

Condition	Gene	Mutations	Population	Detection Frequency	References
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	HMGCL	c.914_915delTT c.206_207delCT c.505_506delTTC 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 c.1330C>T c.1310T>C c.1277T>C c.1225C>T c.1155A>C c.1114C>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.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	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.191delIT c.1276_1277delGT c.372_389delCATGCCCGCCTGGAACCT 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 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	General population	>73%	Ferdinandusse S. et al. (2007)
Aicardi-Goutières Syndrome	SAMHD1	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 c.4709G>C c.4964T>G c.5048G>A c.4707-1G>C c.4708T>C c.4717T>C	General population	>75%	Rice G.I. et al. (2009)

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Alport Syndrome, X-Linked	COL4A5	c.4718G>C	General population	>79%	Pont-Kingdon G. et al. (2009)
		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			
		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					
Alstrom Syndrome	ALMS1		General population	>46%	Marshall J. D. et al. (2015)

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		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 c.2436+1delG c.1478_1485delTTCCCTCT c.901delA c.1584_1585delCTinsG c.2028_2029insT			
Andermann Syndrome	SLC12A6	c.3031C>T c.2023C>T c.619C>T c.2416G>T c.963C>A c.655C>T c.630G>A 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 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 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 c.10709C>G c.10664T>A c.10658T>C	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 c.10709C>G c.10664T>A c.10658T>C	General population Finnish	>70% >95%	Ikonen E. et al. (1991); Saarela J. et al. (2001)

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Autosomal Recessive Polycystic Kidney Disease	PKHD1	c.10444C>T	Ashkenazi Jewish General population Caucasian	>90% >69% >70%	Sweeney, W. E. & Avner, E. D. (1993); Shi, L. et al.(2017)
		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					
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					
c.323C>G					
Bardet-Biedl Syndrome (BBS1-related)	BBS1	c.851delA	General population	>70%	Mykytyn K. et al. (2003); Janssen S. et al. (2011)
		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					
c.323C>G					

Condition	Gene	Mutations	Population	Detection Frequency	References
Bardet Biedl Syndrome (BBS12-related)	BBS12	c.335_337delTAG	European population	>50%	Stoetzel, C. et al. (2007); Burststedt, M. et al. (2013); Burststedt, M. S. et al. (2001); Muller, J. et al. (2010)
		c.760G>T			
		c.865G>C			
		c.1063C>T			
		c.1375C>T			
		c.1483_1484delGA			
		c.1502C>T			
		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	Mediterranean Middle-Eastern Thai Chinese African-American	>95% >95% >95% >95% >80%	Origa, R. (2000)		
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					

Condition	Gene	Mutations	Population	Detection Frequency	References
Biotinidase Deficiency	BTD	c.45dupG	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)
		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			
		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					

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)
		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 c.1095delT			
Choroideremia, X-Linked	CHM	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 c.1095delT	General population	>70%	Simunovic M. P. et al. (2016)
		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			
Citrin Deficiency	SLC25A13	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)

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Combined Oxidative Phosphorylation Deficiency 3	TSFM	c.493C>T	Finnish	>75%	Ahola S. et al. (2014); Shamseldin H. E. et al. (2012)
		c.70-1G>A			
		c.919C>T			
		c.997C>T			
		c.1007G>A			
		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					
c.91delG					
c.125dupG					
c.430dup					
Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	PMM2	c.130_131insA	Caucasian Ashkenazi Jewish General population	>80% >90% >80%	Matthijs G. et al. (2000); Le Bizec C. et al. (2005); Shi L. et al. (2017)
		c.256C>T			
		c.568C>T			
		c.383C>G			
		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					
c.1466C>A					
c.1477_1478delCA					
c.1477C>T					
c.1519_1521delATC					
c.1521_1523delCTT					
c.1520_1522delTCT					
c.1545_1546delTA					
Congenital Neutropenia (HAX1-related)	HAX1	c.130_131insA	General population	>95%	Klein C. et al. (2007); Smith B. N. et al. (2009)
		c.256C>T			
Crigler Najjar Syndrome, Type I	UGT1A1	c.524T>A	Tunisian Sardinian	>99% >70%	Francoual, J. et al. (2002)
		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			
		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	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
		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					

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_3886delATT			
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 c.2741C>T c.2204+6T>C c.2087G>C	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	IKBKAP	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)
		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			
Fanconi Anemia, Type C	FANCC	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	French-Canadian Korean/Japanese Portuguese-Brazilian	>80%	Auerbach, A. D. et al. (2005); Mehta, P. A. & Tolar, J. (2002); Fanconi Anemia: Guidelines for Diagnosis and Management, Fourth Edition (2014)
		c.925-2A>G		>65%	
		c.307+1G>C		>80%	
		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					
Gaucher Disease	GBA	c.1448T>G	Ashkenazi Jewish General population	>97%	Beutler E. et al. (1992); Beutler E. et al. (1993); Shamseddine A. et al. (2004)
		c.1397T>G		>75%	
		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			
Glutaric Acidemia, Type 2A	ETFPA	c.509G>T	General population	>50%	Freneaux E. et al. (1992)
		c.481C>T			
Glycine Encephalopathy (GLDC-related)	GLDC	c.476G>A	Finnish General population	>78% >10%	Van Hove, J., Coughlin, C. & Scharer, G. (2002)
		c.797C>T			
		c.470T>G			
		c.346G>A			
		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					
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	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)
		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					
Glycogen Storage Disease, Type 1B	SLC37A4	c.1042_1043delCT	General population	>50%	Galli L. et al. (1999)
		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					
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	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)
		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					
Glycogen Storage Disease, Type 7	PFKM	c.115C>T	Ashkenazi Jewish	>95%	Sherman, J. B. et al. (1994)
		c.329G>T			
		c.116G>C			
		c.450+1G>A			
		c.496C>T			
GRACILE Syndrome	BCSL1	c.133C>T	Finnish	>95%	de Lonlay, P. et al. (2001); Visapää, I. et al. (2002)
		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					
Hereditary Fructose Intolerance	ALDOB	c.865delC	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)
		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					
c.903+469T>C					
Homocystinuria, Type cbIE	MTRR	c.1361C>T	Caucasian	>72%	Zavadáková P. et al. (2005)
		c.1459G>A			
		c.1573C>T			
c.1379T>G					
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, Hertz Type	LAMC2	c.283C>T 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	Italian	>30%	Posteraro, P. et al. (2004); Castori, M. et al. (2007)

Condition	Gene	Mutations	Population	Detection Frequency	References
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 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	General population	>72%	Fogli A. et al. (2004); van der Lei H. D. et al. (2010)
Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]	LHCGR	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	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)

Condition	Gene	Mutations	Population	Detection Frequency	References
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 c.1528G>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.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 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	Finnish Japanese Italian General population	>95% >80% >80% >70%	Sperandeo M. P. et al. (2008)
Maple Syrup Urine Disease, Type 1B	BCKDHB	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
Methylmalonic Acidemia (MMAA-related)	MMAA	c.970C>T	General population	>80%	Manoli, I., Sloan, J. L. & Venditti, C. P. (2005); Lerner-Ellis, J. P. et al. (2004)
		c.988G>A			
		c.1016C>T			
		c.1022T>A			
		c.1090G>A			
		c.1114G>T			
		c.1149T>A			
		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			
		c.1630_1631delG>insTA			
		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					

Condition	Gene	Mutations	Population	Detection Frequency	References
Methylmalonic Aciduria, Type Mut(O)	MUT	c.1889G>A	Asian General population	>75% >70%	Sakamoto O. et al. (2007); Han L. S. et al. (2015); Forny P. et al. (2016)
		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					
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 cb1C	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 cb1D	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 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 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	General population	unknown	Coelho D. et al. (2008)
Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked	IDS		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)

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%	Hrebíček 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 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	Ashkenazi Jewish	>95%	Shi, L. et al. (2017)
Myotubular Myopathy, X-Linked	MTM1	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 c.88G>C c.70C>G c.610C>T c.1A>G c.2T>C	Navajo population	>99%	El-Hattab, A. W., Scaglia, F., Craigen, W. J. & Wong, L.-J. C. (2012)
Neuronal Ceroid Lipofuscinosis (CLN8-related)	CLN8	c.92G>A c.544-2A>G c.581A>G c.709G>A c.763C>T c.766C>G	Finnish General population	>95% >70%	Ranta S. et al. (1999); Sahin Y. et al. (2017)
Neuronal Ceroid Lipofuscinosis (MFSD8-related)	MFSD8	c.637_639delTGG 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	Newfoundland General population	>95% >85%	Moore S. J. et al. (2008); Sleat D. E. et al. (1999)
		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					
Nijmegen Breakage Syndrome	NBN	c.702+1G>C	Eastern European N. American	100% >70%	Varon, R., Demuth, I. & Chrzanoska, K. H. (1999)
		c.702+1G>T			
		c.702+1G>A			
		c.697A>T			
		c.657_661delACAAA			
c.585-1G>A					
c.585-2A>G					
Omenn Syndrome (RAG2-related)	RAG2	c.686G>A	General population	unknown	Schwarz K. et al. (1996); Corneo B. et al. (2001)
		c.595G>T			
		c.583T>G			
Ornithine Aminotransferase Deficiency	OAT	c.159delC	Finnish General population	>95% unknown	Mitchell G. A. et al. (1989); Doimo M. et al. (2013)
		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					
Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]	SLC25A15	c.91G>C	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.95C>G			
		c.535C>T			
		c.538G>A			
		c.562_564delITTC			
c.564C>G					
c.212T>A					
c.279delIT					
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	Asian General Population	>75% >70%	Chen K. et al. (2014); Tang H. Y. et al. (2015)
		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					
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					
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)	PEX1		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)

Condition	Gene	Mutations	Population	Detection Frequency	References
Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)	PEX2	c.373C>T	Ashkenazi Jewish	>95%	Shi, L. et al. (2017)
		c.355C>T			
		c.304C>T			
		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					

Condition	Gene	Mutations	Population	Detection Frequency	References
Phenylketonurea	PAH	c.865G>A	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)
		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					

Condition	Gene	Mutations	Population	Detection Frequency	References	
		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 c.266G>A c.397C>T c.706G>A c.1072C>T				
Pontocerebellar Hypoplasia, Type 1A	VRK1		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 c.10815delT c.13486C>T c.13338+5G>A	Moroccan Jewish	>99%	Ben-Zeev, B. et al. (2003); Feinstein, M. et al. (2014)	
Primary Ciliary Dyskinesia (DNAH5-related)	DNAH5	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)	
Primary Ciliary Dyskinesia (DNA11-	DNA11	c.48+2dupT c.1612G>A c.2001+1G>A	General population	>67%	Zariwala M.A. et al. (2006); Fedick	

Condition	Gene	Mutations	Population	Detection Frequency	References
related)	DNAAF1	c.1490G>A c.1569+1G>T c.1644G>A	Ashkenazi Jewish	>95%	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 c.6714delT	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.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	General population	>70%	Clark P. A. et al. (1995); Puck J. M. et al. (1997)
		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					
Sickle-Cell Disease	HBB	c.364G>C	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)
		c.364G>A			
		c.79G>A			
		c.20A>T			
		c.19G>A			
Sjögren-Larsson Syndrome	ALDH3A2	c.943C>T	Swedish Dutch	>95% >69%	Jagell, S., Henrik, K. & Holmgren, G. (2008); Willemssen, M. A. A. P. et al. (1999)
		c.1277T>G			
Steroid-Resistant Nephrotic Syndrome	NPHS2	c.1297_1298delGA	General population	>61%	Ruf R. G. et al. (2004); Hinkes B. G. et al. (2007)
		c.1367T>A			
		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					

Condition	Gene	Mutations	Population	Detection Frequency	References
Stuve-Wiedemann Syndrome	LIFR	c.2472_2476delTATGT	General population	>59%	Romeo Bertola D. et al. (2016)
		c.170delC			
		c.2275dupT			
		c.1621dupA			
		c.756dupT			
		c.653dup			
		c.2434C>T			
		c.1789C>T			
		c.1601-1G>A			
		c.2170C>G			
Tay-Sachs Disease	HEXA	c.1330+1G>A	Ashkenazi Jewish General population	>81% >32%	Scott, S. A. et al. (2010); Kaback, M. et al. (1993)
		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					
c.785G>A					
c.733C>T					
Usher Syndrome, Type 1F	PCDH15	c.84T>A	Ashkenazi Jewish	>64%	Ben-Yosef, T. et al. (2003); Brownstein, Z. et al. (2004)
		c.7C>T			
		c.1A>G			
		c.449T>C			
Usher Syndrome, Type 3	CLRN1	c.528T>G	Finnish Ashkenazi Jewish	>95% >95%	Joensuu T. et al. (2001); Fields R.R. et al. (2002)
		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			
		c.1024G>A			
		c.883C>T			
		c.894G>AB			
c.652C>T					
Wolman Disease	LIPA	c.599T>C	General Population Sephardic Jewish - Iranian	>69% >95%	Scott S.A. et al. (2013); Valles-Ayoub Y. et al. (2011)
		c.260G>T			
		c.894+2T>C			
		c.894+1G>A			
		c.892C>T			
		c.656T>G			
		c.253C>T			

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