

Variant report grouped by medical condition

Dermatitis, atopic, 6, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
SPINK5	rs2303067	risk factor	missense variant(NM_006846.3:c.1258A>G)	0/1	A>G

Microvascular complications of diabetes 6

Gene	rs#	Significance	Reference	GenoType	Detected Change
SOD2	rs4880	risk factor	missense variant(NM_001024465.2:c.47T>C)	0/1	A>G

Uric acid nephrolithiasis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
ZNF365	rs7076156	risk factor	missense variant(NM_199452.3:c.184G>A)	1/1	A>G

Multiple sclerosis, susceptibility to, 5

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNFRSF1A	rs1800693	risk factor	intron variant(NM_001065.3:c.625+10A>G)	0/1	T>C

Microvascular complications of diabetes 5

Gene	rs#	Significance	Reference	GenoType	Detected Change
PON1	rs854560	risk factor	missense variant(NM_000446.5:c.163T>A)	1/1	A>T

Frontotemporal dementia

Gene	rs#	Significance	Reference	GenoType	Detected Change
GRN	rs5848	risk factor	3 prime UTR variant(NM_002087.3:c.*78C>T)	0/1	C>T

Fatal familial insomnia

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Myofibrillar myopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

Immunodeficiency 20

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR3A	rs10127939	Pathogenic	missense variant(NM_0011275.93.1:c.197T>A)	0/1	A>T
FCGR3A	rs396991	drug response	missense variant(NM_0011275.93.1:c.526T>G)	0/1	A>C

nevirapine response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Cortisone reductase deficiency 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
H6PD	rs12032814	Likely pathogenic	intron variant(NM_004285.3:c.745+88T>A)	0/1	T>A

Waardenburg syndrome 2 and ocular albinism, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

Exfoliation syndrome, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
LOXL1-AS1	rs1048661	risk factor	missense variant(NM_005576.3:c.422G>T)	0/1	G>T

Breast adenocarcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

High density lipoprotein cholesterol level quantitative trait locus 13

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA1	rs2230806	protective	missense variant(NM_005502.3:c.656G>A)	0/1	C>T

Glaucoma, congenital

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104.3:c.1294C=)	0/1	C>G

Lymphoproliferative disorders, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
EPHX1	rs2234922	drug response	missense variant(NM_0011360.18.3:c.416A>G)	0/1	A>G

EPHX1	rs1051740	risk factor	missense variant(NM_0011360 18.3:c.337T>C)	0/1	T>C
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Abetalipoproteinaemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTTP	rs3816873	protective	missense variant(NM_000253. 3:c.383T>C)	0/1	T>C

Septic shock, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594. 3:c.-488G>A)	0/1	G>A

Thyroid-associated orbitopathy, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
CTLA4	rs231775	risk factor	missense variant(NM_005214. 4:c.49A>G)	0/1	A>G

No MEN2 disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975. 4:c.135G>A)	1/1	A>G

Irido-corneo-trabecular dysgenesis

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104. 3:c.1294C=)	0/1	C>G

Warfarin response

Gene	rs#	Significance	Reference	GenoType	Detected Change

VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T
VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

Stickler syndrome, type 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
COL11A1	rs1676486	risk factor	non-coding transcript variant(NR_134980.1:n.4937T>C)	0/1	A>G

Neural tube defects, folate-sensitive, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTRR	rs1801394	drug response	non-coding transcript variant(NR_134480.1:n.203A>G)	0/1	A>G
MTHFD1	rs2236225	risk factor	missense variant(NM_005956.3:c.1958G>A)	1/1	G>A

risperidone response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510.1:c.2137G>A)	0/1	G>A

Systemic lupus erythematosus, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A
FCGR2B	rs1050501	risk factor	missense variant(NM_004001.4:c.695T>C)	0/1	T>C

CTLA4	rs231775	risk factor	missense variant(NM_005214.4:c.49A>G)	0/1	A>G
PTPN22	rs2476601	risk factor	missense variant(NM_015967.6:c.1858C>T)	1/1	A>G

phenprocoumon response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

Hyperglycinuria

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLC6A20	rs17279437	Pathogenic	missense variant(NM_020208.3:c.596C>T)	0/1	G>A

Hereditary lymphedema type I

Gene	rs#	Significance	Reference	GenoType	Detected Change
FLT4	rs34255532	Pathogenic	missense variant(NM_182925.4:c.2860C>T)	0/1	G>A

Serum amyloid a variant

Gene	rs#	Significance	Reference	GenoType	Detected Change
SAA1	rs1136743	Pathogenic	missense variant(NM_000331.5:c.209C>T)	0/1	C>T

opioids response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change

ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Blau syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOD2	rs5743289	risk factor	intron variant(NM_022162.2:c.2798+158C>T)	0/1	C>T

INFLAMMATORY BOWEL DISEASE 1 (CROHN DISEASE), SUSCEPTIBILITY TO

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOD2	rs5743289	risk factor	intron variant(NM_022162.2:c.2798+158C>T)	0/1	C>T

Wolfram-like syndrome, autosomal dominant

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005.3:c.461-9A>G)	1/1	A>G

Diabetes mellitus, noninsulin-dependent, association with

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005.3:c.461-9A>G)	1/1	A>G

Carcinoma of cervix

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Transferrin variant d1

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T

Sarcoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Polysubstance abuse, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
FAAH	rs324420	risk factor	missense variant(NM_001441.2:c.385C>A)	0/1	C>A

Proline dehydrogenase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRODH	rs450046	risk factor	missense variant(NM_016335.4:c.1562A>G)	1/1	C>T

Carnitine palmitoyltransferase II deficiency, infantile

Gene	rs#	Significance	Reference	GenoType	Detected Change
CPT2	rs1799821	risk factor	missense variant(NM_000098.2:c.1102G>A)	1/1	G>A

Low density lipoprotein cholesterol level quantitative trait locus 6

Gene	rs#	Significance	Reference	GenoType	Detected Change
SORT1	rs12740374	association	N/A	0/1	G>T

Cockayne syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G

Leprosy, early-onset, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
LTA	rs1041981	risk factor	missense variant(NM_001159740.2:c.179C>A)	0/1	C>A

Bietti crystalline corneoretinal dystrophy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP4V2	rs1055138	Pathogenic	missense variant(NM_207352.3:c.64C>G)	0/1	C>G

Thrombophilia due to activated protein C resistance

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

Vitamin b12 plasma level quantitative trait locus 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
FUT2	rs601338	protective	nonsense(NM_000511.5:c.461G>A)	1/1	G>A

Transferrin variant bv

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T

Factor xiii, a subunit, deficiency of

Gene	rs#	Significance	Reference	GenoType	Detected Change
F13A1	rs2815822	Pathogenic	intron variant(NM_000129.3:c.-19+12A=)	1/1	T>G
F13A1	rs5985	protective	missense variant(NM_000129.3:c.103G>T)	0/1	C>A

Atrial septal defect 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
GATA4	rs3729856	Likely pathogenic	missense variant(NM_002052.4:c.1129A>G)	0/1	A>G

Primary dilated cardiomyopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

antineoplastic agents response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Plasminogen activator inhibitor type 1 deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
SERPINE1	rs6092	Pathogenic	missense variant(NM_000602.4:c.43G>A)	0/1	G>A

nicotine response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change

CHRNA3	rs1051730	risk factor	synonymous variant(NM_0011666 94.1:c.645C>T)	0/1	G>A
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Tobacco addiction, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
GABBR2	rs3750344	risk factor	synonymous variant(NM_005458. 7:c.360A>G)	0/1	T>C
GABBR2	rs2184026	risk factor	intron variant(NM_005458. 7:c.460-23G=)	1/1	C>T

Hypertension, pregnancy-induced, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOS3	rs1799983	risk factor	missense variant(NM_000603. 4:c.894T>G)	0/1	T>G

nortriptyline response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106. 5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106. 5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106. 5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106. 5:c.1457G>C)	0/1	G>C

methadone response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927. 4:c.3435T>C)	0/1	A>G
CYP2B6	rs2279345	drug response	intron variant(NM_000767. 4:c.823-197T>A)	0/1	T>C

CYP2B6	rs4803419	drug response	intron variant(NM_000767.4:c.485-18C>T)	0/1	C>T
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Chromophobe renal cell carcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

Gerstmann-Straussler-Scheinker syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Primary progressive aphasia

Gene	rs#	Significance	Reference	GenoType	Detected Change
GRN	rs5848	risk factor	3 prime UTR variant(NM_002087.3:c.*78C>T)	0/1	C>T

Blood group, Dombrock system

Gene	rs#	Significance	Reference	GenoType	Detected Change
ART4	rs11276	Affects	missense variant(NM_021071.2:c.793G>A)	0/1	C>T

Pseudoxanthoma elasticum

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCC6	rs2238472	Pathogenic	missense variant(NM_001171.5:c.3803G>A)	0/1	C>T

Adrenocortical carcinoma, pediatric

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Netherton syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
SPINK5	rs2303067	risk factor	missense variant(NM_006846.3:c.1258A>G)	0/1	A>G

Tangier disease, variant

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA1	rs2230806	protective	missense variant(NM_005502.3:c.656G>A)	0/1	C>T

Malignant lymphoma, non-Hodgkin

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Neuroferritinopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
FTL	rs2230267	Likely pathogenic	synonymous variant(NM_000146.3:c.163T>C)	1/1	T>C

Platelet-type bleeding disorder 16

Gene	rs#	Significance	Reference	GenoType	Detected Change
ITGA2B	rs5911	association	missense variant(NM_000419.4:c.2621T>G)	1/1	A>C

Sandhoff disease, adult type

Gene	rs#	Significance	Reference	GenoType	Detected Change
HEXB	rs820878	Pathogenic	missense variant(NM_000521.3:c.185C>T)	1/1	T>C

Sepsis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASP12	rs497116	risk factor	nonsense(NM_001191016.2:c.373C>T)	1/1	G>A

Clopidogrel response

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

fluorouracil, leucovorin, and oxaliplatin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTHFR	rs1801131	risk factor	missense variant(NM_005957.4:c.1286A>C)	0/1	T>G
MTHFR	rs1801133	drug response	missense variant(NM_005957.4:c.665C>T)	0/1	G>A

warfarin response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

**hormonal contraceptives for systemic use response -
Toxicity/ADR**

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130. 4:c.1601G>A)	1/1	T>C

tegafur response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
DPYD	rs1801265	Pathogenic	missense variant(NM_000110. 3:c.85T>C)	1/1	G>A

celecoxib response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771. 3:c.430C=)	0/1	C>T

cisplatin response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546. 5:c.215C>G)	0/1	G>C
ERCC1	rs11615	drug response	synonymous variant(NM_001983. 3:c.354T>C)	0/1	A>G
XRCC1	rs25487	drug response	missense variant(NM_006297. 2:c.1196A>G)	0/1	T>C

Metachromatic leukodystrophy, mild

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487. 5:c.1178C>G)	1/1	G>C

Preeclampsia/eclampsia 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
STOX1	rs1341667	risk factor	missense variant(NM_0011301 61.2:c.457T>C)	0/1	T>C
STOX1	rs10509305	Pathogenic	missense variant(NM_0011301 61.2:c.1824A>C)	0/1	A>C

Diabetes mellitus type 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
IRS1	rs1801278	risk factor	missense variant(NM_005544. 2:c.2911G>A)	0/1	C>T
KCNJ11	rs5219	risk factor	missense variant(NM_000525. 3:c.67A>G)	0/1	T>C
ENPP1	rs1044498	risk factor	missense variant(NM_006208. 2:c.517A>C)	0/1	A>C

Diabetes mellitus type 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545. 6:c.1720G>A)	1/1	A>G

Migraine without aura, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594. 3:c.-488G>A)	0/1	G>A

Hypocalciuric hypercalcemia, familial, type 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388. 3:c.2956G>T)	0/1	G>T

Plasminogen deficiency, type I

Gene	rs#	Significance	Reference	GenoType	Detected Change
PLG	rs73015965	Pathogenic	missense variant(NM_000301.3:c.112A>G)	0/1	A>G

BAK PLATELET-SPECIFIC ANTIGEN

Gene	rs#	Significance	Reference	GenoType	Detected Change
ITGA2B	rs5911	association	missense variant(NM_000419.4:c.2621T>G)	1/1	A>C

adalimumab response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Aphasia, primary progressive, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Proguanil, poor metabolism of

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

Bernard-Soulier syndrome, type A2, autosomal dominant

Gene	rs#	Significance	Reference	GenoType	Detected Change
GP1BA	rs6065	drug response	missense variant(NM_000173.6:c.482C>T)	0/1	C>T

Maple syrup urine disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
DBT	rs12021720	Pathogenic	synonymous variant(NM_001918.3:c.1150G=)	1/1	T>C

infliximab response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Budd-Chiari syndrome, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

Age-related macular degeneration 12

Gene	rs#	Significance	Reference	GenoType	Detected Change
CX3CR1	rs3732379	Pathogenic	missense variant(NM_0011711 74.1:c.841G>A)	0/1	C>T

Arylsulfatase A pseudodeficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

nicotine response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
COMT	rs4680	drug response	missense variant(NM_000754.3:c.472G>A)	0/1	G>A

SUDDEN INFANT DEATH SYNDROME

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

Carnitine palmitoyltransferase II deficiency, late-onset

Gene	rs#	Significance	Reference	GenoType	Detected Change
CPT2	rs1799821	risk factor	missense variant(NM_000098. 2:c.1102G>A)	1/1	G>A

Efavirenz response

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2B6	rs2279345	drug response	intron variant(NM_000767. 4:c.823-197T>A)	0/1	T>C
CYP2B6	rs4803419	drug response	intron variant(NM_000767. 4:c.485-18C>T)	0/1	C>T

Down syndrome, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTRR	rs1801394	drug response	non-coding transcript variant(NR_134480.1 :n.203A>G)	0/1	A>G

TNF receptor-associated periodic fever syndrome (TRAPS)

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNFRSF1A	rs1800693	risk factor	intron variant(NM_001065. 3:c.625+10A>G)	0/1	T>C

Prostate cancer, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
RNASEL	rs486907	risk factor	missense variant(NM_021133. 3:c.1385G>A)	0/1	C>T

Maple syrup urine disease, thiamine-responsive, type II

Gene	rs#	Significance	Reference	GenoType	Detected Change
DBT	rs12021720	Pathogenic	synonymous variant(NM_001918.3:c.1150G=)	1/1	T>C

Dopamine receptor d2, reduced brain density of

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510.1:c.2137G>A)	0/1	G>A

Muscle AMP deaminase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
AMPD1	rs17602729	Pathogenic	nonsense(NM_000036.2:c.133C>T)	0/1	G>A

Febrile seizures, familial, 3a

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

MEN2A and Unclassified

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Severe combined immunodeficiency disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL7R	rs1494558	Pathogenic	non-coding transcript variant(NR_120485.1:n.300T>C)	1/1	T>C

IL7R	rs1494555	Pathogenic	non-coding transcript variant(NR_120485.1 :n.515G>A)	1/1	G>A
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Rheumatoid arthritis

Gene	rs#	Significance	Reference	GenoType	Detected Change
PTPN22	rs2476601	risk factor	missense variant(NM_015967.6:c.1858C>T)	1/1	A>G

Brachydactyly type A2

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_0013191 38.1:c.-241-34T>C)	0/1	A>G

cyclophosphamide response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
SOD2	rs4880	risk factor	missense variant(NM_0010244 65.2:c.47T>C)	0/1	A>G

Scapuloperoneal syndrome, neurogenic, Kaeser type

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

Adenoma, cortisol-producing

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP21A2	rs6467	Pathogenic	intron variant(NM_000500.7:c.293-13C>G)	0/1	C>A

Malaria, severe, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change

FCGR2A	rs1801274	risk factor	missense variant(NM_021642.3:c.497A>G)	0/1	A>G
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salbutamol response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB2	rs1042713	drug response	synonymous variant(NM_000024.5:c.46A=)	0/1	G>A
ADRB2	rs1042714	risk factor	missense variant(NM_000024.5:c.79C>G)	0/1	G>C

Addison disease, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
PTPN22	rs2476601	risk factor	missense variant(NM_015967.6:c.1858C>T)	1/1	A>G

Leprosy 5

Gene	rs#	Significance	Reference	GenoType	Detected Change
TLR1	rs5743618	protective	missense variant(NM_003263.3:c.1805G>T)	0/1	C>A
TLR1	rs4833095	risk factor	missense variant(NM_003263.3:c.743A>G)	0/1	T>C

Alzheimer disease, late-onset, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOS3	rs1799983	risk factor	missense variant(NM_000603.4:c.894T>G)	0/1	T>G
C10orf55	rs2227564	risk factor	missense variant(NM_002658.4:c.422C>T)	0/1	T>C

**cyclophosphamide and epirubicin response - Efficacy,
Toxicity/ADR**

Gene	rs#	Significance	Reference	GenoType	Detected Change
GSTP1	rs1695	drug response	missense variant(NM_000852.3:c.313A>G)	0/1	A>G

Norwalk virus infection, resistance to

Gene	rs#	Significance	Reference	GenoType	Detected Change
FUT2	rs601338	protective	nonsense(NM_000511.5:c.461G>A)	1/1	G>A

Trimethylaminuria, mild

Gene	rs#	Significance	Reference	GenoType	Detected Change
FMO3	rs2266782	Likely pathogenic	missense variant(NM_001002294.2:c.472G>A)	0/1	G>A

Enzyme activity finding

Gene	rs#	Significance	Reference	GenoType	Detected Change
PON1	rs854560	risk factor	missense variant(NM_000446.5:c.163T>A)	1/1	A>T

WFS1-Related Disorders

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005.3:c.461-9A>G)	1/1	A>G

bupropion response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510.1:c.2137G>A)	0/1	G>A

Mild non-PKU hyperphenylalanemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
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PAH	rs5030860	Pathogenic	missense variant(NM_000277.1:c.1241A>G)	0/1	T>C
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Hemangioma, capillary infantile

Gene	rs#	Significance	Reference	GenoType	Detected Change
FLT4	rs34255532	Pathogenic	missense variant(NM_182925.4:c.2860C>T)	0/1	G>A

Glutaric aciduria, type 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
GCDH	rs9384	Likely pathogenic	non-coding transcript variant(NR_102316.1 :n.1768G>T)	0/1	G>T
GCDH	rs8012	Likely pathogenic	non-coding transcript variant(NR_102316.1 :n.1645A>G)	0/1	A>G

Coronary artery disease, resistance to

Gene	rs#	Significance	Reference	GenoType	Detected Change
CX3CR1	rs3732379	Pathogenic	missense variant(NM_0011711 74.1:c.841G>A)	0/1	C>T

Metachromatic leukodystrophy, adult type

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

CEREBRAL AMYLOID ANGIOPATHY, PRNP-RELATED

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Dilated cardiomyopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

rosuvastatin response - Other

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446.4:c.521T>C)	0/1	T>C

simvastatin response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446.4:c.521T>C)	0/1	T>C

salmeterol response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB2	rs1042713	drug response	synonymous variant(NM_000024.5:c.46A=)	0/1	G>A
ADRB2	rs1042714	risk factor	missense variant(NM_000024.5:c.79C>G)	0/1	G>C

Familial hypocalciuric hypercalcemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388.3:c.2956G>T)	0/1	G>T

High density lipoprotein cholesterol level quantitative trait locus 10

Gene	rs#	Significance	Reference	GenoType	Detected Change
CETP	rs5882	association	missense variant(NM_000078.2:c.1264G>A)	1/1	G>A

Hyperferritinemia cataract syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
FTL	rs2230267	Likely pathogenic	synonymous variant(NM_000146.3:c.163T>C)	1/1	T>C

Central hypoventilation syndrome, congenital, with hirschsprung disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Arylsulfatase a pseudodeficiency, severe

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Acquired immunodeficiency syndrome, slow progression to

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL4R	rs1801275	risk factor	missense variant(NM_000418.3:c.1727A>G)	0/1	A>G

Stargardt disease 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350.2:c.2828G>A)	0/1	C>T

Congenital long QT syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

Neoplasm of stomach

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546. 5:c.215C>G)	0/1	G>C

Diabetes mellitus, insulin-dependent, 5

Gene	rs#	Significance	Reference	GenoType	Detected Change
TAB2	rs237025	risk factor	missense variant(NM_0010022 55.1:c.163G>A)	0/1	G>A

Long QT syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

Hyperphenylalaninemia, non-pku

Gene	rs#	Significance	Reference	GenoType	Detected Change
PAH	rs5030860	Pathogenic	missense variant(NM_000277. 1:c.1241A>G)	0/1	T>C

Ischemic heart disease, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOS3	rs1799983	risk factor	missense variant(NM_000603. 4:c.894T>G)	0/1	T>G

Calcium oxalate urolithiasis

Gene	rs#	Significance	Reference	GenoType	Detected Change
AHSG	rs2070635	association	intron variant(NM_001622.2:c.574-149A>G)	0/1	A>G
CD44	rs7116432	association	intron variant(NM_001001391.1:c.881+779A>G)	0/1	A>G
AHSG	rs4918	risk factor	missense variant(NM_001622.2:c.767G>C)	1/1	G>C
AHSG	rs4917	risk factor	missense variant(NM_001622.2:c.743T>C)	1/1	T>C

Ventricular septal defect 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
GATA4	rs3729856	Likely pathogenic	missense variant(NM_002052.4:c.1129A>G)	0/1	A>G

VON HIPPEL-LINDAU SYNDROME, MODIFIER OF

Gene	rs#	Significance	Reference	GenoType	Detected Change
CCND1	rs9344	risk factor	synonymous variant(NM_053056.2:c.723G>A)	0/1	G>A

antidepressants response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Alzheimer disease, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A
A2M	rs669	risk factor	missense variant(NM_000014.4:c.2998A>G)	0/1	T>C

Deafness, autosomal recessive 77

Gene	rs#	Significance	Reference	GenoType	Detected Change
LOXHD1	rs1450425	Likely pathogenic	intron variant(NM_144612.6:c.4530+107A>G)	1/1	T>C

citalopram response - Metabolism/PK

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

Bardet-Biedl syndrome 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
BBS2	rs4784677	Pathogenic	missense variant(NM_031885.3:c.209A>G)	1/1	C>T

cyclophosphamide response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Asthma-related traits, susceptibility to, 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
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NPSR1	rs324981	risk factor	missense variant(NM_207173.1:c.320A>T)	0/1	A>T
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L-ferritin deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
FTL	rs2230267	Likely pathogenic	synonymous variant(NM_000146.3:c.163T>C)	1/1	T>C

Cataplexy and narcolepsy

Gene	rs#	Significance	Reference	GenoType	Detected Change
PPAN	rs1551570	association	intron variant(NM_020230.5:c.190-151C>T)	0/1	C>T
PPAN-P2RY11	rs12460842	association	2KB upstream variant(NM_002566.4:c.-203A>G)	0/1	A>G
EIF3G	rs2305795	association	3 prime UTR variant(NM_002566.4:c.*638G>A)	0/1	G>A

Osteoarthritis susceptibility 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
FRZB	rs7775	risk factor	missense variant(NM_001463.3:c.970C>G)	0/1	G>C

fluorouracil response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Malignant tumor of esophagus

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

cerivastatin response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446.4:c.521T>C)	0/1	T>C

Asthma, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
SPINK5	rs2303067	risk factor	missense variant(NM_006846.3:c.1258A>G)	0/1	A>G
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A
IL13	rs20541	risk factor	missense variant(NM_002188.2:c.431A>G)	0/1	A>G

acenocoumarol response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

Canavan disease, mild

Gene	rs#	Significance	Reference	GenoType	Detected Change
SPATA22	rs12948217	Pathogenic	nonsense(NM_001128085.1:c.693C>A)	0/1	C>T

**MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA,
WITH HIRSCHSPRUNG DISEASE**

Gene	rs#	Significance	Reference	GenoType	Detected Change
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RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G
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Brachydactyly, type a1, c

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_0013191 38.1:c.-241-34T>C)	0/1	A>G

Myofibrillar myopathy 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

Atransferrinemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T

Atrioventricular septal defect 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
GATA4	rs3729856	Likely pathogenic	missense variant(NM_002052.4:c.1129A>G)	0/1	A>G

Xeroderma pigmentosum, group G

Gene	rs#	Significance	Reference	GenoType	Detected Change
BIVM-ERCC5	rs9514067	Likely pathogenic	nonsense(NM_000123.3:c.3238C>T)	1/1	G>C

Deficiency of xanthine oxidase

Gene	rs#	Significance	Reference	GenoType	Detected Change
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XDH	rs1366813	Likely pathogenic	intron variant(NM_000379.3:c.3276+12A>G)	1/1	T>C
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Xeroderma pigmentosum, group C

Gene	rs#	Significance	Reference	GenoType	Detected Change
XPC	rs2228001	drug response	non-coding transcript variant(NR_027299.1:n.2795C>A)	0/1	G>T

Permanent neonatal diabetes mellitus

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C

Neonatal severe hyperparathyroidism

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388.3:c.2956G>T)	0/1	G>T

efavirenz response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2B6	rs2279345	drug response	intron variant(NM_000767.4:c.823-197T>A)	0/1	T>C
CYP2B6	rs4803419	drug response	intron variant(NM_000767.4:c.485-18C>T)	0/1	C>T

Cole disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
ENPP1	rs1044498	risk factor	missense variant(NM_006208.2:c.517A>C)	0/1	A>C

Parkinson disease, mitochondrial

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADH1C	rs283413	risk factor	nonsense(NM_00066 9.4:c.232G>T)	1/1	A>C

**DFNA6/14/38 Nonsyndromic Low-Frequency
Sensorineural Hearing Loss**

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005. 3:c.461-9A>G)	1/1	A>G

cyclophosphamide response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTHFR	rs1801131	risk factor	missense variant(NM_005957. 4:c.1286A>C)	0/1	T>G
MTHFR	rs1801133	drug response	missense variant(NM_005957. 4:c.665C>T)	0/1	G>A

clopidogrel response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769. 2:c.681G>A)	0/1	G>A

Diabetes mellitus

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525. 3:c.67A>G)	0/1	T>C

Myopathy, variable

Gene	rs#	Significance	Reference	GenoType	Detected Change
CPT2	rs1799821	risk factor	missense variant(NM_000098. 2:c.1102G>A)	1/1	G>A

tramadol response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

clomipramine response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

Glaucoma, early-onset, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104.3:c.1294C=)	0/1	C>G

hmg coa reductase inhibitors response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CETP	rs5882	association	missense variant(NM_000078.2:c.1264G>A)	1/1	G>A

Brachydactyly type C

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_001319138.1:c.-241-34T>C)	0/1	A>G

Asthma-related traits, susceptibility to, 7

Gene	rs#	Significance	Reference	GenoType	Detected Change

CHI3L1	rs4950928	risk factor	5 prime UTR variant(NM_001276.2:c.-131C>G)	0/1	G>C
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Familial hypercholesterolemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
GHR	rs6180	risk factor	missense variant(NM_000163.4:c.1630A>C)	1/1	A>C
LDLR	rs2228671	Pathogenic	nonsense(NM_000527.4:c.81C>A)	0/1	C>T

hmg coa reductase inhibitors response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446.4:c.521T>C)	0/1	T>C

Skin/hair/eye pigmentation, variation in, 5

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLC45A2	rs16891982	association	missense variant(NM_016180.4:c.1122C>G)	1/1	C>G

Alzheimer disease, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Skin/hair/eye pigmentation, variation in, 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

Sandhoff disease, chronic

Gene	rs#	Significance	Reference	GenoType	Detected Change
HEXB	rs820878	Pathogenic	missense variant(NM_000521.3:c.185C>T)	1/1	T>C

Metachromatic leukodystrophy, juvenile type

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Deficiency of steroid 11-beta-monoxygenase

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP11B1	rs61752786	Likely pathogenic	missense variant(NM_000497.3:c.1120C>T)	0/1	G>T

paclitaxel response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104.3:c.1294C=)	0/1	C>G

Diaphyseal dysplasia

Gene	rs#	Significance	Reference	GenoType	Detected Change
TGFB1	rs1800470	risk factor	missense variant(NM_000660.6:c.29C>T)	0/1	G>A

Familial hypoalphalipoproteinemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA1	rs2230806	protective	missense variant(NM_005502.3:c.656G>A)	0/1	C>T

Cone-rod dystrophy 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350.2:c.2828G>A)	0/1	C>T

Spina bifida, folate-sensitive, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTHFD1	rs2236225	risk factor	missense variant(NM_005956.3:c.1958G>A)	1/1	G>A

cisplatin response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G
XPC	rs2228001	drug response	non-coding transcript variant(NR_027299.1:n.2795C>A)	0/1	G>T

Factor xiii, b subunit, deficiency of

Gene	rs#	Significance	Reference	GenoType	Detected Change
F13B	rs6003	risk factor	missense variant(NM_001994.2:c.344G>A)	1/1	C>T

clomipramine response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Asthma, nocturnal, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB2	rs1042713	drug response	synonymous variant(NM_000024.5:c.46A=)	0/1	G>A
ADRB2	rs1042714	risk factor	missense variant(NM_000024.5:c.79C>G)	0/1	G>C

Tyrosinase-negative oculocutaneous albinism

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

simvastatin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Lactase persistence

Gene	rs#	Significance	Reference	GenoType	Detected Change
MCM6	rs182549	association	intron variant(NM_005915.5:c.1362+117G>A)	1/1	C>T

Xeroderma pigmentosum, type 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
XPC	rs2228001	drug response	non-coding transcript variant(NR_027299.1:n.2795C>A)	0/1	G>T

trimipramine response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Seizures

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

Congenital heart defects, multiple types, 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
TAB2	rs237025	risk factor	missense variant(NM_0010022.55.1:c.163G>A)	0/1	G>A

Cutaneous malignant melanoma 8

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

aspirin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
GP1BA	rs6065	drug response	missense variant(NM_000173.6:c.482C>T)	0/1	C>T

Cutaneous malignant melanoma 6

Gene	rs#	Significance	Reference	GenoType	Detected Change
XRCC3	rs861539	risk factor	missense variant(NM_0011001 19.1:c.722C>T)	0/1	G>A

Myocardial infarction

Gene	rs#	Significance	Reference	GenoType	Detected Change
LTA	rs1041981	risk factor	missense variant(NM_0011597 40.2:c.179C>A)	0/1	C>A

Intermediate maple syrup urine disease type 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
DBT	rs12021720	Pathogenic	synonymous variant(NM_001918.3:c.1150G=)	1/1	T>C

Metachromatic leukodystrophy, severe

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Factor V deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

Retinitis pigmentosa 19

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350.2:c.2828G>A)	0/1	C>T

Malaria, resistance to

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR2B	rs1050501	risk factor	missense variant(NM_004001.4:c.695T>C)	0/1	T>C

Severe combined immunodeficiency, autosomal recessive, T cell-negative, B cell-positive, NK cell-positive

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL7R	rs1494558	Pathogenic	non-coding transcript variant(NR_120485.1 :n.300T>C)	1/1	T>C
IL7R	rs1494555	Pathogenic	non-coding transcript variant(NR_120485.1 :n.515G>A)	1/1	G>A

Atopy, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
SPINK5	rs2303067	risk factor	missense variant(NM_006846.3:c.1258A>G)	0/1	A>G
IL4R	rs1801275	risk factor	missense variant(NM_000418.3:c.1727A>G)	0/1	A>G

Congenital adrenal hyperplasia

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP11B1	rs61752786	Likely pathogenic	missense variant(NM_000497.3:c.1120C>T)	0/1	G>T

Glioma susceptibility 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia

Gene	rs#	Significance	Reference	GenoType	Detected Change
TPP1	rs56144125	Pathogenic	splice acceptor variant(NM_000391.3:c.509-1G>A)	0/1	C>T

Prion disease, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Autoimmune lymphoproliferative syndrome, type V

Gene	rs#	Significance	Reference	GenoType	Detected Change
CTLA4	rs231775	risk factor	missense variant(NM_005214.4:c.49A>G)	0/1	A>G

Neuronal ceroid lipofuscinosis

Gene	rs#	Significance	Reference	GenoType	Detected Change
TPP1	rs56144125	Pathogenic	splice acceptor variant(NM_000391.3:c.509-1G>A)	0/1	C>T

Sandhoff disease, infantile

Gene	rs#	Significance	Reference	GenoType	Detected Change
HEXB	rs820878	Pathogenic	missense variant(NM_000521.3:c.185C>T)	1/1	T>C

warfarin response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

Hypertension, salt-sensitive essential, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADD1	rs4961	drug response	missense variant(NM_176801.2:c.1378G>T)	0/1	G>T

paclitaxel response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Lumbar disc disease, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
CILP	rs2073711	risk factor	missense variant(NM_003613.3:c.1184T>C)	0/1	A>G

Soluble interleukin-6 receptor, serum level of, quantitative trait locus

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL6R	rs2228145	association	missense variant(NM_000565.3:c.1073A>C)	0/1	A>C

diclofenac response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

Transferrin variant c1/c2

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T

Pseudofolliculitis barbae, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
KRT75	rs2232387	risk factor	missense variant(NM_004693.2:c.481G>A)	0/1	C>T

Spongy degeneration of central nervous system

Gene	rs#	Significance	Reference	GenoType	Detected Change
ASPA	rs12948217	Pathogenic	nonsense(NM_001128085.1:c.693C>A)	0/1	C>T

Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR2A	rs1801274	risk factor	missense variant(NM_021642.3:c.497A>G)	0/1	A>G

Marshall syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
COL11A1	rs1676486	risk factor	non-coding transcript variant(NR_134980.1:n.4937T>C)	0/1	A>G

Dihydropyrimidine dehydrogenase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
DPYD	rs1801265	Pathogenic	missense variant(NM_000110.3:c.85T>C)	1/1	G>A

sporadic abdominal aortic aneurysm

Gene	rs#	Significance	Reference	GenoType	Detected Change
FTL	rs2230267	Likely pathogenic	synonymous variant(NM_000146.3:c.163T>C)	1/1	T>C
COL11A1	rs1676486	risk factor	non-coding transcript variant(NR_134980.1:n.4937T>C)	0/1	A>G

Laron syndrome with elevated serum GH-binding protein

Gene	rs#	Significance	Reference	GenoType	Detected Change
GHR	rs6180	risk factor	missense variant(NM_000163.4:c.1630A>C)	1/1	A>C

Atypical hemolytic-uremic syndrome 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
CFH	rs800292	risk factor	missense variant(NM_000186.3:c.184G>A)	0/1	G>A
CFH	rs1061170	Pathogenic	synonymous variant(NM_000186.3:c.1204C=)	1/1	C>T
CFH	rs2274700	risk factor	synonymous variant(NM_000186.3:c.1419G>C)	0/1	G>A

rituximab response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR3A	rs10127939	Pathogenic	missense variant(NM_0011275 93.1:c.197T>A)	0/1	A>T
FCGR3A	rs396991	drug response	missense variant(NM_0011275 93.1:c.526T>G)	0/1	A>C

Debrisoquine, poor metabolism of

Gene	rs#	Significance	Reference	GenoType	Detected Change
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CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

TNF receptor binding, altered

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Hypocalcemia, autosomal dominant 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388.3:c.2956G>T)	0/1	G>T

fluorouracil and oxaliplatin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
GSTP1	rs1695	drug response	missense variant(NM_000852.3:c.313A>G)	0/1	A>G

carboplatin response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
XRCC1	rs25487	drug response	missense variant(NM_006297.2:c.1196A>G)	0/1	T>C
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G

Hypoparathyroidism familial isolated

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388.3:c.2956G>T)	0/1	G>T

Retinal dystrophy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350.2:c.2828G>A)	0/1	C>T

Corticosterone methyloxidase type 2 deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP11B2	rs61757294	Pathogenic	missense variant(NM_000498.3:c.1157T>C)	0/1	A>G

pravastatin response - Metabolism/PK

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446.4:c.521T>C)	0/1	T>C

furosemide and spironolactone response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADD1	rs4961	drug response	missense variant(NM_176801.2:c.1378G>T)	0/1	G>T

Marshall/Stickler syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
COL11A1	rs1676486	risk factor	non-coding transcript variant(NR_134980.1:n.4937T>C)	0/1	A>G

**APOLIPOPROTEIN A-IV POLYMORPHISM,
APOA4*1/APOA4*2**

Gene	rs#	Significance	Reference	GenoType	Detected Change
APOA4	rs5110	Pathogenic	missense variant(NM_000482.3:c.1140G>T)	0/1	C>A

Platinum compounds response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
GSTP1	rs1695	drug response	missense variant(NM_000852.3:c.313A>G)	0/1	A>G
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G

Xeroderma pigmentosum group g/Cockayne syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
BIVM-ERCC5	rs9514067	Likely pathogenic	nonsense(NM_000123.3:c.3238C>T)	1/1	G>C

olanzapine response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510.1:c.2137G>A)	0/1	G>A

Transient neonatal diabetes mellitus 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C

Kuru, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Long QT syndrome 2, acquired, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

Stargardt's disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350. 2:c.2828G>A)	0/1	C>T

Colorectal cancer, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
CCND1	rs9344	risk factor	synonymous variant(NM_053056. 2:c.723G>A)	0/1	G>A

Li-Fraumeni-like syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546. 5:c.215C>G)	0/1	G>C

Multiple synostoses syndrome 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_0013191 38.1:c.-241-34T>C)	0/1	A>G

clozapine response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510. 1:c.2137G>A)	0/1	G>A

**Antiinflammatory agents, non-steroids response -
Toxicity/ADR**

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771. 3:c.430C=)	0/1	C>T

pravastatin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446. 4:c.521T>C)	0/1	T>C

Hyperlipidemia, familial combined, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
USF1	rs3737787	risk factor	3 prime UTR variant(NM_007122. 4:c.*187C>T)	1/1	G>A
USF1	rs2073658	risk factor	intron variant(NM_007122. 4:c.561-100G>A)	1/1	C>T

Essential tremor, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
DRD3	rs6280	risk factor	missense variant(NM_000796. 5:c.25G>A)	0/1	C>T

Coronary artery disease, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
IRS1	rs1801278	risk factor	missense variant(NM_005544. 2:c.2911G>A)	0/1	C>T
PON1	rs854560	risk factor	missense variant(NM_000446. 5:c.163T>A)	1/1	A>T

amitriptyline response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Asthma and atopy, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
PLA2G7	rs1051931	risk factor	missense variant(NM_0011683.57.1:c.1136T>C)	1/1	A>G
PLA2G7	rs1805018	risk factor	missense variant(NM_0011683.57.1:c.593T>C)	0/1	A>G

Jakob-Creutzfeldt disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Hashimoto thyroiditis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
AP4B1-AS1	rs2476601	risk factor	missense variant(NM_015967.6:c.1858C>T)	1/1	A>G
CTLA4	rs231775	risk factor	missense variant(NM_005214.4:c.49A>G)	0/1	A>G

Transferrin variant b2

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T

Fasting plasma glucose level quantitative trait locus 5

Gene	rs#	Significance	Reference	GenoType	Detected Change
GCKR	rs1260326	association	missense variant(NM_001486.3:c.1337T>C)	0/1	T>C

Hypertriglyceridemia, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
APOA5	rs2266788	risk factor	3 prime UTR variant(NM_052968.4:c.*158C>T)	1/1	G>A

fluorouracil response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
DPYD	rs1801265	Pathogenic	missense variant(NM_000110.3:c.85T>C)	1/1	G>A

Vitamin k-dependent clotting factors, combined deficiency of, 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

Glycogen storage disease type II, infantile

Gene	rs#	Significance	Reference	GenoType	Detected Change
GAA	rs2278619	Likely pathogenic	intron variant(NM_000152.4:c.1327-18A>G)	1/1	A>G

platinum response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G

Nasopharyngeal carcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Familial medullary thyroid carcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Schizophrenia 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRODH	rs450046	risk factor	missense variant(NM_016335.4:c.1562A>G)	1/1	C>T

Metachromatic leukodystrophy, late-onset

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Cancer progression and tumor cell motility

Gene	rs#	Significance	Reference	GenoType	Detected Change
FGFR4	rs351855	Pathogenic	missense variant(NM_213647.2:c.1162G>A)	0/1	G>A

Vascular dementia, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Unclassified

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Iminoglycinuria, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLC6A20	rs17279437	Pathogenic	missense variant(NM_020208.3:c.596C>T)	0/1	G>A

Hypercalciuric hypercalcemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388.3:c.2956G>T)	0/1	G>T

SERUM HDL CHOLESTEROL LEVEL, MODIFIER OF

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

warfarin response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

Li-Fraumeni syndrome 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Long QT syndrome 2/5

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Basal laminar drusen

Gene	rs#	Significance	Reference	GenoType	Detected Change
CFH	rs800292	risk factor	missense variant(NM_000186.3:c.184G>A)	0/1	G>A
CFH	rs1061170	Pathogenic	synonymous variant(NM_000186.3:c.1204C=)	1/1	C>T
CFH	rs2274700	risk factor	synonymous variant(NM_000186.3:c.1419G>C)	0/1	G>A

Familial colorectal cancer

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Alcoholism, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
GABRA2	rs279871	risk factor	intron variant(NM_000807.2:c.704-104A>G)	1/1	T>C

Pseudoxanthoma elasticum, forme fruste

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCC6	rs2238472	Pathogenic	missense variant(NM_001171.5:c.3803G>A)	0/1	C>T

Long QT syndrome 2/9, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Early infantile epileptic encephalopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

Skin/hair/eye pigmentation 3, blue/green eyes

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

Maple syrup urine disease type 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
DBT	rs12021720	Pathogenic	synonymous variant(NM_001918.3:c.1150G=)	1/1	T>C

Muscular dystrophy, limb-girdle, type 2r

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

Psoriatic arthritis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A
LTA	rs1041981	risk factor	missense variant(NM_001159740.2:c.179C>A)	0/1	C>A

Exercise stress response, impaired, association with

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C

Glanzmann's thrombasthenia

Gene	rs#	Significance	Reference	GenoType	Detected Change
ITGA2B	rs5911	association	missense variant(NM_000419.4:c.2621T>G)	1/1	A>C

Bull's eye maculopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350.2:c.2828G>A)	0/1	C>T

Wolfram syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005.3:c.461-9A>G)	1/1	A>G

Breast cancer, invasive, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change

TGFB1	rs1800470	risk factor	missense variant(NM_000660. 6:c.29C>T)	0/1	G>A
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Li-Fraumeni syndrome 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546. 5:c.215C>G)	0/1	G>C

Celiac disease 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
CTLA4	rs231775	risk factor	missense variant(NM_005214. 4:c.49A>G)	0/1	A>G

carbamazepine response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
EPHX1	rs2234922	drug response	missense variant(NM_0011360 18.3:c.416A>G)	0/1	A>G
EPHX1	rs1051740	risk factor	missense variant(NM_0011360 18.3:c.337T>C)	0/1	T>C
SCN1A	rs3812718	risk factor	intron variant(NM_006920. 4:c.603-91G>A)	1/1	C>T

ethanol response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510. 1:c.2137G>A)	0/1	G>A

Glycogen storage disease, type II

Gene	rs#	Significance	Reference	GenoType	Detected Change
GAA	rs2278619	Likely pathogenic	intron variant(NM_000152. 4:c.1327-18A>G)	1/1	A>G

platinum response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
XRCC1	rs25487	drug response	missense variant(NM_006297. 2:c.1196A>G)	0/1	T>C
ERCC1	rs11615	drug response	synonymous variant(NM_001983. 3:c.354T>C)	0/1	A>G

Genetic prion diseases

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311. 3:c.385A>G)	0/1	A>G

**MACULAR DEGENERATION, AGE-RELATED, 2,
SUSCEPTIBILITY TO**

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350. 2:c.2828G>A)	0/1	C>T

Bernard-Soulier syndrome, type A1

Gene	rs#	Significance	Reference	GenoType	Detected Change
GP1BA	rs6065	drug response	missense variant(NM_000173. 6:c.482C>T)	0/1	C>T

antipsychotics response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ANKK1	rs1800497	drug response	missense variant(NM_178510. 1:c.2137G>A)	0/1	G>A

carboplatin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTHFR	rs1801131	risk factor	missense variant(NM_005957.4:c.1286A>C)	0/1	T>G
MTHFR	rs1801133	drug response	missense variant(NM_005957.4:c.665C>T)	0/1	G>A

Bone mineral density quantitative trait locus 15

Gene	rs#	Significance	Reference	GenoType	Detected Change
CALCR	rs1801197	risk factor	missense variant(NM_0011647.37.1:c.1442T>C)	0/1	A>G

Inborn genetic diseases

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G
GAA	rs2278619	Likely pathogenic	intron variant(NM_000152.4:c.1327-18A>G)	1/1	A>G
MTRR	rs1801394	drug response	non-coding transcript variant(NR_134480.1:n.203A>G)	0/1	A>G
CYP21A2	rs6467	Pathogenic	intron variant(NM_000500.7:c.293-13C>G)	0/1	C>A
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T
LDLR	rs2228671	Pathogenic	nonsense(NM_00052.7.4:c.81C>A)	0/1	C>T
TPP1	rs56144125	Pathogenic	splice acceptor variant(NM_000391.3:c.509-1G>A)	0/1	C>T

ondansetron response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.343T>C)	0/1	A>G

ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C
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Porokeratosis 9, multiple types

Gene	rs#	Significance	Reference	GenoType	Detected Change
FDPS	rs2297480	drug response	intron variant(NM_002004.3:c.-1-98T>G)	0/1	T>G

Maturity-onset diabetes of the young, type 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

clopidogrel response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

21-hydroxylase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP21A2	rs6467	Pathogenic	intron variant(NM_000500.7:c.293-13C>G)	0/1	C>A

Multiple endocrine neoplasia, type 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Beta-2-adrenoreceptor agonist, reduced response to

Gene	rs#	Significance	Reference	GenoType	Detected Change
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ADRB2	rs1042713	drug response	synonymous variant(NM_000024.5:c.46A=)	0/1	G>A
ADRB2	rs1042714	risk factor	missense variant(NM_000024.5:c.79C>G)	0/1	G>C

Basal cell carcinoma, susceptibility to, 7

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Multiple endocrine neoplasia, type 2b

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Multiple endocrine neoplasia, type 2a

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Prekallikrein deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
KLKB1	rs3733402	Pathogenic	missense variant(NM_000892.4:c.428G>A)	0/1	G>A

Thyroid cancer, anaplastic

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

fentanyl response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Acquired long QT syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Clear cell carcinoma of kidney

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

metformin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLC47A2	rs12943590	drug response	5 prime UTR variant(NM_001099646.1:c.-130C>T)	1/1	G>A

Short QT syndrome 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Homocysteinemia due to MTHFR deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTHFR	rs1801131	risk factor	missense variant(NM_005957.4:c.1286A>C)	0/1	T>G

MTHFR	rs1801133	drug response	missense variant(NM_005957.4:c.665C>T)	0/1	G>A
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amitriptyline response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

Carnitine palmitoyltransferase II deficiency, lethal neonatal

Gene	rs#	Significance	Reference	GenoType	Detected Change
CPT2	rs1799821	risk factor	missense variant(NM_000098.2:c.1102G>A)	1/1	G>A

Dysplasminogenemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
PLG	rs73015965	Pathogenic	missense variant(NM_000301.3:c.112A>G)	0/1	A>G

Bardet-biedl syndrome 2/6, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
BBS2	rs4784677	Pathogenic	missense variant(NM_031885.3:c.209A>G)	1/1	C>T

Phenylthiocarbamide tasting

Gene	rs#	Significance	Reference	GenoType	Detected Change
TAS2R38	rs713598	drug response	missense variant(NM_176817.4:c.145G>C)	0/1	C>G
TAS2R38	rs10246939	drug response	missense variant(NM_176817.4:c.886A>G)	0/1	T>C

TAS2R38	rs1726866	drug response	missense variant(NM_176817.4:c.785T>C)	0/1	G>A
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Susceptibility to progression to renal failure in IgA nephropathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
AGT	rs699	risk factor	missense variant(NM_000029.3:c.803T>C)	0/1	A>G

clopidogrel response - Dosage, Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

FRONTOTEMPORAL LOBAR DEGENERATION WITH UBIQUITIN-POSITIVE INCLUSIONS, SUSCEPTIBILITY TO

Gene	rs#	Significance	Reference	GenoType	Detected Change
GRN	rs5848	risk factor	3 prime UTR variant(NM_002087.3:c.*78C>T)	0/1	C>T

escitalopram response - Metabolism/PK

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

Hypertension resistant to conventional therapy

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOS3	rs1799983	risk factor	missense variant(NM_000603.4:c.894T>G)	0/1	T>G

Diabetes mellitus AND insipidus with optic atrophy AND deafness

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005. 3:c.461-9A>G)	1/1	A>G

citalopram response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769. 2:c.681G>A)	0/1	G>A

Adenocarcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546. 5:c.215C>G)	0/1	G>C

Crohn disease, association with

Gene	rs#	Significance	Reference	GenoType	Detected Change
AGT	rs699	risk factor	missense variant(NM_000029. 3:c.803T>C)	0/1	A>G

Atopy, resistance to

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL4R	rs1801275	risk factor	missense variant(NM_000418. 3:c.1727A>G)	0/1	A>G

Serum calcium level

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388. 3:c.2956G>T)	0/1	G>T

nevirapine response - Other

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2B6	rs2279345	drug response	intron variant(NM_000767.4:c.823-197T>A)	0/1	T>C
CYP2B6	rs4803419	drug response	intron variant(NM_000767.4:c.485-18C>T)	0/1	C>T

Methylmalonic aciduria cblB type

Gene	rs#	Significance	Reference	GenoType	Detected Change
MMAB	rs9593	Pathogenic	non-coding transcript variant(NR_038118.1:n.876T>A)	0/1	A>T

Sacral agenesis with vertebral anomalies

Gene	rs#	Significance	Reference	GenoType	Detected Change
T	rs3127334	risk factor	intron variant(NM_003181.3:c.1034+79C>T)	1/1	G>A

Venous thrombosis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
F13B	rs6003	risk factor	missense variant(NM_001994.2:c.344G>A)	1/1	C>T

Colchicine resistance

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Maturity-onset diabetes of the young, type 13

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C

Hirschsprung disease 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

methotrexate response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
MTRR	rs1801394	drug response	non-coding transcript variant(NR_134480.1:n.203A>G)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Congestive heart failure and beta-blocker response, modifier of

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB1	rs1801253	drug response	missense variant(NM_000684.2:c.1165G>C)	1/1	G>C

Skin/hair/eye pigmentation, variation in, 10

Gene	rs#	Significance	Reference	GenoType	Detected Change
TPCN2	rs3829241	association	missense variant(NM_139075.3:c.2201G>A)	0/1	G>A
TPCN2	rs35264875	association	missense variant(NM_139075.3:c.1450A>T)	0/1	A>T

Rotor syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLCO1B1	rs4149056	drug response	missense variant(NM_006446.4:c.521T>C)	0/1	T>C

Prostate cancer, hereditary, 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
RNASEL	rs486907	risk factor	missense variant(NM_021133.3:c.1385G>A)	0/1	C>T

Carnitine palmitoyltransferase II deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
CPT2	rs1799821	risk factor	missense variant(NM_000098.2:c.1102G>A)	1/1	G>A

Metachromatic leukodystrophy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Sandhoff disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
HEXB	rs820878	Pathogenic	missense variant(NM_000521.3:c.185C>T)	1/1	T>C

acenocoumarol response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

Primary open angle glaucoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104.3:c.1294C=)	0/1	C>G

Obesity, association with

Gene	rs#	Significance	Reference	GenoType	Detected Change
SDC3	rs2491132	association	missense variant(NM_014654.3:c.622G>A)	0/1	C>T

Familial type 5 hyperlipoproteinemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
APOA5	rs2266788	risk factor	3 prime UTR variant(NM_052968.4:c.*158C>T)	1/1	G>A

Prostate cancer, hereditary, 13

Gene	rs#	Significance	Reference	GenoType	Detected Change
MSMB	rs10993994	Pathogenic	2KB upstream variant(NM_002443.3:c.-89T=)	0/1	T>C

Spastic paraparesis 75, autosomal recessive

Gene	rs#	Significance	Reference	GenoType	Detected Change
MAG	rs2301600	Pathogenic	missense variant(NM_080600.2:c.399C>G)	0/1	C>T

desipramine response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A

CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Bardet-biedl syndrome 1/2, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
BBS2	rs4784677	Pathogenic	missense variant(NM_031885.3:c.209A>G)	1/1	C>T

Glaucoma, primary open angle, juvenile-onset

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104.3:c.1294C=)	0/1	C>G

Brugada syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

tenofovir response -

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCC4	rs1751034	drug response	synonymous variant(NM_005845.4:c.3348G>A)	1/1	C>T

Hypophosphatemic rickets, autosomal recessive, 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
ENPP1	rs1044498	risk factor	missense variant(NM_006208.2:c.517A>C)	0/1	A>C

Inflammatory bowel disease 10, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
ATG16L1	rs2241880	risk factor	missense variant(NM_030803. 6:c.898A>G)	0/1	A>G

Tetralogy of Fallot

Gene	rs#	Significance	Reference	GenoType	Detected Change
GATA4	rs3729856	Likely pathogenic	missense variant(NM_002052. 4:c.1129A>G)	0/1	A>G
JAG1	rs1131695	Pathogenic	nonsense(NM_00021 4.2:c.765C>G)	0/1	G>A

anthracyclines and related substances response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
HAS3	rs2232228	drug response	synonymous variant(NM_0011992 80.1:c.279A>G)	0/1	A>G

imipramine response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106. 5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106. 5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106. 5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106. 5:c.1457G>C)	0/1	G>C

Hereditary cancer-predisposing syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
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RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Osteoarthritis of hip

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_0013191 38.1:c.-241-34T>C)	0/1	A>G

Venous thrombosis, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
F13A1	rs2815822	Pathogenic	intron variant(NM_000129.3:c.-19+12A=)	1/1	T>G
F13A1	rs5985	protective	missense variant(NM_000129.3:c.103G>T)	0/1	C>A

Deafness, congenital heart defects, and posterior embryotoxon

Gene	rs#	Significance	Reference	GenoType	Detected Change
JAG1	rs1131695	Pathogenic	nonsense(NM_000214.2:c.765C>G)	0/1	G>A

Pheochromocytoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

docetaxel response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP1B1	rs1056836	drug response	synonymous variant(NM_000104.3:c.1294C=)	0/1	C>G

Oculocutaneous albinism type 1, temperature sensitive

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

Hyperalphalipoproteinemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
CETP	rs5882	association	missense variant(NM_000078.2:c.1264G>A)	1/1	G>A

Malaria, cerebral, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Coronary heart disease in familial hypercholesterolemia, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA1	rs2230806	protective	missense variant(NM_005502.3:c.656G>A)	0/1	C>T

Dilated cardiomyopathy 1I

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

Sandhoff disease, infantile type

Gene	rs#	Significance	Reference	GenoType	Detected Change

HEXB	rs820878	Pathogenic	missense variant(NM_000521.3:c.185C>T)	1/1	T>C
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Sandhoff disease, juvenile type

Gene	rs#	Significance	Reference	GenoType	Detected Change
HEXB	rs820878	Pathogenic	missense variant(NM_000521.3:c.185C>T)	1/1	T>C

Bardet-biedl syndrome 2/4, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
BBS2	rs4784677	Pathogenic	missense variant(NM_031885.3:c.209A>G)	1/1	C>T

Oculocutaneous albinism type 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

Oculocutaneous albinism type 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLC45A2	rs16891982	association	missense variant(NM_016180.4:c.1122C>G)	1/1	C>G

4-Hydroxyphenylpyruvate dioxygenase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
HPD	rs1154510	Pathogenic	missense variant(NM_002150.2:c.97G>A)	0/1	T>C

Homocystinuria due to MTHFR deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change

MTHFR	rs1801131	risk factor	missense variant(NM_005957.4:c.1286A>C)	0/1	T>G
MTHFR	rs1801133	drug response	missense variant(NM_005957.4:c.665C>T)	0/1	G>A

Retinitis pigmentosa 74

Gene	rs#	Significance	Reference	GenoType	Detected Change
BBS2	rs4784677	Pathogenic	missense variant(NM_031885.3:c.209A>G)	1/1	C>T

MEN2 phenotype: Unclassified

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Obesity

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB2	rs1042713	drug response	synonymous variant(NM_000024.5:c.46A=)	0/1	G>A
ADRB3	rs4994	risk factor	missense variant(NM_000025.2:c.190T>C)	0/1	A>G
ADRB2	rs1042714	risk factor	missense variant(NM_000024.5:c.79C>G)	0/1	G>C
ENPP1	rs1044498	risk factor	missense variant(NM_006208.2:c.517A>C)	0/1	A>C

Tumor necrosis factor alpha (TNF-alpha) inhibitors response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Recurrent abortion

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

Osteosarcoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Primary familial hypertrophic cardiomyopathy

Gene	rs#	Significance	Reference	GenoType	Detected Change
DES	rs41272699	Pathogenic	missense variant(NM_001927.3:c.638C>T)	0/1	C>T

Fibular hypoplasia and complex brachydactyly

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_001319138.1:c.-241-34T>C)	0/1	A>G

Bisphosphonates response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
FDPS	rs2297480	drug response	intron variant(NM_002004.3:c.-1-98T>G)	0/1	T>G

Neonatal diabetes mellitus

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C



Thyroid carcinoma, sporadic medullary

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Cardiovascular phenotype

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Frontotemporal dementia, ubiquitin-positive

Gene	rs#	Significance	Reference	GenoType	Detected Change
GRN	rs5848	risk factor	3 prime UTR variant(NM_002087.3:c.*78C>T)	0/1	C>T

Interleukin 6, serum level of, quantitative trait locus

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL6R	rs2228145	association	missense variant(NM_000565.3:c.1073A>C)	0/1	A>C

Platinum compounds response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G
XRCC1	rs25487	drug response	missense variant(NM_006297.2:c.1196A>G)	0/1	T>C

Aerodigestive tract cancer, squamous cell, alcohol-related, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change

ADH1B	rs1229984	protective	synonymous variant(NM_000668.5:c.143A=)	1/1	T>C
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Diabetes mellitus, insulin-dependent, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
AP4B1-AS1	rs2476601	risk factor	missense variant(NM_015967.6:c.1858C>T)	1/1	A>G
CTLA4	rs231775	risk factor	missense variant(NM_005214.4:c.49A>G)	0/1	A>G

etanercept response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Diabetes mellitus, insulin-dependent, 20

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

Breast cancer, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
XRCC3	rs861539	risk factor	missense variant(NM_0011001.19.1:c.722C>T)	0/1	G>A

Phenytoin response

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

Cerebrooculofacioskeletal syndrome 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G

Hypercholanemia, familial

Gene	rs#	Significance	Reference	GenoType	Detected Change
EPHX1	rs2234922	drug response	missense variant(NM_0011360.18.3:c.416A>G)	0/1	A>G
EPHX1	rs1051740	risk factor	missense variant(NM_0011360.18.3:c.337T>C)	0/1	T>C

Hypertension, essential, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
AGT	rs699	risk factor	missense variant(NM_000029.3:c.803T>C)	0/1	A>G

efavirenz response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2B6	rs2279345	drug response	intron variant(NM_000767.4:c.823-197T>A)	0/1	T>C
CYP2B6	rs4803419	drug response	intron variant(NM_000767.4:c.485-18C>T)	0/1	C>T

Laron-type isolated somatotropin defect

Gene	rs#	Significance	Reference	GenoType	Detected Change
GHR	rs6180	risk factor	missense variant(NM_000163.4:c.1630A>C)	1/1	A>C

Hepatoblastoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
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TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C
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Alagille syndrome 1

Gene	rs#	Significance	Reference	GenoType	Detected Change
JAG1	rs1131695	Pathogenic	nonsense(NM_000214.2:c.765C>G)	0/1	G>A

oxazepam response - Other

Gene	rs#	Significance	Reference	GenoType	Detected Change
UGT2B15	rs1902023	drug response	missense variant(NM_001076.3:c.253T>G)	1/1	A>C

Choroid plexus carcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

L-ferritin deficiency, autosomal recessive

Gene	rs#	Significance	Reference	GenoType	Detected Change
FTL	rs2230267	Likely pathogenic	synonymous variant(NM_000146.3:c.163T>C)	1/1	T>C

phenytoin response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

Fibrochondrogenesis

Gene	rs#	Significance	Reference	GenoType	Detected Change

COL11A1	rs1676486	risk factor	non-coding transcript variant(NR_134980.1 :n.4937T>C)	0/1	A>G
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Debrisoquine, ultrarapid metabolism of

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

morphine response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Carcinoma of pancreas

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Emery-Dreifuss muscular dystrophy 5, autosomal dominant

Gene	rs#	Significance	Reference	GenoType	Detected Change
SYNE2	rs36215895	Pathogenic	missense variant(NM_182914.2:c.18632C>T)	0/1	C>T

Smoking as a quantitative trait locus 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
CHRNA5	rs16969968	risk factor	missense variant(NM_000745.3:c.1192G>A)	0/1	G>A
CHRNA3	rs1051730	risk factor	synonymous variant(NM_0011666 94.1:c.645C>T)	0/1	G>A

digoxin response - Other

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

Human immunodeficiency virus dementia, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
TNF	rs1800629	drug response	2KB upstream variant(NM_000594.3:c.-488G>A)	0/1	G>A

Myocardial infarction, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
F13A1	rs2815822	Pathogenic	intron variant(NM_000129.3:c.-19+12A=)	1/1	T>G
F13A1	rs5985	protective	missense variant(NM_000129.3:c.103G>T)	0/1	C>A

Long QT syndrome 2/3, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Lung cancer susceptibility 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
CHRNA5	rs16969968	risk factor	missense variant(NM_000745.3:c.1192G>A)	0/1	G>A
CHRNA3	rs1051730	risk factor	synonymous variant(NM_0011666 94.1:c.645C>T)	0/1	G>A

Mephenytoin, poor metabolism of

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C19	rs4244285	drug response	synonymous variant(NM_000769.2:c.681G>A)	0/1	G>A

oxycodone response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCB1	rs1045642	drug response	synonymous variant(NM_000927.4:c.3435T>C)	0/1	A>G
ABCB1	rs2032582	drug response	missense variant(NM_000927.4:c.2677T>G)	1/1	A>C

efavirenz response - Metabolism/PK

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2B6	rs2279345	drug response	intron variant(NM_000767.4:c.823-197T>A)	0/1	T>C
CYP2B6	rs4803419	drug response	intron variant(NM_000767.4:c.485-18C>T)	0/1	C>T

Trimethylaminuria

Gene	rs#	Significance	Reference	GenoType	Detected Change
FMO3	rs2266782	Likely pathogenic	missense variant(NM_0010022 94.2:c.472G>A)	0/1	G>A

Age-related macular degeneration 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
CFH	rs800292	risk factor	missense variant(NM_000186.3:c.184G>A)	0/1	G>A
CFH	rs1061170	Pathogenic	synonymous variant(NM_000186.3:c.1204C=)	1/1	C>T
CFH	rs2274700	risk factor	synonymous variant(NM_000186.3:c.1419G>C)	0/1	G>A

Sarcoidosis, early-onset

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOD2	rs5743289	risk factor	intron variant(NM_022162.2:c.2798+158C>T)	0/1	C>T

Cardiac arrhythmia

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Familial hemiplegic migraine type 3

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

Neural tube defects, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
T	rs3127334	risk factor	intron variant(NM_003181.3:c.1034+79C>T)	1/1	G>A

Resting heart rate

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB1	rs1801253	drug response	missense variant(NM_000684.2:c.1165G>C)	1/1	G>C

cetuximab response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR3A	rs10127939	Pathogenic	missense variant(NM_0011275 93.1:c.197T>A)	0/1	A>T
FCGR3A	rs396991	drug response	missense variant(NM_0011275 93.1:c.526T>G)	0/1	A>C

Neonatal insulin-dependent diabetes mellitus

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C

Hepatitis b virus, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
IFNAR2	rs2229207	risk factor	missense variant(NM_207585.2:c.23T>C)	0/1	T>C

4-Alpha-hydroxyphenylpyruvate hydroxylase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
HPD	rs1154510	Pathogenic	missense variant(NM_002150.2:c.97G>A)	0/1	T>C

Hereditary pancreatitis

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRSS1	rs144422014	Pathogenic	missense variant(NM_002769.4:c.161A>G)	0/1	A>G

PRSS1	rs202003805	Pathogenic	missense variant(NM_002769.4:c.47C>T)	0/1	C>T
PRSS1	rs111033566	Pathogenic	missense variant(NM_002769.4:c.86A>T)	0/1	A>T

Congenital central hypoventilation

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Age-related macular degeneration 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350.2:c.2828G>A)	0/1	C>T

Metabolic syndrome, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB2	rs1042713	drug response	synonymous variant(NM_000024.5:c.46A=)	0/1	G>A
ADRB2	rs1042714	risk factor	missense variant(NM_000024.5:c.79C>G)	0/1	G>C

Grebe syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_0013191.38.1:c.-241-34T>C)	0/1	A>G

trastuzumab response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR3A	rs10127939	Pathogenic	missense variant(NM_0011275.93.1:c.197T>A)	0/1	A>T

FCGR3A	rs396991	drug response	missense variant(NM_0011275 93.1:c.526T>G)	0/1	A>C
FCGR2A	rs1801274	risk factor	missense variant(NM_021642. 3:c.497A>G)	0/1	A>G

Coronary artery spasm 2, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
PON1	rs854560	risk factor	missense variant(NM_000446. 5:c.163T>A)	1/1	A>T

Retinal dystrophy, early-onset severe

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA4	rs1801581	Pathogenic	missense variant(NM_000350. 2:c.2828G>A)	0/1	C>T

Hypocalcemia, autosomal dominant 1, with bartter syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388. 3:c.2956G>T)	0/1	G>T

Metachromatic leukodystrophy, late infantile

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487. 5:c.1178C>G)	1/1	G>C

Long QT syndrome, bradycardia-induced

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

Transferrin variant chi

Gene	rs#	Significance	Reference	GenoType	Detected Change
TF	rs1049296	association	missense variant(NM_001063.3:c.1765C>T)	0/1	C>T

doxepin response - Dosage, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106.5:c.100C>T)	0/1	G>A
CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Hepatocellular carcinoma

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Pseudo von Willebrand disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
GP1BA	rs6065	drug response	missense variant(NM_000173.6:c.482C>T)	0/1	C>T

Oculocutaneous albinism type 1B

Gene	rs#	Significance	Reference	GenoType	Detected Change
TYR	rs1126809	association	missense variant(NM_000372.4:c.1205G>A)	1/1	G>A

Short stature, idiopathic, autosomal

Gene	rs#	Significance	Reference	GenoType	Detected Change
GHR	rs6180	risk factor	missense variant(NM_000163.4:c.1630A>C)	1/1	A>C

Multiple endocrine neoplasia IIA

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Arylsulfatase a pseudodeficiency, intermediate

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Emphysema, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
EPHX1	rs2234922	drug response	missense variant(NM_0011360 18.3:c.416A>G)	0/1	A>G
EPHX1	rs1051740	risk factor	missense variant(NM_0011360 18.3:c.337T>C)	0/1	T>C

Spongiform encephalopathy with neuropsychiatric features

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

warfarin response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change

VKORC1	rs9934438	drug response	intron variant(NM_024006.5:c.174-136C>T)	0/1	G>A
VKORC1	rs8050894	drug response	intron variant(NM_024006.5:c.283+124G>C)	0/1	C>G
VKORC1	rs17708472	drug response	intron variant(NM_024006.5:c.173+525C>T)	1/1	G>A

Sudden cardiac death

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

Pulmonary disease, chronic obstructive, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
EPHX1	rs2234922	drug response	missense variant(NM_0011360 18.3:c.416A>G)	0/1	A>G
EPHX1	rs1051740	risk factor	missense variant(NM_0011360 18.3:c.337T>C)	0/1	T>C

Testicular anomalies with or without congenital heart disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
GATA4	rs3729856	Likely pathogenic	missense variant(NM_002052.4:c.1129A>G)	0/1	A>G

FMTc and Unclassified

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Encephalopathy, acute, infection-induced, 4, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
CPT2	rs1799821	risk factor	missense variant(NM_000098.2:c.1102G>A)	1/1	G>A

Factor H deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
CFH	rs800292	risk factor	missense variant(NM_000186.3:c.184G>A)	0/1	G>A
CFH	rs1061170	Pathogenic	synonymous variant(NM_000186.3:c.1204C=)	1/1	C>T
CFH	rs2274700	risk factor	synonymous variant(NM_000186.3:c.1419G>C)	0/1	G>A

Thrombophilia due to factor V Leiden

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

FMO3 activity, decreased

Gene	rs#	Significance	Reference	GenoType	Detected Change
FMO3	rs2266782	Likely pathogenic	missense variant(NM_0010022.94.2:c.472G>A)	0/1	G>A

Insulin resistance, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
IRS1	rs1801278	risk factor	missense variant(NM_005544.2:c.2911G>A)	0/1	C>T
ENPP1	rs1044498	risk factor	missense variant(NM_006208.2:c.517A>C)	0/1	A>C
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

MEN2A and FMTc

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Symphalangism, proximal, 1b

Gene	rs#	Significance	Reference	GenoType	Detected Change
GDF5	rs143383	risk factor	intron variant(NM_001319138.1:c.-241-34T>C)	0/1	A>G

Corticosterone methyloxidase type 1 deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP11B2	rs61757294	Pathogenic	missense variant(NM_000498.3:c.1157T>C)	0/1	A>G

Long QT syndrome 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Schizophrenia, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTHFR	rs1801131	risk factor	missense variant(NM_005957.4:c.1286A>C)	0/1	T>G
COMT	rs4680	drug response	missense variant(NM_000754.3:c.472G>A)	0/1	G>A
MTHFR	rs1801133	drug response	missense variant(NM_005957.4:c.665C>T)	0/1	G>A
DRD3	rs6280	risk factor	missense variant(NM_000796.5:c.25G>A)	0/1	C>T

Tolbutamide response

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771. 3:c.430C=)	0/1	C>T

Asthma, childhood, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADRB2	rs1042713	drug response	synonymous variant(NM_000024. 5:c.46A=)	0/1	G>A
ADRB2	rs1042714	risk factor	missense variant(NM_000024. 5:c.79C>G)	0/1	G>C

oxaliplatin response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
XRCC1	rs25487	drug response	missense variant(NM_006297. 2:c.1196A>G)	0/1	T>C
ERCC1	rs11615	drug response	synonymous variant(NM_001983. 3:c.354T>C)	0/1	A>G

Slow acetylator due to N-acetyltransferase enzyme variant

Gene	rs#	Significance	Reference	GenoType	Detected Change
NAT2	rs1801280	drug response	missense variant(NM_000015. 2:c.341T>C)	0/1	T>C
NAT2	rs1208	drug response	synonymous variant(NM_000015. 2:c.803G=)	0/1	G>A

tamoxifen response - Efficacy, Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2D6	rs1065852	drug response	missense variant(NM_000106. 5:c.100C>T)	0/1	G>A

CYP2D6	rs16947	drug response	missense variant(NM_000106.5:c.886C>T)	1/1	A>G
CYP2D6	rs3892097	drug response	splice acceptor variant(NM_000106.5:c.506-1G>A)	0/1	C>T
CYP2D6	rs1135840	drug response	missense variant(NM_000106.5:c.1457G>C)	0/1	G>C

Hirschsprung disease, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
RET	rs1800858	risk factor	synonymous variant(NM_020975.4:c.135G>A)	1/1	A>G

Alcohol dependence

Gene	rs#	Significance	Reference	GenoType	Detected Change
ADH1C	rs283413	risk factor	nonsense(NM_000669.4:c.232G>T)	1/1	A>C
ADH1B	rs1229984	protective	synonymous variant(NM_000668.5:c.143A=)	1/1	T>C

Cystic fibrosis

Gene	rs#	Significance	Reference	GenoType	Detected Change
TGFB1	rs1800470	risk factor	missense variant(NM_000660.6:c.29C>T)	0/1	G>A

Thyroid hormone metabolism, abnormal

Gene	rs#	Significance	Reference	GenoType	Detected Change
ERCC1	rs11615	drug response	synonymous variant(NM_001983.3:c.354T>C)	0/1	A>G

Myocardial infarction 1

Gene	rs#	Significance	Reference	GenoType	Detected Change

LRP8	rs5174	risk factor	missense variant(NM_004631.4:c.2855G>A)	1/1	C>T
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Homocystinuria-Megaloblastic anemia due to defect in cobalamin metabolism, cblE complementation type

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTRR	rs1801394	drug response	non-coding transcript variant(NR_134480.1 :n.203A>G)	0/1	A>G

diuretics response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
NEDD4L	rs4149601	drug response	synonymous variant(NM_0011449 68.1:c.24G>A)	0/1	G>A

capecitabine response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
DPYD	rs1801265	Pathogenic	missense variant(NM_000110.3:c.85T>C)	1/1	G>A

Generalized epilepsy with febrile seizures plus, type 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

Pyrimidine analogues response - Toxicity/ADR

Gene	rs#	Significance	Reference	GenoType	Detected Change
DPYD	rs1801265	Pathogenic	missense variant(NM_000110.3:c.85T>C)	1/1	G>A

Ceroid lipofuscinosis neuronal 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
TPP1	rs56144125	Pathogenic	splice acceptor variant(NM_000391.3:c.509-1G>A)	0/1	C>T

DIABETES MELLITUS, TYPE II, SUSCEPTIBILITY TO

Gene	rs#	Significance	Reference	GenoType	Detected Change
HNF1A	rs1169305	Pathogenic	missense variant(NM_000545.6:c.1720G>A)	1/1	A>G

Coronary artery spasm 1, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOS3	rs1799983	risk factor	missense variant(NM_000603.4:c.894T>G)	0/1	T>G

Arylsulfatase a, allele a

Gene	rs#	Significance	Reference	GenoType	Detected Change
ARSA	rs743616	Pathogenic	missense variant(NM_000487.5:c.1178C>G)	1/1	G>C

Platelet-activating factor acetylhydrolase deficiency

Gene	rs#	Significance	Reference	GenoType	Detected Change
PLA2G7	rs1051931	risk factor	missense variant(NM_0011683.57.1:c.1136T>C)	1/1	A>G
PLA2G7	rs1805018	risk factor	missense variant(NM_0011683.57.1:c.593T>C)	0/1	A>G

Leanness, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
AHSG	rs2070635	association	intron variant(NM_001622.2:c.574-149A>G)	0/1	A>G

AHSG	rs4918	risk factor	missense variant(NM_001622.2:c.767G>C)	1/1	G>C
AHSG	rs4917	risk factor	missense variant(NM_001622.2:c.743T>C)	1/1	T>C

Carcinoma, adrenocortical, androgen-secreting

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP21A2	rs6467	Pathogenic	intron variant(NM_000500.7:c.293-13C>G)	0/1	C>A

Islet cell hyperplasia

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C

MACULAR DEGENERATION, AGE-RELATED, 12, SUSCEPTIBILITY TO

Gene	rs#	Significance	Reference	GenoType	Detected Change
CX3CR1	rs3732379	Pathogenic	missense variant(NM_0011711 74.1:c.841G>A)	0/1	C>T

Laron syndrome with undetectable serum GH-binding protein

Gene	rs#	Significance	Reference	GenoType	Detected Change
GHR	rs6180	risk factor	missense variant(NM_000163.4:c.1630A>C)	1/1	A>C

Graves disease 4

Gene	rs#	Significance	Reference	GenoType	Detected Change
CTLA4	rs231775	risk factor	missense variant(NM_005214.4:c.49A>G)	0/1	A>G

Li-Fraumeni syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
TP53	rs1042522	drug response	missense variant(NM_000546.5:c.215C>G)	0/1	G>C

Phenylketonuria

Gene	rs#	Significance	Reference	GenoType	Detected Change
PAH	rs5030860	Pathogenic	missense variant(NM_000277.1:c.1241A>G)	0/1	T>C

Glipizide poor metabolizer

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

Lupus nephritis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
FCGR2A	rs1801274	risk factor	missense variant(NM_021642.3:c.497A>G)	0/1	A>G

Metabolic syndrome, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
MTTP	rs3816873	protective	missense variant(NM_000253.3:c.383T>C)	0/1	T>C

Malignant melanoma of skin

Gene	rs#	Significance	Reference	GenoType	Detected Change
SLC45A2	rs16891982	association	missense variant(NM_016180.4:c.1122C>G)	1/1	C>G

Generalized arterial calcification of infancy 2

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCC6	rs2238472	Pathogenic	missense variant(NM_001171.5:c.3803G>A)	0/1	C>T

Fluorouracil response

Gene	rs#	Significance	Reference	GenoType	Detected Change
DPYD	rs1801265	Pathogenic	missense variant(NM_000110.3:c.85T>C)	1/1	G>A

short QT syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_000238.3:c.1956T>A)	0/1	A>G

Cataract 41

Gene	rs#	Significance	Reference	GenoType	Detected Change
WFS1	rs10010131	association	intron variant(NM_006005.3:c.461-9A>G)	1/1	A>G

Arterial calcification of infancy

Gene	rs#	Significance	Reference	GenoType	Detected Change
ENPP1	rs1044498	risk factor	missense variant(NM_006208.2:c.517A>C)	0/1	A>C

Non-syndromic genetic deafness

Gene	rs#	Significance	Reference	GenoType	Detected Change
LOXHD1	rs1450425	Likely pathogenic	intron variant(NM_144612.6:c.4530+107A>G)	1/1	T>C

Allergic rhinitis, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
IL13	rs20541	risk factor	missense variant(NM_002188.2:c.431A>G)	0/1	A>G

Alzheimer disease, early-onset, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
PRNP	rs1799990	risk factor	missense variant(NM_000311.3:c.385A>G)	0/1	A>G

Bardet-Biedl syndrome

Gene	rs#	Significance	Reference	GenoType	Detected Change
BBS2	rs4784677	Pathogenic	missense variant(NM_031885.3:c.209A>G)	1/1	C>T

Human immunodeficiency virus type 1, rapid progression to AIDS

Gene	rs#	Significance	Reference	GenoType	Detected Change
CX3CR1	rs3732379	Pathogenic	missense variant(NM_0011711.74.1:c.841G>A)	0/1	C>T

Lumbar disc herniation, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
COL11A1	rs1676486	risk factor	non-coding transcript variant(NR_134980.1:n.4937T>C)	0/1	A>G

Diabetes mellitus, permanent neonatal, with neurologic features

Gene	rs#	Significance	Reference	GenoType	Detected Change

KCNJ11	rs5219	risk factor	missense variant(NM_000525.3:c.67A>G)	0/1	T>C
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Ischemic stroke, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
NOS3	rs1799983	risk factor	missense variant(NM_000603.4:c.894T>G)	0/1	T>G
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

Severe myoclonic epilepsy in infancy

Gene	rs#	Significance	Reference	GenoType	Detected Change
SCN1A	rs3812718	risk factor	intron variant(NM_006920.4:c.603-91G>A)	1/1	C>T

Aldosterone to renin ratio, increased

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP11B2	rs61757294	Pathogenic	missense variant(NM_000498.3:c.1157T>C)	0/1	A>G

Tangier disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
ABCA1	rs2230806	protective	missense variant(NM_005502.3:c.656G>A)	0/1	C>T

Hypercholesterolaemia

Gene	rs#	Significance	Reference	GenoType	Detected Change
LDLR	rs2228671	Pathogenic	nonsense(NM_000527.4:c.81C>A)	0/1	C>T

Renal dysplasia

Gene	rs#	Significance	Reference	GenoType	Detected Change
AGT	rs699	risk factor	missense variant(NM_000029.3:c.803T>C)	0/1	A>G

Preeclampsia, susceptibility to

Gene	rs#	Significance	Reference	GenoType	Detected Change
AGT	rs699	risk factor	missense variant(NM_000029.3:c.803T>C)	0/1	A>G
EPHX1	rs2234922	drug response	missense variant(NM_0011360 18.3:c.416A>G)	0/1	A>G
EPHX1	rs1051740	risk factor	missense variant(NM_0011360 18.3:c.337T>C)	0/1	T>C

Leprosy, protection against

Gene	rs#	Significance	Reference	GenoType	Detected Change
TLR1	rs5743618	protective	missense variant(NM_003263.3:c.1805G>T)	0/1	C>A
TLR1	rs4833095	risk factor	missense variant(NM_003263.3:c.743A>G)	0/1	T>C

Congenital heart disease

Gene	rs#	Significance	Reference	GenoType	Detected Change
GATA4	rs3729856	Likely pathogenic	missense variant(NM_002052.4:c.1129A>G)	0/1	A>G

Epilepsy, idiopathic generalized 8

Gene	rs#	Significance	Reference	GenoType	Detected Change
CASR	rs1801725	association	missense variant(NM_000388.3:c.2956G>T)	0/1	G>T

Factor V Hong Kong

Gene	rs#	Significance	Reference	GenoType	Detected Change
F5	rs6025	risk factor	missense variant(NM_000130.4:c.1601G>A)	1/1	T>C

Periventricular nodular heterotopia with syndactyly, cleft palate and developmental delay

Gene	rs#	Significance	Reference	GenoType	Detected Change
NEDD4L	rs4149601	drug response	synonymous variant(NM_0011449 68.1:c.24G>A)	0/1	G>A

celecoxib response - Dosage

Gene	rs#	Significance	Reference	GenoType	Detected Change
CYP2C9	rs1799853	drug response	synonymous variant(NM_000771.3:c.430C=)	0/1	C>T

Long QT syndrome 1/2, digenic

Gene	rs#	Significance	Reference	GenoType	Detected Change
KCNH2	rs1137617	Pathogenic	nonsense(NM_00023 8.3:c.1956T>A)	0/1	A>G

hydrochlorothiazide response - Efficacy

Gene	rs#	Significance	Reference	GenoType	Detected Change
NEDD4L	rs4149601	drug response	synonymous variant(NM_0011449 68.1:c.24G>A)	0/1	G>A

Glycogen storage disease II, adult form

Gene	rs#	Significance	Reference	GenoType	Detected Change
GAA	rs2278619	Likely pathogenic	intron variant(NM_000152.4:c.1327-18A>G)	1/1	A>G