Condition	Gene	Mutations	Population	Detection Frequency	References
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	c.914_915delTT c.206_207delCT c.505_506delTC c.208G>C c.122G>A c.109G>A c.109G>T c.835G>A c.561+1G>A c.804C>A c.528T>G c.528T>A c.501C>G c.499T>A c.121C>T	General population Saudi Arabia Portugese	>65% >95% >85%	Cardoso M.L. et al. (2004); Menao S. et al. (2009)
3-Methylcrotonyl-CoA Carboxylase Deficiency 1	MCCC1	c.1330C>T c.1310T>C c.1277T>C c.1225C>T c.1155A>C c.1114C>T	European population	>28%	Baumgartner, M. R. et al. (2001); Fonseca, H. et al.(2016); Grünert, S. C. et al. (2012)
3-Methylcrotonyl-CoA Carboxylase Deficiency 2	MCCC2	c.295G>C c.302C>A c.380C>G c.463C>T c.464G>A c.499T>C c.538C>T c.569A>G c.803G>C c.838G>T c.929C>G c.994C>T c.1309A>G	General population	>35%	Baumgartner, M. R. et al. (2001); Fonseca, H. et al.(2016); Grünert, S. C. et al. (2012)
Abetalipoproteinemia	MTTP	c.2593G>T	Ashkenazi Jewish	>95%	Benayoun, L. *et al.* (2007)
Acyl-CoA Oxidase I Deficiency	ACOX1	c.1789_1792delACTC c.190_192delGTC c.191delT c.1276_1277delGT c.372_389delCATGCCCGCCTGGAACTT c.167dupG c.1872T>A c.926A>G c.928T>C c.904C>T c.538+1G>A c.431-1G>A c.442C>T	General population	>73%	Ferdinandusse S. et al. (2007)
Aicardi-Goutières Syndrome	SAMHD1	c.359_370delATCCTATCCATG c.649_650insG c.1609-1G>C c.1642C>T c.1503+1G>T c.1411-2A>G c.1324C>T c.1153A>G c.1106T>C c.760A>G c.625G>A c.602T>A c.368A>C c.428G>A c.427C>T c.445C>T c.433C>T c.434G>A c.490C>T	General population	>75%	Rice G.I. et al. (2009)
		c.4709G>C c.4964T>G c.5048G>A c.4707-1G>C c.4708T>C c.4717T>C			

Condition	Gene	Mutations	Population	Detection Frequency	References
Alport Syndrome, X-Linked	COL4A5	c.4718G>C c.4719T>A c.4720G>A c.4769C>T c.4774T>C c.4775G>T c.4784G>T c.4786T>G c.4800G>A c.4805G>T c.4808A>G c.4809T>A c.4811C>T c.4821G>A c.4821+1G>A c.4880T>C c.4912T>G c.4913G>A c.4914T>G c.4931G>C c.4931G>A c.4949C>G c.4994G>A c.4994+2T>C c.4995-2A>G c.5038C>T c.5047C>T c.5048G>T c.5048G>C c.5050T>C c.5052T>G c.5054A>C c.5059T>G c.5060G>T c.5069G>A	General population	>79%	Pont-Kingdon G. et al. (2009)
Alstrom Syndrome	ALMS1	c.2800dupT c.10477C>T c.10769delC c.10819C>T c.11074A>T c.11310_11313delAGAG c.3334delG c.4150dupA c.5305C>T c.5580T>G c.5963C>G c.8158C>T c.8500G>T c.9743C>A c.2723C>G c.2816T>A c.4177C>T c.10543C>T c.10879C>T c.10986G>A c.11201C>A c.2702C>G c.2772dupT c.3294_3295delAA c.3214_3215delAG c.3419C>G c.4139_4140delAC c.4033C>T c.4174C>T c.4199dupT c.4207G>T c.4268T>A c.5129T>G c.5139T>G c.5417C>G c.5449C>T c.5567_5568delCT c.6111_6112delTC c.8002C>T c.8171_8181del c.8218C>T c.8309delC	General population	>46%	Marshall J. D. et al. (2015)

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.8388dupA			
		c.8471delG			
		c.8573C>A			
		c.10382-2A>G			
		c.10477delC			
		c.10555_10558delGACA			
		c.10563_10564delTA			
		c.10573_10574delAT			
		c.10603_10604delGAinsT			
		c.10603_10608delGACAAGinsTCAAA			
		c.10668_10671delAGAA			
		c.10774C>T			
		c.10784_10785delTG			
		c.10843G>T			
		c.10939G>T			
		c.10969C>T			
		c.11110_11128del			
		c.11308dupA			
		c.11379delT			
		c.11408delG			
		c.11410C>T			
		c.11443C>T			
		c.11454C>G			
Andermann Syndrome	SLC12A6	c.2436+1delG c.1478_1485delTTCCCTCT c.901delA c.1584_1585delCTinsG c.2028_2029insT c.3031C>T c.2023C>T c.619C>T c.2416G>T c.963C>A c.655C>T c.630G>A	General population French Canadian	>95% >95%	Uyanik G. et al. (2006); Salin-Cantegrel A. et al. (2007)
Aromatase Deficiency	CYP19A1	c.1224delC c.469delC c.1303C>T c.1310G>A c.1123C>T c.1094G>A c.629-3C>A c.628G>A c.452-1G>A c.1051C>T	General population	>60%	Herrmann B. L. et al. (2002); Marino R. et al. (2015)
Arthrogryposis Mental Retardation Seizures	SLC35A3	c.514C>T	Ashkenazi Jewish	>50%	Edvardson, S. *et al.* (2013)
Asparagine Synthetase Deficiency	ASNS	c.1648C>T c.1193A>G c.1084T>G c.17C>A c.1614G>A c.1556G>A c.1165G>C c.146G>A c.139T>G	General population Sephardic Jewish - Iranian	>94% >95%	Ruzzo E. K. et al. (2013); Ben-Salem S. et al. (2015)
Aspartylglycosaminuria	AGA	c.200_201delAG c.916T>C c.904G>A c.488G>C c.302C>T c.179G>A c.214T>C c.940+1G>A c.503G>A c.490C>T c.473G>A c.439T>C c.404T>C c.346C>T c.319C>T c.299G>A c.281+1G>T c.192T>A	General population Finnish	>70% >95%	Ikonen E. et al. (1991); Saarela J. et al. (2001)
		c.10709C>G c.10664T>A c.10658T>C			

Condition	Gene	Mutations	Population	Detection Frequency	References
Autosomal Recessive Polycystic Kidney Disease	PKHD1	c.10444C>T c.10412T>G c.10219C>T c.10174C>T c.9719G>A c.9718C>T c.9689delA c.9683C>A c.9646C>T c.9530T>C c.9053C>T c.8950+1G>T c.8870T>C c.8824C>T c.8555-2A>G c.8555-2A>C c.8068T>C c.8050C>T c.8011C>T c.7916C>A c.7350+1G>T c.6992T>A c.6910C>T c.5895dupA c.5513A>G c.5485C>T c.5452C>T c.5448T>A c.5236+1G>A c.4991C>T c.4882C>G c.4870C>T c.3761_3762delCCinsG c.3367G>A c.2980C>T c.2854G>A c.2854G>C c.2822-1G>C c.2452C>T c.2414C>T c.2408-2A>G c.2407+1G>A c.2341C>T c.2279G>A c.2264C>T c.2192C>A c.2141-2A>C c.1505A>T c.1486C>T c.1480C>T c.1458C>A c.1397G>A c.1095G>A c.1068dup c.982C>T c.664A>G c.603-1G>A c.603-2A>G c.383delC c.370C>T c.340C>T c.282-2A>T c.107C>T	Ashkenazi Jewish General population Caucasian	>90% >69% >70%	Sweeney, W. E. & Avner, E. D. (1993); Shi, L. et al.(2017)
Bardet-Biedl Syndrome (BBS1-related)	BBS1	c.851delA c.436C>T c.442G>A c.1169T>G c.1645G>T c.913G>A c.433-2A>G c.479G>A c.479+2T>G c.831-3C>G c.855C>A c.871C>T c.1609-2A>T	General population	>70%	Mykytyn K. et al. (2003); Janssen S. et al. (2011)
		c.323C>G			

Condition	Gene	Mutations	Population	Detection Frequency	References
Bardet Biedl Syndrome (BBS12-related)	BBS12	c.335_337delTAG c.760G>T c.865G>C c.1063C>T c.1375C>T c.1483_1484delGA c.1502C>T	European population	>50%	Stoetzel, C. et al. (2007); Burstedt, M. et al. (2013); Burstedt, M. S. et al. (2001); Muller, J. et al. (2010)
Beta Thalassemia	HBB	c.415G>C c.383_385delAGG c.316-2A>G c.316-2A>C c.316-3C>G c.316-3C>A c.316-106C>G c.316-146T>G c.316-197C>T c.315+1G>A c.298G>A c.295G>A c.287dupA c.277C>T c.271G>T c.257T>C c.251delG c.247A>G c.230delC c.217_221delAGTGAinsT c.216_217insA c.216dupT c.203_204delTG c.199A>G c.193delG c.184A>T c.182T>A c.176C>G c.143_144insA c.135delC c.130G>T c.126_129delCTTT c.127_129delTTT c.128T>C c.127T>G c.114_120delGACCCAG c.118C>T c.116_117delCC c.114G>A c.113G>A c.112delT c.108C>A c.108delC c.103G>T c.93-22_95del25 c.93G>T c.93-1G>C c.93-1G>A c.93-21G>A c.92+6T>C c.92+5G>T c.92+5G>A c.92+5G>C c.92+2T>A c.92+2T>C c.92+1G>T c.92+1G>A c.92+1G>C c.92G>C c.86T>A c.85dupC c.82G>T c.80A>G c.75T>A c.70_72delGTT c.70G>T c.59A>G c.52A>T c.51delC c.47G>A	Mediterranean Middle-Eastern Thai Chinese African-American	>95% >95% >95% >95% >80%	Origa, R. (2000)

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.45dupG			
		c.36delT			
		c.34G>A			
		c.27dupG			
		c.25_26delAA			
		c.20delA			
		c.17_18delCT			
		c.4delG			
		c.2T>G			
		c.2T>C			
		c.-29G>A			
		c.-50A>C			
		c.-78A>G			
		c.-50-29A>G_c.-79A>G			
		c.-80T>A			
		c.-81A>G			
		c.-136C>G			
		c.-137C>A			
		c.-137C>G			
		c.-50-88C>T_c.-138C>T			
		c.-138C>A			
Biotinidase Deficiency	BTD	c.98_104delGCGGCTGinsTCC c.100G>A c.235C>T c.511G>A c.528G>T c.755A>G c.1207T>G c.1466A>C c.1368A>C c.1612C>T c.1595C>T c.136G>T c.160G>T c.171T>G c.184G>A c.184G>T c.190G>A c.192G>C c.194A>G c.245C>A c.257T>G c.262C>T c.278A>G c.283C>T c.298G>A c.309+1G>T c.466C>T c.469C>T c.470G>A c.479G>T c.518T>G c.557G>A c.559C>T c.582C>G c.584A>G c.587C>G c.595G>A c.631C>T c.643C>T c.646T>A c.652G>C c.654G>C c.664G>C c.683A>G c.695T>C c.701C>T c.709G>A c.758C>T c.770T>A c.783C>G c.814T>G c.836T>A c.836T>G c.856A>G c.865G>C c.1096T>C	General population	>60%	Wolf, B. (2000); Norrgard K. J. et al. (1999); Norrgard K. J. et al (1997); Mühl A. et al. (2001); Kasapkara Ç. S. et al. (2015); Pomponio R. J. et al. (1998)

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.1126C>T c.1129G>T c.1157G>A c.1158G>A c.1214T>C c.1249G>T c.1253G>C c.1268G>C c.1271G>A c.1271G>C c.1275T>G c.1284C>A c.1313A>G c.1314T>A c.1333G>A c.1334G>T c.1339C>T c.1352G>A c.1460G>A c.1461G>A c.1463G>A c.1489C>T c.1511T>A c.1526C>G c.1557T>G c.1610G>A c.1613G>A c.1627G>C c.1628A>T			
Canavan Disease	ASPA	c.433-2A>G c.454T>C c.640G>T c.654C>A c.692A>G c.693C>A c.731A>G c.820G>A c.854A>C c.859G>A c.867C>A c.914C>A	Ashkenazi Jewish European population	>98% >60%	Feigenbaum, A. et al. (2004); Kaul, R. et al. (1994); Matalon, R. & Matalon, K. M. (Elsevier, 2015)
Carpenter Syndrome	RAB23	c.434T>A	General population	>56%	Jenkins D. et al. (2007); Jenkins D. et al. (2001)
Choreacanthocytosis	VPS13A	c.6059delC	Ashkenazi Jewish	>67%	Rampoldi, L. *et al.* (2001); Dobson-Stone, C. *et al.* (2002); Siegl, C. *et al.* (2013); Lossos, A. *et al.* (2005)
Choroideremia, X-Linked	CHM	c.1584_1587delTGTT c.1609+2dupT c.1131_1133dupATA c.877C>T c.703-1G>C c.715C>T c.1609+2T>A c.1520A>G c.1144G>T c.1138C>T c.1019C>A c.820-2A>G c.808C>T c.799C>T c.757C>T	General population	>70%	Simunovic M. P. et al. (2016)
Citrin Deficiency	SLC25A13	c.1095delT c.852_855delTATG c.1638_1660dup23 c.1399C>T c.1063C>T c.1177+1G>A c.955C>T c.754G>A c.615+5G>A c.74C>A c.127C>T c.1592G>A c.1078C>T c.674C>A c.615+1G>C c.550C>T	Chinese Japanese Korean Taiwanese	>80% >80% >80% >80%	Kobayashi K. et al. (2003)

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.493C>T c.70-1G>A			
Combined Oxidative Phosphorylation Deficiency 3	TSFM	c.919C>T c.997C>T c.1007G>A	Finnish	>75%	Ahola S. et al. (2014); Shamseldin H. E. et al. (2012)
Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	PMM2	c.193G>T c.338C>T c.357C>A c.385G>A c.368G>A c.395T>C c.422G>A c.415G>A c.470T>C c.484C>T c.563A>G c.691G>A c.677C>G c.653A>T c.179-1G>T c.205C>T c.255+1G>A c.255+2T>C c.256-2A>G c.256-1G>C c.323C>T c.337C>A c.347+1G>A c.348-2A>C c.348-1G>C c.349G>C c.355T>C c.359T>C c.367C>T c.421C>T c.430T>C c.432C>A c.442G>A c.447+5G>A c.524-2A>G c.527G>A c.560G>A c.580C>T c.620T>C c.623G>C c.639+1G>A c.647A>T c.652C>G c.669C>G	Caucasian Ashkenazi Jewish General population	>80% >90% >80%	Matthijs G. et al. (2000); Le Bizec C. et al. (2005); Shi L. et al. (2017)
Congenital Neutropenia (HAX1-related)	HAX1	c.91delG c.125dupG c.430dup c.130_131insA c.256C>T c.568C>T c.383C>G	General population	>95%	Klein C. et al. (2007); Smith B. N. et al. (2009)
Crigler Najjar Syndrome, Type I	UGT1A1	c.513_515delCTT c.524T>A c.840C>A c.864+1G>C c.923G>A c.991C>T c.992A>G c.1021C>T c.1069C>T c.1070A>G c.1084+1G>T c.1085-2A>G c.1124C>T c.1198A>G	Tunisian Sardinian	>99% >70%	Francoual, J. et al. (2002)
		c.1466C>A c.1477_1478delCA c.1477C>T c.1519_1521delATC c.1521_1523delCTT c.1520_1522delTCT c.1545_1546delTA			

Condition	Gene	Mutations	Population	Detection Frequency	References
Cystic Fibrosis*	CFTR	c.1558G>T c.1572C>A c.1573C>T c.1585-8G>A c.1585-1G>A c.1624G>T c.1645A>C c.1646G>A c.1647T>G c.1648G>T c.1652G>A c.1654C>T c.1657C>T c.1675G>A c.1679G>A c.1679G>C c.1679+1G>C c.1766+1G>A c.1766+1G>C c.1766+1G>T c.1973_1985del13insAGAAA c.2012delT c.2017G>T c.2051_2052delAAinsG c.2052dupA c.2052delA c.2125C>T c.2128A>T c.2175dupA c.2988+1G>A c.3472C>T c.3484C>T c.3528delC c.3532_3535dupTCAA c.3587C>G c.3611G>A c.3612G>A c.3659delC c.3731G>A c.3744delA c.3747delG c.3752G>A c.3761T>G c.3764C>A c.3764C>G c.3773dupT c.3846G>A c.3873+1G>A c.3909C>G c.1040G>C c.489+1G>T c.170G>A c.171G>A c.175dupA c.178G>T c.223C>T c.233dupT c.254G>A c.262_263delTT c.263T>G c.273+1G>A c.273+3A>C c.274-1G>A c.274G>T c.292C>T c.310delA c.313delA c.325_327delTATinsG c.366T>A c.409delC c.442delA c.579+1G>T c.1397C>A c.1397C>G c.1400T>C c.1418delG c.948delT	General population European population US population Ashkenazi Jewish Finnish	>75% >70% >70% >97% >95%	Farrell, P. M. (2008); Elliott, A. M. et al. (2012); Bobadilla, J. L., et al. (2002); Abeliovich, D., et al. (1992); Kerem, E., et al. (1997); Kinnunen, S., et al. (2005); Macek, M.Jr., et al. (1997); The Clinical and Functional TRanslation of CFTR (CFTR2); available at http://cftr2.org

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.3276C>A c.2668C>T c.2215delG c.3302T>A c.233dupT c.1202G>A c.658C>T c.2464G>T c.3889dup c.580-1G>T c.1393-1G>A c.1029delC c.1923_1931delinsA c.803delA c.1792_1798del c.828C>A c.115C>T c.2908G>C c.3194T>C c.11C>A c.2737_2738insG c.3883_3886delATTT c.3293G>A c.595C>T c.3276C>G c.3196C>T c.2537G>A c.2538G>A			
Factor XI Deficiency	F11	c.326-1G>A c.400C>T c.403G>T c.408C>A c.438C>A c.901T>C c.976C>T	Ashkenazi Jewish Europe	>95% >75%	Peretz, H. et al. (2013); Asakai, R., et al. (1991); Shpilberg, O., et al.(1995); Asselta, R., et al. (2017); Peretz, H., et al. (2013)
Familial Dysautonomia	ELP1	c.2741C>T c.2204+6T>C c.2087G>C	Ashkenazi Jewish	>99%	Chaverra, M. et al. (2017); Shohat, M. & Weisz Hubshman, M. (2003); Peters, N. et al. (Springer Berlin Heidelberg, 2009)
Fanconi Anemia, Type C	FANCC	c.992_995delAGCA c.662delA c.489_490delGA c.487_490delGAGA c.356_360delCTCAT c.377_378delGA c.67delG c.455dupA c.1598_1599insAG c.1661T>C c.1642C>T c.996+1G>A c.996G>A c.844-1G>C c.843+1G>A c.553C>T c.521+1G>A c.520C>T c.456+4A>T c.66G>A c.65G>A c.165+1G>T c.37C>T c.1333C>T c.1628C>A c.1599G>A c.996+1G>T c.946C>T c.896+2T>G c.896+1G>C c.843+1G>C c.808A>T c.686+1G>T c.535C>T c.457-1G>T c.388G>T c.368C>G c.346-1G>A c.3G>T	Ashkenazi Jewish General population	>95% >84%	Verlander P. C. et al. (1995); Verlander P. C. et al. (1994)

Condition	Gene	Mutations	Population	Detection Frequency	References
Fanconi Anemia, Type G	FANCG	c.1480+1G>C c.925-2A>G c.307+1G>C	French-Canadian Korean/Japanese Portuguese-Brazilian	>80% >65% >80%	Auerbach, A. D. et al. (2003); Mehta, P. A. & Tolar, L. (2002); Fanconi Anemia: Guidelines for Diagnosis and Management, Fourth Edition (2014)
Gaucher Disease	GBA	c.84dup c.115+1G>A c.1343A>T c.1342G>C c.1297G>T c.1226A>G c.1246G>A c.475C>T c.1504C>T c.1448T>C c.1505G>A c.1459G>A c.1448T>G c.1397T>G c.1361C>G c.1348T>A c.1319C>T c.1312G>A c.1309G>T c.1279G>T c.1240G>T c.1228C>G c.1227C>A c.1226A>C c.526G>A c.521A>G c.509G>T c.481C>T c.476G>A	Ashkenazi Jewish General population	>97% >75%	Beutler E. et al. (1992); Beutler E. et al. (1993); Shamseddine A. et al. (2004)
Glutaric Acidemia, Type 2A	ETFA	c.797C>T c.470T>G c.346G>A	General population	>50%	Freneaux E. et al. (1992)
Glycine Encephalopathy (GLDC-related)	GLDC	c.2315+2T>A c.2311G>A c.2306C>T c.2293C>T c.2284G>A c.2281G>A c.2281G>C c.2258A>C c.2216G>A c.2203-2A>G c.1691G>T	Finnish General population	>78% >10%	Van Hove, J., Coughlin, C. & Scharer, G. (2002)
		c.79delC c.96_97delGG c.715delG c.793delC c.788delA c.980_982delTCT c.150_151delGT c.262delG c.537delT c.462_466delTTTGT c.16_19dupAATG c.734dupG c.853_854dupAA c.379_380dupTA c.59A>G c.47C>G c.113A>T c.70C>T c.706T>A c.724C>T c.764C>T c.770C>T c.792C>A c.794T>C c.797G>T c.809G>T c.808G>C c.883C>T c.892T>C c.1012G>T c.1022T>A			

Condition	Gene	Mutations	Population	Detection Frequency	References
Glycogen Storage Disease, Type 1A	G6PC	c.1039C>T c.149G>A c.209G>A c.161A>C c.202G>A c.187T>C c.189G>A c.228G>C c.229T>C c.310C>T c.323C>T c.328G>A c.338C>T c.241G>A c.231-1G>A c.248G>A c.247C>T c.384C>A c.370G>A c.356A>T c.361A>G c.562G>A c.562G>C c.551G>T c.551G>A c.536A>C c.516C>A c.509G>A c.508C>T c.497T>C c.497T>G c.467G>T c.563G>A c.626A>G c.632T>C c.648G>T c.664G>A c.664G>C c.79C>T c.230+1G>C c.230+4A>G c.255C>A c.258G>A c.381C>A c.447-1G>A c.560C>G c.707G>A c.731G>A c.969C>A	Ashkenazi Jewish General population	>98% >90%	Ekstein J. et al. (2004); Lei K. J. et al. (1995); Chevalier-Porst F. et al. (1996); Stroppiano M. et al. (1999); Chou J. Y. et al (2008)
Glycogen Storage Disease, Type 1B	SLC37A4	c.1042_1043delCT c.1243C>T c.742C>T c.572C>T c.547T>C c.352T>C c.68T>G c.83G>A c.1099G>A c.1015G>T c.1016G>A c.1124-1G>A c.1123+1G>C c.1063G>T c.381+1G>A c.287G>A c.148+2T>C c.110C>A c.82C>T c.59G>A c.59G>C c.1A>G	General population	>50%	Galli L. et al. (1999)
		c.3G>C c.16C>T c.18_19delGA c.22C>T c.82+1G>A c.1222C>T			

Condition	Gene	Mutations	Population	Detection Frequency	References
Glycogen Storage Disease, Type 3	AGL	c.1282A>G c.1384delG c.1423+1G>T c.2039G>A c.2590C>T c.2605C>T c.2681+1G>A c.2681+1G>T c.3363-1G>A c.3439A>G c.3444C>G c.3589-1G>A c.3613C>T c.3652C>T c.3682C>T c.3965delT c.3980G>A c.4260-12A>G c.4342G>C c.4347+1G>A c.4348G>T c.4353G>T c.4456delT c.4459C>T c.4481+1G>C c.4481+2T>G	Sephardic Jewish - Moroccan Faroese Finnish	>99% >99% >99%	Kishnani, P. S. et al. (2010); Santer, R. et al. (2001); Goldstein, J. L. et al. (2010); Parvari, R. et al. (1997); Rousseau-Nepton, I., et al. (2015)
Glycogen Storage Disease, Type 7	PFKM	c.115C>T c.329G>T c.116G>C c.450+1G>A c.496C>T	Ashkenazi Jewish	>95%	Sherman, J. B. et al. (1994)
GRACILE Syndrome	BCS1L	c.133C>T c.166C>T c.232A>G c.245C>A c.296C>T c.320+1G>T c.464G>C c.547C>T c.548G>A c.550C>T c.556C>T c.598C>T c.655+1G>A	Finnish	>95%	de Lonlay, P. et al. (2001); Visapää, I. et al. (2002)
Hereditary Fructose Intolerance	ALDOB	c.865delC c.360_363delCAAA c.1005C>G c.10C>T c.720C>A c.612T>G c.524C>A c.442T>C c.448G>C c.324+1G>A c.178C>T c.1095G>C c.1013C>T c.911G>A c.888G>A c.800-2A>C c.625-1G>A c.625-2A>G c.612T>A c.380-1G>A c.380-2A>G c.379+1G>A c.325-1G>A c.324+2T>A c.324G>A c.941G>A	Spanish Central European Northern India US	>95% >90% >77% >64%	Sánchez-Gutiérrez, J.C. et al. (2002); Santer R. et al. (2005); Bijarnia-Mahay S. et al. (2015); Coffee E.M. et al. (2010)
Homocystinuria, Type cblE	MTRR	c.903+469T>C c.1361C>T c.1459G>A c.1573C>T c.1379T>G	Caucasian	>72%	Zavadáková P. et al. (2005)
Hydrolethalus Syndrome	HYLS1	c.632A>G	Finnish	>95%	Mee L. et al (2005); Paetau A. et al. (2008)

Condition	Gene	Mutations	Population	Detection Frequency	References
Inclusion Body Myopathy, Type 2	GNE	c.2228T>C c.2215G>A c.1863C>A c.1760T>C c.1820G>A c.1714G>C	Iranian Jewish Asian	>95% >50%	Haghighi, A. et al. (2015); Cho, A. et al. (2014); Park, Y.-E. et al. (2012)
Isovaleric Acidemia	IVD	c.941C>T	General population	>50%	Vockley, J. & Ensenauer, R. (2006); Mohsen, A.-W. A. *et al.* (1998); Moorthie, S., *et al.* (2014); Ensenauer, R. *et al.* (2004)
Joubert Syndrome, Type 2	TMEM216	c.137-1G>A c.218G>A c.218G>T	Ashkenazi Jewish	>99%	Edvardson, S. et al. (2010)
Junctional Epidermolysis Bullosa, Herlitz Type	LAMC2	c.283C>T	Italian	>30%	Posteraro, P. *et al.* (2004); Castori, M. *et al.* (2007)
		c.1223_1227delACACA c.944G>T c.877-2A>G c.866A>C c.788G>A c.424C>T c.425G>A c.401A>G c.1187G>T c.1298+2T>C c.1187G>A c.1175G>A c.1166G>A c.968G>A c.944G>A c.943C>T			

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Lamellar Ichthyosis, Type 1	TGM1	c.919C>G c.919C>T c.910A>T c.876+2T>C c.872G>A c.857G>A c.832G>A c.826T>A c.817G>A c.790C>T c.758-2A>G c.479C>G c.428G>A c.427C>G c.427C>T c.420A>G c.1055C>T c.2226-2A>G c.379C>T c.377G>A	Norwegian General population	>85% >65%	Pigg M. et al. (1998); Herman M. L. et al. (2009); Hackett B. C. et al. (2010); Bourrat E. et al. (2012); Sulák A. et al. (2017)
Leber Congenital Amaurosis (LCA5-related)	LCA5	c.1476dupA c.1151delC c.838C>T c.838C>T c.835C>T	General population Pakistani	unknown >80%	Weleber, R. G. et al. (2004); Mackay, D. S. et al. (2013); Corton, M. et al. (2014)
Leigh Syndrome, French-Canadian Type	LRPPRC	c.3830_3839delGTGGTGCAATinsAG c.1061C>T c.1091C>G	French Canadian - Saguenay Lac-St. Jean General Population	>95% >95%	Debray F.G. et al. (2011)
Leukoencephalopathy with Vanishing White Matter	EIF2B5	c.271A>G c.318A>T c.1157G>T c.338G>A c.584G>A c.925G>C c.1016G>A c.241G>A c.545C>T c.583C>T c.895C>T c.896G>A c.943C>T c.944G>A c.584G>A c.1010A>G c.1030C>T	General population	>72%	Fogli A. et al. (2004); van der Lei H. D. et al. (2010)
Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]	LHCGR	c.1874T>A c.1869T>G c.1847C>A c.1777G>C c.1733A>G c.1732G>T c.1732G>C c.1730C>T c.1715C>T c.1713G>A c.1660C>T c.1635C>A c.1627T>C c.1624A>C c.1505T>C c.1471T>C c.1118C>T c.1103T>C c.1060G>A c.1027T>A c.430G>T c.391T>C	Brazilian	>90%	Latronico, A. C. et al. (1996); Laue, L. et al. (1995)
Limb Girdle Muscular Dystrophy, Type 2E	SGCB	c.552T>G c.452C>G c.391C>T c.341C>T c.334C>T c.323T>G c.299T>A c.272G>C c.272G>T c.271C>T	US Amish Brazilian European population General population	>99% >55% >25% >13%	Duclos, F. et al. (1998); Semplicini, C. et al. (2015); Vainzof, M. et al. (1999); Bönnemann, C. G. et al. (1996)

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Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3]	DLD	c.199-1G>A c.214A>G c.685G>T c.1081A>G c.1123G>A c.1178T>C c.1236+1G>T c.1436A>T c.1444A>G c.1463C>T c.1483A>G	Ashkenazi Jewish	>85%	Scott, S. A. et al. (2010); Quinonez, S. C. & Thoene, J. G. (2014); Shaag, A. et al. (1999)
Lipoprotein Lipase Deficiency	LPL	c.548A>G c.590G>A c.607G>A c.644G>A c.662T>C c.665G>A c.693C>G c.701C>T c.755T>C	General population French-Canadian African	>23% >80% >70%	Monsalve, M. V et al. (1990); Gilbert, B. et al. (2001); Yuanhong, M., et al. (1991); Henderson, H.E., et al. (1992)
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	HADHA	c.1528G>C c.1195C>T c.1195C>T c.1132C>T	European population	>80%	IJlst, L. et al. (1996); Nedoszytko, B. et al. (2017); IJlst, L. et al. (1994)
Lysinuric Protein Intolerance	SLC7A7	c.1185_1188delTTCT c.1381_1384dupATCA c.1228C>T c.895-2A>T c.726G>A c.625+1G>C c.161G>T c.1417C>T c.1402C>T c.1371C>A c.1273T>C c.1158C>A c.1122C>A c.998+1G>T c.998G>T c.895-2A>G c.753G>T c.713C>T c.625+1G>A c.622C>T c.571A>G c.563C>T c.158C>T c.149T>A	Finnish Japanese Italian General population	>95% >80% >80% >70%	Sperandeo M. P. et al. (2008)
Maple Syrup Urine Disease, Type 1B	BCKDHB	c.479T>G c.487G>T c.488A>T c.503G>A c.508C>T c.508C>G c.508C>A c.547C>T c.548G>C c.548G>A c.554C>T c.560G>A c.564T>A c.616C>T c.633+1G>T c.633+1G>C c.633+1G>A c.748G>T c.752T>C c.799C>T c.832G>A c.840+1G>T c.840+1G>A c.840+2T>G c.841-1G>C c.853C>T c.952-2A>G c.952-1G>A c.964A>G	Ashkenazi Jewish	>95%	Baskovich, B. et al. (2016); Edelmann, L. et al. (2001); Scott, S. A. et al. (2010)

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.970C>T c.988G>A c.1016C>T c.1022T>A c.1090G>A c.1114G>T c.1149T>A			
Methylmalonic Acidemia (MMAA-related)	MMAA	c.1A>G c.64C>T c.72C>A c.124C>T c.161G>A c.202C>T c.266T>C c.283C>T c.358C>T c.387C>A c.397C>T c.433C>T c.439+1G>A c.503delC c.562G>C c.562+1G>T c.562+1G>A c.575G>A c.586C>T c.620A>G c.650T>A c.653G>A c.658G>A c.664A>G c.721A>T c.728C>A c.733+1G>A c.970-2A>T c.988C>T c.1025T>G c.1075C>T c.1076G>A c.1084C>T c.1104G>A	General population	>80%	Manoli, I., Sloan, J. L. & Venditti, C. P. (2005); Lerner-Ellis, J. P. et al. (2004)
		c.1630_1631delGGinsTA c.671_678dup c.1022dupA c.2150G>T c.2179C>T c.655A>T c.643G>A c.607G>A c.572C>A c.349G>T c.281G>T c.280G>A c.323G>A c.322C>T c.91C>T c.2080C>T c.1867G>A c.1481T>A c.1280G>A c.1207C>T c.1097A>G c.1105C>T c.1106G>A c.970G>A c.1025C>A c.2200C>T c.2131G>T c.2107G>C c.2099T>A c.2054T>G c.2026G>A c.2020C>G c.1975C>T c.1956+2T>C c.1924G>C c.1898T>G			

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Methylmalonic Aciduria, Type Mut(0)	MUT	c.1889G>A c.1885A>G c.1880A>G c.1874A>G c.1867G>C c.1853T>C c.1846C>T c.1663G>A c.1655C>T c.1599T>A c.1560+1G>T c.1553T>C c.1531C>T c.1489G>T c.1445-2A>G c.1287C>G c.1277G>A c.1271C>T c.1207C>G c.1164T>A c.1142G>T c.1130C>A c.1108A>C c.1084-1G>C c.1084-1G>A c.1084-2A>G c.1083+1G>A c.983T>C c.982C>T c.977G>A c.976A>G c.974G>A c.935G>T c.927G>A c.753+2T>A c.693C>G c.691T>A c.689C>G c.682C>T c.670G>T c.654A>C c.653A>G c.643G>T c.571G>A c.566A>T c.560C>G c.521T>C c.467A>T c.454C>T c.385+5G>A c.385+2T>C c.378C>A c.330T>G c.329A>G c.313T>C c.299A>G c.295A>G c.284C>G c.278G>A c.257C>T c.160A>T c.129G>A c.88C>T c.52C>T c.19C>T c.2T>C c.729_730insTT c.55dup c.360dup c.692dup	Asian General population	>75% >70%	Sakamoto O. et al. (2007); Han L. S. et al. (2015); Forny P. et al. (2016)
		c.658_660delAAG c.271dupA c.394C>T c.347T>C c.331C>T c.440G>C c.482G>A			

Condition	Gene	Mutations	Population	Detection Frequency	References
Methylmalonic Aciduria and Homocystinuria, Type cblC	MMACHC	c.608G>A c.217C>T c.276G>A c.292C>T c.315C>G c.391C>T c.420G>A c.427C>T c.440G>A c.457C>T c.464G>A c.471G>A c.481C>T c.565C>A c.615C>A c.615C>G c.666C>A c.688C>T c.609G>A	General population	>95%	Morel C. F. et al. (2006)
Methylmalonic Aciduria and Homocystinuria, Type cblD	MMADHC	c.696+3_696+6del c.57_64delCTCTTTAG c.455dupC c.419dupA c.307_324dup18 c.748C>T c.746A>G c.776T>C c.545C>A c.160C>T c.472C>T c.683C>G	General population	unknown	Coelho D. et al. (2008)
Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked	IDS	c.1402C>T c.1403G>A c.1403G>T c.1393C>T c.1327C>T c.1264T>G c.1122C>T c.514C>T c.1508T>A c.1505G>C c.1433A>G c.1425G>A c.1403G>C c.1393C>A c.1265G>T c.1265G>A c.1106C>A c.1099A>T c.1046G>A c.1033T>A c.1027G>A c.1025A>C c.1016T>C c.1006+1G>T c.1006G>T c.1003C>T c.998C>T c.613G>C c.592G>A c.587T>C c.508-1G>C c.508-1G>A c.1400C>T c.1400C>T	General population	unknown	Bunge S. et al. (1992); Rathmann M. et al. (1996); Froissart R. et al. (2007); Zhang H. et al. (2011); Zanetti A. et al. (2019)
		c.525dupT c.234+1G>A c.1270G>A c.1411G>A c.1516C>T c.1553C>T c.1622C>T c.372-2A>G c.410T>C c.493+1G>A c.848C>T c.852-1G>A			

Condition	Gene	Mutations	Population	Detection Frequency	References
Mucopolysaccharidosis, Type IIIC [Sanfilippo C]	HGSNAT	c.1030C>T c.1150C>T c.1250+1G>A c.398G>C c.518G>A c.836A>C c.851+1G>T c.851+1G>A c.852-2A>C c.887C>A c.947G>A c.962T>G c.1031G>A c.1102A>T c.1129-2A>T c.1445T>A c.1464+1G>A c.1542+1G>C c.1542+1G>A c.1542+2T>G c.1614-2A>T c.1634C>A c.1674C>G	Caucasians	>67%	Hrebícek M. et al. (2006); Coutinho M. F. et al. (2008); Ruijter G. J. et al. (2008); Fedele A. O. et al. (2007)
Multiple Sulfatase Deficiency	SUMF1	c.463T>C	Ashkenazi Jewish	>95%	Shi, L. *et al.* (2017)
Myotubular Myopathy, X-Linked	MTM1	c.141_144delAGAA c.139_141delAAA c.605delT c.593dupA c.969dupA c.49G>T c.1261-10A>G c.1262G>A c.1261C>T c.1456C>T c.1420C>T c.85C>T c.109C>T c.70C>T c.142G>T c.205C>T c.208C>T c.469G>A c.566A>G c.670C>T c.664C>T c.614C>T c.721C>T c.1190A>G c.2T>G c.3G>A c.63+1G>A c.63+3A>T c.64-1G>A c.137-11T>A c.137-3T>G c.145G>T c.145G>A c.205C>G c.226G>T c.231+1G>A c.231+2T>G c.514G>T c.528+1G>T c.529-2A>G c.535C>T c.549G>A c.550A>G c.557C>T c.611T>G c.629A>G c.637C>T c.676C>A c.678+1G>A c.679-1G>A c.679G>A c.683T>C c.743G>T	General population	>70%	de Gouyon B. M. et al. (1997); Laporte J. et al. (2000); Herman G. E. et al. (2002)

Condition	Gene	Mutations	Population	Detection Frequency	References
		c.757C>T c.779A>C c.791T>G c.958T>C c.1036T>C c.1040T>G c.1053+1G>C c.1120C>G c.1132G>A c.1136G>A c.1137G>A c.1139A>T c.1160C>A c.1180G>C c.1191T>G c.1204G>A c.1205G>C c.1210G>A c.1232G>A c.1233G>T c.1234A>G c.1244G>A c.1260+1G>A c.1261-1G>C c.1262G>T c.1307C>T c.1325T>A c.1328A>C c.1337G>A c.1353+1G>A c.1353+2T>C c.1354-1G>A c.1376A>T c.1378G>T c.1388T>G c.1388T>C c.1427G>T c.1433T>C c.1467+1G>T c.1467+1G>A c.1467+2T>A c.594C>A c.674T>C c.690G>T			
Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome]	MPV17	c.186+2T>C c.149G>A c.148C>T c.106C>T	Navajo population	>99%	El-Hattab, A. W., Scaglia, F., Craigen, W. J. & Wong, L.-J. C. (2012)
Neuronal Ceroid Lipofuscinosis (CLN8-related)	CLN8	c.88G>C c.70C>G c.610C>T c.1A>G c.2T>C c.92G>A c.544-2A>G c.581A>G c.709G>A c.763C>T c.766C>G c.637_639delTGG	Finnish General population	>95% >70%	Ranta S. et al. (1999); Sahin Y. et al. (2017)
Neuronal Ceroid Lipofuscinosis (MFSD8-related)	MFSD8	c.998+1G>A c.929G>A c.894T>G c.881C>A c.754+2T>A c.754+1G>T	General population Turkish	>55% >70%	Kousi, M., Lehesjoki, A.-E. & Mole, S. E. (2012)
		c.1678_1679delCT c.1611_1621del c.622C>T c.616C>T c.509-1G>A c.509-1G>C c.379C>T c.311T>A c.17+1G>C c.1525C>T c.1340G>A			

Condition	Gene	Mutations	Population	Detection Frequency	References
Neuronal Ceroid Lipofuscinosis (TPP1-related)	TPP1	c.1266G>C c.1094G>A c.1093T>C c.1552-1G>A c.1551+1G>T c.1551+1G>C c.1551+1G>A c.1259C>A c.1166G>A c.1145+1G>A c.1098G>A c.1076-1G>A c.1076-2A>G c.1076-2A>T c.617G>A c.605C>T c.509-1G>T c.380+5G>A c.380G>A c.237C>G c.17+1G>A c.1339C>T c.1361C>A	Newfoundland General population	>95% >85%	Moore S. J. et al. (2008); Sleat D. E. et al. (1999)
Nijmegen Breakage Syndrome	NBN	c.702+1G>C c.702+1G>T c.702+1G>A c.697A>T c.657_661delACAAA c.585-1G>A c.585-2A>G	Eastern European N. American	100% >70%	Varon, R., Demuth, I. & Chrzanowska, K. H. (1999)
Omenn Syndrome (RAG2-related)	RAG2	c.686G>A c.595G>T c.583T>G	General population	unknown	Schwarz K. et al. (1996); Corneo B. et al. (2001)
Ornithine Aminotransferase Deficiency	OAT	c.159delC c.1205T>C c.994G>A c.991C>T c.677C>T c.539G>C c.3G>A c.1307T>A c.1276C>T c.1250C>T c.1201G>T c.1192C>T c.1186C>T c.1181G>A c.1180T>C c.1172G>A c.955C>T c.952G>A c.901-2A>G c.749G>C c.748C>T c.734A>G c.722C>T c.710G>A c.627T>A c.596C>A c.542C>T c.533G>A c.163T>C c.162C>A	Finnish General population	>95% unknown	Mitchell G. A. et al. (1989); Doimo M. et al. (2013)
Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]	SLC25A15	c.91G>C c.95C>G c.535C>T c.538G>A c.562_564delTTC c.564C>G c.212T>A	French-Canadian Japanese	>95% >50%	Debray, F.-G. et al. (2008); Miyamoto, T. et al. (2001); Salvi, S. et al. (2001); Salvi, S. et al. (2001); Sokoro, A. A. H. et al. (2010); Camacho, J. & Rioseco-Camacho, N. (2012)
		c.279delT c.412G>T c.-103T>C c.1115C>T c.1151A>G c.1246A>C c.1334T>G			

Condition	Gene	Mutations	Population	Detection Frequency	References
Pendred Syndrome	SLC26A4	c.1540C>A c.2168A>G c.716T>A c.707T>C c.919-2A>G c.1001+1G>A c.165-2A>G c.170C>G c.227C>T c.249G>A c.259G>T c.269C>T c.281C>T c.296C>G c.299T>C c.304+2T>C c.349C>T c.397T>A c.412G>C c.412G>T c.601-1G>A c.619C>T c.626G>T c.706C>G c.765+2T>C c.918+1G>T c.918+2T>C c.946G>T c.1001G>T c.1001+1G>T c.1003T>C c.1079C>T c.1085C>A c.1105A>T c.1105A>G c.1149+3A>G c.1160C>T c.1173C>A c.1174A>T c.1225C>T c.1226G>C c.1226G>A c.1229C>T c.1231G>C c.1238A>G c.1262A>C c.1263+1G>T c.1263+1G>A c.1264-1G>C c.1336C>T c.1337A>G c.1341+1G>C c.1489G>A c.1541A>G c.1544+1G>A c.2090-1G>A c.2118C>A c.2153T>C c.2162C>T c.2171A>G c.2186T>C c.2188C>T c.2206C>T c.2215C>T c.2228T>A c.2235+2T>C c.1343C>T	Asian General Population	>75% >70%	Chen K. et al. (2014); Tang H. Y. et al. (2015)
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)	PEX1	c.2927-2A>G c.2926+2T>C c.2926+1G>A c.2916delA c.2894T>C c.2875C>T c.2528G>A c.2176C>T c.2137C>T c.2097dupT	General population	>70%	Walter, C. et al. (2001); Maxwell, M. A. et al. (1999); Walter, C. et al. (2001); Maxwell, M. A. et al. (2002); Weller, S., Gould, S. J. & Valle, D. P. (2003); Steinberg, S. et al. (2004)

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Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)	PEX2	c.373C>T c.355C>T c.304C>T	Ashkenazi Jewish	>95%	Shi, L. et al. (2017)
		c.1315+1G>A c.1301C>A c.1282C>T c.1256A>G c.1252A>C c.1249T>C c.1243G>A c.1241A>G c.1240T>C c.1238G>C c.1223G>A c.1222C>T c.1220C>T c.1219C>T c.1208C>T c.1200-1G>A c.1199+17G>A c.1199+2T>G c.1199+1G>C c.1199+1G>A c.1199G>C c.1197A>T c.1184C>G c.1180G>T c.1180G>C c.1172G>C c.1171A>G c.1169A>G c.1163T>C c.1162G>A c.1162G>C c.1157A>G c.1147C>T c.1139C>T c.1129delT c.1114A>T c.1097C>A c.1076C>G c.1068C>G c.1068C>A c.1066-2A>T c.1066-3C>T c.1066-11G>A c.1065+1G>A c.1055delG c.1049C>A c.1045T>C c.1042C>G c.1033G>T c.1033G>A c.1028A>G c.1025C>A c.1004A>C c.960G>C c.955G>T c.941C>A c.940C>A c.934G>T c.932T>C c.929C>T c.926C>T c.926C>A c.913-2A>C c.913-7A>G c.912+2T>C c.912+1G>A c.912G>A c.910C>T c.901C>A c.898G>T c.896T>G c.895T>C c.890G>A c.869A>T			

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Phenylalanine Hydroxylase Deficiency (Phenylketonurea)	PAH	c.865G>A c.856G>A c.847A>T c.844G>A c.842+5G>A c.842+4A>G c.842+3G>C c.842+2T>A c.842+1G>A c.842C>T c.841C>T c.841C>G c.839A>G c.838G>A c.836C>T c.835C>G c.833C>T c.833C>A c.830A>G c.829T>G c.826A>G c.824C>T c.824C>G c.823C>T c.818C>T c.814G>T c.809G>A c.806T>A c.796A>C c.785T>G c.782G>A c.782G>T c.782G>C c.781C>T c.781C>G c.776C>T c.775G>A c.770G>T c.764T>C c.757G>A c.755G>A c.754C>T c.754C>G c.745C>T c.740G>T c.739G>C c.737C>A c.734T>C c.734T>A c.733G>C c.731C>T c.728G>A c.727C>T c.724C>T c.722G>A c.722G>T c.721C>T c.694C>T c.691T>C c.689T>C c.688G>A c.682G>A c.673C>A c.662A>G c.653G>T c.648C>G c.638T>C c.635T>C c.632C>T c.631C>A c.618C>A c.618C>G c.612T>G c.611A>G c.591G>C c.581T>C c.569T>C	General population European population Ashkenazi Jewish Ireland	>75% >70% >53% >40%	van Wegberg, A. M. J. et al. (2017); Braun-Falco, M. et al. (Springer Berlin Heidelberg, 2009); O'Neill., C.A., et al. (1994); Bercovich, D., et al. (2008)

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		c.568G>A c.563G>A c.561G>A c.535T>A c.533A>G c.529G>C c.529G>A c.527G>T c.526C>T c.520A>G c.511G>A c.510-2A>G c.508C>G c.506G>C c.506G>A c.505C>T c.504C>A c.500A>T c.498C>G c.490A>G c.482T>C c.473G>A c.472C>T c.464G>C c.464G>A c.442-1G>A c.442-2A>C c.400C>T c.386A>G c.385G>T c.355C>T c.331C>T c.320A>G c.311C>A c.284_286delTCA c.283A>T c.261C>A c.250G>T c.242C>A c.227A>G c.226G>T c.204A>T c.196G>T c.194T>C c.183C>A c.169G>A c.169G>T c.169-2A>G c.143T>C c.140C>T c.136G>A c.127G>T c.121C>T c.116_118delTCT c.117C>G c.110T>C			
Pontocerebellar Hypoplasia, Type 1A	VRK1	c.266G>A c.397C>T c.706G>A c.1072C>T	Ashkenazi Jewish General population	>95% >95%	Renbaum P. et al. (2009); Gonzaga-Jauregui C. et al. (2013); Najmabadi H. et al. (2011)
Pontocerebellar Hypoplasia, Type 2D	SEPSECS	c.1001A>G c.715G>A	Sephardic Jewish - Moroccan, Iraqi General Population	>95% >95%	Agamy O. et al. (2010)
Pontocerebellar Hypoplasia, Type 2E	VPS53	c.2084A>G c.1556+5G>A c.1516C>T	Moroccan Jewish	>99%	Ben-Zeev, B. et al. (2003); Feinstein, M. et al. (2014)
Primary Ciliary Dyskinesia (DNAH5-related)	DNAH5	c.10815delT c.13486C>T c.13338+5G>A c.10384C>T c.4348C>T c.1730G>C c.13426C>T c.13331G>A c.10858C>T	General population Amish or Mennonite	>23% >95%	Hornef N. et al. (2006); Ferkol T.W. et al. (2013)

Condition	Gene	Mutations	Population		References
Primary Ciliary Dyskinesia (DNAI1-related)	DNAI1	c.48+2dupT c.1612G>A c.2001+1G>A c.1490G>A c.1569+1G>T c.1644G>A	General population Ashkenazi Jewish	>67% >95%	Zariwala M.A. et al. (2006); Fedick A.M. et al. (2015)
Primary Hyperoxaluria, Type 3	HOGA1	c.700+5G>T c.860G>T c.700+2T>G	General population	>77%	Williams E. L. et al. (2012); Hopp K. et al. (2015)
Pycnodysostosis	CTSK	c.990A>G c.934C>G c.934C>T c.926T>C	Danish	>88%	Haagerup, A. et al. (2000)
Pyruvate Dehydrogenase Deficiency (PDHB-Related)	PDHB	c.1030C>T c.419T>A c.395A>G	General population	>20%	Brown, R. et al. (2004); Okajima, K. et al. (2008)
Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy]	RLBP1	c.773T>G c.700C>T	Swedish	>99%	Burstedt, M. et al. (2013); Burstedt, M. S. et al. (2001); Burstedt, M. S. et al. (1999)
Retinitis Pigmentosa 25 (EYS-related)	EYS	c.6714delT c.4350_4356delTATAGCT c.4957dupA c.8805C>A c.7919G>A c.5014C>T	Japanese Danish	>57% >34%	Hosono K. et al. (2012); Jespersgaard C. et al. (2019)
Retinitis Pigmentosa 59 (DHDDS-related)	DHDDS	c.124A>G	Ashkenazi Jewish	>95%	Shi, L. *et al.* (2017); Zelinger, L. *et al.* (2011)
Sanfilippo Syndrome, Type D [Mucopolysaccharidosis IIID]	GNS	c.1226dupG c.1169delA c.1168C>T c.1138_1139insGTCCT c.1063C>T	General population	>80%	Jansen, A. C. M. et al. (2007); Mok, A., Cao, H. & Hegele, R. A. (2003)
Severe Combined Immunodeficiency, Type Athabaskan	DCLRE1C	c.597C>A	Navajo and Apache Native Americans	>95%	Li L. et al. (2002)

Condition	Gene	Mutations	Population	Detection Frequency	References
Severe Combined Immunodeficiency, X-Linked	IL2RG	c.359dupA c.865C>T c.854G>A c.722G>T c.703C>T c.676C>T c.677G>A c.670C>T c.458T>A c.579G>A c.515T>A c.341G>A c.421C>T c.266A>G c.202G>A c.186T>A c.924+1G>A c.923C>A c.878T>A c.855-1G>A c.846G>A c.758-1G>A c.720G>A c.718T>C c.710G>A c.664C>T c.662T>C c.602C>G c.562C>T c.522G>A c.467C>T c.460C>T c.455T>C c.454+1G>A c.452T>C c.391C>T c.355A>T c.344G>A c.343T>C c.314A>G c.281C>A c.270G>A c.270-1G>T c.548delT	General population	>70%	Clark P. A. et al. (1995); Puck J. M. et al. (1997)
Sickle-Cell Disease	HBB	c.364G>C c.364G>A c.79G>A c.20A>T c.19G>A	Severe pathogenic alleles	100%	Rees, D. C., Williams, T. N. & Gladwin, M. T. (2010); Stuart, M. J. & Nagel, R. L. (2004); Leung, A. K. C. et al. (Springer Berlin Heidelberg, 2009)
Sjögren-Larsson Syndrome	ALDH3A2	c.943C>T c.1277T>G c.1297_1298delGA c.1367T>A	Swedish Dutch	>95% >69%	Jagell, S., Henrik, K. & Holmgren, G. (2008); Willemsen, M. A. A. P. et al. (1999)
Steroid-Resistant Nephrotic Syndrome	NPHS2	c.948delT c.419delG c.503G>A c.412C>T c.413G>A c.353C>T c.964C>T c.874-1G>A c.874-2A>C c.873+2T>A c.873+1G>A c.871C>T c.868G>A c.859C>T c.851C>T c.795-1G>A c.502C>T c.479A>G c.452-1G>A c.451+2T>A c.385C>T c.378+1G>A c.275-2A>G	General population	>61%	Ruf R. G. et al. (2004); Hinkes B. G. et al. (2007)

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Stuve-Wiedemann Syndrome	LIFR	c.2472_2476delTATGT c.170delC c.2275dupT c.1621dupA c.756dupT c.653dup c.2434C>T c.1789C>T c.1601-1G>A c.2170C>G	General population	>59%	Romeo Bertola D. et al. (2016)
Tay-Sachs Disease	HEXA	c.1330+1G>A c.1307_1308delTA c.1305C>T c.1302C>G c.1274_1277dupTATC c.1260G>C c.1259G>A c.1178G>C c.1177C>T c.1176G>A c.1168C>T c.1150C>T c.1073+1G>A c.1073+1G>T c.987G>A c.805+1G>C c.805+1G>A c.805G>A c.805G>C c.788C>T c.772G>C c.749G>A c.709C>T	Ashkenazi Jewish General population	>81% >32%	Scott, S. A. et al. (2010); Kaback, M. et al. (1993)
Usher Syndrome, Type 1F	PCDH15	c.785G>A c.733C>T c.84T>A c.7C>T c.1A>G	Ashkenazi Jewish	>64%	Ben-Yosef, T. et al. (2003); Brownstein, Z. et al. (2004)
Usher Syndrome, Type 3	CLRN1	c.449T>C c.528T>G c.359T>A c.144T>G c.619C>T c.541C>T c.461T>G c.368C>A c.189C>A c.184C>T c.98G>A	Finnish Ashkenazi Jewish	>95% >95%	Joensuu T. et al. (2001); Fields R.R. et al. (2002)
Wolman Disease	LIPA	c.1024G>A c.883C>T c.894G>AB c.652C>T c.599T>C c.260G>T c.894+2T>C c.894+1G>A c.892C>T c.656T>G c.253C>T	General Population Sephardic Jewish - Iranian	>69% >95%	Scott S.A. et al. (2013); Valles-Ayoub Y. et al. (2011)

*The VERAgene 100 panel tests for mutations that cause the classic Cystic Fibrosis phenotype.