InheriGen Plus: Pan-Ethnic Carrier Screen

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					
Achromatopsia, CNGB3-associated	CNGB3	European	83%	1 in 123	< 1 in 700
		Pingelapese (Micronesian)	99%	1 in 3	< 1 in 189
p.Glu336Ter, p.Thr383Ilefs, p.Ser435Phe, p.Arg274Valfs, c.991-3T>G					
Adrenal Hyperplasia, Congenital, due to 17-Alpha-hydroxylase Deficiency	CYP17A1	Brazilian	53%	1 in 501	1 in 1064
		Canadian Mennonite	93%	1 in 501	1 in 7137
		Chinese	19%	1 in 501	1 in 618
p.Trp406Arg, c.1243+5G>A, p.Ile479Hisfs, p.Asp487_Phe489del, p.Tyr329Asp					
Agenesis of the Corpus Callosum with Peripheral Neuropathy (Andermann Syndrome)	SLC12A6	French Canadian	99%	1 in 23	1 in 2200
p.Thr754Profs					
Albinism, Oculocutaneous, Type I	TYR	Chinese	11%	1 in 81	1 in 91
		Indian	56%	1 in 101	1 in 227
p.Arg278Ter					
Albinism, Oculocutaneous, Type IV	SLC45A2	Japanese	39%	1 in 146	1 in 239
p.Asp157Asn					
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					
Antley-Bixler Syndrome	POR	European Caucasian	40%	Unknown	< 1 in 500
		General Population	50%	Unknown	< 1 in 500
		Japanese	60%	Unknown	< 1 in 500
p.Arg457His, p.Ala287Pro					
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					
Arthrogryposis, mental retardation, and seizures	SLC35A3	Ashkenazi Jewish	>95%	1 in 453	1 in 9033
p.Gln172Ter, p.Ser338Gly					
Aspartylglucosaminuria (AGU)	AGA	Finnish	100%	1 in 69	1 in 13503
p.Glu67Alafs, p.Ser72Pro, p.Cys163Ser					
Ataxia Neuropathy Spectrum (ANS)	POLG	Scandinavian	59%	1 in 100	1 in 244
p.Ala467Thr, p.Trp748Ser					
Ataxia with isolated Vitamin E Deficiency	TTPA	Italian	35%	1 in 266	1 in 410
		North African	94%	1 in 159	1 in 2672

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
p.Glu249Asnfs					
Ataxia-Telangiectasia	ATM	Amish	99%	Unknown	Unknown
		Costa Rican	56%	1 in 101	1 in 226
		Italian	7%	1 in 101	1 in 108
		North African Jewish	97%	1 in 82	1 in 2728
		Norwegian	57%	1 in 101	1 in 232
		Polish	54%	1 in 101	1 in 111
p.Arg35Ter, p.Glu522Ilefs, p.His1082Leufs, p.Lys1192Lys, p.Ala1299Cysfs, p.Gln1970Ter, c.7630-2A>C, p.Arg2547_Ser2549del					
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					
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)	SACS	Northeastern Quebec	95%	1 in 22	1 in 431
p.Arg2502Ter, p.Ile2949Phefs					
Bardet-Biedl Syndrome 1	BBS1	General Population	65%	< 1 in 250	< 1 in 700
p.Met390Arg					
Bardet-Biedl Syndrome 10	BBS10	General Population	48%	< 1 in 250	< 1 in 500
p.Cys91Leufs					
Bardet-Biedl Syndrome 12	BBS12	General Population	14%	1 in 501	1 in 582
		Spanish	33%	1 in 949	1 in 1423
p.Phe372Ter					
Bardet-Biedl syndrome 2	BBS2	Ashkenazi Jewish	>95%	1 in 136	<1 in 2693
		General Population	29%	<1 in 500	<1 in 700
p.Arg632Pro, p.Val75Gly, p.Asp104Ala, p.Tyr24Ter, p.Ile314Serfs					
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, Glu122Gln, p.Glu122Lys, 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					
Bilateral Frontoparietal Polymicrogyria (BFPP)	GPR56	General Population	Unknown	Unknown	<1 in 500
p.Cys346Ser, p.Arg38Trp, p.Arg565Trp, p.Cys91Ser, p.Cys91Ser, p.Gln247Cysfs					
Bloom Syndrome	BLM	Ashkenazi Jewish	97%	1 in 108	1 in 3559
		European and North American	35%	Unknown	<1 in 500

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		Japanese	40%	1 in 1646	1 in 2743
p.Trp428Ter, p.Trp567Ter, p.Gln645Ter, p.Tyr736Leufs, p.Trp803Leufs, p.Arg836Glyfs, p.Arg899Ter, p.Gln975Lysfs, p.Ser186Ter					
Bruck syndrome-1	FKBP10	Alaskan Eskimo	99%	1 in 29	1 in 2839
p.Tyr293del					
Canavan Disease	ASPA	Ashkenazi Jewish	98%	1 in 55	1 in 2715
		General Population	50%	< 1 in 100	< 1 in 200
c.433-2A>G, p.Tyr231Ter, p.Glu285Ala, p.Ala305Glu					
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_Ile186delinsPhe, p.Pro227Leu, p.Asp328Gly					
Carpenter Syndrome	RAB23	General Population	67%	< 1 in 500	< 1 in 1500
		Northern European	75%	< 1 in 500	< 1 in 2000
p.Leu145Ter					
Cerebrotendinous Xanthomatosis (CTX)	CYP27A1	General Population	11%	1 in 71	1 in 80
p.Arg395Cys, p.Arg395Ser					
Ceroid Lipofuscinosis, Neuronal, 1	PPT1	Finnish	98%	1 in 70	< 1 in 3000
		General Population (US)	59%	< 1 in 139	< 1 in 300
p.Thr75Pro, p.Arg122Trp, p.Arg151Ter					
Ceroid Lipofuscinosis, Neuronal, 2	TPP1	European Caucasian	63%	1 in 139	< 1 in 350
		Newfoundland	67%	1 in 53	1 in 159
c.509-1G>A, c.509-1G>C, p.Arg208Ter, p.Gly284Val					
Ceroid Lipofuscinosis, Neuronal, 3	CLN3	Finnish	90%	1 in 74	1 in 733
		General Population	85%	1 in 198	1 in 1317
		Portuguese	88%	1 in 528	1 in 4213
c.677+358_677+382del					
Ceroid Lipofuscinosis, Neuronal, 5	CLN5	Finnish	94%	1 in 100	< 1 in 1700
p.Tyr392Ter					
Ceroid Lipofuscinosis, Neuronal, 6	CLN6	Costa Rican	93%	1 in 60	1 in 836
		Pakistani	75%	1 in 139	1 in 554
		Portuguese	81%	1 in 139	1 in 692
p.Glu72Ter, p.Arg106Profs, p.Ile154del					
Ceroid Lipofuscinosis, Neuronal, 8	CLN8	Finnish	99%	1 in 135	< 1 in 13,000
p.Arg24Gly					
Charcot-Marie-Tooth Disease, Type 4D (CMT4D)	NDRG1	Gypsy/Romani	>99%	1 in 11	< 1 in 989
p.Arg148Ter					
Choroideremia (CHM)	CHM	Finnish	90%	< 1 in 5000	< 1 in 57000
c.1609+2dupT					
Ciliary Dyskinesia, Primary, 1	DNAI1	Ashkenazi Jewish	95%	1 in 357	1 in 7118
		Caucasian	67%	1 in 201	1 in 606
		Polish,Slavic	79%	1 in 201	1 in 960

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
p.Tyr408Ter, p.Trp440Ter, p.Gly501Asp, p.Ala542Thr, p.Trp552Ter, c.48+2dupT					
Ciliary Dyskinesia, Primary, 3	DNAH5	Caucasian	15%	1 in 120	1 in 141
p.Pro3606Hisfs					
Citrin Deficiency	SLC25A13	Japanese	>30%	1 in 70	< 1 in 100
c.1180+1G>A, p.Met285Profs					
Coenzyme Q10 deficiency, primary, 7	CoQ4	Ashkenazi Jewish	>95%	1 in 151	1 in 2991
p.Arg240Cys					
Cohen Syndrome	VPS13B	Amish (Ohio)	>95%	1 in 12	1 in 1071
		Finnish	75%	1 in 163	1 in 647
p.Cys1117Phefs, p.Leu3087Phefs					
Congenital Amegakaryocytic Thrombocytopenia (CAMT)	MPL	Ashkenazi Jewish	>95%	1 in 57	<1 in 1128
		General Population	33%	Unknown	Unknown
p.Arg43Ter, p.Arg102Pro, c.79+2T>A					
Congenital Disorder of Glycosylation, Type Ia	PMM2	European Caucasian	53%	1 in 71	1 in 150
p.Phe119Leu, p.Arg141His					
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					
CRB1-associated Retinal Dystrophies	CRB1	European Caucasian	20%	1 in 175	1 in 220
p.Cys948Tyr					
Crigler-Najjar Syndrome	UGT1A1	Dutch	34%	1 in 500	1 in 750
		Tunisian	84%	1 in 500	< 1 in 3000
p.Gln357Arg, p.Lys407Argfs					
Cystic Fibrosis	CFTR	African American	77%	1 in 61	1 in 262
		Ashkenazi Jewish	99%	1 in 24	1 in 2301
		Asian	55%	1 in 94	1 in 205
		Caucasian	92%	1 in 25	1 in 301
		Hispanic	83%	1 in 58	1 in 336

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
c.1393-1G>A, p.Gly551fs, p.Val1360fs, p.Gln359Lys;Thr360Lys , p.Met1Val, p.Arg334Trp (R334W), p.Ile336Lys, p.Thr338Ile, p.Ser341Pro, p.Phe342fs, p.Cys343X , p.Arg347His (R347H), p.Arg347Pro (R347P), p.Arg352Gln, p.Trp361fs, p.Ser364Pro, p.Ser4X, c.1116+1G>A, p.Gln378fs, p.Gln39X, p.Glu384fs, p.Asn386fs, p.Trp401X , c.1209+1G>A, p.Gln414X , p.Ile444fs, p.Lys447fs, p.Ala455Glu (A455E), p.Ala46Asp, p.Ser466X, p.Ser466X, p.Leu467Pro, p.Gly473fs, p.Gly480Cys, p.Ser489X , p.Ser492Phe , p.Gln493X (Q493X), p.Gln493fs, p.Ile506Ser , p.Ile507del (deltaI507), p.Phe508del (deltaF508), p.Tyr515X , p.Val520Phe (V520F), p.Cys524X , p.Gln525X , c.1585-1G>A, c.1585-8G>A, p.Gly542Ter (G542X), p.Ser549Arg (S549R A>C), p.Ser549Asn (S549N), p.Ser549Ile, p.Ser549Arg (S549R T>G), c.165-1G>A, p.Gly551Ser , p.Gly551Asp (G551D), p.Gln552X , p.Arg553Ter (R553X), p.Glu56Lys , p.Ala559Thr, p.Arg560Lys, p.Arg560Thr (R560T), c.1679+1G>C, c.1679+1.6kb A>G, c.1680-1 G>A, p.Ala561Glu , p.Asp565fs, p.Pro574His, p.Val580fs, p.Arg59fs, p.Glu585X , c.1766+1G>A, c.1766+1G>C, c.1766+1 G>T, c.1766+3 A>G, c.1766+5 G>T, p.Glu60X (E60X), p.Met607_Gln634del, p.Gln637fs, p.Ser641fs, p.Arg658fs, p.Asn659fs, p.Thr663fs, p.Pro67Leu, p.Leu671X , p.Lys684fs (c.2183 AA>G), p.Lys684fs (c.2184delA), p.Gln685fs, p.Arg709X , p.Lys710X , c.2175_2176 insA, p.Leu732X, p.Val739fs, p.Arg75X , p.Arg764X , p.Arg785X, p.Arg792X, p.Ser809fs, p.Leu818fs, p.Ser821fs, p.Glu822X, c.2490+1 G>A, p.Glu831X, p.Trp846X , p.Gly85Glu (G85E), p.Tyr849X , p.Arg851X, p.Phe861fs, p.Leu88fs (394delTT), c.2657+5G>A, c.2658-1G>C, p.Gln890X , c.273+1G>A, c.273+3A>C, p.Ser912X , p.Tyr913X , p.Tyr913X , p.Glu92Lys, c.274-1 G>A, p.Glu92X , p.Val922fs, p.Leu927Pro, p.Ser945Leu, p.Ala959fs, p.Gly970Arg, p.Gln98X, c.2988+1G>A, c.2988 G>A, c.2989-1G>A, p.Val1001fs, p.Tyr1014fs, p.Ile1023_Val1024del, p.Gln1042X , p.Ile105fs, c.3140-26 A>G, p.His1054Asp, c.3181G>C, p.Leu1065Pro , p.Arg1066Cys, p.Arg1066His , p.Arg1070Gln, p.Phe1074Leu, p.Leu1077Pro, p.Tyr109fs, p.Trp1089X , p.Tyr1092Ter (Y1092X C>A), p.Tyr1092Ter (Y1092X C>G), p.Asp110His, p.Met1101Lys, p.Glu1104X, p.Asp1152His (D1152H), p.Arg1158X , p.Arg1162Ter (R1162X), p.Arg117Cys, p.Arg117His (R117H), p.Lys1177fs (c.3659delC), p.Thr1179IlefsX17, p.Thr1179fs, p.Ser1196X , p.Asp1202fs, p.Trp1204X , p.Trp1204X , p.Thr1220fs, p.Tyr122X (Y122X), c.3691 delT, p.Gln1238X , c.3717+12191C >T, c.3717+4A>G, c.3718-1G>A, p.Gly1244Glu, p.Lys1250fs (c.3876delA), p.Ser1251Asn, p.Ser1255Pro, p.Ser1255X , p.Leu1258fs (c.3905insT), p.Trp1282Ter (W1282X), c.3873+1G>A, c.3873+2T>C, p.Val1293fs, p.Ile1295fs, p.Ser1297fs, p.Asn1303Lys (N1303K), p.Gln1313X, c.3964-78_4242+577del, p.Gly1349Asp, p.Glu1371X , p.Cys1400X , p.Gln1412X , p.Ile142fs, c.4242+1G>A, p.Glu1418fs, p.Ser1435fs, p.Ile148fs, c.489+1G>T, p.Ile177fs, p.Glu178Arg, p.Ser18fs (CFTRdele2,3), p.Leu183fs, c.579+1G>T, c.579+3A>G, c.579+5 G>A, c.580-1 G>T, p.His199Tyr, p.Pro205Ser , p.Leu206Trp, p.Gln220X , p.Glu241fs, p.Asn268fs, p.Ile269fs, p.Cys276X , p.Gln30X , p.Phe312del , p.Phe316fs (1078delT), p.Glu330X , IVS8-5T, IVS8-7T, IVS8-9T					
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					
Dihydrolipoamide Dehydrogenase Deficiency	DLD	Ashkenazi Jewish	95%	< 1 in 80	< 1 in 1500
p.Tyr35Ter, p.Gly229Cys					
Dihydropyrimidine Dehydrogenase Deficiency	DPYD	General Population	52%	1 in 51	1 in 104
c.1905+1G>A					
Dyskeratosis congenita, autosomal recessive 5	RTEL1	Ashkenazi Jewish	80%	1 in 204	1 in 1014
p.Met492Ile, p.Arg1264His					
Ehlers-Danlos syndrome, type VIIC	ADAMTS2	Ashkenazi Jewish	95%	1 in 187	1 in 3722
p.Gln225Ter					
Ethylmalonic Encephalopathy	ETHE1	General Population	11%	Very rare	< 1 in 500
p.Arg163Trp					
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

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p.Cys56Arg, p.Gln134Ter, p.Glu135Ter, p.Phe301Leu					
Familial Dysautonomia	IKBKAP	Ashkenazi Jewish	>99%	1 in 30	1 in 3000
p.Arg696Pro, c.2204+6T>C, p.Pro914Leu					
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
		Mediterranean Jewish	69%	1 in 4	1 in 12
		Turkish	76%	1 in 5	1 in 18
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

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		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					
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					
GRACILE Syndrome	BCS1L	Finnish	>99%	1 in 109	< 1 in 10,000
p.Ser78Gly					

InheriGen Plus: Pan-Ethnic Carrier Screen

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
Hermansky-Pudlak Syndrome 3	HPS3	Ashkenazi Jewish	89%	1 in 235	1 in 2109
		Puerto Rican	98%	1 in 32	1 in 1483
c.1163+1G>A					
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
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					
Hypophosphatasia	ALPL	Canadian Mennonite	93%	1 in 25	1 in 367
		European Caucasian	17%	1 in 40	1 in 48
		Japanese	53%	Unknown	Unknown
p.Gly334Asp, p.Asp378Val, p.Leu520Argfs, p.Glu191Lys, p.Phe327Leu					
Inclusion body myopathy 2	GNE	Iranian Jewish	99%	1 in 20	< 1 in 1800
		Japanese	73%	Unknown	< 1 in 500
		Korean	80%	Unknown	< 1 in 500
p.Cys44Ser, p.Val603Leu, p.Met743Thr					
Joubert Syndrome 2	TMEM216	Ashkenazi Jewish	99%	1 in 92	1 in 9122
p.Arg73His, p.Arg73Leu					
Junctional Epidermolysis Bullosa, Herlitz type, LAMA3-associated	LAMA3	Pakistani	99%	Unknown	< 1 in 500
p.Arg2270Ter					
Junctional Epidermolysis Bullosa, Herlitz type, LAMB3-associated	LAMB3	General Population	55%	1 in 316	1 in 696
		Italian	17%	1 in 388	1 in 470
		Netherlands	71%	1 in 264	1 in 909

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
957_958ins77, p.Arg42Ter, p.Arg635Ter, p.Arg1009Glyfs, p.Gln1083Ter, p.Gln243Ter					
Junctional Epidermolysis Bullosa, LAMC2-associated	LAMC2	Italian	33%	Unknown	< 1 in 500
p.Arg95Ter					
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					
Lamellar Ichthyosis, Type 1	TGM1	General Population	28%	1 in 224	< 1 in 300
		Norwegian	80%	1 in 151	< 1 in 750
c.877-2A>G					
Leber Congenital Amaurosis, CEP290-associated	CEP290	Northern European	48%	1 in 224	1 in 430
c.2991+1655A>G					
Leber Congenital Amaurosis, RDH12-associated	RDH12	General Population	20%	1 in 501	1 in 626
p.Thr49Met, p.Arg62Ter, p.Gln189Ter, p.Tyr226Cys, p.Ala269Glyfs					
Leigh Syndrome, French Canadian Type	LRPPRC	French Canadian	95%	1 in 23	< 1 in 400
p.Ala354Val					
Leukoencephalopathy with Vanishing White Matter	EIF2B5	General Population	34%	Unknown	< 1 in 500
p.Arg113His					
Limb-Girdle Muscular Dystrophy, Type 2A	CAPN3	Bulgarian	58%	1 in 100	1 in 246
		Croatian	76%	1 in 133	1 in 550
		Italian (Northeastern)	38%	1 in 163	1 in 263
		Russian	45%	< 1 in 100	< 1 in 180
		Turkish	35%	1 in 100	1 in 160
p.Arg490Gln, p.Thr184Argfs					
Limb-Girdle Muscular Dystrophy, Type 2C	SGCG	General Population	Unknown	1 in 350	1 in 350
		Gypsy/Romani	99%	< 1 in 50	< 1 in 5000
p.Phe175Leufs, p.Glu263Lys, p.Cys283Tyr, p.Gly30Trpfs					
Limb-Girdle Muscular Dystrophy, Type 2D	SGCA	Brazilian	64%	1 in 250	1 in 694
		European Caucasian	23%	1 in 250	1 in 325
p.Arg77Cys					
Limb-Girdle Muscular Dystrophy, Type 2E	SGCB	Amish	99% (Indiana)	Unknown	< 1 in 500
		General Population	Unknown	Unknown	< 1 in 500
p.Ser114Phe, p.Val127Serfs, p.Thr151Arg					
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

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		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					
Meckel-Gruber Syndrome, Type 1	MKS1	European	12%	1 in 188	1 in 212
		Finnish	55%	1 in 48	1 in 106
		German	47%	1 in 184	1 in 344
c.1408-35_1408-7del29					
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					
Megalencephalic Leukoencephalopathy with Subcortical Cysts	MLC1	Japanese	71%	Unknown	Unknown
		Libyan Jewish	99%	1 in 40	< 1 in 4000
p.Gly59Glu, p.Ser93Leu					
Metachromatic Leukodystrophy (MLD)	ARSA	Austrian	72%	1 in 101	1 in 355
		European	56%	1 in 101	1 in 228
		Habbanite Jewish	>95%	1 in 5	1 in 391
		Israeli-Arab	30%	1 in 45	1 in 64
		Japanese	18%	Unknown	Unknown
		Polish	54%	1 in 79	1 in 170
		Spanish	56%	1 in 101	1 in 228
p.Pro379Leu, c.1210+1G>A, p.Thr411Ile, p.Pro428Leu, c.465+1G>A, p.Ile181Ser, p.Gly247Arg, p.Asp257His, p.Thr276Met					
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					
Mitochondrial complex 1 deficiency	NDUFAF5	Ashkenazi Jewish	>95%	1 in 290	<1 in 5778
p.Gly250Val					
Mitochondrial DNA depletion syndrome 6	MPV17	Navajo	99%	1 in 20	1 in 1950
p.Arg50Gln					
Mucolipidosis, Type IV	MCOLN1	Ashkenazi Jewish	95%	1 in 96	<1 in 1900

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
c.406-2A>G, g.4127_10560del6434					
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					
Multiple sulfatase deficiency	SUMF1	Ashkenazi Jewish	95%	1 in 279	1 in 5561
p.Ser155Pro					
Nemaline Myopathy 2	NEB	Ashkenazi Jewish	>95%	1 in 108	< 1 in 2100
c.7432-2025_7536+372del2502					
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					
Niemann-Pick Disease, Type A/B	SMPD1	Ashkenazi Jewish	95%	1 in 70	1 in 1405
		Canary Island	>95%	1 in 159	<1 in 3100
		General Population	29%	1 in 197	1 in 277
		North African	79%	Unknown	Unknown
		Saudi Arabian	85%	Unknown	Unknown
p.His423Tyr, p.Arg498Leu, p.Gly579Ser, p.Arg610del, p.Leu304Pro, p.Phe333Serfs					
Niemann-Pick Disease, Type C	NPC1	General Population	>15%	>1 in 174	<1 in 200
p.Gly992Arg, p.Gly992Trp, p.Pro1007Ala, p.Ile1061Thr					
Nijmegen Breakage Syndrome	NBN	Eastern European	85%	1 in 155	< 1 in 1000
p.Lys219Asnfs					
Odonto-onycho-dermal dysplasia/Schopf-Schulz-Passarge Syndrome	WNT10A	General Population	>36%	Unknown	< 1 in 500
p.Cys107Ter, p.Glu233Ter					
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					
Peroxisome biogenesis disorder 1A (Zellweger)	PEX1	General Population	>80%	1 in 140	< 1 in 700
p.Ile700Tyrfs, p.Gly843Asp, p.Gly973Alafs					
Peroxisome biogenesis disorder 5A (Zellweger)	PEX2	Ashkenazi Jewish	95%	1 in 227	1 in 4516
p.Arg119Ter					

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
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
		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					
Phosphoglycerate dehydrogenase deficiency	PHGDH	Ashkenazi Jewish	95%	1 in 453	1 in 9033
p.Val490Met					
Polycystic Kidney Disease, Autosomal Recessive (ARPKD)	PKHD1	Ashkenazi Jewish	90%	1 in 105	1 in 1039
		Finnish	79%	1 in 101	1 in 475
		General Population	30%	1 in 71	1 in 101
		Spanish	40%	1 in 134	1 in 222
p.Gln3392Ter, p.Val3471Gly, p.Arg3482Cys, p.Ile3553Thr, p.Thr36Met, p.Arg496Ter, p.Pro805Leu, p.Ala1254Glyfs, p.Leu1966Thrfs, p.Leu1966Profs, p.Ile222Val, p.Ile2331Lys, c.7350+653A>G, p.Arg2671Ter, p.Ile2944Hisfs, p.Ile2957Thr, p.Ile3177Thr, p.Asp3230Valfs					
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					

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
Primary Hyperoxaluria, Type 2	GRHPR	European Caucasian	30%	1 in 500	< 1 in 715
p.Asp35Thrfs					
Progressive Pseudorheumatoid Dysplasia (PPD)	WISP3	Middle Eastern	57%	Unknown	< 1 in 500
p.Cys70Ter, p.Cys132Tyr, p.Cys197Ter					
Prolidase Deficiency	PEPD	Druze	67%	1 in 21	1 in 62
p.Ser202Phe					
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.Gly407Argfs, p.Arg430Trp, p.Val412Cysfs, p.Thr448Ile, p.Arg185Trp, p.Glu188Lys					
Pseudoxanthoma Elasticum	ABCC6	European	28%	1 in 80 to 1 in 160	< 1 in 110
p.Arg1141Ter					
Pycnodysostosis	CTSK	Danish	90%	1 in 76	1 in 746
p.Gly79Glu, p.Leu309Pro					
Pyridoxine-Dependent Epilepsy	ALDH7A1	Dutch	64%	< 1 in 260	< 1 in 725
		European Caucasian	33%	< 1 in 260	< 1 in 390
p.Glu427Gln					
Pyruvate Carboxylase Deficiency	PC	Canadian Indian	> 99%	1 in 10	< 1 in 850
		General Population	13%	1 in 250	1 in 288
p.Ala610Thr, p.Arg631Gln					
Retinal dystrophies, RLBP1-associated	RLBP1	Newfoundland	99%	Unknown (Newfoundland dystrophy)	< 1 in 500
		Northern Swedish	94%	1 in 60 (Bothnia dystrophy)	< 1 in 900
p.Lys47Lys, c.141+2T>C, p.Arg234Trp					
Retinitis Pigmentosa 25	EYS	Moroccan Jewish	Unknown	1 in 94	Unknown
p.Thr135Leufs					
Retinitis pigmentosa 59	DHDDS	Ashkenazi Jewish	95%	1 in 117	1 in 2317
p.Lys42Glu					
Retinoschisis 1, X-Linked, Juvenile	RS1	European Caucasian	35%	< 1 in 2500	< 1 in 3800
		Finnish	95%	< 1 in 7500	< 1 in 150,000
p.Glu72Lys, p.Gly74Val, p.Arg102Trp, p.Gly109Arg, p.Arg141Cys, p.Gln154Ter, p.Pro192Ser, p.Arg200Cys, p.Arg213Trp					
Rhizomelic Chondrodysplasia Punctata, Type 1	PEX7	European Caucasian	72%	< 1 in 159	< 1 in 550
p.Gly217Arg, p.Ala218Val, p.Leu292Ter, c.903+1G>C					
Salla Disease	SLC17A5	Finnish	97%	1 in 100 to 1 in 200	< 1 in 3000
p.Leu336Trpfs, p.Arg39Cys					
Sandhoff Disease	HEXB	Argentinian	78%	1 in 26	1 in 115
		Cypriot Maronites	86%	1 in 7	1 in 43
.445+1G>A, p.Met26Cysfs					
Sanfilippo, Type A	SGSH	European Caucasian	48%	1 in 147	1 in 281
		Italian	33%	1 in 147	1 in 220

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DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
p.Val361Serfs, p.Ser66Trp, p.Arg74Cys, p.Arg245His					
Sanfilippo, Type B	NAGLU	Japanese	42%	1 in 200	1 in 345
		Spanish Portuguese	38%	1 in 187	1 in 300
p.Arg565Trp, p.Arg565Gln, p.Arg565Pro, p.Arg234Cys					
Sanfilippo, Type C	HGSNAT	Dutch	69%	1 in 346	1 in 1105
		European Caucasian	55%	1 in 346	1 in 762
		Portuguese	83%	1 in 457	1 in 2683
		Spanish/Moroccan	70%	1 in 384	1 in 1278
p.Arg344Cys, p.Arg384Ter, p.Ser518Phe, p.Ser541Leu, c.234+1G>A, c.372-2A>G, c.493+1G>A, p.Val176Cysfs, c.852-1G>A					
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					
Sialidosis, type II	NEU1	Northern Spain (Seville)	99%	Very rare	< 1 in 500
p.Leu270Phe					
Sjogren-Larsson Syndrome (SLS)	ALDH3A2	European	28%	1 in 250	1 in 345
		Swedish	79%	1 in 50 to 1 in 200	<1 in 236
p.Glu433Argfs, p.Pro315Ser					
Smith-Lemli-Opitz Syndrome	DHCR7	Ashkenazi Jewish	>75%	1 in 37	1 in 155
		European Caucasian	63%	1 in 30 to 1 in 70	< 1 in 80
		General Population	>75%	1 in 69	1 in 294
p.Arg352Trp, p.Arg352Gln, p.Cys380Tyr, p.Arg404Cys, p.Gly410Ser, p.Arg446Gln, p.Glu448Lys, p.Thr93Met, p.Leu109Pro, p.His119Leu, p.Trp151Ter, p.Trp151Ter, p.Leu157Pro, p.Arg242Cys, p.Arg242His, p.Trp248Cys, p.Phe302Leu, c.964-1G>C, p.Val326Leu					
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	SLC1A4	Ashkenazi Jewish	90%	1 in 118	1 in 1171
p.Glu256Lys					
Spinal Muscular Atrophy	SMN1	African American	71%	1 in 66	1 in 121 (2 copies)
		Ashkenazi Jewish	90%	1 in 41	1 in 350 (2 copies)
		Asian	93%	1 in 53	1 in 628 (2 copies)
		Caucasian	95%	1 in 35	1 in 632 (2 copies)
		Hispanic	91%	1 in 117	1 in 1061 (2 copies)
Exon 7					
Stargardt disease, type 1	ABCA4	General Population	15%	1 in 50	1 in 60
p.Gly863Ala, p.Ala1038Val, c.5018+2T>C, p.Gly1961Glu					
Stuve-Wiedemann syndrome (Schwartz-Jampel Syndrome Type 2)	LIFR	General Population	26%	Unknown	< 1 in 500
		United Arab Emirates	99%	1 in 70	< 1 in 6900
p.Glu219Glyfs					
Sulfate Transporter-Related Osteochondrodysplasia	SLC26A2	Finnish	96%	1 in 100	1 in 2473
		General Population	60%	Unknown	Unknown

InheriGen Plus: Pan-Ethnic Carrier Screen

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		Portuguese	89%	Unknown	Unknown
c.-26+2T>C, p.Val341del, p.Cys653Ser, p.Arg178Ter, p.Arg279Trp					
Tay-Sachs Disease	HEXA	Ashkenazi Jewish	95%	1 in 30	1 in 548
		Cajun	99%	1 in 27	1 in 2601
		French Canadian	64%	1 in 73	1 in 205
		General Population	43%	1 in 264	1 in 458
		Japanese	64%	1 in 127	1 in 355
		Moroccan Jewish	80%	1 in 60	1 in 290
c.-2564_253+5128delinsG, p.Ile335Phe, c.1073+1G>A, p.Ser426Ilefs, p.Cys458Tyr, c.1421+1G>C, p.Arg499His, p.Arg504Cys, c.346+1G>C, p.Arg170Trp, p.Arg170Gln, p.Arg178His, p.Arg178Leu, c.571-1G>T, p.Leu205Trpfs, p.Arg247Trp, p.Arg249Trp, p.Gly269Ser, c.805+1G>A, p.Phe305del					
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					
Usher Syndrome, Type 1B	MYO7A	General Population	10%	1 in 139	1 in 154
		North African	64%	Unknown	Unknown
p.Met599Ile, p.Ala826Thr, p.Arg1240Gln, p.Arg1861Ter, p.Ala2009Profs					
Usher Syndrome, Type 1C	USH1C	Acadian	>95%	Unknown	<1 in 500
		French Canadian	78%	1 in 91	1 in 406
		General Population	41%	1 in 416	1 in 704
p.Val72=, p.Arg80Profs, c.497-2delA					
Usher Syndrome, Type 1D	CDH23	General Population	18%	1 in 186	1 in 227
p.Arg1746Gln, c.6050-9G>A					
Usher Syndrome, Type 1F	PCDH15	Ashkenazi Jewish	75%	1 in 147	1 in 585
p.Tyr684Ter, p.Leu1424Phefs, p.Arg3Ter, p.Arg245Ter					
Usher Syndrome, Type 2A	USH2A	French Canadian	61%	Unknown	Unknown
		General Population	21%	1 in 78	1 in 98
		Spanish	8%	Unknown	Unknown
p.Arg334Trp, p.Cys419Phe, p.Glu767Serfs, p.Cys1447Glnfs, p.Ser307Argfs					
Usher Syndrome, Type 3	CLRN1	Ashkenazi Jewish	95%	1 in 106	1 in 2188
		Finnish	>95%	1 in 98	<1 in 1951.
p.Asn48Lys, p.Tyr189Ter					
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
p.Arg389His, p.Phe443Profs, p.Val88Trpfs, c.589+1G>A					
Walker-Warburg Syndrome	FKTN	Ashkenazi Jewish	>95%	1 in 144	1 in 14179
		Japanese	86%	1 in 52	1 in 367
c.*4287_*4288ins3062, p.Phe390Ilefs					
Werner Syndrome	WRN	General Population	26%	1 in 224	1 in 302

InheriGen Plus: Pan-Ethnic Carrier Screen

DISEASE INFORMATION & MUTATIONS TESTED (cont)

Disease	Gene	Ethnicity	Detection Rate	Carrier Frequency	Residual Risk
		Japanese	71%	1 in 71	1 in 241
p.Arg369Ter, p.Arg389Glufs, p.Cys727Leufs, c.3139-1G>C, p.Asn1197Thrfs					
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					