InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
3-Hydroxy-3-Methylglutaryl CoA lyase deficiency	HMGCL	Iberian Peninsula	84%	Unknown	< 1 in 500
		Saudi Arabian	94%	< 1 in 50	< 1 in 800
p.Glu37Ter, p.Arg41Gln, c.561+1G>A, p.Phe305Tyrfs					
Abetalipoproteinemia	MTP/MTTP	Ashkenazi Jewish	75%	1 in 131	< 1 in 500
p.Gly865Ter					
Achalasia-Addisonianism-Alacrima syndrome	AAAS	General Population	Unknown	Unknown	< 1 in 500
		Puerto Rican	Unknown	Unknown	< 1 in 500
c.1331+1G>A, p.Arg478Ter, p.Gln15Lys, p.Ser263Pro					
Alport syndrome, autosomal recessive	COL4A3	Ashkenazi Jewish	>95%	1 in 183	<1 in 3643
		Chinese	15%	Unknown	Unknown
		Northern European	16%	Unknown	Unknown
p.Leu14_Leu21del, p.Leu1474Cysfs, p.Arg1481Ter, p.Ser1524Ter					
Argininosuccinic Aciduria	ASL	Dutch	56%	Unknown	1 in 300
		Saudi Arabian	52%	1 in 80	1 in 165
p.Gln354Ter, p.Arg385Cys, p.Gln116Ter, c.446+1G>A					
Ataxia with isolated Vitamin E Deficiency	TTPA	Italian	35%	1 in 266	1 in 410
		North African	94%	1 in 159	1 in 2672
p.Glu249Asnfs					
Autoimmune Polyglandular Syndrome, Type 1	AIRE	Finnish	89%	1 in 80	1 in 715
		Iranian Jewish	>95%	1 in 48	<1 in 940
p.Tyr85Cys, p.Arg257Ter					
Bernard-Soulier Syndrome, Type A1	GP1BA	General Population	Unknown	Very rare	< 1 in 500
p.Leu145Pro					
Bernard-Soulier Syndrome, Type C	GP9	General Population	Unknown	Very rare	<1 in 500
p.Asn61Ser					
Beta Hemoglobinopathies	HBB	African American	98%	1 in 9	1 in 378
		Chinese	99%	1 in 21	1 in 2011
		Indian	97%	1 in 20	1 in 420
		Mediterranean	98%	1 in 6	1 in 350
		Middle Eastern	86%	1 in 15	1 in 98
		Thai	97%	1 in 11	1 in 328
c.*110T>C, c.*111A>G, c.*113 A>G, c.*185_*209del619, c.-136C>G, c.-137C>G, c.-138C>A, c.-138C>G, c.-138C>T, c.-140C>T, c.-151C>T, c.-29G>A, c.-50A>C, c.-78A>G, c.-79A>G, c.-80T>A, c.-80T>C, c.-81A>G, p.Met1?, p.Trp38Glyfs, p.Trp38Ter, p.Gln39Ter, p.Phe42Leufs , p.Phe46Leufs, p.Pro52Leufs, p.Pro6Argfs, p.Glu7Lys , p.Met1?, p.Glu7Glyfs, p.Glu7Val , p.Val68Alafs , p.Ser73Ter, p.Ser73Lysfs, p.Leu76Trpfs, p.Ala77Valfs, p.Lys9Valfs, p.Gly84Alafs, p.Lys9Asnfs*15, p.Glu91Ter , p.Leu97Alafs, c.315+1G>A, c.315+2T>A, c.315+2T>C, c.316-1G>A, c.316-1G>C, c.316-1G>T, c.316-106C>G, c.316-146T>G, c.316-197C>T, c.316-2A>C, c.316-2A>G, c.316-3C>A, c.316-3C>G, p.Thr13Leufs, p.Glu122Lys , p.Glu122Ter, p.Gln128_Ala129delin, p.Val2Cysfs, p.Trp16Valfs, p.Trp16Glyfs, p.Trp16Ter, p.Trp16Ter, p.Lys18Argfs, p.Lys18Ter, p.Asn20Ser, p.Glu23Valfs, p.Gly25Gly, p.Glu27Lys, p.Glu27Ter, p.Ala28Ser, p.Leu29Profs, p.Arg31Thr, c.92+1G>A, c.92+1G>T, c.92+2T>A, c.92+2T>C, c.92+5G>A, c.92+5G>C, c.92+5G>T, c.92+6T>C, c.93-1G>A, c.93-1G>C, c.93-15T>G, c.93-2A>G, c.93-21G>A					
Carnitine Palmitoyltransferase Deficiency, Type 1A	CPT1A	Hutterite	95%	1 in 16	< 1 in 300
p.Gly710Glu					
Carnitine Palmitoyltransferase Deficiency, Type 2	CPT2	Ashkenazi Jewish	>95%	1 in 43	<1 in 846
		Caucasian	71%	1 in 181	1 in 621
p.Arg37Profs, p.Phe383Tyr, p.Lys414Thrfs, p.Pro50His, p.Gly549Asp, p.Tyr628Ser, p.Arg631Cys, p.Ser113Leu, p.Tyr120Cys, p.Arg124Ter, p.Arg151Gln, p.Glu174Lys, p.Leu178_lle186delinsPhe, p.Pro227Leu, p.Asp328Gly					
Cerebrotendinous Xanthomatosis (CTX)	CYP27A1	General Population	11%	1 in 71	1 in 80

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
p.Arg395Cys, p.Arg395Ser					
Citrin Deficiency	SLC25A13	Japanese	>30%	1 in 70	< 1 in 100
c.1180+1G>A, p.Met285Profs					
Congenital Disorder of Glycosylation, Type Ib	MPI	General Population	Unknown	Very rare	< 1 in 400
p.Arg295His					
Congenital Myasthenic Syndrome, CHRNE-associated	CHRNE	European/Gypsy	>50%	< 1 in 20	< 1 in 39
		North African	>44%	Unknown	< 1 in 400
p.Glu443Lysfs, p.Asn452Glufs					
Congenital Myasthenic Syndrome, RAPSN-associated	RAPSN	General Population	70%	Unknown	< 1 in 500
p.Asn88Lys					
Crigler-Najjar Syndrome	UGT1A1	Dutch	34%	1 in 500	1 in 750
		Tunisian	84%	1 in 500	< 1 in 3000
p.Gln357Arg, p.Lys407Argfs					
Cystinosis	CTNS	French Canadian	54%	1 in 39	1 in 84
		General Population (US)	62%	1 in 159	1 in 416
		Italian	17%	1 in 159	1 in 191
c.-36008_c.848del57119, p.Trp138Ter, p.Leu158Pro, p.Asp205Asn					
Deafness, autosomal recessive 1A	GJB2	African (Ghana)	76%	1 in 226	1 in 941
		Ashkenazi Jewish	93%	1 in 21	1 in 286
		East Asian	68%	1 in 23	1 in 68
		General Population	80%	1 in 43	1 in 210
		Middle Eastern	75%	1 in 77	1 in 307
		South Asian (Indian/Pakistani)	75%	1 in 100	1 in 397
p.Val37Ile , p.Leu56Argfs, p.Trp77Arg, p.Trp77Ter, p.Leu79Cysfs, p.Leu90Pro, p.Lys105Glyfs, p.Gly12Valfs, p.Glu120del, p.Gln124Ter, p.Arg143Trp, p.Arg184Pro , p.Trp24Ter					
Factor XI Deficiency (Hemophilia C)	F11	Ashkenazi Jewish	94%	1 in 11	1 in 157
		French Basque	64%	1 in 101	1 in 280
		General Population	12%	1 in 501	1 in 569
		Italian	31%	1 in 501	1 in 667
		Turkish	12%	Unknown	Unknown
p.Cys56Arg, p.Gln134Ter, p.Glu135Ter, p.Phe301Leu					
Familial Hypercholesterolemia, homozygous, LDLR-associated	LDLR	Ashkenazi Jewish	35%	1 in 67	1 in 103
p.Gly219del					
Familial Hypercholesterolemia, LDLRAP1 associated	LDLRAP1	Sardinian	54%	< 1 in 100	< 1 in 200
p.His144Glnfs					
Familial Hyperinsulinism	ABCC8	Ashkenazi Jewish	88%	< 1 in 52	< 1 in 424
		Finnish	43%	1 in 100	< 1 in 175
c.3992-9G>A, p.Phe1388del, p.Val187Asp					
Familial Mediterranean Fever	MEFV	Arab	53%	1 in 4	1 in 8
		Armenian	78%	1 in 5	1 in 18
		Ashkenazi Jewish	52%	1 in 5	1 in 9
		Mediterranean	68%	1 in 5	1 in 15

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		Mediterranean Jewish	69%	1 in 4	1 in 12
		Turkish	76%	1 in 5	1 in 18
p.Met680Ile , p.Met680Ile, p.Met694Val, p.Met694Ile , p.Val726Ala					
Familial Neurohypophyseal Diabetes Insipidus (FNDI), Autosomal Recessive	AVP	General Population	Unknown	Unknown	Unknown
p.Pro26Leu					
Fanconi anemia, complementation group C	FANCC	Ashkenazi Jewish	99%	1 in 89	1 in 8801
p.Arg548Ter, p.Leu554Pro, p.Gln13Ter, c.456+4A>T, p.Arg185Ter, p.Asp23Ilefs					
Fanconi anemia, complementation group G	FANCG	Brazilian	99%	Very rare	< 1 in 25000
		Japanese	65%	Very rare	< 1 in 1000
p.Gln356Ter, p.Glu395Trpfs, c.1480+1G>C, p.Trp599Profs, c.307+1G>C					
Fructose intolerance	ALDOB	General Population	3%	1 in 71	1 in 74
		Spanish	17%	1 in 71	1 in 86
p.Asn120Lysfs					
Galactosemia	GALT	African American	68%	1 in 86	1 in 256
		Ashkenazi Jewish	95%	1 in 156	1 in 3100
		General Population	85%	1 in 110	1 in 728
		Hispanic	68%	Unknown	Unknown
		Irish	93%	1 in 107	1 in 1231
		Irish Travelers	>95%	1 in 11	<1 in 200
c.-1040_+790del5576ins129, c.253-2A>G, p.Ser135Leu, p.Thr138Met, p.Phe171Ser, p.Gln188Arg, p.Leu195Pro, p.Tyr209Cys, p.Tyr209Ser, p.Lys285Asn					
Gaucher Disease	GBA	Ashkenazi Jewish	96%	1 in 15	1 in 354
		Brazilian	74%	Unknown	Unknown
		Chinese	33%	Unknown	Unknown
		General Population (non-Jewish)	70%	< 1 in 100	< 1 in 331
		Indian	73%	Unknown	Unknown
		Irish	67%	Unknown	Unknown
		Italian	67%	Unknown	Unknown
		Japanese	49%	Unknown	Unknown
		Korean	22%	Unknown	Unknown
		Portuguese	76%	Unknown	Unknown
		Romanian	81%	Unknown	Unknown
		Spanish	74%	1 in 274	1 in 1052
		Turkish	80%	Unknown	Unknown
c.115+1G>A, p.Asn409Ser, p.Val433Leu, p.Asp448His, p.Leu483Pro, p.Arg502Cys, p.Arg535His, p.Leu29Alafs					
Glaucoma 3 Primary Congenital	CYP1B1	Caucasian	13%	1 in 51	1 in 58
		Indian	7%	1 in 29	1 in 31
		Saudi Arabian	15%	1 in 26	1 in 30
		Slovakian Gypsy (Rom)	99%	1 in 9	1 in 832
p.Glu387Lys, p.Arg390Cys, p.Arg390His , p.Arg469Trp					
Glutaric Acidemia, Type I	GCDH	Amish	99%	1 in 12	< 1 in 1000
		Caucasian	>40%	1 in 112	< 1 in 187
p.Val400Met, p.Arg402Trp, p.Ala421Val, p.Arg227Pro, p.Ala293Thr					
Glutaric Acidemia, Type IIA	ETFA	European Caucasian	25%	Very rare	< 1 in 500
p.Thr266Met					

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
Glutaric Acidemia, Type IIC	ETFDH	European Caucasian	17%	Very rare	< 1 in 500
p.Leu334Pro, p.Met1?					
Glycogen Storage Disease, Type Ia (GSDIa)	G6PC	Ashkenazi Jewish	>99%	1 in 71	1 in 7022
		Caucasian	63%	1 in 159	1 in 420
		Chinese	80%	1 in 159	1 in 789
		General Population	67%	1 in 159	1 in 479
		Hispanic	78%	1 in 159	1 in 710
		Italian	70%	1 in 159	1 in 523
		Japanese	90%	1 in 159	1 in 1577
		Korean	75%	1 in 159	1 in 631
		Turkish	67%	1 in 159	1 in 485
p.Gln347Ter, p.Asp38Val, p.Arg83Cys, p.Arg83His, p.Tyr127Ilefs, p.Gly188Arg, p.Leu216Leu, p.Gln27Argfs					
Glycogen Storage Disease, Type Ib (GSDIb)	SLC37A4	European Caucasian	44%	1 in 354	1 in 629
		Japanese	43%	1 in 354	1 in 617
p.Gly361Cys, p.Leu370Valfs, p.Trp118Arg					
Glycogen Storage Disease, Type III (GSDIII)	AGL	Caucasian	27%	1 in 159	1 in 215
		Dutch	15%	Unknown	Unknown
		Faorese	99%	1 in 28	1 in 2736
		General Population	51% (GSD IIIb)	1 in 159	< 1 in 300 (GSD IIIb)
		Irish	13%	Unknown	Unknown
		North African Jewish	99%	1 in 37	1 in 3626
p.Arg408Ter, p.Gln6Ter, p.Gln6Hisfs, p.Ser1486Profs					
Glycogen Storage Disease, Type V (GSDV)	PYGM	Caucasian	71%	1 in 159	1 in 550
		Japanese	68%	Unknown	Unknown
		Spanish	74%	1 in 205	1 in 779
p.Arg50Ter, p.Phe710del, p.Trp798Arg, p.Tyr85Ter, p.Gly205Ser					
Holocarboxylase Synthetase Deficiency	HLCS	Faorese	>95%	1 in 51	1 in 991
		General Population	49%	1 in 148	1 in 286
		Japanese	77%	<1 in 159	<1 in 677
c.1519+5G>A, p.Arg508Trp, p.Val550Met, p.Leu237Pro, p.Gly261Valfs					
Homocystinuria (CBS Deficiency)	CBS	African American	63%	1 in 294	1 in 782
		Australian	39%	Unknown	Unknown
		Central European	68%	1 in 145	1 in 444
		Dutch	52%	1 in 251	1 in 525
		Iberian Peninsula	52%	1 in 167	1 in 348
		Irish	68%	1 in 128	1 in 393
		Italian	35%	Unknown	Unknown
		Norwegian	55%	1 in 41	1 in 89
		Qatari	95%	1 in 22	1 in 392
		South American	63%	Unknown	Unknown
		United Kingdom	57%	Unknown	Unknown
		Unites States (Caucasian)	33%	1 in 294	1 in 440
p.Arg336Cys, p.Thr353Met, c.1224-2A>C, p.Ala114Val, p.Thr191Met, p.Thr262Met, p.Arg266Lys, p.Ile278Thr, c.844_845c.844_845ins68, p.Gly307Ser, p.Val320Ala					
Homocystinuria, cblE type	MTRR	European	60%	Very rare	< 1 in 500

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
p.Ser481Leu, c.903+469T>C					
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome	SLC25A15	French Canadian	96%	1 in 20	1 in 472
p.Phe188del					
Hyperphenylalaninemia, BH4-Deficient, A	PTS	Chinese	70%	1 in 182	< 1 in 600
p.Asn52Ser, p.Pro87Ser, p.Asp136Val					
Krabbe Disease	GALC	European Caucasian	60%	1 in 159	1 in 395
		Japanese	38%	1 in 159	1 in 255
30Kb DEL, p.Glu385Ter, p.Lys491fs, p.Thr529Met, p.Tyr567Ser, p.Thr668Pro, p.Asn228_Ser232delin, p.Gly286Asp					
Lipoprotein Lipase Deficiency	LPL	French Canadian	29%	1 in 44	1 in 61
		General Population	24%	Unknown	< 1 in 500
p.Gly215Glu					
Long-Chain 3-Hydroxyacyl-Coenzyme A Dehydrogenase (LCHAD) Deficiency	HADHA	General Population	87%	1 in 125	1 in 970
p.Gln378Ter, p.Glu510Gln					
Lysinuric Protein Intolerance	SLC7A7	Finnish	99%	1 in 138	< 1 in 10,000
		Italian	44%	< 1 in 120	< 1 in 200
		Japanese	64%	1 in 120	1 in 330
p.Arg410Ter, p.Ile461Asnfs, p.Trp242Ter, c.895-2A>G					
Maple Syrup Urine Disease, Type 1A	BCKDHA	General Population	11%	1 in 216	1 in 242
		Mennonite	>95%	1 in 7	<1 in 124
		Portuguese Roma/Gypsy	>95%	1 in 71	< 1 in 1400
p.Arg40Glyfs, p.Tyr438Asn					
Maple Syrup Urine Disease, Type 1B	BCKDHB	Ashkenazi Jewish	99%	1 in 80	1 in 7900
p.Glu372Ter, p.Arg183Pro, p.Gly278Ser					
Medium-chain acyl-coenzyme A dehydrogenase (MCAD) deficiency	ACADM	European Caucasian	>80%	1 in 66	1 in 324
		Saudi Arabian	95%	1 in 68	< 1 in 1300
p.Thr154Ile, p.Gly300Arg, p.Lys362Glu					
Methylmalonic Aciduria and Homocystinuria, Cobalamin C (cblC) type	MMACHC	Chinese	54%	Very rare	< 1 in 500
		General Population	65%	Very rare	< 1 in 500
		Italian	75%	Very rare	< 1 in 500
		Portuguese	91%	Very rare	< 1 in 500
p.Arg91Lysfs, p.Arg111Ter, p.Arg132Ter, p.Trp203Ter, p.Trp203Ter					
Methylmalonic Aciduria due to Methylmalonyl-CoA Mutase Deficiency	MUT	African American	36%	Unknown	Unknown
		Caucasian (European)	37%	1 in 142	1 in 223
		Chinese	17%	Unknown	Unknown
		Hispanic-Spanish-Latin American	49%	Unknown	Unknown
		Japanese	42%	1 in 112	1 in 193
p.Arg369Cys, p.Arg369His, p.Gly544Ter, p.Arg694Trp, p.Gly717Val, p.Arg108Cys, p.Glu117Ter, p.Gly203Arg, p.Asn219Tyr					
Methylmalonic Aciduria, MMAA-associated (cblA type)	MMAA	Caucasian	45%	Unknown	< 1 in 400
p.Arg145Ter					

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
Mucopolysaccharidosis type I (MPS I)	IDUA	Chinese	11%	Unknown	Unknown
		General Population	63%	1 in 159	1 in 427
		Irish Travelers	>95%	1 in 10	1 in 916
		Italian	48%	Unknown	Unknown
		Japanese	22%	Unknown	Unknown
		Moroccan	92%	Unknown	Unknown
		Scandinavian	85%	1 in 159	1 in 416
		Tunisian	44%	1 in 200	1 in 358
p.Leu346Arg, p.Trp402Ter, p.Gly51Asp, p.Pro533Arg, p.Gln70Ter, p.Arg89Gln, c.590-7G>A, p.Cys205Alafs, p.Ala327Pro					
Nephrotic Syndrome, Congenital Finnish	NPHS1	Finnish	16%	1 in 46	1 in 54
p.Arg1109Ter					
Nephrotic Syndrome, Steroid-Resistant Type 2	NPHS2	European	< 20%	Unknown	< 1 in 300
		Israeli-Arab	55%	Unknown	< 1 in 500
p.Arg138Ter, p.Arg138Gln					
Omenn Syndrome	DCLRE1C	Navajo and Apache (Athabascan-speaking)	98%	1 in 23	< 1 in 1000
p.Tyr199Ter					
Osteopetrosis, autosomal recessive 1	TCIRG1	Ashkenazi Jewish	>95%	1 in 350	<1 in 6878
		General Population	6%	1 in 354	1 in 377
c.117+4A>T					
Pendred Syndrome	SLC26A4	European Caucasian	20%	1 in 58	1 in 73
p.Leu236Pro					
Phenylketonuria (PKU)	PAH	Ashkenazi Jewish	15%	1 in 225	1 in 265
		Belgian	40%	1 in 51	1 in 84
		Brazilian	37%	1 in 71	1 in 113
		Bulgarian	81%	1 in 71	1 in 375
		Caucasian	47%	1 in 50	1 in 94
		Chech	74%	1 in 48	1 in 185
		Chinese	4%	1 in 53	1 in 56
		Croatian	77%	1 in 46	1 in 200
		Danish	67%	1 in 55	1 in 168
		English	57%	1 in 55	1 in 129
		Estonian	91%	1 in 37	1 in 406
		German	82%	1 in 45	1 in 251
		Hungarian	61%	1 in 47	1 in 122
		Icelandic	10%	1 in 51	1 in 56
		Irish	68%	1 in 34	1 in 104
		Latvian	86%	1 in 46	1 in 329
		Lithuanian	82%	1 in 48	1 in 266
		Norwegian	61%	1 in 58	1 in 147
		Polish	74%	1 in 45	1 in 174
		Portuguese	29%	1 in 62	1 in 87
		Romanian	57%	1 in 45	1 in 105
		Spanish	28%	1 in 51	1 in 70
		Swedish	45%	1 in 71	1 in 129
		Taiwanese	29%	1 in 118	1 in 166

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		Turkish	41%	1 in 26	1 in 44
		Welsh	59%	1 in 58	1 in 140
		West Scotland	53%	1 in 44	1 in 93
c.1066-11G>A, p.Phe39Leu, p.Arg408Trp, p.Arg408Gln, c.1315+1G>A, p.Leu48Ser, p.Ile65Thr, p.Arg158Gln, p.Arg261Gln, p.Gly272Ter, p.Phe299Cys					
Pompe Disease	GAA	African descent	49%	1 in 60	1 in 115
		Brazilian	34%	1 in 101	1 in 152
		Caucasian	50%	1 in 101	1 in 199
		Chinese	29%	1 in 112	1 in 158
		Dutch	72%	1 in 101	1 in 353
		Italian	48%	1 in 123	1 in 236
c.-32-13T>G, p.Asp645Glu, p.Gly828_Asn882del, p.Arg854Ter, p.Glu176fs					
Primary Hyperoxaluria, Type 1	AGXT	General Population	50%	1 in 354	1 in 700
p.Lys12Glnfs, p.Phe152Ile, p.Gly170Arg, p.Ile244Thr					
Primary Hyperoxaluria, Type 2	GRHPR	European Caucasian	30%	1 in 500	< 1 in 715
p.Asp35Thrfs					
Propionic Acidemia, PCCA-associated	PCCA	General Population	26%	1in 159	1 in 214
		Japanese	46%	1 in 93	1 in 173
c.1644-6C>G, c.184-618_300+3930del4779, p.Leu308Phefs					
Propionic Acidemia, PCCB-associated	PCCB	Japanese	42%	< 1 in 66	< 1 in 114
		Spanish/Latin American	50%	< 1 in 159	< 1 in 316
p.Arg430Trp, p.Val412Cysfs, p.Gly427Argfs, p.Thr448Ile, p.Arg185Trp, p.Glu188Lys					
Pyridoxine-Dependent Epilepsy	ALDH7A1	Dutch	64%	< 1 in 260	< 1 in 725
		European Caucasian	33%	< 1 in 260	< 1 in 390
p.Glu427Gln					
Segawa Syndrome	TH	Chinese	25%	Very rare	1 in 225
		Dutch	70%	Very rare	1 in 225
p.Arg233His					
Severe Combined Immunodeficiency due to Adenosine Deaminase Deficiency	ADA	General Population	41%	1 in 501	1 in 848
p.Leu107Pro, p.Pro297Gln, p.Ala329Val					
Short/Branched Chain Acyl-CoA Dehydrogenase (SBCAD) Deficiency	ACADSB	Hmong	99%	1 in 8	1 in 792
p.Met389Val, c.303+3A>G					
Tyrosinemia, Type I	FAH	Ashkenazi Jewish	99%	1 in 100	< 1 in 9900
		Finnish	88%	1 in 123	< 1 in 975
		French Canadian	80%	1 in 64	< 1 in 300
		Scandinavian	24%	1 in 112	< 1 in 147
p.Gly337Ser, c.1062+5G>A, p.Glu357Ter, p.Asp233Val, c.707-1G>C, p.Pro261Leu, p.Trp262Ter					
Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	ACADVL	General Population	21%	1 in 87	1 in 109
p.Gly464Asp, p.Arg492Trp, c.1679-6G>A, p.Thr283Met, p.Val306Ala					
Vitamin D hydroxylation-deficient Rickets, Type IA	CYP27B1	French Canadian	>95%	1 in 25	1 in 2380
		General Population	26%	Unknown	Unknown
		Japanese	17%	Unknown	Unknown
		Korean	50%	Unknown	Unknown

InheriGen Tx

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
p.Arg389His, p.Phe443Profs, p.Val88Trpfs, c.589+1G>A					
Wilson Disease	ATP7B	Ashkenazi Jewish	83%	1 in 101	1 in 597
		Eastern European	62%	1 in 87	1 in 229
		Mediterranean	17%	1 in 87	1 in 92
		Western European	29%	1 in 87	1 in 122
p.Arg778Gly, p.Trp779Ter, p.Glu1064Ala, p.His1069Gln					
X-Linked Severe Combined Immunodeficiency	IL2RG	General Population	25%	1 in 25001	1 in 33468
p.Cys62Ter, p.Gly114Asp, c.454+1G>A, p.Ile153Asn, p.Leu172Pro, p.Arg222Cys, p.Arg226Cys, p.Arg226His, p.Arg285Gln, p.Arg289Ter, p.Leu293Gln, p.Ser308Ter					