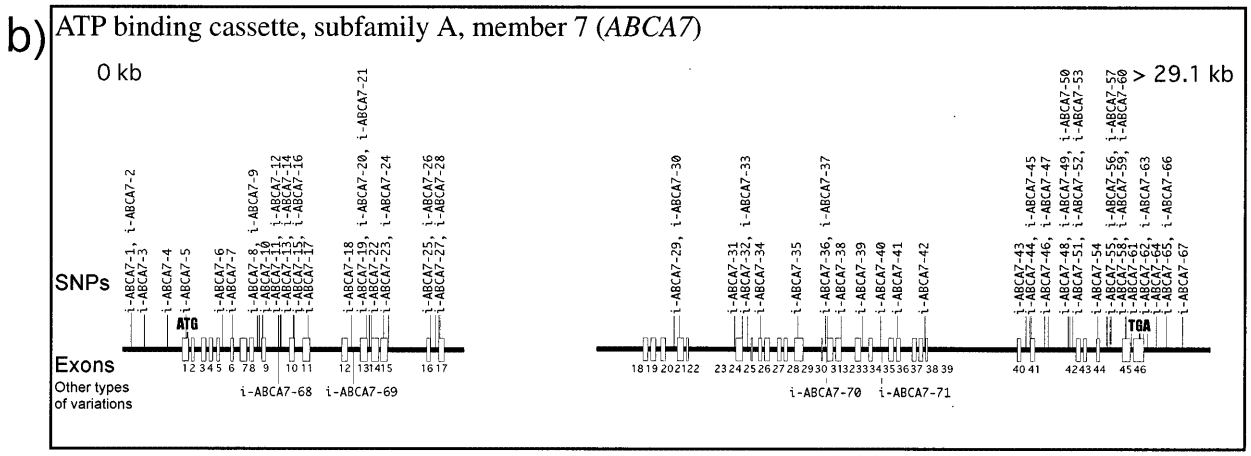


Fig. 1a–l. Genomic organizations and locations of single-nucleotide polymorphisms (SNPs) in 13 regions containing ATP-binding cassette transporter genes. Exons and introns are represented by *rectangles* and *horizontal lines*, respectively. SNPs are indicated *above* the genes (designations correspond 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 316 bp 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



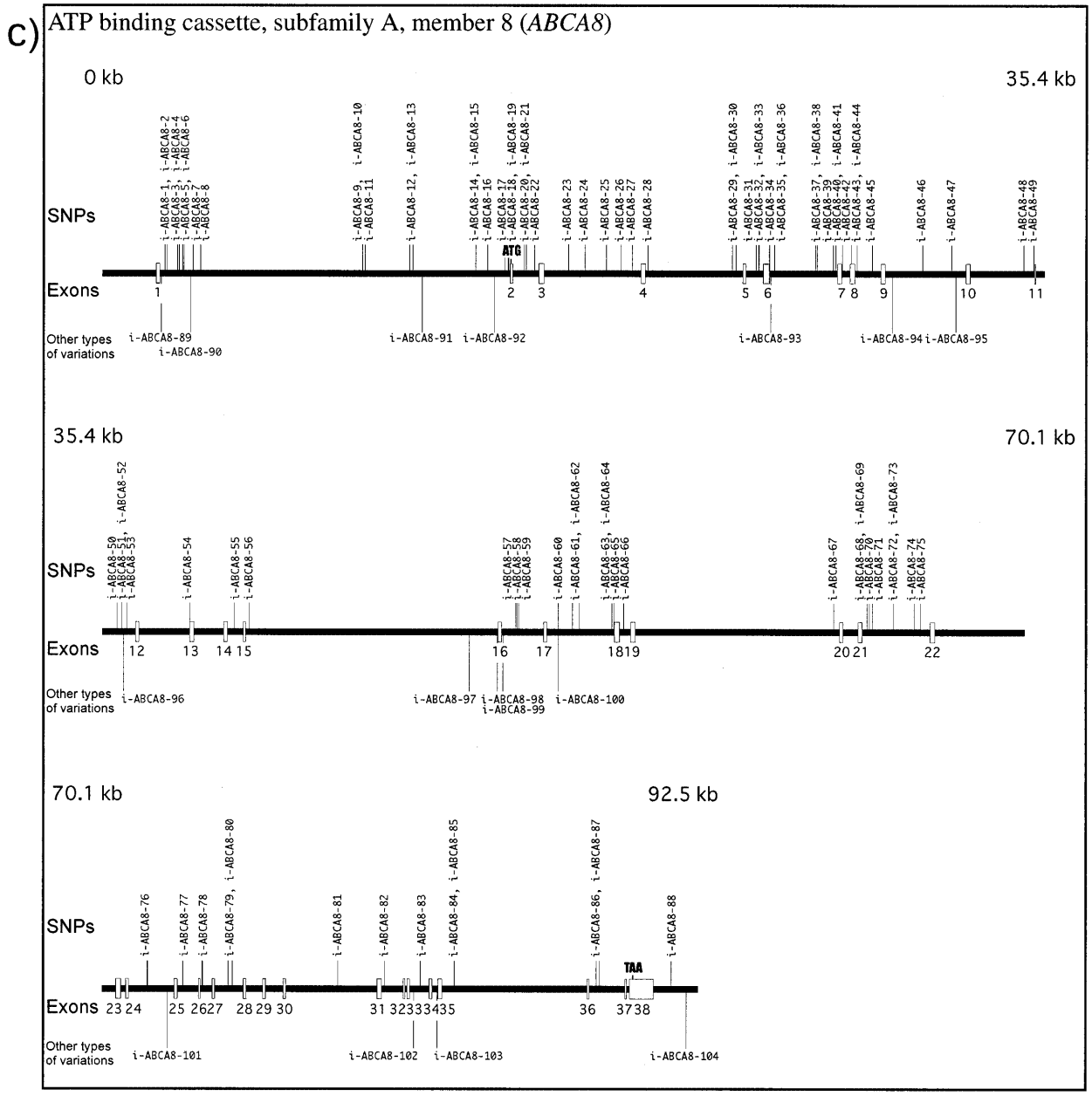
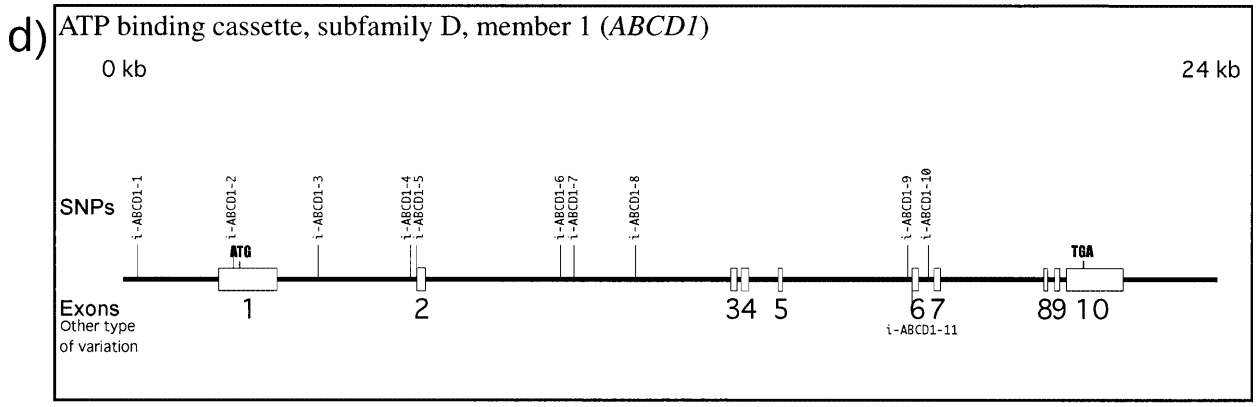
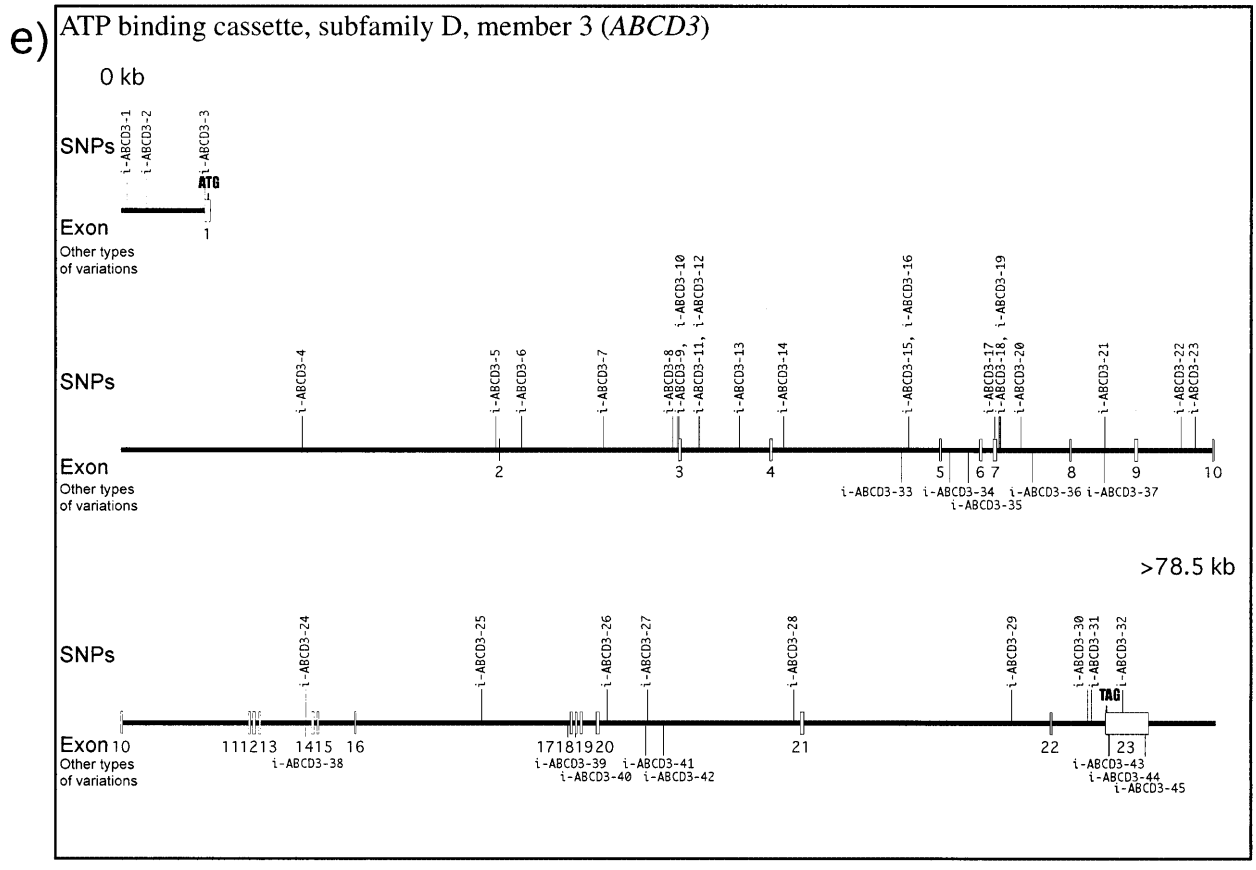


Fig. 1a–l. Continued





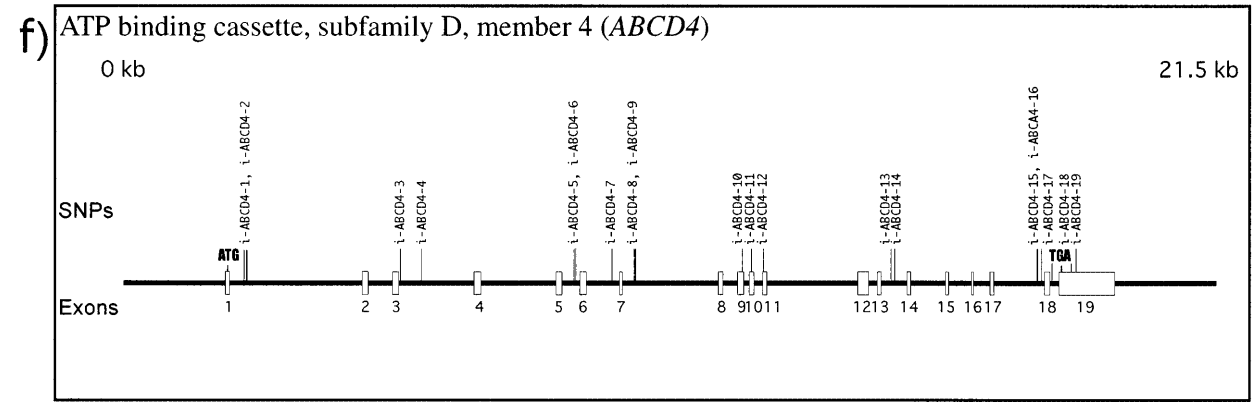


Fig. 1a–l. Continued

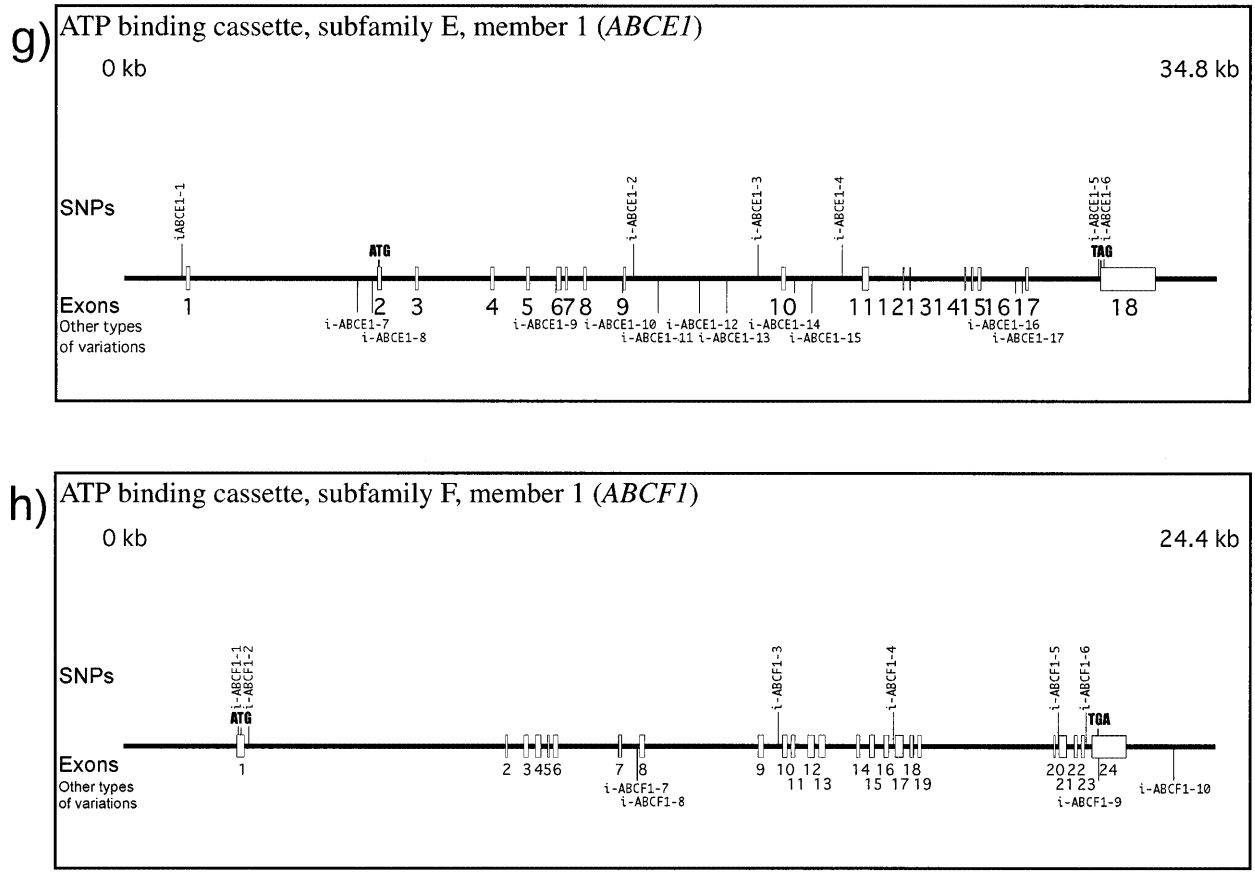


Fig. 1a–l. Continued

as a lipid transporter, but that has not been proven. Screening a region of approximately 57.7 kb 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 656 bp 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.

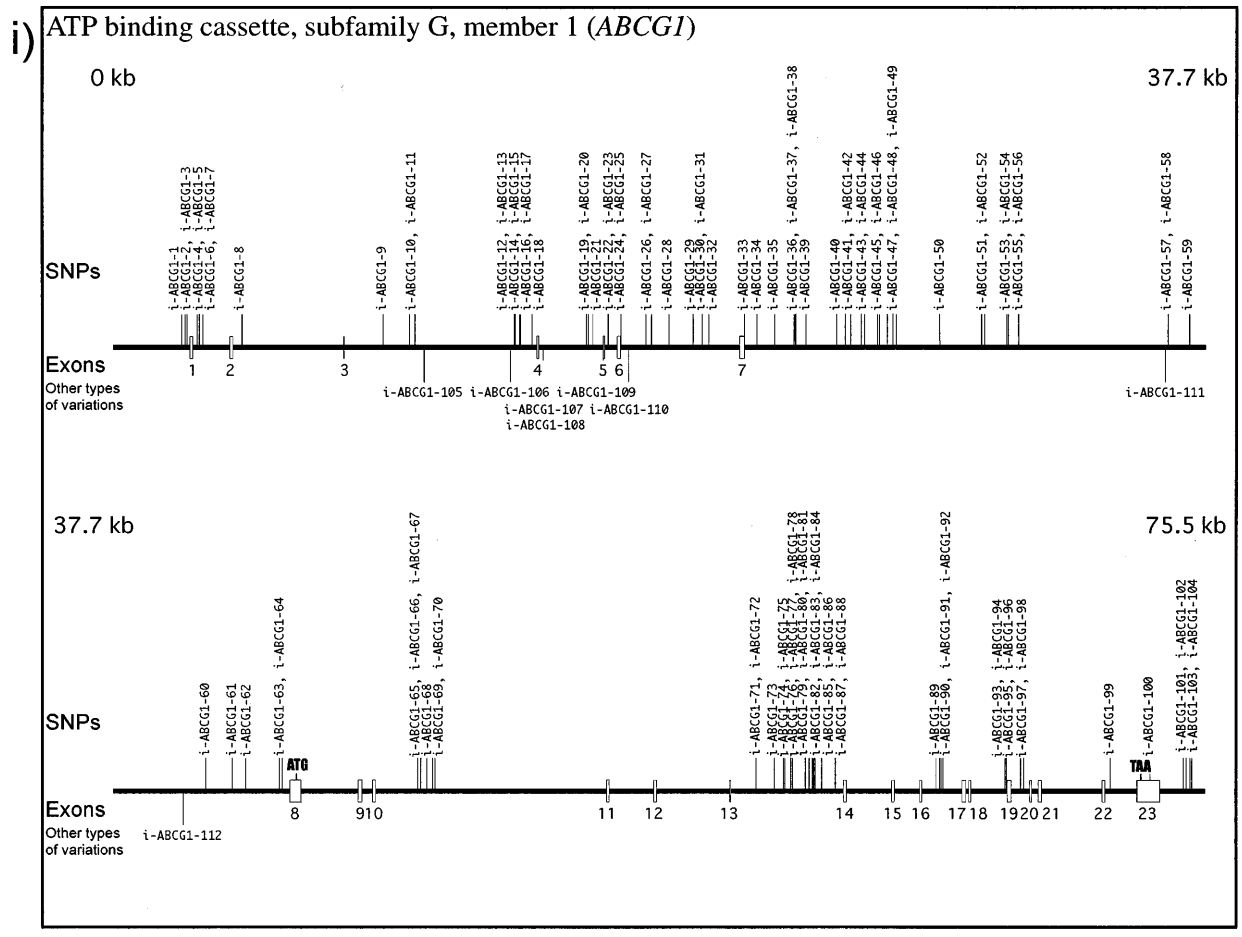
ABCD genes

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 37.1 kb 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 13.4 kb around the *ABCD4* gene detected a



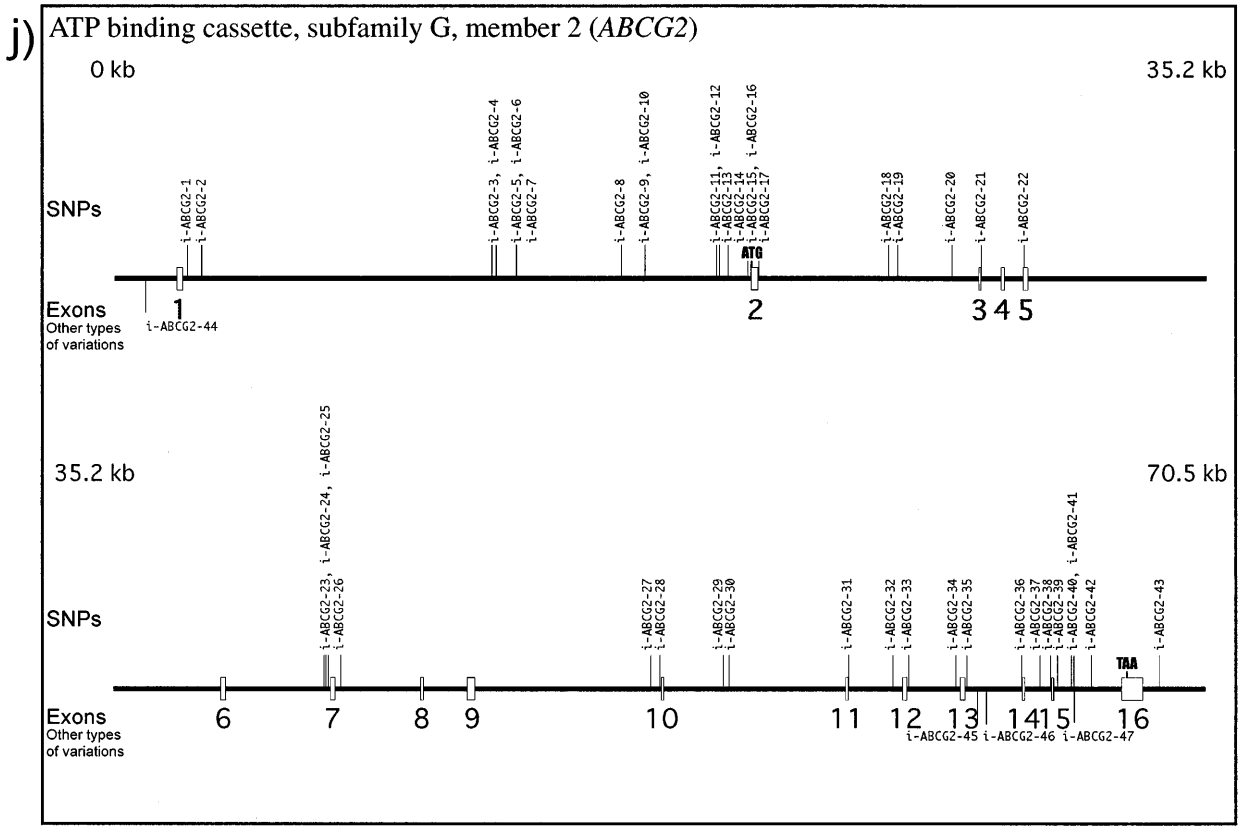


Fig. 1a–l. Continued

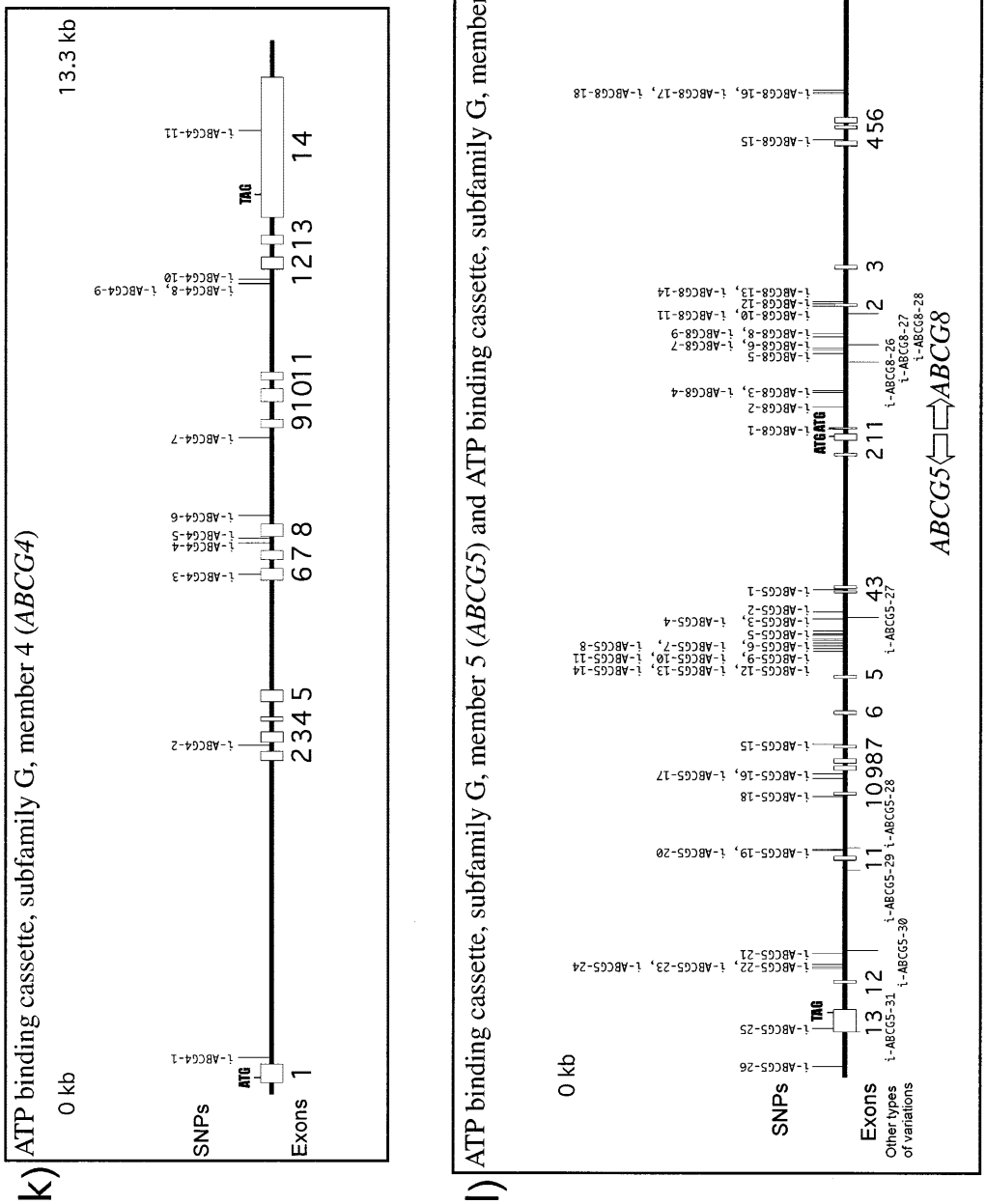


Fig. 1a–l. Continued

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

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
ATP binding cassette, subfamily A, member 4 (*ABCA4*)									
i-ABCA4-1	5' Flanking region		-1005	tgccatcataagcagaaact	A/C	tctctctcttcttggaagct			
i-ABCA4-2	5' Flanking region		-819	gtctagagtctttcaaagag	A/T	acacattctgagatttgagg			
i-ABCA4-3	5' Flanking region		-680	agcaccaccccattgcaggg	C/A	tggaatgacagtaatgggcc			
i-ABCA4-4	Intron 1		208	tgcccttcccaggaagatgt	G/A	tttctctgtcctcagccaca			
i-ABCA4-5	Intron 1		234	ctgtcctcagccacatgaaa	A/G	tcttttgcctaccgtgcctg			
i-ABCA4-6	Intron 1		510	agctcacgatcaagtcacag	T/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	atgccgctctggggagggga	A/C	gctgagcatgattttggaag			
i-ABCA4-14	Intron 1		4309	ctgagcatgattttggaagc	C/T	ggcagaagaggctattgtga			
i-ABCA4-15	Intron 1		4416	tgcagcaaccgcccccgccc	C/T	ccgccaaaaacaaacacact			
i-ABCA4-16	Intron 1		4996	tttacccctggaacaggcag	G/A	ccaagctggcT/Cggtcccctc			
i-ABCA4-17	Intron 1		5007	aacaggcagG/Accaagctggc	T/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	A/G	tcaagagtccttaagctcca			
i-ABCA4-21	Intron 1		7290	gatttttgttggctttgcaa	T/A	ggatcacagtcatttattca			
i-ABCA4-22	Intron 1		7483	tctgagcctctttccttaac	T/C	gcagagtgagtggC/Ttacaga		+	
i-ABCA4-23	Intron 1		7497	cttaacT/Cgcagagtgagtgg	C/T	tacagagaaatctttactac			
i-ABCA4-24	Intron 2		1067	tcaagcagcagcagcaactg	C/A	gtggagtcttcttgaactaa			
i-ABCA4-25	Intron 2		1106	aacactcctatgcccctctc	G/A	gcacaaaatgacG/Atgtcccc			dbSNP ID:rs1889404
i-ABCA4-26	Intron 2		1119	ccctctcG/Agcacaaaatgac	G/A	tgtccccccttgcttcccct			dbSNP ID:rs1889405
i-ABCA4-27	Intron 2		1243	cacccagcacagggactggc	A/T	cacatgagatgctcctgctt		+	
i-ABCA4-28	Intron 3		26	tgttgagatccctaccatgc	A/G	ggggaggaagttgcacaccc			
i-ABCA4-29	Intron 3		101	agcatggagcactgagtgtt	C/T	ttgtggctttgctgagcccc			
i-ABCA4-30	Intron 3		330	tgcttgggtggagtgaatca	T/C	tgtaggagaaaaactcagtt			
i-ABCA4-31	Intron 3		470	tgaagtcaggtttacaaagt	C/G	aagtttacttcttgggagaa			
i-ABCA4-32	Intron 3		634	tgaaaaccaatgacccctct	T/C	ccaagaaaaatggccacata			
i-ABCA4-33	Intron 3		1016	ccttgggggagctcagtatg	A/G	ttcttccaggagaagcctgc			
i-ABCA4-34	Intron 3		1554	gaaagttgggtttcatgttt	T/C	gcactcacattatgagtgaa			
i-ABCA4-35	Intron 3		1686	ctagacattctcacagagcc	A/G	agggcagcaaggcggggctc			
i-ABCA4-36	Intron 3		1823	ttcacctctctccatggacc	A/G	gtctcccctgctcctcaatg			
i-ABCA4-37	Intron 3		1938	caaattcctgggaacaaatc	G/A	ggttgacccagcT/Gttattct			
i-ABCA4-38	Intron 3		1951	acaaatcG/Aggttgacccagc	T/G	ttattctccctgtcccatca			
i-ABCA4-39	Intron 3		2063	ggctgtcagagcctacctgc	G/T	tgaatgggtggaaggG/Acagg			
i-ABCA4-40	Intron 3		2079	ctgcG/Ttgaatgggtggaagg	G/A	caggtctcagagaattgggt			
i-ABCA4-41	Intron 3		2186	agacacacagagcatgggac	C/T	gagaggcgagcagaccctgc			
i-ABCA4-42	Intron 3		2214	gagcagaccctgccaaaact	G/A	ggagactgaatagatcgctc			
i-ABCA4-43	Intron 4		2717	cgtgcttctgcacagccacc	T/C	gggaaggtatgccgatggtt			dbSNP ID:rs1211213
i-ABCA4-44	Intron 4		2802	attctcagcagggaggatta	A/G	tggtaaaagcccaggaatgg			dbSNP ID:rs1209515
i-ABCA4-45	Intron 4		3182	cccccagagccacagcagcc	C/G	tgtctcctgggtggtcttgt			
i-ABCA4-46	Intron 4		3515	agtataataaaagcaggagc	C/T	atagcccccaactctcaaga			
i-ABCA4-47	Intron 4		3907	aggggagtgacagtgggcac	C/A	actctcagggaacccA/Gttac			dbSNP ID:rs570878
i-ABCA4-48	Intron 4		3923	gcacC/Aactctcagggaaccc	A/G	ttactgtgagagaagccact			dbSNP ID:rs570926
i-ABCA4-49	Intron 4		3952	agagaagccactgtgccact	G/C	tgtggtcgaacttcaagacc			
i-ABCA4-50	Intron 4		4125	ggctgtccagcacacagggg	C/A	aggcctcttggccactgggg			dbSNP ID:rs481931
i-ABCA4-51	Intron 4		4637	aatcacttgccccaaggtca	C/T	cttaactgttaggtgttctt		+	

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA4-52	Intron 4		5319	acctctaggggctcccagag	A/G	ccccaagaacagaaccttcc			
i-ABCA4-53	Intron 6		2266	caccettgcagacctcagac	G/A	ggtcctgggggcttgctttc			
i-ABCA4-54	Intron 6		2857	ccagaggagaaagctctgcc	G/A	tagT/Ccggcctcagttaacca			
i-ABCA4-55	Intron 6		2861	aggagaaagctctgccG/Atag	T/C	cggcctcagttaaccacgga			
i-ABCA4-56	Intron 6		3078	gcaggcattaaaatgggact	T/G	tgcctttattgctcctgggc			
i-ABCA4-57	Intron 6		3375	ttaaatgccaaatgagttct	C/G	attaacaaagaaagagggaa			
i-ABCA4-58	Intron 6		3412	ggaaaatctcagtaaaccac	C/T	gtgacggcatctacccactt			
i-ABCA4-59	Intron 6		4635	ctttcgggtggatattgcta	C/T	gtcaagtgtctgggaaagcc			
i-ABCA4-60	Intron 6		5576	ccactaatatgcattcttta	G/C	taagcggtctcaatatacac			dbSNP ID:rs538880
i-ABCA4-61	Intron 6		5925	aaaaagcattttgctcttat	A/G	aaagcacagcctcttttgag			dbSNP ID:rs952499
i-ABCA4-62	Intron 6		6916	cccagacaacccaagcagag	A/G	cctcttagggccggaatcat			dbSNP ID:rs483904
i-ABCA4-63	Intron 6		6993	agcacaggatcaaggcctaa	A/G	ggccccttagactgacctca			dbSNP ID:rs554931
i-ABCA4-64	Intron 6		7242	ttgccattttgatctgtgac	T/C	ttttttccagaaatagttt			dbSNP ID:rs1191237
i-ABCA4-65	Intron 6		7454	atggagggctccctcgggac	T/C	aggcagtattcagagatgta			dbSNP ID:rs1191238
i-ABCA4-66	Intron 6		-264	aaacagcaattagaatcact	T/C	tgaaatagtgatagtattta			
i-ABCA4-67	Intron 6		-86	aggaggggggagttttcaa	A/G	catataggagatcagactgt			dbSNP ID:rs574741
i-ABCA4-68	Intron 6		-32	tatacctacaaacatatata	T/C	atttaaaaaattgttttact			dbSNP ID:rs526016
i-ABCA4-69	Intron 7		828	gatgtgggaaagttagagaa	G/C	agcccattgtactaatgctc			
i-ABCA4-70	Intron 7		1019	aggcttcttgactgtctaga	T/C	agcaagtctaatcatttgtg			
i-ABCA4-71	Intron 8		374	gtaaacacggctgtgggatg	C/A	ttttacaaacacaatatcgt			
i-ABCA4-72	Intron 8		874	tgatgagcttgttattggtg	G/A	ggtacagcctattaatttag			
i-ABCA4-73	Intron 9		605	tcgtgtctctgtcttgatct	C/T	tgtctggttttaggccaact			
i-ABCA4-74	Coding region	exon 10	1268	aacttttgaagaactggaac	G/A	C/Tgttaggaagttggtcaaag	Arg/His423His		
i-ABCA4-75	Coding region	exon 10	1269	acttttgaagaactggaacG/A	C/T	gttaggaagttggtcaaagc	Arg/His423Arg/His		
i-ABCA4-76	Intron 11		5236	ggcctggcacagatgaaata	C/T	tattcagagttcacagtgta			dbSNP ID:rs1761375
i-ABCA4-77	Intron 11		5270	cagtgtattttcatttcata	A/G	tatatttgattttcaggtct			dbSNP ID:rs1932015
i-ABCA4-78	Intron 11		5687	atcatgtaatgtactttaga	C/G	tcagatatataaatatttgt			
i-ABCA4-79	Intron 11		7136	gacttcccaacttaccttag	T/C	ggagctgtagtcacatagaa			
i-ABCA4-80	Intron 11		7180	acgctcataaatgcttctct	G/A	ggctgtaaaggttgaatttt			
i-ABCA4-81	Intron 11		7701	gttagacgcaggcattacct	C/T	gtggctttgccccagtgtga			
i-ABCA4-82	Intron 11		8073	gggatgtttgcccacatcca	T/C	tggcatttctcaaaaggaac			
i-ABCA4-83	Intron 11		8586	cagctgcctgcgctggagag	G/A	gctcaaacctcttccgccag			
i-ABCA4-84	Intron 11		8893	agcaaagatgccctttgact	C/T	cttttcccactagtggtgct			dbSNP ID:rs549114
i-ABCA4-85	Intron 11		9257	gaatgaggtcacttgctgca	T/A	ggcaggtggcttccccatga			dbSNP ID:rs581244
i-ABCA4-86	Intron 11		11234	cccaaataattttgtttttc	G/A	ttttaggaattaaatttcag			
i-ABCA4-87	Intron 11		11641	aagaaacaaacatttattga	C/G	aacttttggtgtgtgacctg			
i-ABCA4-88	Intron 11		11808	tggtatttcttaaagaaata	C/T	caattccatttccttttaac			
i-ABCA4-89	Intron 11		11923	aagatcattattaatatctc	A/G	tcagcgtggtgtcacttaag			
i-ABCA4-90	Intron 11		12055	tgagaacattacatgggacc	T/C	gcccccagggcatggaggct			dbSNP ID:rs1191232
i-ABCA4-91	Intron 12		305	tcaccctgtggtcgggaggt	G/A	tgagtgagctatccaagccc			
i-ABCA4-92	Intron 13		1461	ttgggtttcagtgtcagcat	G/A	tagctgtctactcagatccc			
i-ABCA4-93	Intron 14		1237	aagggcaccaaagttctaag	A/G	gatgaggggaggagctgagc			dbSNP ID:rs549848
i-ABCA4-94	Intron 14		1268	ggagctgagccccttgtcct	T/C	atctaggtttcccttgttct			
i-ABCA4-95	Intron 14		1309	ttcccatccctcagtctgct	T/C	cttttcccagtaccaacatg			dbSNP ID:rs550060
i-ABCA4-96	Intron 14		2979	tcacctgtgtgggtagcaaa	C/T	ctcagaaaatcaagtataga			dbSNP ID:rs497511
i-ABCA4-97	Intron 17		23	gagtcctttaaaacacaaat	C/G	ttaatgtttgaaatcaactc			
i-ABCA4-98	Intron 17		204	tgctgggccctgtgtgatca	T/G	gaatggctgatcatggatga			dbSNP ID:rs1191231
i-ABCA4-99	Intron 17		715	gggactcccctagagctgaa	G/A	tactctcccatctgtttgtt			
i-ABCA4-100	Intron 18		1282	ggaagatgaagaacctaagc	C/T	gcttccagaaattcatgagg			
i-ABCA4-101	Intron 18		1531	gtctacccctaggaccatt	G/A	taagagtacattgaggtaat		+	dbSNP ID:rs544830
i-ABCA4-102	Intron 19		1802	actgctcacccaggaggcaa	C/A	gcctcgagtcatgcaccgaa			dbSNP ID:rs2039446
i-ABCA4-103	Intron 20		-195	acagattattccattgtatg	C/A	atgaactatgtaagccatcc			
i-ABCA4-104	Intron 23		755	ctggctgccgctggggtttc	C/T	tatgtccatccacggggagg			
i-ABCA4-105	Intron 26		497	ctgagttaggtctagatggg	G/A	acactttggatgaatgagga			dbSNP ID:rs1320502

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA4-106	Intron 26		702	tatcaaatacaactcagacg	T/G	cagtctcctggccctttga			
i-ABCA4-107	Intron 27		156	cctgctttccaaaccttat	C/T	ttgattcttggtaacatgaa			
i-ABCA4-108	Intron 27		385	tttaaagaacagtgagtcac	G/A	tgacttgctctttgaaatgc			
i-ABCA4-109	Intron 28		299	gacatgccatcagaccactg	C/T	gagtgttcaggcagcctacc			
i-ABCA4-110	Intron 29		168	ctccttccacacttgtgtgc	A/G	gggacattcactacctccta			
i-ABCA4-111	Intron 29		497	gctgtcaataaggaccaaaa	C/T	agactaatttcaaattcctc			
i-ABCA4-112	Intron 29		567	agctgctaggaataaaaagg	G/A	agacaaaacG/Aatccacaagc			
i-ABCA4-113	Intron 29		577	aataaaaaggG/Aagacaaaac	G/A	atccacaagctagagatggt			
i-ABCA4-114	Intron 30		-2494	aatcacagctcatctgctgc	A/G	tcatagggatcccaaaagaa			
i-ABCA4-115	Intron 30		-2169	aatgtaacagccaaagtcct	A/G	gaaaaaggcaagccagttcc			
i-ABCA4-116	Intron 31		535	ctaactgtgaattatcatct	T/G	tgatcactgccctttgagat			
i-ABCA4-117	Intron 31		957	gagttctcagcagcaaatct	C/A	cagtatgaaattttggattt			dbSNP ID:rs1889407
i-ABCA4-118	Intron 32		445	tccagaggtttagaacctca	C/T	caagtgggactctaggagcc			dbSNP ID:rs1932014
i-ABCA4-119	Intron 33		48	aggatttttgacttgcttaa	C/T	taccatgaatgagaaactct			dbSNP ID:rs472908
i-ABCA4-120	Intron 35		129	tgtttagtcaggcacatatg	A/C	acatccgactttcaaataag			dbSNP ID:rs933073
i-ABCA4-121	Intron 35		209	tctccccaacatttatgtgg	C/A	aagtaagtttacatttggtt			
i-ABCA4-122	Intron 36		3209	ttgaggcctccacaccccac	G/A	gcaggttgccccctgaggaa			dbSNP ID:rs915199
i-ABCA4-123	Intron 36		3542	cttggcagggaggtagggca	T/C	ggggtggggtaggaggacta			dbSNP ID:rs915201
i-ABCA4-124	Intron 37		304	ctgggggcagccattcccca	A/G	cccctcacccagctctgact			dbSNP ID:rs914957
i-ABCA4-125	Intron 37		525	taaatttgaatgagtaattc	A/G	tccatctcggcctcagtttc		+	
i-ABCA4-126	Intron 37		766	tgttgcaggctggagaaccc	T/G	cctatgaattgtacagggct			
i-ABCA4-127	Intron 37		856	aaaaccccatgaagtggtca	A/G	ggcaggcatcattatctcca		+	
i-ABCA4-128	Intron 38		62	tagtagagtatgtgttggtc	G/A	agcagagccaggggcaagca			
i-ABCA4-129	Intron 38		761	tccttgggcaagttaatctt	G/A	atgaagagactgggtgttct			
i-ABCA4-130	Intron 38		1315	cagagtcagactctggaaag	G/T	C/Agggggataagaacacagc			
i-ABCA4-131	Intron 38		1316	agagtcagactctggaaagG/T	C/A	gggggataagaacacagcc			
i-ABCA4-132	Intron 38		1526	ccaacatttgctaagcaccc	G/A	ccttcaaaaacctggtattt			dbSNP ID:rs567370
i-ABCA4-133	Intron 38		1561	gtattttcatgtaaattatc	C/A	G/Aatacacagctgctatggaa			
i-ABCA4-134	Intron 38		1562	tattttcatgtaaattatcC/A	G/A	atacacagctgctatggaaa			
i-ABCA4-135	Intron 38		1674	ccagctgaacaccacgtgcc	G/A	ggtgtgtgctgatataaaca		+	dbSNP ID:rs486479
i-ABCA4-136	Intron 38		2867	tgcctggctagacaaagggg	A/C	agctccC/Tgcccactagaaac			dbSNP ID:rs565155
i-ABCA4-137	Intron 38		2874	ctagacaaaggggA/Cagctcc	C/T	gcccactagaaacttgcagg			
i-ABCA4-138	Intron 39		123	gaggggaccttgttgggctg	G/A	aggtgtcctgccagctggag			dbSNP ID:rs2065711
i-ABCA4-139	Intron 40		1904	gacactgtacagccagccca	A/C	tcctgacccctttttcttcat			
i-ABCA4-140	Coding region	exon 41	5814	ggaaataaaactgacatctt	A/G	aggctacatgaactaaccaa	Leu1938Leu		
i-ABCA4-141	Intron 41		122	atttggttcccagttttatg	T/G	agggtcatcatccctgtgtt			
i-ABCA4-142	Intron 41		287	tctgcagagcatgggtcagc	C/T	tcgagatgtctcagtactca			dbSNP ID:rs1191234
i-ABCA4-143	Intron 41		411	cctcttcccctccttgctct	C/A	accctgtctcagttctcagt			
i-ABCA4-144	Intron 41		443	gttctcagtccggtttcttc	G/A	tatcttgcagatttatccA/Gg			
i-ABCA4-145	Coding region	exon 42	5844	cG/Atatcttgcagatttatcc	A/G	ggcacctccagcccagcagt	Pro1948Pro		Maugeri et al. (1999)
i-ABCA4-146	Intron 43		328	tttgtagcctattcctataa	A/G	aatgcaccattgcttcC/Gcat			
i-ABCA4-147	Intron 43		345	taaA/Gaatgcaccattgcttc	C/G	cattacctccctccacacat			
i-ABCA4-148	Intron 43		370	acctccctccacacattttt	A/G	caaaaC/Tgtttcagggagttt			
i-ABCA4-149	Intron 43		376	ctccacacatttttA/Gcaaaa	C/T	gtttcagggagtttactgag			
i-ABCA4-150	Intron 43		670	ttaaacagactggtccccta	T/C	gggcaggacagagaggatga			
i-ABCA4-151	Intron 43		701	gagaggatgagctctcactc	A/G	tctgcctctttcctggctgc			dbSNP ID:rs557026
i-ABCA4-152	Intron 43		822	gttaggtgctgctgacatct	G/A	tccagcatctgcttgactgg			
i-ABCA4-153	Intron 43		915	ggcaggacgagtcctgagca	C/T	gcttcactggctcagacagg			
i-ABCA4-154	Intron 43		1242	actgagctggacgctagaaa	G/T	aaactataggcttaagacac			
i-ABCA4-155	Intron 43		1671	tagagaagtttacttccatc	G/A	ggacacatgcatcttttcta			
i-ABCA4-156	Intron 43		2036	ttgaaggatactcagtaatt	G/A	ctttttttcttgcagtattt			
i-ABCA4-157	Intron 45		176	gtgtttggttcacacagctc	C/T	ggagaaaaacaagtcaC/Tggc			
i-ABCA4-158	Intron 45		193	ctcC/Tggagaaaaacaagtca	C/T	ggcacagccttgacttggga			
i-ABCA4-159	Intron 47		238	cccaagtctctggatggggc	A/G	tctgatcaggatgcatgcag			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA4-160	Intron 47		269	atgcatgcagagcctggctg	G/A	gatgagggagggctgctacc			
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	C/T	caccttatctatgctgaggt		+	
i-ABCA4-164	Intron 47		931	ggacactgcatagatatcta	T/C	agaaatagcagcatgtcagg			
i-ABCA4-165	Intron 47		1260	acactctctggtggaccatc	A/C	ctcatccaagagagggtaac			
i-ABCA4-166	Intron 48		1663	tctcgctcttctcttacctc	T/C	aggtgtttgtaaattttgct			Maugeri et al. (1999)
i-ABCA4-167	Intron 49		127	agagagccccacccacacca	C/T	ggtccctaccaagtccccac			
i-ABCA4-168	Intron 49		-1545	gcagttaattccaaactttt	C/A	tcccttattggatgagatca			dbSNP ID:rs1932016
i-ABCA4-169	5' Flanking region		(-1441)–(-1400)	gtaaatctcagttgaatcag	(TCA)14~16	atttttcagtctggttcctg			
i-ABCA4-170	Intron 1		4712–4720	gaggggcggggactataggc	(A)8~10	cagcctaattcaaggatgag			
i-ABCA4-171	Intron 1		7295–7304	ttgttggctttgcaaT/Aggat	CACAGTCAT/del	ttattcactcattcattcac			
i-ABCA4-172	Intron 2		951–952	cctgtccatcagactcttct	TT/del	acctctcccgaggagccca			
i-ABCA4-173	Intron 3		2642–2653	cctgggtgacagagcgagat	(A)10~12	tagcatgagatattattact		+	
i-ABCA4-174	Intron 4		5202	cacaaagcatctgacacccc	C/del	atccagccctggctaacttt			
i-ABCA4-175	Intron 6		3029–3044	cactaaaaacaaaaatttac	(A)16~18	cctgaaagaaattgcaggca		+	
i-ABCA4-176	Intron 6		5138–5139	ttcatgacagatcagatgtt	G/ins	cttttatggatttacaaaga			
i-ABCA4-177	Intron 6		5985	tttccttcttcaaacccccc	C/del	agactaggagaaggtctgtc			
i-ABCA4-178	Intron 6		6094	gggacggacagaaaaagacc	T/del	agtttctgttgagccaaaga			
i-ABCA4-179	Intron 6		-161	tatttttttcaattaaataaa	A/del	gagtttttgtttctaaaag			
i-ABCA4-180	Intron 7		809–810	gggccgagtatgcacactga	TG/ins	tgtgggaaagttagagaaG/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	A/del	ccaaaagttctctctcactt			
i-ABCA4-184	Intron 10		11	catgatcagagtaaggggggg	G/del	ttggaggatggggaggggag			
i-ABCA4-185	Intron 11		4242	ggagaggaaatgatgttagt	G/del	cctcctgtaaataggcccag			
i-ABCA4-186	Intron 11		13743–13753	tgctcttttgtgggtaatgg	(T)9~11	cctcttccaggagaagaaaa			
i-ABCA4-187	Intron 13		636–637	cggggtggagggttgggagg	G/ins	ctcatttgtcattatagatg			
i-ABCA4-188	Intron 18		569–570	tgctgccctcatcttctctc	TT/del	aaactagttctgtatttctc			
i-ABCA4-189	Intron 20		(-304)–(-297)	tataacctgacttttttttc	(A)7~9	ggattgctttttaaacata			
i-ABCA4-190	Intron 22		1236–1246	gctgaattagttcccttggg	(T)9~11	agttaactcctgatttttgc			
i-ABCA4-191	Intron 26		4626–4635	gataatcaatgctgtaaggg	(A)9~10	tggcattagagatccagacc			
i-ABCA4-192	Intron 33		115–116	taaaaccgtcttgtttgttt	GT/del	ttacatggtttttagggccc			
i-ABCA4-193	Intron 36		1078	taagcagctatcacttaaca	A/del	tacaaaaccagagattatca			
i-ABCA4-194	Intron 37		290–291	ccttgaccaaagcctggggg	T/ins	cagccattccccaA/Gcccctc			
i-ABCA4-195	Intron 38		896	ataaaaagaggggaaaaaa	A/del	gaaggcagtcgctgcagggc			
i-ABCA4-196	Intron 38		1209–1210	gtggacccctgagactgact	CT/del	ttccagatcttgttagggtt			
i-ABCA4-197	Intron 38		1322	agactctggaaagG/TC/Aggggg	G/del	ataagaacacagccccagca			
i-ABCA4-198	Intron 38		3107	gggccccacctgctgaagag	A/del	gggggggtgggtttgcccc			
i-ABCA4-199	Intron 40		152	ttttctccaataatacaagt	A/del	gaggatcgggttaaaatagg			
i-ABCA4-200	Intron 43		330	tgtagcctattcctataaA/Ga	A/del	tgcaccattgcttcC/Gcatta			
i-ABCA4-201	intron 43		1354	tttaattggcccagccatgc	C/del	tttggtggcttttgtcattg			
i-ABCA4-202	Intron 47		1305–1308	catcctgctgaaggagaaag	AAAG/del	caccaatggcccaagcccta			
ATP binding cassette, subfamily A, member 7 (*ABCA7*)									
i-ABCA7-1	5' Flanking region		-1596	agaatgttggccccctcccc	C/T	tC/Tctgcatcctctgcagaag			
i-ABCA7-2	5' Flanking region		-1594	aatgttggcccctccccC/Tt	C/T	ctgcatcctctgcagaagcc			
i-ABCA7-3	5' Flanking region		-1180	ggccagtgagtgacgggcag	G/A	tcgcccaaatagcagcgtgc			
i-ABCA7-4	5' Flanking region		-460	agagctggggtcgtgcctcc	A/G	gctgggcaactgcctgtctc			
i-ABCA7-5	5' Untranslated region	exon 1	-9	ctctgtcccgtcccctgccc	A/G	gtctcaccatggccttctgg			
i-ABCA7-6	Intron 5		91	ccccgggccaaggacctccc	G/A	ttccaggcatccaggctgtc			
i-ABCA7-7	Coding region	exon 6	563	cagcttgttggaggccgctg	A/G	ggacctggcccaggaggtac	Glu188Gly		
i-ABCA7-8	Intron 8		103	gccggagggtcacggaaact	A/G	tttgaagaagtaggagttag			
i-ABCA7-9	Intron 8		166	tgcggaggatcagaggcaca	C/T	gcaggagcaaggcagagggg			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA7-10	Coding region	exon 9	955	accggaccttcgaggagctc	A/G	ccctgctgagggatgtccgg	Thr319Ala		
i-ABCA7-11	Intron 9		421	ttttttttttttttttttt	T/A	taagagatggagtctcactc		+	
i-ABCA7-12	Intron 9		463	gttgcccaggctggactgca	G/A	tggC/Tgagatcttggctcact		+	
i-ABCA7-13	Intron 9		467	cccaggctggactgcaG/Atgg	C/T	gagatcttggctcactgcaa		+	
i-ABCA7-14	Intron 9		488	gagatcttggctcactgcaa	C/T	ctccgcctcctggattcaag		+	
i-ABCA7-15	Coding region	exon 10	1184	cgcacacgctgatgtggggc	A/G	cctggtgggcacgctgggcc	His395Arg		
i-ABCA7-16	Intron 10		10	gagtgacggaggtgagggcc	T/C	gtccacctgcggggtctgtt			
i-ABCA7-17	Coding region	exon 11	1388	cctgggccccggccacgtgc	G/A	catcaaaatccgcatggaca	Arg463His		
i-ABCA7-18	Intron 12		115	caggctgcgaactttgcacc	T/G	ttacaccactccacgtgacc			
i-ABCA7-19	Coding region	exon 13	1824	cccttcctgctcagcgccgc	A/G	ctgctggttctggtgctcaa	Ala608Ala		
i-ABCA7-20	Intron 13		55	ggtgcgctggagggtgacag	A/G	caggggcggccccacgtggg			
i-ABCA7-21	Intron 13		78	ggggcggccccacgtgggtg	C/A	gcgcccccaggccaatccag			
i-ABCA7-22	Coding region	exon 14	1851	cgttgcctctcacagctggg	A/G	gacatcctcccctacagcca	Gly617Gly		
i-ABCA7-23	Coding region	exon 15	2153	cgagggcgcgcagtggcaca	A/C	cgtgggcacccggcctacgg	Asn718Thr		
i-ABCA7-24	Intron 15		34	ggcggggctccgggccgggt	C/G	gcacctgctttgcgggaggc			
i-ABCA7-25	Intron 16		8	ctggacccaaagggtgaggc	A/C	ctacgaggcttaatagctgg			
i-ABCA7-26	Intron 16		161	tcccgcagcttttataggcc	C/T	cggcccagcaggtcccggat			
i-ABCA7-27	Coding region	exon 17	2385	caccccatctctgcagtgct	G/A	gtagaagaggcaccgcccgg	Leu795Leu		
i-ABCA7-28	Coding region	exon 17	2421	cccggcctgagtcctggcgt	C/A	tccgttcgcagcctggagaa	Val807Val		
i-ABCA7-29	Intron 20		166	cgagacagtaagagttgggg	A/G	tagacagaggttcccctgga			
i-ABCA7-30	Coding region	exon 21	3027	ctgctgggagaccgtgtggc	C/T	gtggtggcaggtggccgctt	Ala1009Ala		
i-ABCA7-31	Intron 22		1386	gggtggggcgtgagccgggg	C/T	tccctgaagcaccccttttgt			
i-ABCA7-32	Coding region	exon 23	3417	gggatctccgacaccagcct	C/G	gaggaggtgtgaggcctggg	Leu1139Leu		
i-ABCA7-33	Intron 23		147	ggagctctggtggctcagat	G/A	tcccttgggaaggcctgggg			
i-ABCA7-34	Coding region	exon 25	3528	gctggcctagacgtaaccct	A/G	cggctcaagatgccgccaca	Leu1176Leu		
i-ABCA7-35	Coding region	exon 29	4046	cccagcctgccagtgtagcc	G/A	gcccggtgcccggcgcctgc	Arg1349Gln		
i-ABCA7-36	Intron 30		81	ccccctgggagctctcccgg	C/A	ccccccggccctcagctccc			
i-ABCA7-37	Coding region	exon 31	4239	ctgcctgcatggccccacag	A/G	tacggaggcttctcgctggg	Arg1413Arg		dbSNP ID:rs881768
i-ABCA7-38	Intron 32		1	caaggagcagctgtctgagg	G/C	tgcactgtgagtccctccac			
i-ABCA7-39	Intron 33		54	ccactgcttgccactgccct	G/A	tctggccccttgtaggcagg			
i-ABCA7-40	Intron 34		245	cagtactttgggaggccgag	G/A	caggaggactgcttgtggcc		+	
i-ABCA7-41	Coding region	exon 36	5057	ggtgagccggatcttgaaac	A/G	ggtcttccttatcttccccc	Gln1686Arg		
i-ABCA7-42	Intron 38		65	ggcccactcacctttctgaa	A/G	gacctgcactctcccaggta			
i-ABCA7-43	Intron 40		154	ctctacctcccacacgcgga	C/G	caggccctgagacacccctg			
i-ABCA7-44	Intron 40		277	ctgagcccccggcgccccca	T/C	ccccagcgtggcccgggaac			
i-ABCA7-45	Coding region	exon 41	5592	gtggcccgggaacccagtgc	T/C	gcgcacctcagcatgggata	Ala1864Ala		
i-ABCA7-46	Intron 41		286	ctccttgactctgccttctg	T/C	ggccctgcccacttgctcct			
i-ABCA7-47	Intron 41		389	tggccgttcccagtttgcag	C/T	cgtttcactgcctcttccat			
i-ABCA7-48	Intron 41		991	cacactatggccctgcccca	C/T	acC/TcatC/GccA/Ggctccaccca		+	
i-ABCA7-49	Intron 41		994	actatggccctgccccaC/Tac	C/T	catC/GccA/Ggctccacccacac		+	
i-ABCA7-50	Intron 41		998	tggccctgccccaC/TacC/Tcat	C/G	ccA/Ggctccacccacaccatg		+	
i-ABCA7-51	Intron 41		1001	ccctgccccaC/TacC/TcatC/Gcc	A/G	gctccacccacaccatggcc		+	
i-ABCA7-52	Intron 41		1051	actcatgctggctccaccca	C/T	accatggcccgccccatac		+	
i-ABCA7-53	Intron 41		1131	tgccctgccccatgcccatt	A/G	tgcccctgctccacactcaa		+	
i-ABCA7-54	Coding region	exon 44	5985	gaagcgctctgctcgcgcct	G/A	gccatcatggtgaatgggcg	Leu1995Leu		
i-ABCA7-55	Intron 44		201	ggcgcaggaccaggaggcgt	G/C	agccgggggctctgggtgga			
i-ABCA7-56	Intron 44		233	ctgggtggatttagaagaca	C/T	aatcaggtgtgcgttggagt			
i-ABCA7-57	Intron 44		313	agttaggggagggcctggtt	A/G	gtgggcggggccataggaaa			
i-ABCA7-58	Intron 44		337	ggcggggccataggaaagtg	G/C	ggcgggggtatttattgtgt			dbSNP ID:rs1609436
i-ABCA7-59	Coding region	exon 45	6133	tggcggccgagttccctggg	G/T	cggagctgcgcgaggcacat	Ala2045Ser		
i-ABCA7-60	Coding region	exon 45	6159	ctgcgcgaggcacatggagg	C/T	cgcctgcgcttccagctgcc	Gly2053Gly		
i-ABCA7-61	Intron 45		27	acggcgccggggtcgggctg	G/C	gggaggcaggctgggggcca			
i-ABCA7-62	3' Untranslated region	exon 46	6580	aaggctggagagaagccgtg	G/C	tggtgaaaccgtgtgcatgt			dbSNP ID:rs1134007
i-ABCA7-63	3' Flanking region		108	caagctgagtgtgcacatac	G/A	ggccaagtggcgattcatag			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA7-64	3' Flanking region		376	cttacaggagcccggtgtcc	C/T	ggagcacaggccagggccgg			
i-ABCA7-65	3' Flanking region		687	cagcagggagacttggggag	G/A	G/Agggagagagttcacactgc			
i-ABCA7-66	3' Flanking region		688	agcagggagacttggggagG/A	G/A	gggagagagttcacactgcg			
i-ABCA7-67	3' Flanking region		1169	cctcgacctgacccacttca	C/T	ggggctgcagggcgggtgat			
i-ABCA7-68	Intron 9		398–422	cgtgaactaccacgtcctgc	$(T)_{22-26}$	aagagatggagtctcactct		+	
i-ABCA7-69	Intron 12		175–184	ggggactctgagggtctggt	$(G)_{8-10}$	actctgagggtctgggggcc			
i-ABCA7-70	Intron 30		81–87	ccccctgggagctctcccgg	$(C)_{6-7}$	ggccctcagctccccttccc			
i-ABCA7-71	Intron 34		349–361	agaaagagaaagagagaaag	$(A)_{12-14}$	cagaaatgtgctttgggtga		+	
ATP binding cassette, subfamily A, member 8 (*ABCA8*)									
i-ABCA8-1	Intron 1		204	ctggtaattaatattagata	A/G	ataaaaacattgagttagaa			
i-ABCA8-2	Intron 1		266	aacattatgttgttttaaac	A/G	taactgagtgtagaaataag			
i-ABCA8-3	Intron 1		674	gttctgaattttacaacagt	A/G	taagaggataaaaatgagtc			dbSNP ID:rs2041321
i-ABCA8-4	Intron 1		733	ttgccatatgtataataaag	T/A	attcatgtttttgctagcct			
i-ABCA8-5	Intron 1		861	agactggagtttgcatgcta	C/T	ctaagactgtagctgattcc			
i-ABCA8-6	Intron 1		907	gaggagatcatcctcttggc	C/T	aatgtctattaacttcgcca			
i-ABCA8-7	Intron 1		1262	cagaaacttttgccctctct	G/A	taggctagctcactgtgaaa			
i-ABCA8-8	Intron 1		1537	agctctcttaaaagtatcca	T/C	gctgaattttctgcacctta			
i-ABCA8-9	Intron 1		7622	tcgttaacagcaatgataat	T/C	tagcccatccttatccC/Taga			
i-ABCA8-10	Intron 1		7639	aatT/Ctagcccatccttatcc	C/T	agaaacaacaggctcataag			
i-ABCA8-11	Intron 1		7720	tccatgtgttacaaactgcc	C/T	tggagaacagaaaaagagaa			
i-ABCA8-12	Intron 1		9397	cataatatatatacatatgc	G/A	cacacacacacatatacaca			
i-ABCA8-13	Intron 1		9519	agtagttcatgttggaacaa	T/C	atgcttgagaaatgcagaaa			
i-ABCA8-14	Intron 1		11880	acacggagatcatatatatg	G/C	aaataC/Tgtgtgtgtattttt			dbSNP ID:rs1860445
i-ABCA8-15	Intron 1		11886	agatcatatatatgG/Caaata	C/T	gtgtgtgtatttttcctgat			dbSNP ID:rs1860446
i-ABCA8-16	Intron 1		12341	ctgggtgcaatctggggaca	G/T	aaactacatagtaattaaat			dbSNP ID:rs887380
i-ABCA8-17	Intron 1		12973	ttgataacaggcacagggca	T/C	cacaaataaatgatggaaca		+	
i-ABCA8-18	Intron 1		13100	cattggagtattaggctacg	T/C	ttttttgttgtttgcaggat			
i-ABCA8-19	Intron 1		13128	ttgtttgcaggatatttctt	T/C	ttcttaagaacttcatatta			
i-ABCA8-20	Intron 2		420	caattagttttcttcaaaaa	A/G	gtagaaaagttggaattgta			
i-ABCA8-21	Intron 2		505	catataaaaaatcttgatta	A/T	actttggtatattttaaaaa			
i-ABCA8-22	Intron 2		819	gcaatgccttggaactatct	C/T	ttaaaacacattgactttca			
i-ABCA8-23	Intron 3		915	ttgtgttcgatagatcagta	G/A	ggtgactagttaacaatgat		+	
i-ABCA8-24	Intron 3		1539	aaagggaaatctgtggtgat	C/T	gccctgtcattcattcatag			
i-ABCA8-25	Intron 3		2341	ttcctttctttgtcaacttc	C/T	gtccaaattccactcaagct			
i-ABCA8-26	Intron 3		2882	tattctatattctgtactct	A/G	ttaatattctataataataa			
i-ABCA8-27	Intron 3		3314	atttaaatatctatctctct	A/G	tatttaccatttcaaattta			
i-ABCA8-28	Intron 4		89	gaggttagtatgccaaatta	G/A	agcatcactatctgtcataa			
i-ABCA8-29	Intron 4		3264	ttccattggcctattatgcc	C/T	gtgttatatccagtgttaga			
i-ABCA8-30	Intron 4		3403	aagagaccaacaaaattctt	C/G	atcagcagaaaagcacagga			
i-ABCA8-31	Intron 5		389	gcttactgaatatataaatt	G/C	agaaaagccatgccaagcaa			
i-ABCA8-32	Intron 5		479	tgagagtggtgagtaactca	A/G	aatgcctggactccG/Aaggtc			
i-ABCA8-33	Intron 5		494	actcaA/Gaatgcctggactcc	G/A	aggtcccagcaggtcaatga			
i-ABCA8-34	Coding region	exon 6	792	atgggtcttcgggattcagc	G/A	ttctggtgagtcaaacgcag	Ala264Ala		
i-ABCA8-35	Intron 6		200	cctcccaagtagctgggact	G/A	caggtgccgA/Gccaccatgcc		+	
i-ABCA8-36	Intron 6		210	agctgggactG/Acaggtgccg	A/G	ccaccatgcctggataattt		+	
i-ABCA8-37	Intron 6		1751	gtgagttattattgtgttgg	C/T	tttgcagctgttttgttttt			
i-ABCA8-38	Intron 6		1808	atttcattatagttttcaaa	G/T	aatattgtaaaacaaaagaa			
i-ABCA8-39	Intron 6		2412	tattcctaattctaaagaat	T/C	ctgcccaaaactttacctt			
i-ABCA8-40	Intron 6		2506	tggatgaataagtgaatgaa	G/A	agttatcttagaA/Gtccattt			
i-ABCA8-41	Intron 6		2519	gaatgaaG/Aagttatcttaga	A/G	tccatttcaggtcttccttt			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA8-42	Intron 7		28	agtgaattaaatatctttcc	A/G	tccacctatagcctaaaaat			
i-ABCA8-43	Coding region	exon 8	991	taaagaaatctttcctcacc	G/A	gcctggtcgtgttcctcctc	Gly331Ser		
i-ABCA8-44	Intron 8		74	tggaatccataggctgtaat	C/T	atttacaaactcagcattgt			
i-ABCA8-45	Intron 8		635	ccagaaattctccctcatgc	C/T	gccaatttacatggatcacc			dbSNP ID:rs767690
i-ABCA8-46	Intron 9		1417	acacatacttaaatatattt	T/C	ctctgttctacttttgtttt			
i-ABCA8-47	Intron 9		2504	agaggaaaattatggtttgg	G/A	aatgaaataaagcagaaata			
i-ABCA8-48	Intron 10		2013	tggccaaagatctttccaac	C/T	tgtgccagtggttcacagga			
i-ABCA8-49	Intron 10		2378	ctgaagaaaattgtcacttt	G/A	aagtatcttttctttttttc			
i-ABCA8-50	Intron 11		-697	aaaaaaaaaaaaaaagagag	A/G	gagaaagaaaatatttgtta			
i-ABCA8-51	Intron 11		-528	tataaaagttagaaaaaaat	G/T	aA/Gtatgttttagaaatagat			
i-ABCA8-52	Intron 11		-526	taaaagttagaaaaaaatG/Ta	A/G	tatgttttagaaatagatgt			
i-ABCA8-53	Intron 11		-342	ctcaaaggagttttagccat	G/A	taataacttactattaatct			
i-ABCA8-54	Coding region	exon 13	1632	ggttcagtcaccatctataa	C/T	aataagctttcagaaatggc	Asn544Asn		
i-ABCA8-55	Intron 14		252	cttattgcaaaataagtgaa	G/A	ttgagtttctaagagatcaa			
i-ABCA8-56	Intron 15		130	ttttgtttttgagacggagt	A/C	tcgatcatctcggctcactg		+	
i-ABCA8-57	Intron 16		534	acatatacattcattcaaat	A/G	cacattttatggtgacaaca			
i-ABCA8-58	Intron 16		588	gaatcatcaggaaagtgtta	C/T	gcaaattctgattagtactt			
i-ABCA8-59	Intron 16		645	atttaaagaaaatttgtaga	C/T	gttttaggtggaatgaagaa			
i-ABCA8-60	Intron 17		431	tgtcaggtttttcttttttt	T/A	ttctttatgttagaaattgg			
i-ABCA8-61	Intron 17		970	ttttgcacaggttttattag	A/C	ttcagtccactcccagcttc			dbSNP ID:rs1373068
i-ABCA8-62	Intron 17		1390	gctgtaaactcgttttgtga	C/A	ttaggtaccccatgattcta			
i-ABCA8-63	Intron 17		2452	cacgttatacctatagtaac	G/A	cggaagaG/Ctctaatcatgag			
i-ABCA8-64	Intron 17		2460	acctatagtaacG/Acggaaga	G/C	tctaatcatgagatG/Ccttag			
i-ABCA8-65	Intron 17		2475	gaagaG/Ctctaatcatgagat	G/C	cttagcagagccaatctcta			
i-ABCA8-66	Intron 18		152	gaagaagcacaggagagagg	C/T	agaatcttgacatccaaagg			
i-ABCA8-67	Intron 19		7477	aaaatctattttgaaagaca	C/T	ttggaactaaaaaaatcttt			
i-ABCA8-68	Intron 21		196	ttgtttaaagtaaaataaaa	T/C	G/Caacaaaacatttttcaaag			
i-ABCA8-69	Intron 21		197	tgtttaaagtaaaataaaaT/C	G/C	aacaaaacatttttcaaaga			
i-ABCA8-70	Intron 21		287	actgtggtggggtgggggga	G/T	gggggagggatagcattggg		+	
i-ABCA8-71	Intron 21		403	cctgcacaatgtgcacatgt	A/G	ccctaaaacctaaagtataa		+	
i-ABCA8-72	Intron 21		1194	tctcgctctgccacccagcc	G/A	gagtgcagtggcA/Gggatcat		+	dbSNP ID:rs1156340
i-ABCA8-73	Intron 21		1207	cccagccG/Agagtgcagtggc	A/G	ggatcatagctcactgtaac		+	
i-ABCA8-74	Intron 21		1968	taatcaaagaagcaactatg	A/C	gaatcatgaaaataacatgg			dbSNP ID:rs991210
i-ABCA8-75	Intron 21		2215	atgttggatatttttgagt	T/C	aagctgtcacatttgagata			dbSNP ID:rs991209
i-ABCA8-76	Intron 24		692	ctcctagatatagacaaaaa	A/C	caaggtgcacaatggccatg			
i-ABCA8-77	Intron 25		212	cctgattaatatatgggaag	G/A	aagggtaaggggtagtggga			
i-ABCA8-78	Intron 26		67	aataattttcagttctgtac	A/G	cactgtgaaacttctttttat			
i-ABCA8-79	Intron 27		515	gtgtctcccaaaccacatca	G/T	tttcatcttttgctattaca			
i-ABCA8-80	Intron 27		661	cctggatattatcagactta	G/A	aatggagaggaaaagtcaat			
i-ABCA8-81	Intron 30		1967	caaaaattagatacaagggg	G/C	tgaaattgactttaattgta			
i-ABCA8-82	Intron 31		112	ctctaaatgctgacccaggt	C/G	acactgggtagatttacaac			
i-ABCA8-83	Intron 33		401	cttctcactaggttgtgaga	C/T	gctgttgttaaattttatgt			
i-ABCA8-84	Intron 35		480	ctggtaacagcatcatcctg	A/T	tgtA/Gtttattttcatagaca			dbSNP ID:rs1373065
i-ABCA8-85	Intron 35		484	taacagcatcatcctgA/Ttgt	A/G	tttattttcatagacagaaa			
i-ABCA8-86	Intron 36		258	tttgcatgtatgttggtaaa	A/G	cctaagtcaaaactcagtta			
i-ABCA8-87	Intron 36		375	atattattttactgtcttag	C/G	ctgtatattaagaaactgac			
i-ABCA8-88	3' Flanking region		674	gcggtggacatagaaagccc	G/A	gaagcttcttgatgtgctta			
i-ABCA8-89	Intron 1		56–57	ttttgcttttgtgtgtgagt	TT/del	gtttcagaggttttgtcttt			
i-ABCA8-90	Intron 1		1180–1191	taaagtataataataaaacg	$(A)_{9\sim11}$	gaaattcctcctgtacagag			
i-ABCA8-91	Intron 1		9877–9885	ctcctgcaaataggtatgac	$(A)_{8\sim12}$	tcaactgagtacaaaaagct			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCA8-92	Intron 1		12588	gtactagagtgcactccttt	T/del	gcaacaggacggccaaagga			
i-ABCA8-93	Intron 6		78	tcaatgcatctttttttttt	T/del	gaaatggagtctcgctctgt			
i-ABCA8-94	Intron 9		265	gtatatggtatttttttttt	T/del	agacctcttagaaagctagt			
i-ABCA8-95	Intron 9		2666	attttttttaaaggtatcca	A/del	tagtcattctcaatttcttc			
i-ABCA8-96	Intron 11		-447	ggatattctgggttttttt	T/del	ctacaaactcaagttttttg			
i-ABCA8-97	Intron 15		8407	gtggaataatttttgactta	T/del	gcatttggtcaaataaaatt			
i-ABCA8-98	Intron 15		9458–9470	tatgtcgagtaacatatgtc	$(T)_{11\sim15}$	ctgaatgccagcttgcagtt			
i-ABCA8-99	Intron 16		54–56	tgaataatagtcatcatcat	CAT/del	aattattatcattacaacta			
i-ABCA8-100	Intron 17		433	tcaggtttttctttttttT/At	T/del	ctttatgttagaaattggac			
i-ABCA8-101	Intron 24		1462	actccatctcaaaaaaaaaa	A/del	gagagaaaaaaattctgcat			
i-ABCA8-102	Intron 33		155	caatactttgcaaaaaaaaa	A/del	gatctttccctgatgatatt			
i-ABCA8-103	Intron 34		184	atactgaatggttttttttt	T/del	ctcctttctcatatgacctc			
i-ABCA8-104	3' Flanking region		1240	atccttggaccaaaaaaaaa	A/del	ctttatctgtgctttgcgtg			
ATP binding cassette, subfamily D, member 1 (*ABCD1*)									
i-ABCD1-1	5' Flanking region		-1772	agtcccagggctagggcaca	G/A	gcaccctcctgcctaactcg			
i-ABCD1-2	5' Untranslated region	exon 1	-59	acaatccttccagccacctg	C/T	ctcaactgctgccccaggca			Dvorakova et al. (2001)
i-ABCD1-3	Intron 1		906	gggcacaatggcatccatcc	C/T	ccgaaggcctgtgtgtgctc			
i-ABCD1-4	Intron 1		2924	gagacctggcccacccaat	C/T	gtaacctctggctctcggcc			
i-ABCD1-5	Intron 1		3056	aagcctctctgtgtctgtca	C/T	cccccgcaggtggagctggc			rs2269365
i-ABCD1-6	Intron 2		2972	agaagtttcccttgctttcc	G/A	tcaagcttggctctgctcga			
i-ABCD1-7	Intron 2		3258	gcgagacagcacctgcagcc	G/A	cttcgctccatggctgccat			
i-ABCD1-8	Intron 2		4612	ggtccttcacaggacattcc	C/T	accacttcagccacacccca			
i-ABCD1-9	Intron 5		2748	aatggcctgcgtgctggcct	C/T	gggcattgggagcctctcaa			rs2071120
i-ABCD1-10	Intron 6		212	atctgtgtggggtgtgtgca	C/T	gggcggcgatgtgagcgtgt			
i-ABCD1-11	Intron 5		2835	ggcgtcagcggctgttgccc	C/del	tgcaggtggaggaaggcatg			
ATP binding cassette, subfamily D, member 3 (*ABCD3*)									
i-ABCD3-1	5' Flanking region		-2834	acatccctttcttgcctggc	A/G	gatttgaactctttgagtca			
i-ABCD3-2	5' Flanking region		-2118	tacagaatcacctttgtcaa	G/A	cctttaagccttttattgaaag			
i-ABCD3-3	5' Untranslated region	exon 1	-40	gtagccgccgccgccgccgc	C/T	gccgcgtcccctcgccggct			
i-ABCD3-4	Intron 1		-6763	atactttgccatttgagata	T/C	cagtttggagttgatagctg			
i-ABCD3-5	Intron 1		-117	atatgttctatctcactatg	A/G	tgtgaaaaagaagaaacgct			dbSNP:rs2147794
i-ABCD3-6	Intron 2		731	ctttggacctatactagttt	C/T	cttaggcattgtgcttagaa			
i-ABCD3-7	Intron 2		3551	accacagtggtctttttttt	A/G	tatttaaaaaaattattggg			
i-ABCD3-8	Intron 2		5936	cagaactcacttccttattc	A/G	gtttttagataacattgttt		+	
i-ABCD3-9	Intron 2		6083	tggttctttaattttatgat	A/G	tgtttgttatagctatctta			
i-ABCD3-10	Coding region	exon 3	162	atgcagaaagaggggaaaaa	G/A	gagcgagctgtggtggacaa	Lys54Lys		dbSNP:rs16946
i-ABCD3-11	Intron 3		614	tctcttgtttctgaagtatt	A/T	tttcattttattttatgtga			
i-ABCD3-12	Intron 3		651	gtgaaatgctagggtactgc	C/T	atacagctaccctaaatggt			
i-ABCD3-13	Intron 3		2005	ggctaatttggccagcttcc	C/T	accctctgcaggccttcttc			dbSNP:rs1041282
i-ABCD3-14	Intron 4		395	aaagcatttcaaagaatcac	G/A	ttgagcatgtttattagaag			
i-ABCD3-15	Intron 4		4708	tttcccaaatttatccatct	A/C	gtccctcacagaaaagaaat			dbSNP:rs1158254
i-ABCD3-16	Intron 4		4708	tttcccaaatttatccatct	A/G	gtccctcacagaaaagaaat			
i-ABCD3-17	Coding region	exon 7	555	gacaacagaatagctaatcc	A/G	gaccagctgcttacacaaga	Pro185Pro		
i-ABCD3-18	Intron 7		83	atttatatagaattgatttt	G/A	tggacataacagcagcatgt			dbSNP:rs681187
i-ABCD3-19	Intron 7		124	aaatatttaatgcttttata	A/G	gaaaattagagttgttgtaa			
i-ABCD3-20	Intron 7		838	ggtcacagttgacctagata	T/C	acagttttgagacaaaagaa			
i-ABCD3-21	Intron 8		1150	aatcttgaatacttactagc	A/C	catatattgtgctagatagt			
i-ABCD3-22	Intron 9		1493	tcatcttcttccataggctt	A/G	ggtgtggagaggagatagaa			
i-ABCD3-23	intron 9		1971	ctcttcatctataaaatacc	T/C	ttttttttctagtcgtgatcc			dbSNP:rs2147795
i-ABCD3-24	intron 13		1534	tctgttgagttggggatcct	A/G	tggaaacctcttccttcatc			
i-ABCD3-25	Intron 16		4310	gaaaagtgaatgctgagtag	G/T	ttagccaggcttgatttaga			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCD3-26	Intron 20		273	ttctaaaagttcagagaaac	T/A	ctgtagctcattattcctgg			
i-ABCD3-27	Intron 20		1664	ctcaaaagaaaaaaaaaaa	A/C	aaaaaacacatgatccataa		+	
i-ABCD3-28	Intron 20		6693	cttaaggtttgtgttttact	C/T	tgagcaattagtatttccca			
i-ABCD3-29	Intron 21		7171	atcataaacagagaaataat	A/G	tcttaaatgagctctgaaaa			
i-ABCD3-30	Intron 22		1220	ctagaaatcaaaggcattta	A/G	aatatagccaagcctttatg			
i-ABCD3-31	Intron 22		1358	agtagcaaaataatcatcac	G/A	ccagtgatcatgtgaaggag			
i-ABCD3-32	3' Untranslated region	exon 23	2499	cagcgtttattatacagtgg	T/C	agatttctttagctgccaca			dbSNP:rs698951
i-ABCD3-33	Intron 4		4448–4461	taactttcttgtagttagcg	(T)11~14	aactgttttacttttttaggg			
i-ABCD3-34	Intron 5		268	gttttttggcatttttttt	T/del	aaccttcagtccaggttttc			
i-ABCD3-35	Intron 5		891–902	ttggtgtaaaacctgtagtg	(T)10~13	aacaaatgcaaatatagtgt			
i-ABCD3-36	Intron 7		1226–1227	gggaatgggggtgtatcta	T/ins	tacaactttccatgtaattt			
i-ABCD3-37	Intron 8		1129	cagatttacttttttttttt	T/del	aatcttgaatacttactagc			
i-ABCD3-38	Intron 13		1595–1596	tgaaacataataaagcacac	TA/ins	gttatcattaatactttatg			
i-ABCD3-39	Intron 16		7337–7351	ttaattacttcacagactga	(T)13~15	caggttcgatctggggctaa			
i-ABCD3-40	Intron 18		12	gttcctcaggtaagacctag	C/del	ttgagttatctttgatctaa			
i-ABCD3-41	Intron 20		1652–1670	gcaagactctgtctcaaaag	(A)17~20	cacatgatccataatagagg		+	
i-ABCD3-42	Intron 20		2262–2273	ttaatccatttttgtaaatc	(T)11~13	accttaaattagcaactatc			
i-ABCD3-43	3' Untranslated region	exon 23	2072–2079	taaaataaagttgagcttag	(T)8~9	aaaaaaaaaacaaagcaaca			
i-ABCD3-44	3' Untranslated region	exon 23	2080–2091	gttgagcttagttttttttt	(A)10~11	caaagcaacaaattaactag			
i-ABCD3-45	3' Untranslated region	exon 23	3349–3368	acttattttctgttcagatt	(A)16~19	ctcagatatcctatacaacc			
ATP binding cassette, subfamily D, member 4 (*ABCD4*)									
i-ABCD4-1	Intron 1		276	tggcattctttttttgaaaa	G/A	aagaacctcaggtgcacaaa			
i-ABCD4-2	Intron 1		329	cttctcagttcttgacaccc	T/C	gtgggccaatgcaaggctcc			
i-ABCD4-3	Intron 3		171	ttaagcacgttgatcttgct	A/G	ttggcccacgtgggactgat			
i-ABCD4-4	Intron 3		449	cctacccctcattcagtagg	G/A	gggctaccacctgctcactc			
i-ABCD4-5	Intron 5		238	ataggagcagagggcccatt	G/A	ggccactcagattggacagg			dbSNP ID:rs2074945
i-ABCD4-6	Intron 5		273	gacaggggctacctgagagg	G/T	aacaggagtcagggctgagg			
i-ABCD4-7	Intron 6		511	ggggttttctaccccataac	A/C	gtccaggaaagggtcagact			dbSNP ID:rs2074946
i-ABCD4-8	Intron 7		240	tagtcttagtggcctagcgt	G/A	gggcctgaaattgtcaaatg			
i-ABCD4-9	Intron 7		267	gaaattgtcaaatgaatgaa	T/C	gcctcatcctcttgctggtg			
i-ABCD4-10	Coding region	exon 9	910	tctatggagacctgagtccc	G/A	cagagcttagcaccctggtc	Ala304Thr		
i-ABCD4-11	Coding region	exon 10	981	atcagctgcttcacccagct	C/A	atcgacctgtccacgacgct	Leu327Leu		
i-ABCD4-12	Coding region	exon 11	1102	gcgagatcctgggcgagagc	G/A	agtggggcttggacacgtga	Glu367Lys		
i-ABCD4-13	Intron 13		191	tggattgggcccactactca	T/C	agcagctcctgaggcaggta			
i-ABCD4-14	Intron 13		262	acgcgtatgtcaaacaccca	A/G	ggtcggattctggggcccct			
i-ABCD4-15	Intron 17		848	cctctgctcctctggcccat	C/G	cttctccctgaggcagggct			
i-ABCD4-16	Intron 17		946	gtgggaggagaagcagcggc	G/A	gcagagggcagggctttgat			
i-ABCD4-17	Intron 18		41	ggcctgaggaggagaaa_aa	C/T	ccaaaggctcagcctggcca			
i-ABCD4-18	3' Untranslated region	exon 19	2001	gcccaggtctaggtttctgt	G/A	ggggacactgaatctcccag			
i-ABCD4-19	3' Untranslated region	exon 19	2083	catatgcttgaagtgctgat	T/G	acctacaaatgatttcagat			dbSNP ID:rs2466
ATP binding cassette, subfamily E, member 1 (*ABCE1*)									
i-ABCE1-1	5' Flanking region		-158	aactcagattctcggcacct	C/T	cagcagctggcttcgccaac			
i-ABCE1-2	Intron 9		237	ctgaaattatatgcaaattc	C/T	gtagctttataggaagcaga			
i-ABCE1-3	Intron 9		4203	ttgtgtaggaagctgataca	T/G	taatttgacatatgagatgt			
i-ABCE1-4	Intron 10		1811	ccaagaaacttcagctttct	C/T	ttcacttaaatataggaaac			
i-ABCE1-5	Intron 17		2301	atatccagaaacagatggta	T/C	gtgcagaacaggttgtacag			
i-ABCE1-6	3' Untranslated region	exon 18	1810	tggatgattagactgactct	G/C	agaatattgataagccattt			
i-ABCE1-7	Intron 1		5349–5363	tttgtctgggttggttgggg	(T)13~16	gagactgggtctgactctca			
i-ABCE1-8	Intron 1		5845–5854	tacatttgtcaaaatttata	(T)9~10	gcagataatcatttcatctc			
i-ABCE1-9	Intron 5		836–851	taaattcacatgattctgta	(T)14~16	aggatcctcctgactggcag			
i-ABCE1-10	Intron 8		1153–1169	tctttcaaacttatatttgc	(T)13~17	catagtttcatgtttgatga			
i-ABCE1-11	Intron 9		1023–1024	ttgctctgtttcaaatctct	T/ins	attcatgggccagcagctcg			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	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	aatcttggaggaatcttttt			
i-ABCE1-16	Intron 16		1163–1172	gattagaaatccaggttaaa	(T)9~10	gttttgcacaaaaatattac			
i-ABCE1-17	Intron 16		1372–1382	taaaatttaatcaaaattga	(T)10~11	ctcttagtcctcaaaccctt			
ATP binding cassette, subfamily F, member 1 (*ABCF1*)									
i-ABCF1-1	5' Untranslated region	exon 1	-60	gccagccccatcggggttcc	C/T	cgccgccggaagcggaaata			
i-ABCF1-2	Intron 1		101	gcacgagactgaccgggccc	C/G	tgcgggagttactgcgcatg			
i-ABCF1-3	Intron 9		321	tggaggtagcggtttgtcag	A/G	ggcttccctgcagggagaaa			dbSNP ID:rs1264440
i-ABCF1-4	Intron 16		103	ctacatttcaaggactgccg	C/T	gcagggctcaggtttctctt			dbSNP ID:rs1264437
i-ABCF1-5	Intron 20		69	tgactttaaccgaccacctc	C/T	ctctctttctcgggcagaaaa			
i-ABCF1-6	Intron 23		35	agtgtgccctcatccctgct	C/A	catggggaccaagctgtagt			
i-ABCF1-7	Intron 7		342–354	acagagcgagactccgtctc	(A)10~14	gaaaaaaaaaaaaacattt			
i-ABCF1-8	Intron 7		356–369	cgtctcaaaaaaaaaaaag	(A)13~15	catttcatcagacctgtctt			
i-ABCF1-9	3' Untranslated region	exon 24	2425	tcagccggccccgagagtga	A/del	gctttccttcccagaagtct			
i-ABCF1-10	3' Flanking region		1067–1068	attaatttgatcaattgtct	T/ins	aatatgtcgtactctagatt			
ATP binding cassette, subfamily G, member 1 (*ABCG1*)									
i-ABCG1-1	5' Flanking region		-386	gcaataatcattggctagag	G/A	tattgtgatatgatgtcatt			
i-ABCG1-2	5' Flanking region		-206	aatctgaccaaaagtgagag	A/C	tgggtagattttcctacttg			dbSNP ID:rs1893590
i-ABCG1-3	5' Flanking region		-136	gctttacgcccagtgacttg	T/G	gagggaacagaactgccctt			dbSNP ID:rs1378577
i-ABCG1-4	Intron 1		199	caccaaatattggtgagctg	C/T	ctggatttgggagatgcagt			
i-ABCG1-5	Intron 1		291	acttggggtccggtgtgagg	A/C	tcctgcactcggtttctgtg			
i-ABCG1-6	Intron 1		318	actcggtttctgtgatggtg	T/A	gtgcaggggagtcacaagtt			
i-ABCG1-7	Intron 1		468	ggtcccaacgggtttctaga	T/C	ccctccagagaagcctttgg			
i-ABCG1-8	Intron 2		434	ctgggtacaggttttgttcc	G/A	gttggtctgctattgagtat			
i-ABCG1-9	Intron 3		1839	ttaaaatgagttgtttttct	C/G	ctaaagcctttagggagttg			
i-ABCG1-10	Intron 3		3076	tttgtcacttccttcgtctc	C/T	ggctctacttccctgggggt			
i-ABCG1-11	Intron 3		3352	gttccttggaggaaacgtgg	G/A	gtacacagtggttccagtta			
i-ABCG1-12	Intron 3		8030	acagtgaagcacaaggcagc	C/T	gaagacacagcaggcaggtc			
i-ABCG1-13	Intron 3		8066	aggtcaggtctgtgtgcaca	T/C	tggcaggctgcA/Gtgcagacc			
i-ABCG1-14	Intron 3		8078	tgtgcacaT/Ctggcaggctgc	A/G	tgcagaccagcctC/Tggccca			dbSNP ID:rs692383
i-ABCG1-15	Intron 3		8092	ggctgcA/Gtgcagaccagcct	C/T	ggcccaggtggagaagcaga			
i-ABCG1-16	Intron 3		8285	ctggacatgtgactcccctg	C/T	acccaccctcacaagcacca			
i-ABCG1-17	Intron 3		8342	gccagaggccaagtcctcag	T/G	gtgccccttttctcccccttca			dbSNP ID:rs915845
i-ABCG1-18	Intron 3		8860	cagggtgatagggagtccaa	T/C	tggacacaggttcagtttgc			
i-ABCG1-19	Intron 4		2246	cttatacagttccatgaaca	T/C	ggctctggaaacacctctgt			dbSNP ID:rs564010
i-ABCG1-20	Intron 4		2319	gggggtgaacagagggcaga	G/A	gcctgggcatcttcactcag			
i-ABCG1-21	Intron 4		2557	gaagggaagaagcagcagca	A/G	gaaagaagccccctggccct			
i-ABCG1-22	Intron 5		139	tgacccagggcaccctagag	T/A	ggcgcccggctccgatcgct			
i-ABCG1-23	Intron 5		177	gctgcccctgcccctccgcc	A/C	gggccacctggagcctcggg			
i-ABCG1-24	Intron 6		13	cagttactgtaagtgctgtt	T/C	ccaggggtggtcaG/Agaatct			
i-ABCG1-25	Intron 6		27	gctgttT/Cccaggggtggtca	G/A	gaatctccctttctggtttt			
i-ABCG1-26	Intron 6		1191	gctaagcagagttaggcccc	G/A	gctagtccttgaatgagaga		+	
i-ABCG1-27	Intron 6		1449	atgctggagcccctgagttc	G/A	gtgggcatacaaggggtggc			
i-ABCG1-28	Intron 6		2282	ctcgcatcacgcagttttca	C/T	gatcctattaattgggtgag			
i-ABCG1-29	Intron 6		3435	ataggcacacatggggaaaa	G/T	gcacccaccaagagtcctca			dbSNP ID:rs2032260
i-ABCG1-30	Intron 6		3853	cctgggcttcagcaggggcc	T/C	cacacctgcaatgggtgC/Tct			
i-ABCG1-31	Intron 6		3871	ccT/Ccacacctgcaatgggtg	C/T	ctggggagagggtgcagatg			
i-ABCG1-32	Intron 6		4175	tccaaagcccagatttggtg	T/C	ttttggggctcttttggaat			
i-ABCG1-33	Intron 7		4	ctggtggaggaagaaaggta	G/A	ggagggcggctgctttgtgt			
i-ABCG1-34	Intron 7		576	agctcaggaggtgtctggaa	C/T	gccacacagtgcaggagttt			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCG1-35	Intron 7		1426	aattctccttctcaacttaa	A/G	gaaatattttatagaaaaat			
i-ABCG1-36	Intron 7		2342	agagcctgcaatgggccgcc	G/A	agggacctgcccatgactca			
i-ABCG1-37	Intron 7		2399	gaggggttgacagacaggat	A/G	tgtctgC/Gtgtgttccagctg			
i-ABCG1-38	Intron 7		2406	tgacagacaggatA/Gtgtctg	C/G	tgtgttccagctgctggttt			
i-ABCG1-39	Intron 7		2911	ccctctctgtgcccactgtt	G/C	tcccaacaccagcctgttct			
i-ABCG1-40	Intron 7		4363	tataatagattcctagcaga	A/G	aacataattgtgagaggaac			
i-ABCG1-41	Intron 7		4752	gctttcagagcccattcaca	C/T	aagggtctcattttattagg			
i-ABCG1-42	Intron 7		5026	ccaggtctgtgggatttcag	G/A	ccaaaaaggagcgtagcaag			
i-ABCG1-43	Intron 7		5532	gggttaaatattccgggcag	C/T	gccaagtcagattatctgta			
i-ABCG1-44	Intron 7		5681	gctaaagtgcatggaaggca	T/C	catgaataaatcctttcagg			
i-ABCG1-45	Intron 7		6290	tcacagcagattcatgagag	T/A	tgaatgtttagccgccatgt			dbSNP ID:rs881394
i-ABCG1-46	Intron 7		6386	agatgctcccctccagccag	C/T	acattttctccctgtgagca			dbSNP ID:rs881395
i-ABCG1-47	Intron 7		6758	acctgcatggtgggtgcccc	C/G	ctgccttcctctactgcctt			dbSNP ID:rs915846
i-ABCG1-48	Intron 7		7029	tgggtcagattaaatatatc	C/T	tgaaggactaaaccgtaaaa			dbSNP ID:rs225440
i-ABCG1-49	Intron 7		7176	ttgctcacattgtgaaaaaa	C/G	gcaaaaagatgggttttcag			dbSNP ID:rs225441
i-ABCG1-50	Intron 7		9243	gcctgagagcgctggcagta	G/A	gaagggtcgccagtgtggac			
i-ABCG1-51	Intron 7		11224	tctggtttagagaggaaaat	G/A	ggcagcatcattttgtcacc			dbSNP ID:rs915847
i-ABCG1-52	Intron 7		11371	gggctctcttggagcccttt	T/G	tctctcccagccctgcgtct			
i-ABCG1-53	Intron 7		12420	gggatttcgaatctcaacac	T/C	ctgagctctgtgctttcccc			
i-ABCG1-54	Intron 7		12484	gagttgtcctccaagagaat	C/T	tttgtatggttccttttctg			dbSNP ID:rs225446
i-ABCG1-55	Intron 7		12955	ctggggttggtgggagccac	A/G	gtctcacacctattggcagg			dbSNP ID:rs225447
i-ABCG1-56	Intron 7		12985	ctattggcaggtcgtgaaca	T/C	tgttcttggatttgcaaata			
i-ABCG1-57	Intron 7		20041	acatggccggcttcccttct	T/C	cctcG/Agaatggcctggaatt			
i-ABCG1-58	Intron 7		20046	gccggcttcccttctT/Ccctc	G/A	gaatggcctggaattcgatc			
i-ABCG1-59	Intron 7		21058	acaagacttagaatttgacc	G/A	tgattttaaaactattctaa			
i-ABCG1-60	Intron 7		26189	ttcttggatgtggccatgca	C/T	gggggcaagggtttgatgag			
i-ABCG1-61	Intron 7		27453	atcatgtggtttgggggaaa	G/C	ctgggaccccacttggtaca			
i-ABCG1-62	Intron 7		28098	caggaaggagacagctgctg	G/C	tgctgcttagagttaggcgc			dbSNP ID:rs225376
i-ABCG1-63	Intron 7		29670	ccttcagttgtaataggcag	A/G	aggagcgcacgaggaggctg			dbSNP ID:rs225378
i-ABCG1-64	Intron 7		29810	attgtttctcctggttttgt	T/C	tgtgttgactttccctttaa			
i-ABCG1-65	Intron 10		1995	cagatcccttggttgctggg	C/T	aggtagtaggagaggttttt			dbSNP ID:rs225383
i-ABCG1-66	Intron 10		2116	aaacagggcttgagtcctcc	G/A	taagggacaggagaccttcc			
i-ABCG1-67	Intron 10		2145	aggagaccttcccacatcct	G/A	gcaagaattcttctttttttc			dbSNP ID:rs225384
i-ABCG1-68	Intron 10		2437	cagactaaatgcacaattct	G/A	gattgagctgactgtattga			dbSNP ID:rs225385
i-ABCG1-69	Intron 10		2689	tgtaaaagatggagaagaac	A/G	cagtagtcgcttgctgtgag		+	dbSNP ID:rs225386
i-ABCG1-70	Intron 10		2804	tgtgactcatggcctctgcc	A/G	ggggactgggctggccctgc			dbSNP ID:rs225387
i-ABCG1-71	Intron 13		1196	tgaaaagaaaatggatgagt	G/A	gaaA/Cccaaaagagagaaaat			
i-ABCG1-72	Intron 13		1200	aagaaaatggatgagtG/Agaa	A/C	ccaaaagagagaaaatgtgg			
i-ABCG1-73	Intron 13		2041	aagcagaggcttttccaccc	G/A	gagactcaagaagctgctcc			
i-ABCG1-74	Intron 13		2490	gtggtgaagtagagctgagc	A/T	cacgggggagccctccatcc			
i-ABCG1-75	Intron 13		2552	atggccttgggccactgcct	G/A	ctgtgccccgagccgagctt			dbSNP ID:rs170441
i-ABCG1-76	Intron 13		2822	cagcaggctccgtgctgaag	T/C	cacagcaagccaggcccttg			
i-ABCG1-77	Intron 13		2850	agccaggcccttggcctgcc	G/A	gagctggaagacccagaaca			
i-ABCG1-78	Intron 13		2919	gcctcccaggagtagctaca	C/T	gggacccgaaggcagatggc			
i-ABCG1-79	Intron 13		3506	ggcagcctgggctgccgaga	T/C	cctccctggagcgcccgccg			
i-ABCG1-80	Intron 13		3538	cgcccgccgggaagccccag	G/A	ggggctggagctacaA/Ggtgg			
i-ABCG1-81	Intron 13		3554	ccagG/Aggggctggagctaca	A/G	gtggccttgcaggttttttg			
i-ABCG1-82	Intron 13		3721	ccagctcatgggcaggggtg	C/T	ggagggaaaggcacccacag			
i-ABCG1-83	Intron 13		3852	caccagagccactcagtcgg	C/T	caagagcgtcgcccagtggt			dbSNP ID:rs225410
i-ABCG1-84	Intron 13		3921	gaagaccagcagtcgatgcc	A/G	gctgggaagagggctctgcc			
i-ABCG1-85	Intron 13		3979	acccaccagccttttccaga	C/T	agccttccagaagctgtttc			
i-ABCG1-86	Intron 13		4291	gagccgctggagtagggtcc	G/A	cttgctatggctcccagggg			
i-ABCG1-87	Intron 13		4922	gaaaccaccagaaattgtgc	A/G	tcctctcatgtgtccattca			dbSNP ID:rs492338
i-ABCG1-88	Intron 13		4968	tattgactggacaccttctc	C/T	gtatggggcactgggctagg		+	

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCG1-89	Intron 16		672	atcagtaacgggtcactaac	G/A	gatgctgctgagtggggcag			
i-ABCG1-90	Intron 16		840	atttcatttcctcaatgtcg	T/C	ctgaccagagagcgggaggt			dbSNP ID:rs450808
i-ABCG1-91	Intron 16		891	tggcccactgttgagggtgt	G/A	ggtgaccagaggggcctgga			
i-ABCG1-92	Intron 16		997	tgtgtcctggtttgtggctt	C/G	atctaggaggtgtggtggcc			dbSNP ID:rs425215
i-ABCG1-93	Intron 18		1616	ctggaggagaagacaggata	A/C	agtctaagacgtgC/Ttgtcac			
i-ABCG1-94	Intron 18		1630	aggataA/Cagtctaagacgtg	C/T	tgtcacagagttcagggtcc			
i-ABCG1-95	Intron 18		1674	gcttccaaaggccgcatccg	G/T	gttgttctctgagcC/Tgagga			
i-ABCG1-96	Intron 18		1689	atccgG/Tgttgttctctgagc	C/T	gaggacggctttgcgaacgc			
i-ABCG1-97	Intron 19		446	tggctgacagtgaacacagc	G/A	gctgcttctccagaacttta			
i-ABCG1-98	Intron 19		581	atgcagagtttcagaagagg	C/G	agactcaggaagagtaaggc			dbSNP ID:rs914189
i-ABCG1-99	Intron 22		243	acccggagagccatggcagg	A/C	ccaagtgttctggacgttgc			
i-ABCG1-100	3' Untranslated region	exon 23	2760	gcctctcagctgatggctgc	A/G	cagtcagatgtctggtggca			dbSNP ID:rs1044317
i-ABCG1-101	3' Flanking region		1129	ctcagaactacatcgagtga	G/A	gtcagtgttgaaaacgccca			dbSNP ID:rs1541290
i-ABCG1-102	3' Flanking region		1257	atggggcccacagccctgcc	T/C	cagaagcagctttggtctcg			
i-ABCG1-103	3' Flanking region		1438	gggggaagagcttgggaacc	A/G	tgagggctgttaggctgcaa			
i-ABCG1-104	3' Flanking region		1518	tgaagggtgaactggagtag	G/C	tgaggattctgcagttgacg			
i-ABCG1-105	Intron 3		3754–3755	ctccaccctgcacctccctg	G/ins	cctccttgatttccctcatc			
i-ABCG1-106	Intron 3		78487854	cagtttccagaatttggggg	(A)6-7	tcccataagctgtcatactt			
i-ABCG1-107	Intron 4		190–191	tgtcgagagctccccttgcc	C/ins	tggttgatcctcagggttct			
i-ABCG1-108	Intron 4		198–206	agctccccttgcctggttga	TCCTCAGGG/del	ttctacttagaatgcctcga			
i-ABCG1-109	5' Untranslated region	exon 5	(-713)–(-741)	cgcagctcaagcctcgtccc	(CGC)8-10	ccccggggcatggcctgtct			
i-ABCG1-110	Intron 6		376–387	tcttgccttgagctcaagag	(A)10-12	tagccaggtttctgcgcatg			
i-ABCG1-111	Intron 7		19944–19945	ctgatgaggaggggaggggg	CACCAGGCAGCAGACTCTGATGAGGAGGGGAGGGGG/ins	caccaggcagcagactctga			
i-ABCG1-112	Intron 7		25136–25137	catgaacttgcctgaccata	G/ins	ccctgtgaggagctagggct			
ATP binding cassette, subfamily G, member 2 (*ABCG2*)									
i-ABCG2-1	Intron 1		152	tcatttgaaagtgggtatgc	G/A	gtttaaaactgacagttcaa			
i-ABCG2-2	Intron 1		614	agctagtcataaataaatac	G/A	ccagagtagtaaggaagaga			
i-ABCG2-3	Intron 1		10002	cctcatgaatggtatacatg	T/A	cccaacatatctctttcgat			
i-ABCG2-4	Intron 1		10123	acagtggtccctttgggtgc	G/A	tatacccaaatccctgcata		+	
i-ABCG2-5	Intron 1		10768	ataggaataattgagaacag	G/A	gtctgaagaactctgcagga			
i-ABCG2-6	Intron 1		10791	ctgaagaactctgcaggaaa	T/C	G/Aaaaatagttccctgctttt			
i-ABCG2-7	Intron 1		10792	tgaagaactctgcaggaaaT/C	G/A	aaaatagttccctgctttta			
i-ABCG2-8	Intron 1		14183	tcacttaaggctttgcaggg	T/G	gtctaggacacagaaagaga			
i-ABCG2-9	Intron 1		14934	aaagtgtctttaaaatttcc	A/G	tcttgagtcagtgagctatt			
i-ABCG2-10	Intron 1		14955	tcttgagtcagtgagctatt	G/T	aaattcaagcaataagttat			
i-ABCG2-11	Intron 1		17251	ctgtttgggaacagcaactc	A/C	atcataggcagagagaaagt			
i-ABCG2-12	Intron 1		17347	atttcaaacctgtttcacaa	G/A	ttgttaagctcatcttaagg			
i-ABCG2-13	Intron 1		17626	gaaggtgcataacaacttcc	T/G	acataaagtctggagctata			
i-ABCG2-14	Intron 1		18271	aaatgaagctgctcattgcc	A/G	cacatttaaaaatggacttg			dbSNP ID:rs1564481
i-ABCG2-15	Intron 1		18369	ctattgcttttctgtctgca	G/T	aaagataaaaactctccaga			
i-ABCG2-16	Coding region	exon 2	34	atgtcgaagtttttatccca	G/A	tgtcacaaggaaacaccaat	Val12Met		
i-ABCG2-17	Intron 2		36	tgtaaaaagacagcttttta	A/G	tttacctacagtgaacctca			
i-ABCG2-18	Intron 2		4230	caaccctaaattggagggcc	C/T	gggcgtggtgattgagaaag			
i-ABCG2-19	Intron 2		4518	gttgacagacttttatagtg	A/C	gggacactgacctgcatgca			
i-ABCG2-20	Intron 2		6278	atgtatgtaccacgtcttca	T/C	attcttaaaggatgacccta			
i-ABCG2-21	Intron 3		10	ggcaaatcttcgtgagtata	A/G	gagagtataagtaagcgttt			
i-ABCG2-22	Coding region	exon 5	421	tgacggtgagagaaaactta	C/A	agttctcagcagctcttcgg	Gln141Lys		
i-ABCG2-23	Intron 6		3158	actattctagttgattctag	A/G	ttgtcaatacaacacactga			dbSNP ID:rs1871744
i-ABCG2-24	Intron 6		3203	tcctattctgttttaataaa	A/G	gcattgaatttaggtttgct			
i-ABCG2-25	Intron 6		3287	gtcaggctgaactagagcaa	A/G	caatctaaaggcaagaatag			
i-ABCG2-26	Intron 7		179	ttcatttttgtagcaccagc	T/C	tgttatttaggtatcttttct			dbSNP ID:rs1481012
i-ABCG2-27	Intron 9		5677	gcacttggactttgctttgc	T/C	acatacttgcattgctctgc			dbSNP ID:rs2054576
i-ABCG2-28	Intron 9		5974	tatactaataaatggtgtgt	A/T	taagttttatctctaattg			
i-ABCG2-29	Intron 10		1908	gacgcttatgtgcagcctat	G/T	ttgatgtctggaaaggctga			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCG2-30	Intron 10		2094	ccctgagggctgaggtatct	G/A	gattatttccagacttgcta			
i-ABCG2-31	Intron 11		20	tgtgagtaggtctttgttct	A/G	ggaacggggctgtccagcag			
i-ABCG2-32	Intron 11		1447	tgttcttcaaggaaagcccc	C/T	gtcaaagaaggaaaagaagc			
i-ABCG2-33	Intron 12		49	atgtctttagtcttgcctat	G/T	ggtgaagtcagttgcacctt			
i-ABCG2-34	Intron 12		1566	tatgcagttacatggacaga	C/T	acaacattggagaccgaggg			
i-ABCG2-35	Intron 13		40	gctctgataaggaattgttt	C/T	tttccttcatttcttcctgc			
i-ABCG2-36	Intron 13		1823	ttactcaagcaggcctgact	C/T	ttagtatttgctttttgtag			
i-ABCG2-37	Intron 14		497	ctaatgaaaacaaacaagaa	T/C	gaaagattgtcactgtaaat			
i-ABCG2-38	Intron 14		815	taactctttggaaacttctt	A/G	aaatttaaaactgtttacct			
i-ABCG2-39	Intron 15		110	ccaggggcactgaatttttc	C/T	gagcctacgttttctcatcc			
i-ABCG2-40	Intron 15		566	gccgcatagtcatgtgttgt	T/A	gtttttaaattaacttggaa			
i-ABCG2-41	Intron 15		639	aacaagaaacacttgaataa	G/A	ttgagaaaaaaccccgtttt			
i-ABCG2-42	Intron 15		1197	tgagtagctgggattacagg	C/T	gcccaccaccacacctggct		+	
i-ABCG2-43	3' Flanking region		520	catcaattcaggtcaagaaa	T/C	agaagattgtagcacacaaa			dbSNP ID:rs1448784
i-ABCG2-44	5' Flanking region		(-998)–(-995)	gttgggatggctacactcac	TCAC/del	aaagcctgatggcccgtttc			
i-ABCG2-45	Intron 13		405	ctgctagtttatttttttt	T/del	aacattttaatttatgttt			
i-ABCG2-46	Intron 13		692–702	tcaatatgtttctgcttatc	$(T)_{9\sim11}$	aatggttacttaatcctaat			
i-ABCG2-47	Intron 15		645–650	aaacacttgaataaG/Attgag	$(A)_{7\sim8}$	ccccgttttcacataatgtt			
ATP binding cassette, subfamily G, member 4 (*ABCG4*)									
i-ABCG4-1	Intron 1		84	ggcctgggtgtcccatgttc	G/A	gaaagtcctgcaccagtggg			
i-ABCG4-2	Intron 2		77	gaacacagaaggtattctga	A/G	agggcattgacccccatcct			
i-ABCG4-3	Coding region	exon 6	679	tggtgtccctcatgaagtcc	C/T	tggcacaggggggccgtacc	Leu227Leu		
i-ABCG4-4	Intron 7		95	ggcctcctaggggtagagat	C/T	tcaccgtcgcctgccttccc			
i-ABCG4-5	Intron 7		158	cttgcccttgggaagtgagt	G/A	tgaatctaaactgagctctc			
i-ABCG4-6	Intron 8		106	ccccagaggcattgcaacca	A/G	tgggtgctaggaagaaccta			
i-ABCG4-7	Intron 8		1089	aggtacacaacttaatggta	C/G	aagattctctgtagacctgg			dbSNP ID:rs626776
i-ABCG4-8	Intron 11		1113	acgtgagacgagataagtga	T/C	ggtcatA/Gtggccagggagga			dbSNP ID:rs674424
i-ABCG4-9	Intron 11		1120	acgagataagtgaT/Cggtcat	A/G	tggccagggaggaaggggac			
i-ABCG4-10	Intron 11		1173	gggggacagcttgaacaaga	A/G	tgtggaggcaggatggacac			
i-ABCG4-11	3' Untranslated region	exon 14	2758	gagtgacaggcacatacatg	A/C	gaacaggccatctcagccct			
ATP binding cassette, subfamily G, member 5 (*ABCG5*)									
i-ABCG5-1	Intron 3		40	ccctggccccccgcccgcc	C/A	cgggggcttaggctacactg			
i-ABCG5-2	Intron 4		841	gcttggaggcatcttgaatg	C/T	gcctcatccaaactggactg			
i-ABCG5-3	Intron 4		1145	gagcaaatccagcccacagc	G/A	tgtaaaatC/Actgataagtaa			
i-ABCG5-4	Intron 4		1154	cagcccacagcG/Atgtaaaat	C/A	ctgataagtaattcagtggg			
i-ABCG5-5	Intron 4		1690	acagagatgagaaggaggct	T/C	gggaatctaccctggctggt			
i-ABCG5-6	Intron 4		1806	tcttttgttccagaatatat	T/C	tatatctagtttatttatgc			
i-ABCG5-7	Intron 4		1878	atttcagatatgtccattct	C/T	tgggtgggtcaaagctacat			
i-ABCG5-8	Intron 4		2052	gggtgtctggaaacaaaact	C/T	attaccatatgagtatcttc			
i-ABCG5-9	Intron 4		2108	tccccctggggtttctgcag	A/T	tagaggtaatcagtacaggg			
i-ABCG5-10	Intron 4		2230	agcttcttgattagaaattc	G/A	gtaaagaatttttttagtc			
i-ABCG5-11	Intron 4		2318	ggagttacaggctttaagta	G/C	agcgaagagaattggaagaa			
i-ABCG5-12	Intron 4		2367	ttaaatgtggctgggggtta	C/T	aaattgggtccccattaaag			
i-ABCG5-13	Intron 4		2464	gattatatgtctttgatgtg	A/G	actcacactgagattgtacc			
i-ABCG5-14	Intron 4		2586	aaagcatttatgataataaa	G/A	tttcaaaacccaaacactta			
i-ABCG5-15	Intron 6		1318	cagagacattcaaagtgcat	C/T	gctacccttgtgatcacaca			
i-ABCG5-16	Intron 9		164	caactattgagttaccaaca	T/C	gttaatatgaatgagctcac			
i-ABCG5-17	Intron 9		365	gtaccgttagcttctctttg	A/G	agctgattttaggacagcca			
i-ABCG5-18	Intron 10		64	tcatggagctagtgggactc	G/A	tgcagggagagctccagggt			
i-ABCG5-19	Intron 10		2406	tcaacaagcctgcttactgc	G/A	gttagttgtgaccattgtct			
i-ABCG5-20	Intron 10		2442	tgtctaagtaatttaatgtt	T/G	tcctatgagagctgaaggag			
i-ABCG5-21	Intron 11		4150	aaggccctgaaatggctgtt	G/T	ctggctattgttccgagctc			

Table 1. Continued

ID	Region	Exon	Position[a]	Flanking sequence[b]	Variation[c]	Flanking sequence[b]	Substitution	Repetitive sequence[d]	dbSNP/Previous report
i-ABCG5-22	Intron 11		4623	caaacagaaagaattttata	C/T	cttttgattgacagaaaata			
i-ABCG5-23	Intron 11		4737	attttcacaatgaatgttgg	T/G	tcggtctctccttccttttt			
i-ABCG5-24	Intron 11		4791	ggttagttctaactttctac	G/A	ttggtaccttcaactttctg			
i-ABCG5-25	3' Untranslated region	exon 13	2578	tgaggattaaaataaaaaac	C/T	gtaggaatgggctcaacagt			
i-ABCG5-26	3' Flanking region		1560	catagcactcagcaagaaac	G/C	tgtgctaaagactgaggttc			
i-ABCG5-27	Intron 4		1078–1080	gggcacagctccctgggagc	AGG/del	agaactcccgatagcagagt			
i-ABCG5-28	Intron 10		2321–2327	agcgggttgggtgagccctt	TAACATT/del	aggtaggtgtggtgttggct			
i-ABCG5-29	Intron 11		422–433	ggaattaagactagtcagac	(A)10~12	gcctgcaggataaaagactg			
i-ABCG5-30	Intron 11		3988–4004	ctttttttgtagtctggtcc	(T)15~17	cttttcctgttcttactctg			
i-ABCG5-31	3' Untranslated region	exon 13	2719–2731	taccctaaaacttaaagtat	(A)11~13	cctaccgaaaaaaaaaaaa			
ATP binding cassette, subfamily G, member 8 (*ABCG8*)									
i-ABCG8-1	5' Untranslated region	exon 1	-19	aagagagctgcagcccaggg	G/T	cacagacctgtgggccccat			Lu et al. (2001)
i-ABCG8-2	Intron 1		898	cctttgactgaattcgggat	A/G	tggcaggatttgaagcagga			
i-ABCG8-3	Intron 1		1548	cctcacaacctgaaaggcca	G/T	gtgtaaattgagaaattcta			
i-ABCG8-4	Intron 1		1611	tggtacgggggagccacttc	C/T	agcccgagccacaacctgtc			
i-ABCG8-5	Intron 1		3245	tgggacaatgaagcaatgtg	T/C	acagtgacagcggagagggc			
i-ABCG8-6	Intron 1		3430	gggttgaggtgggaatggaa	A/C	tctggagttctactcactgg			
i-ABCG8-7	Intron 1		3509	tacacaaatcagcttaaaga	T/A	ctctcatgtacacaccacca			
i-ABCG8-8	Intron 1		3980	gaaaataaaccctggtcaga	C/T	gcttgaggtcagcctccctc			
i-ABCG8-9	Intron 1		4123	aagggtgttctgggctcccc	G/A	taagtgtttgttgggtgcat			
i-ABCG8-10	Intron 1		5354	cagcttctaaaggagcccct	A/C	atctctcctgtctT/Cccacag			Lu et al. (2001)
i-ABCG8-11	Intron 1		5368	gcccctA/Catctctcctgtct	T/C	ccacagggcctccaggatag			Lu et al. (2001)
i-ABCG8-12	Coding region	exon 2	161	ggaggtcagagacctcaact	G/A	ccaggtagaggcacgcctgg	Cys54Tyr		Hubacek et al. (2001)
i-ABCG8-13	Intron 2		86	gaaatataaagggtgggccca	C/A	cttgcaggccctctgcccC/Gc			
i-ABCG8-14	Intron 2		105	aC/Acttgcaggccctctgccc	C/G	caaggacagagtccagtcca			
i-ABCG8-15	Intron 4		43	gacccccaggtccaagaagc	C/T	acagtgtccatgccccgctc			
i-ABCG8-16	Intron 6		1035	caggaggacaggccgcccct	C/T	gccctctgtactcacattct			
i-ABCG8-17	Intron 6		1085	cacagaaaggtcacctccct	C/A	cctgtgctcaggtggcagcc			
i-ABCG8-18	Intron 6		1184	gcacctgccgacctggccat	C/T	ggggaataatttaaagtaac			
i-ABCG8-19	Coding region	exon 8	1199	tggggcggtgcagcagttta	C/A	gacgctgatccggtaattat	Thr400Lys		Hubacek et al. (2001)
i-ABCG8-20	Intron 8		137	gaaaaaaacagcatccagca	G/A	ggcgttggtggcttatgcct		+	
i-ABCG8-21	Intron 9		412	ttctcttttccttccctta	T/C	tttttaggttactcagagag			
i-ABCG8-22	Intron 10		343	aggaagcagaggttcagaga	G/A	gctacgtggctctccaaggc		+	
i-ABCG8-23	Intron 10		614	cttttaaacgtttataataa	T/C	ggcagtgaaggtgctggctt			Lu et al. (2001)
i-ABCG8-24	Coding region	exon 11	1695	gcctccttcttcagcaatgc	C/T	ctctacaactccttctacct	Ala565Ala		Lee et al. (2001)
i-ABCG8-25	Intron 11		82	tgctttcatctggagatgga	C/T	acttatcacttagatccaac			
i-ABCG8-26	Intron 1		2882–2893	tctcttagaaatggataaga	(T)11~13	gacagagtctcacgctgtgg		+	
i-ABCG8-27	Intron 1		3654	tttatctttcccatttttt	T/del	ctgtataatttgggtcttt			
i-ABCG8-28	Intron 1		5045	tcagagcacagaggtttttt	T/del	atagaactctctccggtcca			
i-ABCG8-29	Intron 9		292–302	tggctttactgtgcctattt	(A)10~12	tgagagacttgggcaatatg			
i-ABCG8-30	Intron 9		417–418	tttccttttcccttaT/Cttttt	T/ins	aggttactcagagagggcaa			
i-ABCG8-31	Intron 10		28–34	ggcagggttgagagcaagtg	(C)7~9	acccaccagggtgggggtaa			Lu et al. (2001)
i-ABCG8-32	3' Untranslated region	exon 13	2118	tcctggggacagtgaggaca	A/del	tgaccctacagatgctcagc			

ins, Insertion polymorphism; del, deletion polymorphism; SNP, single-nucleotide polymorphism

[a] Nucleotide numbering is according to the mutation nomenclature (Dunnen and Antonarakis 2000)

[b] 5′ and 3′ sequences to each variation are denoted by small letters

[c] Variation is shown by capital letter

[d] + indicates the variation located with a repetitive sequence

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

Gene	5′ flanking region	5′ UTR[a]	Coding region	Intron	3′ UTR[b]	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
[a]5′ Untranslated region
[b]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 4374 bp 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 10.8 kb 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' half-transporters 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 98 kb 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 637 bp 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 29.1 kb 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 12.6 kb 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 N-terminal 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 head-to-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 1249 bp 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.

References

Allikmets R, Gerrard B, Hutchinson A, Dean M (1996) Characterization of the human ABC superfamily: isolation and mapping of 21 new genes using the expressed sequence tags database. Hum Mol Genet 5:1649–1655

Allikmets R, Singh N, Sun H, Shroyer NF, Hutchinson A, Chidambaram A, Gerrard B, Baird L, Stauffer D, Peiffer A, Rattner A, Smallwood P, Li Y, Anderson KL, Lewis RA, Nathans J, Leppert M, Dean M, Lupski JR (1997) A photoreceptor cell-specific ATP-binding transporter gene (*ABCR*) is mutated in recessive Stargardt macular dystrophy. Nat Genet 15:236–246

Allikmets R, Schriml LM, Hutchinson A, Romano-Spica V, Dean M (1998) A human placenta-specific ATP-binding cassette gene (*ABCP*) on chromosome 4q22 that is involved in multidrug resistance. Cancer Res 58:5337–5339

Berge KE, Tian H, Graf GA, Yu L, Grishin NV, Schultz J, Kwiterovich P, Shan B, Barnes R, Hobbs HH (2000) Accumulation of dietary cholesterol in sitosterolemia caused by mutations in adjacent ABC transporters. Science 290:1771–1775

Bisbal C, Martinand C, Silhol M, Lebleu B, Salehzada T (1995) Cloning and characterization of a RNAse L inhibitor. A new component of the interferon-regulated 2-5A pathway. J Biol Chem 270:13308–13317

Broccardo C, Luciani M, Chimini G (1999) The ABCA subclass of mammalian transporters. Biochim Biophys Acta 1461:395–404

Chen H, Rossier C, Lalioti MD, Lynn A, Chakravarti A, Perrin G, Antonarakis SE (1996) Cloning of the cDNA for a human homologue of the *Drosophila* white gene and mapping to chromosome 21q22.3. Am J Hum Genet 59:66–75

Croop JM, Tiller GE, Fletcher JA, Lux ML, Raab E, Goldenson D, Son D, Arciniegas S, Wu RL (1997) Isolation and characterization of a mammalian homolog of the *Drosophila* white gene. Gene 185:77–85

Dean M, Rzhetsky A, Allikmets R (2001) The human ATP-binding cassette (ABC) transporter superfamily. Genome Res 11:1156–1166

Dodd A, Rowland SA, Hawkes SL, Kennedy MA, Love DR (1997) Mutations in the adrenoleukodystrophy gene. Hum Mutat 9:500–511

Doyle LA, Yang W, Abruzzo LV, Krogmann T, Gao Y, Rishi AK, Ross DD (1998) A multidrug resistance transporter from human MCF-7 breast cancer cells. Proc Natl Acad Sci USA 95:15665–15670

Dunnen JT, Antonarakis SE (2000) Mutation nomenclature extensions and suggestions to describe complex mutations: a discussion. Hum Mutat 15:7–12

Dvorakova L, Storkanova G, Unterrainer G, Hujova J, Kmoch S, Zeman J, Hrebicek M, Berger J (2001) Eight novel *ABCD1* gene mutations and three polymorphisms in patients with X-linked adrenoleukodystrophy: the first polymorphism causing an amino acid exchange. Hum Mutat 18:52–60

Engel T, Lorkowski S, Lueken A, Rust S, Schluter B, Berger G, Cullen P, Assmann G (2001) The human *ABCG4* gene is regulated by oxysterols and retinoids in monocyte-derived macrophages. Biochem Biophys Res Commun 288:483–488

Floyd-Smith G, Slattery E, Lengyel P (1981) Interferon action: RNA cleavage pattern of a (2′–5′) oligoadenylate-dependent endonuclease. Science 212:1030–1032

Gartner J, Moser H, Valle D (1992) Mutations in the 70K peroxisomal membrane protein gene in Zellweger syndrome. Nat Genet 1:16–23

Gray IC, Campbell DA, Spurr NK (2000) Single nucleotide polymorphisms as tools in human genetics. Hum Mol Genet 9:2403–2408

Hassel BA, Zhou A, Sotomayor C, Maran A, Silverman RH (1993) A dominant negative mutant of 2-5A-dependent RNase suppresses antiproliferative and antiviral effects of interferon. EMBO J 12:3297–3304

Holzinger A, Kammerer S, Roscher AA (1997) Primary structure of human PMP69, a putative peroxisomal ABC-transporter. Biochem Biophys Res Commun 237:152–157

Hubacek JA, Berge KE, Cohen JC, Hobbs HH (2001) Mutations in ATP-cassette binding proteins G5 (ABCG5) and G8 (ABCG8) causing sitosterolemia. Hum Mutat 18:359–360

Iida A, Sekine A, Saito S, Kitamura Y, Kitamoto T, Osawa S, Mishima C, Nakamura Y (2001a) Catalog of 320 single nucleotide polymorphisms (SNPs) in 20 oxidoreductase and sulfotransferae genes. J Hum Genet 46:225–240

Iida A, Saito S, Sekine A, Kitamoto T, Kitamura Y, Mishima C, Osawa S, Kondo K, Harigae S, Nakamura Y (2001b) Catalog of 434 single nucleotide polymorphisms (SNPs) in the alcohol dehydrogenase genes, glutathione S transferase genes, and NADH ubiquinone oxidoreductase genes. J Hum Genet 46:385–407

Iida A, Saito S, Sekine A, Kitamura Y, Kondo K, Mishima C, Osawa S, Harigae S, Nakamura Y (2001c) High-density single-nucleotide polymorphism (SNP) map of the 150-kb region corresponding to the human ATP-binding cassette transporter A1 (*ABCA1*) gene. J Hum Genet 46:522–528

Iida A, Saito S, Sekine A, Harigae S, Osawa S, Mishima C, Kondo K, Kitamura Y, Nakamura Y (2001d) Catalog of 46 single-nucleotide polymorphisms (SNPs) in the microsomal glutathione S-transferase 1 (*MGST1*) gene. J Hum Genet 46:590–594

Iida A, Saito S, Sekine A, Mishima C, Kondo K, Kitamura Y, Harigae S, Osawa S, Nakamura Y (2001e) Catalog of 258 single-nucleotide polymorphisms (SNPs) in genes encoding three organic anion transporters, three organic anion-transporting polypeptides, and three NADH:ubiquinone oxidoreductase flavoproteins. J Hum Genet 46:668–683

Iida A, Saito S, Sekine A, Mishima C, Kitamura Y, Kondo K, Harigae S, Osawa S, Nakamura Y (2002a) Catalog of 77 single-nucleotide polymorphisms (SNPs) in the carbohydrate sulfotransferase 1 (*CHST1*) and carbohydrate sulfotransferase 3 (*CHST3*) genes. J Hum Genet 47:14–19

Iida A, Saito S, Sekine A, Kondo K, Mishima C, Kitamura Y, Harigae S, Osawa S, Nakamura Y (2002b) Thirteen single-nucleotide polymorphisms (SNPs) in the alcohol dehydrogenase 4 (*ADH4*) gene locus. J Hum Genet 47:74–76

Kaminski WE, Orso E, Diederich W, Klucken J, Drobnik W, Schmitz G (2000) Identification of a novel human sterol-sensitive ATP-binding cassette transporter (ABCA7). Biochem Biophys Res Commun 273:532–538

Kennedy MA, Venkateswaran A, Tarr PT, Xenarios I, Kudoh J, Shimizu N, Edwards PA (2001) Characterization of the human *ABCG1* gene: liver X receptor activates an internal promoter that produces a novel transcript encoding an alternative form of the protein. J Biol Chem 276:39438–39447

Klein I, Sarkadi B, Varadi A (1999) An inventory of the human ABC proteins. Biochim Biophys Acta 1461:237–262

Klucken J, Buchler C, Orso E, Kaminski WE, Porsch-Ozcurumez M, Liebisch G, Kapinsky M, Diederich W, Drobnik W, Dean M, Allikmets R, Schmitz G (2000) *ABCG1* (*ABC8*), the human homolog of the *Drosophila* white gene, is a regulator of macrophage cholesterol and phospholipid transport. Proc Natl Acad Sci USA 97:817–822

Lee MH, Lu K, Patel SB (2001) Genetic basis of sitosterolemia. Curr Opin Lipidol 12:141–149

Litman T, Brangi M, Hudson E, Fetsch P, Abati A, Ross DD, Miyake K, Resau JH, Bates SE (2000) The multidrug-resistant phenotype associated with overexpression of the new ABC half-transporter, MXR (*ABCG2*). J Cell Sci 113:2011–2021

Lu K, Lee MH, Hazard S, Brooks-Wilson A, Hidaka H, Kojima H, Ose L, Stalenhoef AF, Mietinnen T, Bjorkhem I, Bruckert E, Pandya A, Brewer HB Jr, Salen G, Dean M, Srivastava A, Patel SB (2001) Two genes that map to the STSL locus cause sitosterolemia: genomic structure and spectrum of mutations involving sterolin-1 and sterolin-2, encoded by *ABCG5* and *ABCG8*, respectively. Am J Hum Genet 69:278–290

Maugeri A, van Driel MA, van de Pol DJ, Klevering BJ, van Haren FJ, Tijmes N, Bergen AA, Rohrschneider K, Blankenagel A, Pinckers AJ, Dahl N, Brunner HG, Deutman AF, Hoyng CB, Cremers FP (1999) The 2588G→C mutation in the *ABCR* gene is a mild frequent founder mutation in the Western European population and allows the classification of *ABCR* mutations in patients with Stargardt disease. Am J Hum Genet 64:1024–1035

McCarthy JJ, Hilfiker R (2000) The use of single-nucleotide polymorphism maps in pharmacogenomics. Nat Biotechnol 18:505–508

Meyer UA (2000) Pharmacogenetics and adverse drug reactions. Lancet 356:1667–1671

Miyake K, Mickley L, Litman T, Zhan Z, Robey R, Cristensen B, Brangi M, Greenberger L, Dean M, Fojo T, Bates SE (1999) Molecular cloning of cDNAs which are highly overexpressed in mitoxantrone-resistant cells: demonstration of homology to *ABC* transport genes. Cancer Res 59:8–13

Mosser J, Douar AM, Sarde CO, Kioschis P, Feil R, Moser H, Poustka AM, Mandel JL, Aubourg P (1993) Putative X-linked adrenoleukodystrophy gene shares unexpected homology with ABC transporters. Nature 361:726–730

Nagase T, Ishikawa K, Suyama M, Kikuno R, Hirosawa M, Miyajima N, Tanaka A, Kotani H, Nomura N, Ohara O (1998) Prediction of the coding sequences of unidentified human genes. XII. The complete sequences of 100 new cDNA clones from brain which code for large proteins in vitro. DNA Res 5:355–364

Nickerson DA, Tobe VO, Taylor SL (1997) PolyPhred: automating the detection and genotyping of single nucleotide substitutions using fluorescence-based resequencing. Nucleic Acids Res 25:2745–2751

Repa JJ, Turley SD, Lobaccaro JA, Medina J, Li L, Lustig K, Shan B, Heyman RA, Dietschy JM, Mangelsdorf DJ (2000) Regulation of absorption and ABC1-mediated efflux of cholesterol by RXR heterodimers. Science 289:1524–1529

Richard M, Drouin R, Beaulieu AD (1998) ABC50, a novel human ATP-binding cassette protein found in tumor necrosis factor-alpha-stimulated synoviocytes. Genomics 53:137–145

Robey RW, Medina-Perez WY, Nishiyama K, Lahusen T, Miyake K, Litman T, Senderowicz AM, Ross DD, Bates SE (2001) Overexpression of the ATP-binding cassette half-transporter, ABCG2 (Mxr/BCrp/ABCP1), in flavopiridol-resistant human breast cancer cells. Clin Cancer Res 7:145–152

Roses AD (2000) Pharmacogenetics and the practice of medicine. Nature 405:857–865

Sachidanandam R, Weissman D, Schmidt SC, Kakol JM, Stein LD, Marth G, Sherry S, Mullikin JC, Mortimore BJ, Willey DL, Hunt SE, Cole CG, Coggill PC, Rice CM, Ning Z, Rogers J, Bentley DR, Kwok PY, Mardis ER, Yeh RT, Schultz B, Cook L, Davenport R, Dante M, Fulton L, Hillier L, Waterston RH, McPherson JD, Gilman B, Schaffner S, Van Etten WJ, Reich D, Higgins J, Daly MJ, Blumenstiel B, Baldwin J, Stange-Thomann N, Zody MC, Linton L, Lander ES, Altshuler D (2001) A map of human genome sequence variation containing 1.42 million single nucleotide polymorphisms. Nature 409:928–933

Saito S, Iida A, Sekine A, Eguchi C, Miura Y, Nakamura Y (2001a) Seventy genetic variations in human microsomal and soluble epoxide hydrolase genes (*EPHX1* and *EPHX2*) in the Japanese population. J Hum Genet 46:325–329

Saito S, Iida A, Sekine A, Miura Y, Sakamoto T, Ogawa C, Kawauchi S, Higuchi S, Nakamura Y (2001b) Identification of 197 genetic variations in six human methyltranferase genes in the Japanese population. J Hum Genet 46:529–537

Saito S, Iida A, Sekine A, Miura Y, Ogawa C, Kawauchi S, Higuchi S, Nakamura Y (2002a) Three hundred twenty-six genetic variations in genes encoding nine members of ATP-binding cassette, subfamily B (ABCB/MDR/TAP), in the Japanese population. J Hum Genet 47:38–50

Saito S, Iida A, Sekine A, Miura Y, Ogawa C, Kawauchi S, Higuchi S, Nakamura Y (2002b) Identification of 779 genetic variations in eight genes encoding members of the ATP-binding cassette, subfamily C (ABCC/MRP/CFTR). J Hum Genet 47:147–171

Sekine A, Saito S, Iida A, Mitsunobu Y, Higuchi S, Harigae S, Nakamura Y (2001) Identification of single-nucleotide polymorphisms (SNPs) of human N-acetyltransferase genes *NAT1*, *NAT2*, *AANAT*, *ARD1* and *L1CAM* in the Japanese population. J Hum Genet 46:314–319

Shani N, Jimenez-Sanchez G, Steel G, Dean M, Valle D (1997) Identification of a fourth half ABC transporter in the human peroxisomal membrane. Hum Mol Genet 6:1925–1931

Tyzack JK, Wang X, Belsham GJ, Proud CG (2000) ABC50 interacts with eukaryotic initiation factor 2 and associates with the ribosome in an ATP-dependent manner. J Biol Chem 275:34131–34139

Venkateswaran A, Repa JJ, Lobaccaro JM, Bronson A, Mangelsdorf DJ, Edwards PA (2000) Human white/murine ABC8 mRNA levels are highly induced in lipid-loaded macrophages. A transcriptional role for specific oxysterols. J Biol Chem 275:14700–14707

Williams BR (1991) Transcriptional regulation of interferon-stimulated genes. Eur J Biochem 200:1–11

Zhou A, Hassel BA, Silverman RH (1993) Expression cloning of 2-5A-dependent RNase: a uniquely regulated mediator of interferon action. Cell 72:753–765