
by ThermoFisher
CarrierScan
Expanded Perconception Screening
AMGLAB

CarrierScan™ (by Thermo Fisher Scientific) Carrier Screening Israel

Various genetic variations causing genetic diseases occur with increased frequencies in the various Jewish and Non-Jewish populations in Israel. For these genetic variations, carrier screening campaigns are the essence of preventive healthcare in the field of medical genetics. Thus it is highly recommended by the Israeli Ministry of Health (IMOH), the Israeli Society of Medical Genetics (ISMG), and other professional organizations to test at the preconception (i.e. before pregnancy) or prenatal (i.e. during early stages of pregnancy) stage for a well-defined list of genetic diseases that would put children at risk for being born with a genetic disease. The Applied Biosystems CarrierScan™ assay (by Thermo Fisher Scientific) is currently testing for these diseases and much more.

Traditionally limited to a relatively small number of founder genetic variations in specific populations at risk, the emerging field of genomic medicine is revolutionizing common preconception carrier screening practices. The CarrierScan™ screening test is the most comprehensive test designed for the Israeli market covering 1696 genetic variations in 363 genes representing a total of 359 inherited diseases (OMIM phenotypes). By genotyping thousands of genetic variations across the genome, missense, nonsense, splice site and small intragenic deletion/insertion genetic variations are simultaneously assessed alongside multiple intragenic deletion and duplication genetic variations. It is expected that individuals screened by the test will have a 1 in 2 to 1 in 4 chance of being a carrier for at least one of the included diseases. The test is made available to the general public only through an approved health care provider.

The Panel

The mutation list comprising the CarrierScan™ test adheres to recommended regulatory agency guidelines, curated databases and peer-reviewed manuscripts. It is relevant pan-ethnically with an enrichment on genetic variations known to affect the various Israeli populations. As such, CarrierScan™ strives to include all recommended genetic variations by the IMOH or the ISMG in the first and second categories. All founder genetic variations currently recommended for the various Jewish populations and over 95% of the genetic variations recommended for non-Jewish populations are screened within CarrierScan™. In addition, an expanded panel of *CFTR* genetic variations that cause cystic fibrosis, includes all mutations recommended for screening in the various Israeli populations and is augmented by the *CFTR2* curated database content. Moreover, our innovative design making use of Thermo Fisher bead array genotyping platform allows for the detection of *DMD* gene deletions and duplications, known to cause Duchenne muscular dystrophy, and variations in the SMN1 gene causing Spinal Muscular Athrophy (SMA) Finally, additional genetic variations reported to cause a broad range of disease phenotypes is included.

Validation Methodology

CarrierScan™ assay is an innovative, comprehensive and high- throughput for the reliable and robust detection of sequence and structural variation for preconception expanded carrier screening developed in collaboration with experts across the field of carrier screening research. It is designed to provide detection of both sequence and structural variants simultaneously, including biallelic and multiallelic mutations such as single nucleotide

variants (SNV's), insertion-deletion variants (in-dels) and structural genomic variants such as microdeletions or microduplications- copy number variants (CNV's). For the purpose of validation, thousands of samples and positive controls were included in the validation process to verify the accuracy and precision of the included assays. Thermo Fisher is constantly striving to enlarge its repertoire of positive controls from genomic DNA. Of the 1696 genetic variations screened by the CarrierScan™ assay a total of 1047 genetic variations were confirmed by positive controls from cases representing either heterozygote or homozygote controls. Genetic variations lacking a positive control appear in brackets. The number of positive controls used for validation is continuously increasing and routinely updated.

Testing Methodology

DNA was prepared according to CarrierScan™ Assay 96-Array Format manual workflow, for target preparation that includes DNA amplification, fragmentation, purification and resuspension of target in hybridization cocktail. The hyb-ready targets are then transferred to the Applied Biosytems Gene Titan™ Multi- Chanel (MC) instrument for automated hands-free processing including hybridization, staining, washing and imaging. The resulting CEL files are analyzed by Axiom™ analysis suite 3.1 or higher or assessed for carrier status using Applied Biosytems CarrierScan Reporter software. Data is then uploaded into the analysis and lab report software by Igentify which creates single or couple reports with calculated risks based on the test results, and the information provided about ethnicity, disease recurrence, and the genetic changes as described in the medical literature.

Testing Limitations

AMG is a certified clinical laboratory holding a license from the Israeli Ministry of health and an ISO 9001:2015 Certificate (IQC). This test was validated with success by AMG Lab using positive controls in a double-blind test. In cases of detection of a mutation that is lacking a clinical positive control, verification will be made by Sanger sequencing in the Gene by Gene Lab in Houston Texas. Gene by Gene is a College of American Pathologists (CAP: 7212851) accredited and Clinical Laboratory Improvement Amendments (CLIA: 45D1102202) certified clinical laboratory qualified to perform high-complexity testing and approved by the Israeli Ministry of Health. Only the genetic variations listed in the appended table are tested; there is a possibility that the tested individual is a carrier for additional genetic variations not screened in this test. Although molecular tests are highly accurate, it is a screening test only and not diagnostic. Rare analytic errors may occur that interfere with reporting. Sources of these errors include sample mix-up, trace contamination, and other technical errors. The presence of additional variants nearby may interfere with mutation detection. Individuals with certain histories or ethnicities may experience better carrier detection with other testing methods, and it is recommended that these options be reviewed with patients by their healthcare provider. For example, in the case of an individual who has a partner that identifies as a carrier of Tay-Sachs, sequencing the HEXA gene or performing hexosaminidase A enzyme analysis may be warranted. CarrierScan™ results must always be interpreted by a medical geneticist or genetic counselor in the context of clinical, familial and ancestral data. Genetic counseling is recommended to properly review and explain these results to the tested individual.

CarrierScan™ R3.0.1 Assay Panel Contains 359 OMIM Phenotypes

- Screens for all founder genetic variations currently recommended for the various Jewish populations.
- Screens for over 95% of genetic variations recommended in Israel for non-Jewish populations.
- Screens for additional genetic variations prevalent in the general world population.
- Screens for an expanded list of genetic variations found within the *CFTR* gene that causes cystic fibrosis.
- Screens for exonic deletions and duplications found in the *DMD* gene causing Duchenne muscular dystrophy and related disorders.
- Screens for mutations and copy number variations of exons 7 and 8 in the SMN1 gene.
- All conditions tested follow an autosomal recessive pattern of inheritance except for a minority of diseases such as Duchenne muscular dystrophy which follow an X-linked recessive pattern of inheritance.
- Genetic variations lacking a positive control appear in brackets (649).
- Diseases recommended by the Israeli Ministry of Health and the Israeli Society of Medical Geneticists.

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Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Alkaptonuria	HGD	No	1	c.1102A>G	General Population	1 in 500	10%	1 in 555
Carnitine deficiency, systemic primary	SLC22A5	No	6	c.136C>T; c.1400C>G; c.1463G>A; c.497+1G>T; c.760C>T; c.844C>T	General Population	1 in 500	10%	1 in 555
Gitelman syndrome	SLC12A3	No	2	c.179C>T; c.2883+1G>T	General Population	1 in 500	10%	1 in 555
Hemophilia A	F8	No	3	c.1172G>A; c.5399G>A; c.6563G>A	General Population	1 in 500	10%	1 in 555
Hemophilia B/Thrombophilia, X-linked, due to factor IX defect	F9	No	1	c.1135C>T	General Population	1 in 500	10%	1 in 555
Methylmalonic aciduria and homocystinuria, cblC type	MMACHC	No	4	c.271dupA; c.347T>C; c.394C>T; c.609G>A	General Population	1 in 500	10%	1 in 555
Methylmalonic aciduria, mut(0) type	MUT	No	5	c.1106G>A; c.1280G>A; c.1630_1631delGGinsTA; c.607G>A; c.655A>T	General Population	1 in 500	10%	1 in 555
Pseudohermaphroditism, male, with gynecomastia	HSD17B3	No	2	c.608C>T; c.803G>A	General Population	1 in 500	10%	1 in 555
Segawa syndrome, recessive	TH	No	1	c.698G>A	General Population	1 in 500	10%	1 in 555
Bare lymphocyte syndrome, type II, complementation group A	CIITA	No	1	c.2885T>C	General Population	1 in 500	10%	1 in 555
17,20-lyase deficiency, isolated/17-alpha-hydroxylase/17,20-lyase deficiency	CYP17A1	No	1	c.1216T>C	General Population	1 in 500	10%	1 in 555
2-methylbutyrylglycinuria	ACADSB	No	3	c.1165A>G; c.303+3A>G; c.443C>T	General Population	1 in 500	10%	1 in 555
3-Methylcrotonyl-CoA carboxylase 1 deficiency	MCCC1	No	1	c.1526delG	General Population	1 in 500	10%	1 in 555
3-Methylcrotonyl-CoA carboxylase 2 deficiency	MCCC2	No	2	c.1015G>A; c.538C>T	General Population	1 in 500	10%	1 in 555
3-methylglutaconic aciduria, type III	OPA3	X	2	c.143-1G>C; (c.322_339delCAGCGCCACAAGGAGGAG)	Ashkenazi Jewish	1 in 500	10%	1 in 555
					General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	1 in 10	91%	1 in 108
3-phosphoglycerate dehydrogenase deficiency	PHGDH	No	1	c.1468G>A	Ashkenazi Jewish	1 in 400	99%	1 in 39901
					General Population	1 in 500	10%	1 in 555
4 optional diseases	CEP290	No	1	c.384_387delTAGA	General Population	1 in 500	10%	1 in 555
5-fluorouracil toxicity/Dihydropyrimidine dehydrogenase deficiency	DPYD	No	3	c.1905+1G>A; c.299_302delTCAT; c.557A>G	General Population	1 in 500	10%	1 in 555
Abetalipoproteinemia	MTTP	No	4	(c.2212delT); c.2593G>T; c.307A>T; (c.62-2A>G)	Ashkenazi Jewish	1 in 131	99%	1 in 13101
					General Population	1 in 447	10%	1 in 500
Achalasia-addisonianism-alacrimia syndrome	AAAS	No	2	c.1331+1G>A; c.787T>C	General Population	1 in 500	10%	1 in 555
Achondrogenesis Ib/Atelosteogenesis, type II/De la Chapelle dysplasia/Diastrophic dysplasia/Diastrophic dysplasia, broad bone-platyspondylic variant/Epiphyseal dysplasia, multiple, 4	SLC26A2	No	3	c.1957T>A; c.532C>T; c.835C>T	General Population	1 in 500	10%	1 in 555
Achromatopsia 3/Macular degeneration, juvenile	CNGB3	No	4	c.1006G>T; c.1148delC; c.1208G>A; c.819_826delCAGACTCC	General Population	1 in 500	10%	1 in 555
Achromatopsia, type 2	CNGA3	No	1	(c.1585G>A)	Bukharian Jewish	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
					Iranian Jewish	N/A	99%	1 in 555
					Iraqi Jewish	N/A	99%	1 in 555
Acromesomelic dysplasia, Demirhan type	BMPR1B	No	1	c.101G>A	General Population	1 in 500	10%	1 in 555
Acyl-CoA dehydrogenase, short-chain, deficiency of	ACADS	No	1	c.319C>T	Ashkenazi Jewish	1 in 63	90%	1 in 621
					General Population	1 in 94	10%	1 in 104
Adams–Oliver syndrome	EOGT	No	1	(c.1074delA)	General Population	1 in 500	10%	1 in 555
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	CYP11B1	No	2	(c.1342C>T); c.1343G>A	General Population	1 in 158	10%	1 in 176
					Moroccan Jewish	1 in 60	90%	1 in 586
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency/Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	CYP21A2	No	3	c.1174G>A; c.1360C>T; c.719T>A	General Population	1 in 500	10%	1 in 555
Adrenocorticotropic hormone deficiency	TBX19	X	1	(c.573_576delCATA)	General Population	1 in 500	10%	1 in 555
Adrenoleukodystrophy	ABCD1	No	50	(c.1028G>T); (c.1165C>G); c.1202G>A; (c.1213T>C); (c.1252C>T); (c.1390C>T); (c.139C>T); c.1415_1416delAG; (c.1429G>T); (c.1451C>G); (c.1520G>A); (c.1523C>T); (c.1526A>T); (c.1529G>A); (c.1544C>T); (c.1552C>G); (c.1552C>T); c.1553G>A; (c.1559T>A); (c.1586_1588delGTG); (c.1634+1G>A); (c.1635-2A>G); (c.1661G>A); (c.1772G>C); (c.1792_1793delAT); (c.1817C>T); (c.1822G>A); (c.1849C>T); (c.1850G>A); (c.1865+1G>A); (c.1866-10G>A); (c.1937delC); (c.1938_1939dupGG); (c.310C>T); (c.346G>T); (c.396G>A); (c.421G>A); (c.442A>G); (c.443A>G); (c.520T>G); (c.541_542delTA); (c.686T>C); (c.761C>T); (c.796G>A); (c.839G>T); (c.847C>G); (c.848A>G); (c.871G>A); (c.874_876delGAG); (c.901-1G>A)	General Population	1 in 500	12%	1 in 570
					Moroccan Jewish	1 in 500	60%	1 in 1250
Aicardi-Goutieres syndrome, type 5	SAMHD1	No	18	(c.1106T>C); (c.649_650insG); (c.676C>G); Exon1; Exon10; Exon11; Exon12; Exon13; Exon14; Exon15; Exon16; Exon2; Exon3; Exon4; Exon5; Exon6; Exon7; Exon8-9	Ashkenazi Jewish	1 in 111	75%	1 in 441
					General Population	1 in 500	10%	1 in 555
Albinism, oculocutaneous, type IA	TYR	X	14	(c.1037-1G>A); c.1037-7T>A; c.1118C>A; (c.1204C>T); c.1299C>G; c.140G>A; (c.149C>G); c.242C>T; c.325G>A; c.454C>T; c.649delC; (c.757G>A); c.832C>T; c.896G>A	Asian	1 in 80	18%	1 in 98
					General Population	1 in 100	10%	1 in 111
					Moroccan Jewish	1 in 29	83%	1 in 168
Albinism, oculocutaneous, type II/Albinism, brown oculocutaneous/	OCA2	No	1	c.1044+13634_c.2080-6294del122625	General Population	1 in 500	10%	1 in 555
Albinism, oculocutaneous, type IV	SLC45A2	No	2	c.469G>A; c.904A>T	General Population	1 in 500	10%	1 in 555
Alport syndrome	COL4A5	No	14	(c.2014G>A); (c.2039delC); (c.2164G>C); (c.2555G>T); (c.2696_2705delGTATGATGGG); (c.4235delG); (c.430G>C); (c.4691G>C); (c.476delG); (c.4942dupT); (c.4946T>G); (c.5030G>A); (c.64C>T); (c.81+1G>T)	General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Alport syndrome 2, autosomal recessive	COL4A3	No	1	c.4420_4424delCTTTT	General Population	1 in 500	10%	1 in 555
Alport syndrome 2, autosomal recessive	COL4A4	No	2	c.4129C>T; c.4715C>T	General Population	1 in 500	10%	1 in 555
Alstrom syndrome	ALMS1	No	1	c.10825C>T	General Population	1 in 500	10%	1 in 555
Anauxetic dysplasia 1/Cartilage-hair hypoplasia/Metaphyseal dysplasia without hypotrichosis	RMRP	No	2	n.263G>T; n.71A>G	General Population	1 in 500	10%	1 in 555
Argininosuccinic aciduria	ASL	X	6	c.1135C>T; (c.346C>T); c.35G>A; c.446+1G>A; c.532G>A; c.556C>T	General Population	1 in 132	30%	1 in 189
Arterial calcification, generalized, of infancy, 2/Pseudoxanthoma elasticum	ABCC6	No	2	c.3421C>T; c.4015C>T	General Population	1 in 500	10%	1 in 555
Arthrogryposis, autism spectrum disorder, and epilepsy	SLC35A3	No	2	c.514C>T; (c.886A>G)	Ashkenazi Jewish	1 in 205	99%	1 in 20401
					General Population	1 in 500	10%	1 in 555
Arthropathy, progressive pseudorheumatoid, of childhood	WISP3	X	2	c.156C>A; (c.536_537delGT)	Arab Muslim	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Asparagine synthetase deficiency	ASNS	No	1	(c.1084T>G)	General Population	1 in 500	10%	1 in 555
					Iranian Jewish	1 in 80	99%	1 in 7901
Aspartylglucosaminuria	AGA	No	1	c.214T>C	Arab Palestenan (Jerusalem)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia	APTX	No	1	c.837G>A	General Population	1 in 500	10%	1 in 555
Ataxia-telangiectasia	ATM	X	41	(c.103C>T); c.1339C>T; c.1564_1565delGA; (c.2284_2285delCT); (c.2839-579_2839-576delGTAA); c.3245_3247delATCinsTGAT; (c.3576G>A); c.368delA; c.4852C>T; (c.5763-1050A>G); c.5908C>T; (c.6672_6680delGGCTCTACGinsCTC); (c.7241_7244delAAGC); c.7630-2A>C; c.7638_7646delTAGAATTTC; Exon10-11; Exon12-16; Exon17-18; Exon19-22; Exon2-3; Exon23-24; Exon25-26; Exon27; Exon29-31; Exon32-33; Exon34; Exon35-36; Exon38; Exon4; Exon40-43; Exon44-45; Exon46-49; Exon50; Exon51-54; Exon55-56; Exon5-6; Exon57; Exon58-59; Exon62-63; Exon7; Exon8-9	General Population	1 in 100	10%	1 in 110
					Moroccan Jewish	1 in 81	97%	1 in 2668
					Tunisian Jewish	1 in 81	97%	1 in 2668
					Yemenite Jewish	1 in 81	97%	1 in 2668
Ataxia-telangiectasia-like disorder	MRE11A	No	1	(c.290A>G)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	AIRE	No	5	(c.1163_1164insA); (c.247A>G); c.254A>G; c.769C>T; c.967_979delCTGTCCCCTCCGC	General Population	1 in 500	35%	1 in 769
					Iranian Jewish	1 in 27	99%	1 in 2600
Bardet-Biedl syndrome 1	BBS1	No	1	c.1169T>G	General Population	1 in 342	79%	1 in 1625
Bardet-Biedl syndrome 10	BBS10	No	1	c.271dupT	General Population	1 in 354	46%	1 in 655
Bardet-Biedl syndrome 11	TRIM32	No	1	c.388C>T	Beduim Arab (Negev)	1 in 59	99%	1 in 5801
					General Population	1 in 500	10%	1 in 555
Bardet-Biedl syndrome 12	BBS12	No	2	c.1115_1116delTT; c.1589T>C	General Population	1 in 500	10%	1 in 555
Bardet-Biedl syndrome 13/Joubert syndrome 28/Meckel syndrome 1	MKS1	No	2	c.1408-35_1408-7del29; c.417G>A	General Population	1 in 500	10%	1 in 555
Bardet-Biedl syndrome 2	BBS2	No	3	c.1895G>C; c.224T>G; c.311A>C	Ashkenazi Jewish	1 in 136	99%	1 in 13501
					General Population	1 in 500	10%	1 in 555
Bardet-Biedl syndrome 3	ARL6	No	1	(c.364C>T)	General Population	1 in 500	10%	1 in 555
Bardet-Biedl syndrome 4	BBS4	No	17	c.77-220delA; (c.884G>C); Exon1; Exon10; Exon11-12; Exon13; Exon14; Exon15; Exon16; Exon2; Exon3; Exon4; Exon5; Exon6; Exon7; Exon8; Exon9	General Population	1 in 500	10%	1 in 555
Bartter syndrome, type 3	CLCNKB	X	1	c.1313G>A	General Population	1 in 500	10%	1 in 555
Bartter syndrome, type 4a	BSND	No	2	c.139G>A; (c.28G>A)	General Population	1 in 500	10%	1 in 555
Basel-Vanagait-Smirin-Yosef syndrome	MED25	X	1	c.116A>G	Arab Muslim (Jisr az-Zarqa)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Bernard-Soulier syndrome, type B/Giant platelet disorder, isolated	GP1BB	No	1	c.124_145del22	General Population	1 in 500	10%	1 in 555
Bernard-Soulier syndrome, type C	GP9	No	1	c.182A>G	General Population	1 in 500	10%	1 in 555
Biotinidase Deficiency	BTD	X	8	(c.100G>A); c.1330G>C; c.1368A>C; c.1595C>T; c.1612C>T; c.511G>A; c.528G>T; c.98_104delGCGGCTGinsTCC	Arab Muslim (Mashhad)	N/A	99%	1 in 133
					General Population	1 in 120	10%	1 in 133
Bjornstad syndrome/Leigh syndrome/Mitochondrial complex III deficiency, nuclear type 1	BCS1L	No	2	c.232A>G; c.548G>A	General Population	1 in 500	10%	1 in 555
Blood group, ABO system	ABO	No	1	c.802G>A	General Population	1 in 500	10%	1 in 555
Bloom syndrome	BLM	X	7	c.1284G>A; c.1933C>T; c.2207_2212delATCTGAinsTAGATTC; (c.2407dupT); c.2506_2507delAG; c.2695C>T; (c.3510T>A)	Ashkenazi Jewish	1 in 111	99%	1 in 11001
					General Population	1 in 500	10%	1 in 555
Bothnia retinal dystrophy/Fundus albipunctatus/Retinitis punctata albescens	RLBP1	No	2	c.141+2T>C; c.700C>T	General Population	1 in 500	10%	1 in 555
Brittle cornea syndrome 1	ZNF469	No	2	(c.5943delA); (c.9531delG)	General Population	1 in 500	10%	1 in 555
					Tunisian Jewish	N/A	99%	1 in 555
Canavan disease	ASPA	X	4	(c.433-2A>G); c.693C>A; c.854A>C; c.914C>A	Ashkenazi Jewish	1 in 55	99%	1 in 5401
					General Population	1 in 500	66%	1 in 1470
Carbamoylphosphate synthetase I deficiency	CPS1	X	3	(c.3265C>T); (c.3558+1G>C); (c.4101+2T>C)	Arab Christian	N/A	99%	1 in 555
					Druze Arab (Yarka)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Cardiomyopathy, dilated, 1GG	SDHA	X	1	(c.1664G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Carnitine palmitoyltransferase deficiency, hepatic, type IA	CPT1A	X	2	(c.1361A>G); c.2129G>A	Druze Arab (Abu Snan)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Carnitine palmitoyltransferase deficiency, hepatic, type II, infantile,lethal neonathal	CPT2	No	9	(c.110_111dupGC); (c.1148T>A); c.1239_1240delGA; c.149C>A; c.1646G>A; c.359A>G; c.452G>A; c.641T>C; c.680C>T	Ashkenazi Jewish	1 in 51	99%	1 in 5001
					General Population	1 in 274	80%	1 in 1366
Carnitine-acylcarnitine translocase deficiency	SLC25A20	X	1	(c.713A>G)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Cerebral creatine deficiency syndrome 1	SLC6A8	No	2	c.1540C>T; c.321_323delCTT	General Population	1 in 500	10%	1 in 555
Cerebral creatine deficiency syndrome 2	GAMT	No	1	c.506G>A	General Population	1 in 500	10%	1 in 555
Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome	SNAP29	X	1	(c.223delG)	Arab Muslim (Daburiyya)	N/A	99%	1 in 555
					Arab Muslim (Mashhad)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Cerebrotendinous xanthomatosis	CYP27A1	X	5	(c.1016C>T); (c.355delC); c.819delT; (c.844+1G>A); c.845-1G>A	Druze Arab (Yarka)	N/A	99%	1 in 124
					General Population	1 in 112	10%	1 in 124
					Moroccan Jewish	1 in 76	99%	1 in 7500
Ceroid lipofuscinosis, neuronal, 2/Spinocerebellar ataxia, autosomal recessive 7	TPP1	No	2	c.509-1G>C; c.622C>T	General Population	1 in 500	10%	1 in 555
Ceroid lipofuscinosis, neuronal, 3	CLN3	No	1	c.461-280_677+382del966	General Population	1 in 500	10%	1 in 555
Ceroid lipofuscinosis, neuronal, 5	CLN5	No	1	c.1175_1176delAT	General Population	1 in 500	10%	1 in 555
Cholestasis; progressive familial intrahepatic 2	ABCB11	X	1	(c.3268C>T)	Arab Muslim (Deir al-Asad)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Choreoacanthocytosis	VPS13A	No	64	5pExon48; c.3889C>T; (c.6059delC); Exon1; Exon10; Exon11-12; Exon13; Exon14; Exon15; Exon16; Exon17; Exon18-19; Exon2; Exon20; Exon21; Exon22; Exon23; Exon24; Exon25; Exon26; Exon27; Exon28-29; Exon3; Exon30-31; Exon32; Exon33; Exon34; Exon35; Exon36-37; Exon38; Exon39; Exon4; Exon40; Exon41; Exon42; Exon43-44; Exon45; Exon46; Exon47; Exon48-49; Exon5; Exon50; Exon51; Exon52; Exon53; Exon54; Exon55; Exon56; Exon57; Exon58; Exon59; Exon6; Exon60; Exon61; Exon62; Exon63; Exon64-67; Exon68; Exon69; Exon7; Exon70; Exon71; Exon72; Exon8-9	Ashkenazi Jewish	N/A	99%	1 in 124
					General Population	1 in 112	10%	1 in 124
Chronic granulomatous disease (cytochrome b-negative)	CYBA	No	2	c.171dupG; (c.71G>A)	General Population	1 in 500	10%	1 in 555
					Moroccan Jewish	1 in 13	83%	1 in 76
					Yemenite Jewish	1 in 13	83%	1 in 76
Chronic granulomatous disease (cytochrome b-positive, type 1)	NCF1	X	2	(c.153+1G>A); c.579G>A	General Population	1 in 447	10%	1 in 497
					Kavkazi (Caucasus) Jewish	N/A	99%	1 in 497
Chronic granulomatous disease, X-linked	CYBB	No	25	(c.1016dupC); (c.1081T>C); (c.1166G>C); (c.1244C>A); (c.1499A>G); (c.217C>T); (c.252+5G>A); (c.252G>A); (c.271C>T); (c.301C>T); (c.302A>G); (c.388C>T); (c.388delC); (c.45+6T>C); (c.466G>A); (c.469C>T); (c.483+978G>T); (c.625C>T); (c.676C>T); (c.742dupA); (c.868C>T); (c.8dupA); (c.90_92delCCGinsGGT); (c.907C>A); (c.911C>G)	Ashkenazi Jewish	1 in 102	99%	1 in 10101
					General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	1 in 102	99%	1 in 10101
					Moroccan Jewish	1 in 102	99%	1 in 10101
					Yemenite Jewish	1 in 102	99%	1 in 10101
Ciliary dyskinesia, primary, 1, with or without situs inversus	DNAI1	No	4	c.1212T>G; c.1490G>A; c.1612G>A; c.48+2dupT	General Population	1 in 500	10%	1 in 555
Ciliary dyskinesia, primary, 16	DNAL1	No	1	(c.449A>G)	General Population	1 in 500	10%	1 in 555
Citrullinemia	ASS1	No	5	c.1087C>T; c.1168G>A; c.535T>C; c.787G>A; c.970G>A	General Population	1 in 500	10%	1 in 555
Citrullinemia, adult-onset type II/Citrullinemia, type II, neonatal-onset	SLC25A13	No	1	c.1336A>C	General Population	1 in 500	10%	1 in 555
Clopidogrel, impaired responsiveness to/Mephenytoin poor metabolizer/Omeprazole poor metabolizer/Proguanil poor metabolizer	CYP2C19	No	2	c.358T>C; c.636G>A	General Population	1 in 500	10%	1 in 555
Cockayne syndrome	ERCC6	X	1	(c.1034-1035insT)	Druze Arab (Kisra-Sumei)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Cockayne, type A	ERCC8	X	1	c.966C>A	Arab Christian	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Coenzyme Q10 deficiency, primary, 7	COQ4	No	1	c.718C>T	General Population	1 in 500	10%	1 in 555
Combined malonic and methylmalonic aciduria	ACSF3	No	2	c.1411C>T; c.1672C>T	General Population	1 in 500	10%	1 in 555
Complement factor H deficiency	CFH	No	1	(c.3674A>T;3675_3699del24TCCAACTTGTGCAAAAAGATAGAA)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Complex hereditary spastic paraparesis	PLAA	X	1	(c.2254C>T)	Arab Muslim (Nachef)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Congenital adrenal insufficiency with 46,XY sex reversal	CYP11A1	No	2	(c.644T>C); (c.694C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Congenital amegakaryocytic thrombocytopenia	MPL	No	2	c.127C>T; c.79+2T>A	Ashkenazi Jewish	1 in 75	99%	1 in 7401
					General Population	1 in 500	10%	1 in 555
Congenital arthrogryposis with anterior horn cell disease/Lethal congenital contracture syndrome 1	GLE1	No	1	c.1706G>A	General Population	1 in 500	10%	1 in 555
Congenital disorder of deglycosylation	NGLY1	X	1	(c.1294G>T)	Druze Arab (Julis)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Congenital disorder of glycosylation type Ia	PMM2	No	6	(c.338C>T); c.357C>A; c.415G>A; c.422G>A; c.470T>C; (c.691G>A)	Ashkenazi Jewish	1 in 61	99%	1 in 6001
					General Population	1 in 71	44%	1 in 126
Congenital disorder of glycosylation, type Ib	MPI	No	1	c.305C>T	General Population	1 in 500	10%	1 in 555
Congenital disorder of glycosylation, type Im	DOLK	X	1	(c.912G>T)	Bedouin Arab (Aramsha)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Cornelia de Lange like (Birk Flusser) syndrome	FRMD4A	No	1	(c.2134_2146dupCTGGAGTCCCAGG)	Bedouin Arab (Negev)	1 in 17	99%	1 in 1601
					General Population	1 in 500	10%	1 in 555
Crigler-Najjar syndrome, type I/Crigler-Najjar syndrome, type II/Hyperbilirubinemia, familial transient neonatal/Gilbert syndrome	UGT1A1	No	1	c.211G>A	General Population	1 in 500	10%	1 in 555
Cystic fibrosis	CFTR	X	381	c.1000C>T; c.1001G>A; (c.1006_1007insG); (c.1007T>A); c.1013C>T; c.1021_1022dupTC; c.1021T>C; (c.1029delC); c.1040G>A; c.1040G>C; (c.1040G>T); (c.1055G>A); c.1075C>A; (c.1079C>A); (c.1081delT); (c.1090T>C); c.1116+1G>A; (c.1117-1G>A); (c.1128dupA); c.1135G>T; c.1152delA; c.1155_1156dupTA; (c.115C>T); (c.11C>A); (c.1202G>A); (c.1203G>A); (c.1209+1G>A); c.1210-7_1210-6del[5T]; (c.1240C>T); (c.1327_1330dupGATA); (c.1340delA); c.1364C>A; c.1365_1366delGG; (c.137C>A); (c.1393-1G>A); (c.1393-2A>G); (c.1397C>A); (c.1397C>G); (c.1400T>C); (c.1418delG); c.1438G>T; (c.1439G>A); c.1466C>A; (c.1475C>T); (c.1477_1478delCA); (c.1477C>T); (c.1487G>A); (c.14C>T); c.1519_1521delATC; c.1521_1523delCTT; (c.1545_1546delTA); c.1550A>G; c.1558G>T; (c.1572C>A); (c.1573C>T); c.1584+1G>A; c.1585-1G>A; c.1585-8G>A; c.1624G>T; (c.1625G>A); c.164+12T>C; (c.164+1G>A); (c.164+1G>T); (c.164+2T>C); c.1645_1648delAGTG; (c.1645A>C); c.1646G>A; (c.1646G>T); c.1647T>G; (c.1648G>T); (c.1650delA); (c.1651G>A); (c.165-1G>A); c.1652G>A; (c.1654C>T); c.1657C>T; (c.166G>A); (c.1670delC); (c.1673T>C); (c.1675G>A); (c.1679+1643G>T); (c.1679+1G>A); c.1679+1G>C; c.1679G>A; c.1679G>C; (c.1680-1G>A); (c.1680-886A>G); (c.1682C>A); c.168delA; (c.1692delA); (c.1703delT); (c.1705T>G); (c.170G>A); (c.171G>A); (c.1721C>A); (c.1736A>G); (c.174_177delTAGA); (c.1753G>T); (c.175dupA); c.1766+1G>A; c.1766+1G>C; (c.1766+1G>T); c.1766+3A>G; c.1766+5G>T; (c.178G>T); (c.1792_1798delAAAACTA); (c.1817_1900del84); (c.1826A>G); (c.1841A>G); c.1865G>A; (c.1911delG); (c.1923_1931delCTCAAAACTinsA); c.1966G>T; (c.1973_1985delGAAATTCAATCCTinsAGAAA); (c.1976delA); c.1986_1989delAACT; (c.1A>G); c.200C>T; (c.2012delT); (c.2017G>T); c.2051_2052delAAinsG; c.2052delA; c.2052dupA; (c.2053C>T); (c.2053dupC); c.2058_2061dupTTTT; c.2125C>T; (c.2128A>T); (c.2143C>T); c.2158C>T; c.2175dupA; c.2188G>T; (c.2195T>G); c.220C>T; c.2215delG; c.223C>T; (c.2241_2248del8); (c.2290C>T); (c.233dupT); (c.2353C>T); (c.2374C>T); c.2421A>G; (c.2423_2424dupAT); (c.2453delT); (c.2463_2464delTG); (c.2464G>T); (c.2490+1G>A); (c.2491G>T); (c.2537G>A); (c.2547C>A); c.254G>A; (c.2551C>T); (c.2583delT); (c.2589_2599delAATTTGGTGCT); c.259T>A; (c.2601dupA); (c.2619+1G>A); (c.2619+2dupT); (c.262_263delTT); (c.263T>A); c.263T>G; (c.2645G>A); (c.2657+2_2657+3insA); c.2657+5G>A; (c.2658-1G>C); c.2668C>T; c.273+1G>A; (c.273+3A>C); c.2735C>A; c.2737_2738insG; c.2739T>A; (c.274-1G>A); (c.274-2A>G); (c.274G>A); (c.274G>T); (c.2763_2764dupAG); (c.2780T>C); c.2810dupT; (c.2825delT); c.2834C>T; (c.2859_2890delACATTCTGTTCTTCAAGCACCTATGTCAACCC); (c.2875delG); (c.2896delA); (c.2908G>C); (c.292C>T); c.2930C>T; c.2932A>T; c.2988+1173_3468+2111del8600; c.2988+1G>A; (c.2988G>A); c.2989-1G>A; (c.2989-2A>G); (c.2989-977_3367+248del); (c.3002_3003delTG); (c.3039delC); (c.3039dupC); c.3041A>G; (c.3067_3072delATAGTG); (c.310delA); (c.3124C>T); (c.3139_3139+1delGG); (c.313delA); c.3140-1G>A; (c.3140-26A>G); c.3154T>G; (c.3160C>G); c.3179A>C; (c.3181G>C); (c.3194T>C); c.3196C>T; c.3197G>A; (c.3205G>A); (c.3208C>T); (c.3209G>A); (c.3222T>A); (c.3230T>C); (c.325_327delTATinsG); c.3266G>A; (c.3276C>A); c.3276C>G; c.328delG; c.328G>C; (c.3292T>C); (c.3293G>A); (c.3294G>A); c.3299A>C; (c.3302T>A); (c.3304A>T); (c.330C>A); c.3310G>T; c.3364delA; (c.3368-2A>G); c.3382A>T; (c.3435G>A); (c.343G>T); c.3454G>C; c.3468+2T>C; (c.3469-2A>G); (c.3472C>T); c.3484C>T; c.349C>T; c.350G>A; c.3528delC; (c.3529A>T); c.3530delA; (c.3535_3536insTCAA); (c.3536_3539delCCAA); (c.3587C>G); (c.3600delA); (c.3605delA); (c.3611G>A); (c.3612G>A); c.3659delC; (c.366T>A); c.3691delT; c.3700A>G; (c.3712C>T); c.3717+1G>A; (c.3717+40A>G); (c.3717+4A>G); (c.3718-1G>A); c.3718-2477C>T; c.3718-3T>G; (c.3731G>A); (c.3744delA); (c.3747delG); (c.3752G>A); (c.3761T>G); (c.3763T>C); c.3764C>A; c.3773dupT; (c.3808delG); (c.3808G>A); c.3846G>A; (c.3848G>T); (c.3873+1G>A); (c.3873+2T>C); (c.3883_3886delATTT); (c.3883delA); (c.3889dupT); (c.3891dupT); (c.3899dupT); (c.3908delA); (c.3908dupA); c.3909C>G; c.3937C>T; (c.3964-78_4242+577del); (c.4036_4042delCTAAGCC); c.4046G>A; (c.4077_4080delTGTTinsAA); (c.4086dupT); (c.409delC); (c.4111G>T); c.413_415dupTAC; c.4140delA; c.4143C>A; c.4144C>T; (c.4147dupA); c.4168C>T; c.416A>T; c.4170delA; (c.4197_4198delCT); (c.422C>A); (c.4231C>T); (c.4234C>T); (c.4242+1G>A); (c.4242+1G>T); (c.4251delA); (c.4300_4301dupAG); c.4364C>G; (c.442delA); c.443T>C; (c.487A>G); c.489+1G>T; c.489+2T>C; (c.489+3A>G); (c.50delT); c.523A>G; (c.53+1G>T); (c.531delT); (c.532G>A); (c.543_546delTAGT); c.54-5940_273+10250del21Kb; c.575A>G; c.577G>T; c.578_579+5delAAGTATG; c.579+1G>T; (c.579+3A>G); (c.579+5G>A); (c.57G>A); (c.580-1G>T); (c.595C>T); c.613C>T; c.617T>G; (c.647G>A); (c.658C>T); c.675T>A; (c.680T>G);	African American	1 in 61	77%	1 in 262
					Ashkenazi Jewish	1 in 24	97%	1 in 768
					Asian	1 in 94	54%	1 in 203
					Caucasian	1 in 25	94%	1 in 401
					General Population	1 in 25	94%	1 in 401

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
				c.720_741delAGGGAGAATGATGATGAAGTAC; c.743+1G>A; (c.761delA); (c.79G>T); (c.803delA); (c.805_806delAT); (c.825C>G); (c.828C>A); (c.850dupA); c.861_865delAACTT; (c.870-2A>G); (c.88C>T); (c.-9_14delCGAGAGACCATGCAGAGGTCGCC); c.935_937delTCT; (c.948delT); (c.987delA); (c.988G>T); Exon1; Exon10; Exon11; Exon12; Exon13; Exon14; Exon15; Exon16; Exon17; Exon18; Exon19-20; Exon2; Exon21; Exon22; Exon23; Exon24; Exon25; Exon26; Exon27; Exon3; Exon4; Exon5; Exon6; Exon7; Exon8; Exon9	Hispanic	1 in 58	74%	1 in 220
Cystinosis, nephropathic	CTNS	No	4	c.1015G>A; c.-39273_842del57232; (c.530A>C); c.613G>A	General Population	1 in 158	10%	1 in 175
					Moroccan Jewish	1 in 100	92%	1 in 1240
Cystinuria	SLC3A1	No	1	c.1136+2T>C	General Population	1 in 500	10%	1 in 555
D-bifunctional protein deficiency/Perrault syndrome 1	HSD17B4	No	1	c.1369A>T	General Population	1 in 500	10%	1 in 555
Deafness, autosomal recessive 1	GJB2	X	20	c.109G>A; c.167delT; c.176_191delGCTGCAAGAACGTGTG; c.229T>C; (c.-23+1G>A); (c.230G>A); c.231G>A; c.235delC; c.269T>C; c.299_300delAT; c.313_326delAAGTTCATCAAGGG; c.358_360delGAG; c.35delG; (c.370C>T); c.427C>T; (c.51_62delCACCAGCATTGGinsA); (c.551G>C); c.613C>G; c.71G>A; c.95G>A	Ashkenazi Jewish	1 in 21	95%	1 in 401
					Asian	1 in 76	73%	1 in 279
					Bukharian Jewish	43.478260869	95%	1 in 909
					Caucasian	1 in 42	79%	1 in 196
					General Population	1 in 43	71%	1 in 146
					Iraqi Jewish	43.478260869	95%	1 in 909
	GJB6	X	4	3pDel1000kb; 5p300kbMicrodeletion; 5pDel700kb; Exon1-3	General Population	1 in 500	90%	1 in 5000
Deafness, autosomal recessive 12	CDH23	No	2	c.5237G>A; c.7903G>T	Algerian Jewish	N/A	99%	1 in 218
					General Population	1 in 197	10%	1 in 218
Deafness, autosomal recessive 16	STRC	No	3	5p100kbDeletion; c.4171C>G; Exon1-29	Ashkenazi Jewish	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Deafness, autosomal recessive 22	OTOA	No	1	(c.1025A>T)	General Population	1 in 500	10%	1 in 555
Deafness, autosomal recessive 3	MYO15A	No	3	(c.373_374delCG); (c.4240G>A); (c.8183G>A)	Ashkenazi Jewish	1 in 80	90%	1 in 791
					General Population	1 in 500	10%	1 in 555
Deafness, autosomal recessive 59	DFNB59	X	1	(c.406C>T)	Arab Muslim	1 in 12	99%	1 in 1101
					General Population	1 in 500	10%	1 in 555
Deafness, autosomal recessive 7	TMC1	X	5	(c.100C>T); (c.1165C>T); c.1210T>C; c.1810C>T; c.1939T>C	Bedouin Arab (Negev)	N/A	99%	1 in 943
					General Population	1 in 500	47%	1 in 943
					Moroccan Jewish	1 in 18	99%	1 in 1701
Deafness, autosomal recessive 76	SYNE4	No	1	c.228_229delAT	General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	1 in 39	99%	1 in 3801
Deafness, autosomal recessive 77	LOXHD1	No	1	(c.4714C>T)	Ashkenazi Jewish	1 in 180	99%	1 in 17901
					General Population	1 in 500	10%	1 in 555
Deafness, autosomal recessive 8/10	TMPRSS3	No	1	(c.989delA)	General Population	1 in 500	10%	1 in 555
Desmosterolosis	DHCR24	No	1	(c.307C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Diabetes insipidus, nephrogenic	AQP2	No	2	(c.298G>T); (c.83T>C)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Diabetes insipidus, neurohypophyseal	AVP	No	1	c.77C>T	General Population	1 in 500	10%	1 in 555
Diaphanospondylodysostosis	BMPER	No	1	(c.310C>T)	Arab Muslim (East Jerusalem)	1 in 123	99%	1 in 12201
					General Population	1 in 500	10%	1 in 555
Diarrhea 7, protein-losing enteropathy type	DGAT1	No	1	c.751+2T>C	General Population	1 in 500	10%	1 in 555
Dihydrolipoamide dehydrogenase deficiency	DLD	No	4	(c.104dupA); c.1123G>A; (c.1436A>T); c.685G>T	Ashkenazi Jewish	1 in 107	95%	1 in 2121
					Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Disordered steroidogenesis due to cytochrome P450 oxidoreductase	POR	X	1	(c.1615G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Duchenne muscular dystrophy	DMD	No	105	c.10018T>C; c.10108C>T; c.10126delC; (c.10492_10493insAAT); c.10546G>T; c.1238C>G; (c.2002delG); c.2268delC; c.2971G>T; c.3172C>T; (c.4351_4352insA); c.4611delT; (c.4675-2A>G); (c.4856_4857delAA); (c.4960dupA); (c.6638delT); (c.6808_6811delTTAA); c.7402G>T; (c.7814C>G); (c.8027+2T>C); c.8038C>T; (c.8608C>T); (c.8656C>T); c.8713C>T; c.883C>T; c.9457delT; c.9739C>T; Exon1; Exon10; Exon11; Exon12; Exon13; Exon14-15; Exon16; Exon17; Exon18; Exon19; Exon2; Exon20; Exon21; Exon22; Exon23; Exon24; Exon25; Exon26; Exon27; Exon28; Exon29; Exon3; Exon30; Exon31; Exon32; Exon33; Exon34; Exon35; Exon36; Exon37; Exon38; Exon39; Exon4; Exon40; Exon41; Exon42; Exon43; Exon44; Exon45; Exon46; Exon47; Exon48; Exon49; Exon5; Exon50; Exon51; Exon52; Exon53; Exon54; Exon55; Exon56; Exon57; Exon58; Exon59; Exon6; Exon60; Exon61; Exon62; Exon63; Exon64; Exon65; Exon66; Exon67; Exon68; Exon69; Exon7; Exon70; Exon71; Exon72; Exon73; Exon74; Exon75; Exon76; Exon77; Exon78; Exon79; Exon8; Exon9	General Population	1 in 500	75%	1 in 833
Dysautonomia, familial	IKBKAP	X	3	c.2087G>C; c.2204+6T>C; (c.2741C>T)	Ashkenazi Jewish	1 in 31	99%	1 in 3001
					General Population	1 in 500	10%	1 in 555
Dyserythropoietic anemia, congenital, type Ia	CDAN1	No	2	(c.2605G>A); (c.3124C>T)	Arab	1 in 180	99%	1 in 17901
					Bedouin Arab	1 in 125	99%	1 in 12401
					General Population	1 in 500	10%	1 in 555
Dyserythropoietic anemia, congenital, type II	SEC23B	No	2	(c.2129C>T); c.325G>A	General Population	1 in 500	10%	1 in 555
					Moroccan Jewish	1 in 102	99%	1 in 10101

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Dyskeratosis congenita, autosomal recessive 4 (TERT-related)	TERT	No	1	(c.2701C>T)	General Population	1 in 500	10%	1 in 555
					Iranian Jewish	N/A	99%	1 in 555
Dyskeratosis congenita, autosomal recessive 5 (RTEL1-related)	RTEL1	No	1	c.3791G>A	Ashkenazi Jewish	N/A	67%	1 in 555
					General Population	1 in 500	10%	1 in 555
Dysprothrombinemia/Hypoprothrombinemia	F2	No	1	c.*97G>A	General Population	1 in 500	10%	1 in 555
Ectodermal dysplasia 1, hypohidrotic, X-linked	EDA	No	6	c.1045G>A; c.457C>T; c.463C>T; c.466C>T; c.730C>T; c.895G>A	General Population	1 in 500	10%	1 in 555
Ehlers-Danlos syndrome, type VII-C	ADAMTS2	No	2	(c.2384G>A); c.673C>T	Ashkenazi Jewish	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Encephalopathy, neonatal severe/Mental retardation, X-linked syndromic, Lubs type/Mental retardation, X-linked, syndromic 13	MECP2	No	3	Exon2; Exon3; Exon4	General Population	1 in 500	10%	1 in 555
Enhanced S-cone syndrome	NR2E3	No	2	c.119-2A>C; c.932G>A	Arab Muslim	N/A	99%	1 in 261
					Ashkenazi Jewish	1 in 20	99%	1 in 20000
					General Population	1 in 204	58%	1 in 484
					Moroccan Jewish	N/A	99%	1 in 261
					Portuguese Jewish	N/A	99%	1 in 261
					Spanish Jewish	N/A	99%	1 in 261
					Tunisian Jewish	N/A	99%	1 in 261
Epidermolysis bullosa, junctional, Herlitz type (LAMA3-related)	LAMA3	X	3	(c.2975delA); (c.4815G>T); (c.6808C>T)	Arab Muslim (Bi'ina)	N/A	99%	1 in 555
					Arab Muslim (Deir al-Asad)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Epidermolysis bullosa, junctional, Herlitz type (LAMB3-related)	LAMB3	X	7	c.1903C>T; (c.2166C>A); (c.2914C>T); c.3024delT; (c.3247C>T); (c.430C>T); c.727C>T	Arab Muslim (Bi'ina)	1 in 15	99%	1 in 1401
					Arab Muslim (Deir al-Asad)	1 in 15	99%	1 in 1401
					General Population	1 in 781	76%	1 in 3251
Epidermolysis bullosa, junctional, Herlitz type (LAMC2-related)	LAMC2	No	1	(c.1756C>T)	Arab Muslim	N/A	50%	1 in 222
					General Population	1 in 781	10%	1 in 868
Epidermolysis bullosa; junctional; with pyloric atresia	ITGB4	No	1	c.3279_3739+180del2284	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Epilepsy, progressive myoclonic 1B	PRICKLE1	No	1	c.311G>A	Arab Muslim (Galilee)	N/A	99%	1 in 555
					Arab Muslim (Triangle)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Epilepsy, progressive myoclonic 2A (Lafora)	EPM2A	No	1	Exon1-4	General Population	1 in 500	10%	1 in 555
Epilepsy, pyridoxine-dependent	ALDH7A1	No	1	c.1279G>C	General Population	1 in 500	10%	1 in 555
Fabry disease	GLA	No	3	c.124A>C; c.194G>C; c.644A>G	General Population	1 in 500	10%	1 in 555
Factor VII deficiency	F7	X	1	(c.1256C>T)	Arab Muslim (Majd al-Krum)	N/A	99%	1 in 393
					General Population	1 in 354	10%	1 in 393
Fanconi anemia, complementation group A	FANCA	X	16	c.2172dupG; c.3788_3790delTCT; c.4275delT; (c.891_893+1delCTGG); Exon10; Exon12-14; Exon23; Exon25-26; Exon28; Exon30; Exon32; Exon37; Exon39-40; Exon41-42; Exon43; Exon4-5	Druze Arab (Kisra-Sumei)	N/A	99%	1 in 383
					General Population	1 in 345	10%	1 in 383
					Moroccan Jewish	1 in 133	99%	1 in 13234
					Tunisian Jewish	1 in 133	99%	1 in 13234
Fanconi anemia, complementation group C	FANCC	X	9	c.1642C>T; (c.1661T>C); c.37C>T; c.456+4A>T; c.553C>T; c.65G>A; c.66G>A; c.67delG; c.844-1G>C	Ashkenazi Jewish	1 in 89	99%	1 in 8900
					General Population	1 in 417	30%	1 in 595
Fanconi anemia, complementation group G	FANCG	No	2	c.1480+1G>C; c.307+1G>C	General Population	1 in 500	10%	1 in 555
Fanconi-Bickel syndrome	SLC2A2	No	1	(c.901C>T)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Fatty liver, acute, of pregnancy/HELLP syndrome, maternal, of pregnancy/LCHAD deficiency/Trifunctional protein deficiency	HADHA	No	1	c.1528G>C	General Population	1 in 500	10%	1 in 555
Fructose intolerance, hereditary	ALDOB	No	5	c.1005C>G; c.-11-2042_624+62del644; c.442T>C; c.448G>C; c.524C>A	General Population	1 in 500	10%	1 in 555
Fumarase deficiency	FH	No	1	c.935T>G	General Population	1 in 500	10%	1 in 555
Fundus albipunctatus	RDH5	No	1	c.71_74delTGCC	Ashkenazi Jewish	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	N/A	10%	1 in 555
Galactosemia	GALT	No	16	c.1030C>A; c.-1039_753del3162; c.-119_-116delGTCA; (c.152G>A); c.221T>C; c.253-2A>G; c.292G>A; c.404C>T; c.413C>T; c.425T>A; c.512T>C; c.563A>G; c.584T>C; (c.626A>G); c.855G>T; c.997C>T	Ashkenazi Jewish	1 in 127	99%	1 in 12601
					General Population	1 in 110	60%	1 in 273
Gaucher disease, type I	GBA	X	15	c.115+1G>A; c.1226A>G; c.1263_1317del55; (c.1294T>A); c.1297G>T; c.1342G>C; c.1343A>T; c.1448T>C; (c.1448T>G); c.1504C>T; c.1505G>A; c.1604G>A; c.259C>T; c.721G>A; c.84dupG	Ashkenazi Jewish	1 in 15	95%	1 in 281
					General Population	1 in 158	60%	1 in 394
Glanzmann thrombasthenia (ITGA2B-related)	ITGA2B	No	1	(c.409-2_419delAGGCCTGCGCCCC)	General Population	1 in 500	10%	1 in 555
Glanzmann thrombasthenia (ITGB3-related)	ITGB3	No	3	c.2031_2041delTGCAGTGAATT; (c.428T>G); Exon1-15	General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	1 in 100	99%	1 in 9901
Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset	CYP1B1	No	3	c.1405C>T; (c.1568G>A); (c.182G>A)	General Population	1 in 500	10%	1 in 555
Glutaric acidemia IIA	ETFA	No	1	c.797C>T	General Population	1 in 500	10%	1 in 555
Glutaric aciduria type IIC	ETFDH	X	2	(c.1084G>A); c.250G>A	Arab Muslim (Ein Mahil)	N/A	99%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
					General Population	1 in 500	10%	1 in 555
Glutaricaciduria, type I	GCDH	No	9	c.1198G>A; c.1204C>T; c.1240G>A; (c.1247C>T); c.1262C>T; c.301G>A; c.464A>G; (c.505+1G>A); c.91+5G>T	General Population	1 in 158	40%	1 in 263
Glycine encephalopathy (AMT-related)	AMT	X	1	(c.125A>G)	Arab Muslim (Ilut)	N/A	99%	1 in 351
					General Population	1 in 316	10%	1 in 351
Glycine encephalopathy (GLDC-related)	GLDC	X	5	c.2216G>A; c.2311G>A; (c.2405C>T); c.2607C>A; (c.2T>C)	Arab Muslim (At-Tur Mount of Olives Jerusalem)	1 in 14	99%	1 in 1301
					Bedouin Arab (Negev)	N/A	99%	1 in 130
					General Population	1 in 117	10%	1 in 130
Glycogen storage disease Ia (von Gierke disease)	G6PC	X	15	c.1022T>A; c.1039C>T; c.113A>T; c.247C>T; c.248G>A; c.379_380dupTA; (c.497T>G); c.508C>T; c.562G>A; c.562G>C; c.648G>T; (c.724C>T); (c.79delC); (c.809G>T); (c.979_981delTTC)	Ashkenazi Jewish	1 in 71	99%	1 in 7001
					Asian	1 in 192	79%	1 in 911
					Caucasian	1 in 177	77%	1 in 766
					General Population	1 in 261	60%	1 in 651
					Hispanic	1 in 177	28%	1 in 245
Glycogen storage disease Ib	SLC37A4	X	3	c.1015G>T; c.1042_1043delCT; (c.83G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 416
					General Population	1 in 354	15%	1 in 416
Glycogen storage disease II (Pompe disease)	GAA	X	13	(c.1064T>C); (c.1210G>A); c.1843G>A; c.1935C>A; c.2238G>C; c.2482_2646del165; c.2560C>T; c.2815_2816delGT; c.-32-13T>G; c.525delT; c.670C>T; c.872T>C; c.953T>C	Ashkenazi Jewish	1 in 58	67%	1 in 174
					Bedouin Arab (Hussniyya)	N/A	99%	1 in 398
					Druze Arab (Maghar)	N/A	99%	1 in 398
					General Population	1 in 132	67%	1 in 398
Glycogen storage disease IIIa/IIIb (Cori or Forbes disease)	AGL	X	4	c.1222C>T; c.18_19delGA; c.4260-12A>G; c.4456delT	General Population	1 in 158	10%	1 in 175
					Moroccan Jewish	1 in 35	99%	1 in 3401
Glycogen storage disease IV	GBE1	No	2	c.986A>C; c.986A>G	Ashkenazi Jewish	71.428571428	99%	1 in 7143
					General Population	1 in 500	10%	1 in 555
Glycogen storage disease VII (Tarui disease)	PFKM	No	3	(c.116G>T); (c.283C>T); c.450+1G>A	Ashkenazi Jewish	1 in 250	39%	1 in 409
					General Population	1 in 500	10%	1 in 555
GM1-gangliosidosis, type I/typeII/typeIII/Mucopolysaccharidosis type IVB (Morquio)	GLB1	No	2	c.442C>A; c.601C>T	General Population	1 in 500	10%	1 in 555
Gray platelet syndrome	NBEAL2	X	1	(c.2701C>T)	Arab Muslim (Ayn Hawd)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Growth hormone deficiency, isolated, type IA (GH1-related)	GH1	No	4	(c.456+5G>C); Exon1-2; Exon2-3; Exon4-5	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Growth hormone deficiency, isolated, type IB (GHRHR - related)	GHRHR	No	1	(c.1069C>T)	Arab Muslim (Israel)	1 in 50	99%	1 in 4901
					General Population	1 in 500	10%	1 in 555
Growth retardation, developmental delay, facial dysmorphism (GDFD)	FTO	No	1	(c.947G>A)	Arab (Palestinian)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Haim-Munk syndrome	CTSC	No	1	(c.857A>G)	General Population	1 in 500	10%	1 in 555
					Indian Jewish (Cochin)	N/A	99%	1 in 555
Hemochromatosis, type 2A	HFE2	No	1	c.959G>T	General Population	1 in 500	10%	1 in 555
Hemolytic anemia, with or without immune-mediated polyneuropathy	CD59	No	1	(c.266G>A)	Egyptian Jewish	1 in 66	99%	1 in 6468
					General Population	1 in 500	10%	1 in 555
					Libyan Jewish	1 in 66	99%	1 in 6468
					Moroccan Jewish	1 in 66	99%	1 in 6468
Hermansky-Pudlak syndrome 3	HPS3	No	4	c.1163+1G>A; c.1-2993_c.217+690del3900; (c.1691+2T>G); (c.2482-2A>G)	Ashkenazi Jewish	1 in 235	90%	1 in 2341
					General Population	1 in 500	10%	1 in 555
Hermansky-Pudlak syndrome 6	HPS6	No	1	(c.1065dupG)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
HMG-CoA lyase deficiency	HMGCL	X	2	c.122G>A; c.914_915delTT	Arab Muslim (Bu'eine Nujeidat)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Homocystinuria due to MTHFR deficiency	MTHFR	X	1	(c.474A>T)	Bukharian Jewish	1 in 39	99%	1 in 3821
					General Population	1 in 500	10%	1 in 555
Homocystinuria, B6-responsive and nonresponsive types	CBS	No	5	c.1006C>T; c.1224-2A>C; c.341C>T; c.572C>T; c.919G>A	General Population	1 in 500	10%	1 in 555
Homocystinuria, cblD type, variant 1/Methylmalonic aciduria and homocystinuria, cblD type/Methylmalonic aciduria, cblD type, variant 2	MMADHC	No	2	c.160C>T; c.748C>T	General Population	1 in 500	10%	1 in 555
Hyperinsulinemic hypoglycemia, familial, 1	ABCC8	No	5	(c.2509C>T); c.2857C>T; c.3992-9G>A; (c.4163_4165delTCT); c.560T>A	Ashkenazi Jewish	1 in 52	99%	1 in 5101
					Bedouin Arab (Negev)	N/A	99%	1 in 185
					Finnish	1 in 100	43%	1 in 175
					General Population	1 in 167	10%	1 in 185
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	SLC25A15	No	1	c.95C>G	General Population	1 in 500	10%	1 in 555
					Arab Muslim (Abu Ghosh)	N/A	99%	1 in 175

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Hyperoxaluria, primary, type 1	AGXT	X	12	(c.121G>A); (c.33dupC); c.466G>A; c.508G>A; (c.584T>G); c.613T>C; (c.680+1G>A); c.697C>T; (c.727G>C); c.731T>C; (c.837T>G); (c.997A>T)	Arab Muslim (Bu'eine Nujeidat)	N/A	99%	1 in 175
					Druze Arab (Kisra-Sumei)	N/A	99%	1 in 175
					Druze Arab (Yanuh-Jat)	N/A	99%	1 in 175
					General Population	1 in 158	10%	1 in 175
Hyperphenylalaninemia, BH4-deficient, A	PTS	No	2	c.155A>G; c.259C>T	General Population	1 in 500	10%	1 in 555
Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis	SARS2	X	1	c.1175A>G	Arab Muslim (Sur Baher)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Hypoaldosteronism, congenital, due to CMO I deficiency/Hypoaldosteronism, congenital, due to CMO II deficiency	CYP11B2	No	1	c.763G>T	General Population	1 in 500	10%	1 in 555
Hypomagnesemia 1, intestinal	TRPM6	X	1	(c.2009+1G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Hypoparathyroidism-retardation-dysmorphism syndrome	TBCE	X	2	(c.155_166delGCCACGAAGGGA); (c.208-209delAT)	Bedouin Arab (Ras al-Ein)	N/A	99%	1 in 555
					Bedouin Arab (Kammana Sallama)	N/A	99%	1 in 555
					Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Hypophosphatasia, infantile	ALPL	X	5	c.1001G>A; c.1133A>T; (c.1348C>T); c.331G>A; c.571G>A	Arab Muslim (Kfar Manda)	N/A	99%	1 in 383
					General Population	1 in 345	10%	1 in 383
Hypophosphatemic rickets with hypercalciuria	SLC34A3	No	1	(c.228delC)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Hypothyroidism, congenital, nongoitrous, 1	TSHR	No	2	(c.1825C>T); (c.1957C>G)	Arab Muslim (Jish)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Hypotonia-cystinuria syndrome	2p21	No	6	3pPPM1B-5pSLC3A; CAMKMT_Exon1-10; CAMKMT-3p; PPM1B_Exon2-6; PREPL_Exon1-15; SLC31A_Exon2-9	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Inclusion body myopathy, autosomal recessive	GNE	No	2	c.1225G>T; c.2228T>C	General Population	1 in 500	10%	1 in 555
					Iranian Jewish	1 in 10	99%	1 in 1007
Infantile neuroaxonal dystrophy 1	PLA2G6	X	2	(c.2070_2072delTGT); (c.2251G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Inflammatory bowel disease 28; early onset; autosomal recessive	IL10RA	No	1	(c.537G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Insensitivity to pain, congenital	SCN9A	X	1	(c.2687G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Insensitivity to pain, congenital, with anhidrosis	NTRK1	X	3	(c.1842_1843insT); (c.1976C>T); (c.207_208delTG)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
					Moroccan Jewish	N/A	99%	1 in 555
Isovaleric acidemia	IVD	No	1	c.941C>T	General Population	1 in 158	47%	1 in 297
Joubert syndrome 2	TMEM216	X	3	(c.218G>A); c.218G>T; (c.230G>C)	Ashkenazi Jewish	1 in 92	99%	1 in 9101
					General Population	1 in 500	10%	1 in 555
Kohlschutter-Tonz syndrome	ROGDI	X	1	(c.469C>T)	Druze Arab (Yanuh-Jat)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Krabbe disease	GALC	X	6	c.1162-6080_*9569del31Kb; c.1586C>T; (c.1630G>A); c.1700A>C; (c.1796T>G); c.857G>A	Arab Muslim (Jabel Mukaber)	1 in 6	99%	1 in 501
					Arab Muslim (Sur Baher)	1 in 6	99%	1 in 501
					Druze Arab (Daliyat al-Karmel)	1 in 6	99%	1 in 501
					General Population	1 in 158	10%	1 in 175
Laron dwarfism	GHR	No	5	c.11G>A; (c.594A>G); c.686G>A; (c.703C>T); (c.744delT)	General Population	1 in 387	10%	1 in 430
					Iraqi Jewish	N/A	50%	1 in 222
					Yemenite Jewish	N/A	50%	1 in 222
Leber congenital amaurosis 1	GUCY2D	No	1	(c.389delC)	General Population	1 in 500	10%	1 in 555
					Libyan Jewish	N/A	99%	1 in 555
					Moroccan Jewish	N/A	99%	1 in 555
					Tunisian Jewish	N/A	99%	1 in 555
Leber congenital amaurosis 13	RDH12	No	2	c.146C>T; c.184C>T	General Population	1 in 500	10%	1 in 555
Leber congenital amaurosis 2	RPE65	No	2	c.394G>A; c.95-2A>T	Algerian Jewish	1 in 90	99%	1 in 8901
					General Population	1 in 228	10%	1 in 253
					Moroccan Jewish	1 in 90	99%	1 in 8901
					Tunisian Jewish	1 in 90	99%	1 in 8901
Leber congenital amaurosis 4	AIPL1	No	1	c.211G>T	General Population	1 in 500	10%	1 in 555
					Libyan Jewish	N/A	99%	1 in 555
					Moroccan Jewish	N/A	99%	1 in 555
Leber congenital amaurosis 5	LCA5	No	1	c.835C>T	Ashkenazi Jewish	1 in 96	99%	1 in 9501
					General Population	1 in 500	10%	1 in 555
Leigh syndrome, due to COX deficiency	SURF1	No	3	c.312_321delTCTGCCAGCCinsAT; (c.574_575insCTGC); (c.845_846delCT)	General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Leprechaunism	INSR	X	1	(c.167T>C)	Druze Arab (Peki'in/Buqei'a)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Lethal congenital contractural syndrome 2	ERBB3	No	1	(c.1184-9A>G)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Lethal congenital contractural syndrome 3	PIP5K1C	X	1	(c.757G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Lethal congenital contracture syndrome 4	MYBPC1	No	1	(c.952C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Leukodystrophy and acquired microcephaly with or without dystonia	PLEKHG2	No	1	c.610C>T	General Population	1 in 500	10%	1 in 555
Leukodystrophy, hypomyelinating, 3	AIMP1	No	1	(c.292_293delCA)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Leukodystrophy, hypomyelinating, 4	HSPD1	X	1	(c.86A>G)	Bedouin Arab (Kammana Sallama)	N/A	99%	1 in 555
					Bedouin Arab (Ras al-Ein)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Lipoprotein lipase deficiency	LPL	No	1	c.644G>A	General Population	1 in 500	10%	1 in 555
Liver failure, transient infantile	TRMU	No	1	c.229T>C	General Population	1 in 500	10%	1 in 555
					Yemenite Jewish	1 in 40	75%	1 in 157
Lysinuric protein intolerance	SLC7A7	No	2	c.1228C>T; c.726G>A	General Population	1 in 500	10%	1 in 555
Mandibuloacral dysplasia	LMNA	No	1	c.1580G>A	General Population	1 in 500	10%	1 in 555
					Italian	N/A	95%	1 in 501
Maple syrup urine disease, type Ia	BCKDHA	No	3	c.1312T>A; (c.859C>T); c.861_868delAGGCCCCG	Bedouin Arab (Negev)	N/A	99%	1 in 321
					General Population	1 in 289	10%	1 in 321
Maple syrup urine disease, type Ib	BCKDHB	X	4	(c.1016C>T); (c.1114G>T); c.548G>C; c.832G>A	Ashkenazi Jewish	1 in 113	99%	1 in 11201
					Druze Arab (Abu Snan)	N/A	99%	1 in 11201
					Druze Arab (Peki'in/Buqei'a)	N/A	99%	1 in 11201
					General Population	1 in 327	10%	1 in 363
Maple syrup urine disease, type II	DBT	X	3	(c.581C>G); c.827T>G; Exon1-11	Druze Arab (Maghar)	N/A	99%	1 in 534
					General Population	1 in 481	10%	1 in 534
McArdle disease	PYGM	No	4	c.148C>T; c.2392T>C; (c.632delG); (c.808C>T)	General Population	1 in 158	10%	1 in 175
					Kavkazi (Caucasus) Jewish	1 in 84	99%	1 in 8301
					Yemenite Jewish	1 in 84	99%	1 in 8301
Meconium ileus, familial	GUCY2C	No	1	(c.1160A>G)	Bedouin Arab	1 in 81	92%	1 in 1001
					General Population	1 in 500	10%	1 in 555
Medium-chain acyl-CoA dehydrogenase deficiency	ACADM	No	5	c.199T>C; c.250C>T; c.362C>T; c.799G>A; c.997A>G	General Population	1 in 70	67%	1 in 210
Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	X	4	(c.135dupC); c.176G>A; c.274C>T; (c.278C>T)	General Population	1 in 500	16%	1 in 595
					Libyan Jewish	1 in 40	99%	1 in 3901
					Turkish Jewish	1 in 40	99%	1 in 3901
Megaloblastic anemia-1, Norwegian type (Imerslund-Gräsbeck syndrome)	AMN	No	1	c.208-2A>G	General Population	1 in 500	37%	1 in 793
					Tunisian Jewish	1 in 20	99%	1 in 1901
Mental retardation; autosomal recessive 15	MAN1B1	X	1	(c.1863G>A)	Arab Muslim (Jabel Mukaber)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Mental retardation; autosomal recessive 3	CC2D1A	X	15	Exon1; Exon12; Exon13-14; Exon14; Exon15-17; Exon18-20; Exon2; Exon21-22; Exon23-24; Exon25-26; Exon27-29; Exon3-4; Exon5-6; Exon7; Exon8-11	Arab Muslim (Jisr az-Zarqa)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Metachromatic leukodystrophy	ARSA	X	7	c.1136C>T; (c.1283C>T); (c.292_293delTCinsCT); c.465+1G>A; (c.576G>C); c.763G>A; c.827C>T	General Population	1 in 100	10%	1 in 111
					Yemenite Jewish	1 in 75	99%	1 in 7401
Microcephaly 9, primary, autosomal recessive	CEP152	X	1	(c.2281-2A>G)	Arab Muslim (Jisr az-Zarqa)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Microcephaly, postnatal progressive, with seizures and brain atrophy	MED17	X	1	c.1112T>C	Bukharian Jewish	1 in 20	99%	1 in 1876
					General Population	1 in 500	10%	1 in 555
					Kavkazi (Caucasus) Jewish	1 in 20	99%	1 in 1876
Minicore myopathy with external ophthalmoplegia	RYR1	X	2	(c.3263A>G); (c.9623C>T)	Druze Arab (Julis)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Mitochondrial complex I deficiency (NDUFA11-related)	NDUFA11	No	1	c.97+5G>A	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Mitochondrial complex I deficiency (NDUFAF5-related)	NDUFAF5	No	1	c.749G>T	Ashkenazi Jewish	1 in 290	99%	1 in 28901
					General Population	1 in 500	10%	1 in 555
Mitochondrial complex I deficiency (NDUFS4-related)	NDUFS4	No	1	c.462delA	Ashkenazi Jewish	1 in 1000	99%	1 in 99901
					General Population	1 in 500	10%	1 in 555
					General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Mitochondrial complex I deficiency (NDUFS6-related)	NDUFS6	X	1	(c.344G>A)	Kavkazi (Caucasus) Jewish	1 in 24	99%	1 in 2300
Mitochondrial complex I deficiency, nuclear type 20	ACAD9	No	1	c.976G>C	General Population	1 in 500	10%	1 in 555
Mitochondrial complex III deficiency, nuclear type 4	UQCRQ	X	1	(c.134C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Mitochondrial DNA depletion syndrome 1 (MNGIE type)	TYMP	No	1	c.433G>A	General Population	1 in 500	10%	1 in 555
					Iranian Jewish	N/A	99%	1 in 555
Mitochondrial DNA depletion syndrome 2 (myopathic type)	TK2	X	1	(c.635T>A)	Arab Muslim (Maghar)	1 in 35	99%	1 in 3401
					General Population	1 in 500	10%	1 in 555
Mitochondrial DNA depletion syndrome 3 (hepatocerebral type)	DGUOK	X	1	(c.255delA)	Druze Arab (Sajur)	1 in 5	99%	1 in 401
					General Population	1 in 500	10%	1 in 555
Mitochondrial DNA depletion syndrome 4A (Alpers type)/Mitochondrial DNA depletion syndrome 4B (MNGIE type)/Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE)	POLG	No	3	c.1399G>A; c.2243G>C; c.2542G>A	General Population	1 in 500	10%	1 in 555
Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	SUCLA2	No	1	(c.789del43insATAAA)	Arab Muslim	1 in 61	99%	1 in 6001
					General Population	1 in 500	10%	1 in 555
Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	MPV17	No	1	c.149G>A	General Population	1 in 500	10%	1 in 555
Mitochondrial myopathy and sideroblastic anemia 1	PUS1	No	1	(c.430C>T)	General Population	1 in 500	10%	1 in 555
					Iranian Jewish	N/A	99%	1 in 555
Mitochondrial myopathy, episodic, with optic atrophy and reversible leukoencephalopathy	FDX1L	No	1	c.10A>T	General Population	1 in 500	10%	1 in 555
Molybdenum cofactor deficiency A	MOCS1	X	2	(c.722delT); (c.971G>A)	Arab Muslim (Bu'eine Nujeidat)	1 in 11	95%	1 in 201
					General Population	1 in 223	10%	1 in 248
Mucolipidosis II alpha/beta/Mucolipidosis III alpha/beta	GNPTAB	No	3	c.3335+6T>G; c.3503_3504delTC; c.3565C>T	General Population	1 in 500	10%	1 in 555
Mucolipidosis III gamma	GNPTG	X	1	(c.499dupC)	Arab Muslim	N/A	67%	1 in 555
					Druze Arab (Beit Jann)	1 in 61	91%	1 in 668
					General Population	1 in 500	10%	1 in 555
					Tunisian Jewish	N/A	99%	1 in 555
Mucolipidosis IV	MCOLN1	X	5	c.1-874_c.788del6433bp; c.302_303delTC; c.304C>T; c.406-2A>G; g.4127_10560del6434	Ashkenazi Jewish	1 in 112	99%	1 in 11101
					General Population	1 in 500	10%	1 in 555
Mucopolysaccharidisis type IIIA (Sanfilippo A)	SGSH	No	7	c.1080delC; c.1093C>T; (c.1298G>A); c.197C>G; (c.544C>T); c.734G>A; (c.812C>T)	General Population	1 in 415	10%	1 in 461
Mucopolysaccharidosis Ih	IDUA	X	4	(c.1096A>C); (c.1205G>A); (c.1598C>G); c.208C>T	Druze Arab (Maghar)	1 in 327	60%	1 in 79
					General Population	1 in 144	67%	1 in 434
Mucopolysaccharidosis type IIIB (Sanfilippo B)	NAGLU	No	2	c.1558C>T; c.419A>G	General Population	1 in 500	10%	1 in 555
Mucopolysaccharidosis type IIIC (Sanfilippo C)/Retinitis pigmentosa 73	HGSNAT	No	2	c.1030C>T; c.1150C>T	General Population	1 in 500	10%	1 in 555
Mucopolysaccharidosis type IIID	GNS	No	1	c.1019A>G	General Population	1 in 500	10%	1 in 555
Multiple congenital anomalies-hypotonia-seizures syndrome 1	PIGN	No	1	c.2126G>A	Arab Muslim	N/A	88%	1 in 555
					General Population	1 in 500	10%	1 in 555
Multiple sulfatase deficiency	SUMF1	No	1	c.463T>C	Ashkenazi Jewish	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Muscular dystrophy, limb-girdle, autosomal recessive 1	CAPN3	No	2	c.1469G>A; c.550delA	General Population	1 in 500	10%	1 in 555
Muscular dystrophy, limb-girdle, autosomal recessive 3	SGCA	No	1	c.229C>T	General Population	1 in 500	10%	1 in 555
Muscular dystrophy, limb-girdle, type 2B	DYSF	X	6	(c.2372C>G); c.2779delG; c.4872_4876delGCCCGinsCCCC; (c.5038_5057+3insCTCCCACAGACCTACTGTGTGTA); (c.5057+5G>A); (c.5429G>A)	General Population	1 in 311	10%	1 in 345
					Kavkazi (Caucasus) Jewish	1 in 25	99%	1 in 2401
					Yemenite Jewish	1 in 25	99%	1 in 2401
Muscular dystrophy, limb-girdle, type 2C	SGCG	X	1	(c.525delT)	Bedouin Arab (Shibli-Umm al-Ghanam)	1 in 32	99%	1 in 3101
					General Population	1 in 439	10%	1 in 488
Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4	FKTN	X	1	c.1167dupA	Ashkenazi Jewish	1 in 150	99%	1 in 14901
					General Population	1 in 500	10%	1 in 555
Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	FKRP	X	4	c.1073C>T; (c.160C>G); (c.160C>T); c.826C>A	Bedouin Arab (Tuba-Zangariyye)	N/A	99%	1 in 455
					General Population	1 in 410	10%	1 in 455
Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	RAPSN	No	4	c.-210A>G; c.264C>A; (c.-27C>G); c.41T>C	General Population	1 in 252	10%	1 in 280
					Iranian Jewish	1 in 100	99%	1 in 9901
					Iraqi Jewish	1 in 100	99%	1 in 9901
					Yemenite Jewish	1 in 100	99%	1 in 9901
Myasthenic syndrome, congenital, 23, presynaptic/Combined D-2- and L-2-hydroxyglutaric aciduria	SLC25A1	No	1	c.845G>A	General Population	1 in 500	10%	1 in 555
Myotonia congenita; recessive	CLCN1	No	2	(c.1444G>A); (c.1586C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Nemaline myopathy 2, autosomal recessive	NEB	X	1	c.7432-2025_7536+372del2502	Ashkenazi Jewish	1 in 108	99%	1 in 10701
					General Population	1 in 224	10%	1 in 249
Nephronophthisis 2, infantile	INVS	No	1	(c.2719C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Nephronophthisis 2, infantile	INVS	No	1	(c.2713C>T)	General Population	1 in 500	10%	1 in 555
Nephrotic syndrome, type 1	NPHS1	X	7	c.1138C>T; (c.121_122delCT); c.1358A>G; (c.1481delC); (c.1707C>G); (c.2160dupC); (c.3325C>T)	Arab Muslim (Abu Ghosh)	N/A	99%	1 in 361
					Finnish	1 in 45	81%	1 in 233
					General Population	1 in 325	10%	1 in 361
Nephrotic syndrome, type 2	NPHS2	No	2	c.412C>T; c.413G>A	Arab Muslim (Several regions)	N/A	56%	1 in 222
					General Population	1 in 500	10%	1 in 555
Neuronopathy, distal hereditary motor, type VI	IGHMBP2	X	2	c.114delA; (c.707T>G)	Arab Muslim (Bi'ina)	N/A	99%	1 in 555
					Arab Muslim (Deir al-Asad)	N/A	99%	1 in 555
					Arab Muslim (Jisr az-Zarqa)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Neutropenia, severe congenital 4, autosomal recessive	G6PC3	No	1	(c.785G>A)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Niemann-Pick disease, type C1	NPC1	No	6	(c.1211G>A); c.2932C>T; c.3019C>G; c.3182T>C; c.3467A>G; c.3557G>A	Bedouin Arab (Negev)	N/A	99%	1 in 222
					General Population	1 in 200	10%	1 in 222
Niemann-pick disease, type C2	NPC2	No	1	c.115G>A	General Population	1 in 500	10%	1 in 555
Niemann-Pick disease, types A/B	SMPD1	X	8	c.1172A>C; c.1426C>T; c.1493G>A; c.1493G>T; c.1828_1830delCGC; (c.573delT); c.911T>C; c.996delC	Arab Muslim (Galilee)	N/A	99%	1 in 278
					Arab Muslim (Triangle)	N/A	99%	1 in 278
					Ashkenazi Jewish	1 in 115	97%	1 in 3801
					Caucasian	1 in 250	20%	1 in 312
					General Population	1 in 250	10%	1 in 278
					North African	1 in 250	87%	1 in 1916
Nijmegen breakage syndrome	NBN	No	1	c.657_661delACAAA	General Population	1 in 500	10%	1 in 555
Odontoonychodermal dysplasia/Schopf-Schulz-Passarge syndrome	WNT10A	No	1	c.321C>A	General Population	1 in 500	10%	1 in 555
Omenn syndrome / T- B- severe combined immunodeficiency	RAG1	X	1	(c.1361T>A)	General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	N/A	88%	1 in 555
	RAG2	X	4	(c.104G>T); (c.193G>T); (c.379A>T); c.685C>T	General Population	1 in 500	10%	1 in 555
Omenn syndrome/Severe combined immunodeficiency, Athabascan type	DCLRE1C	No	1	c.241C>T	General Population	1 in 500	10%	1 in 555
Ornithine transcarbamylase deficiency	OTC	No	3	c.275G>A; c.533C>T; c.829C>T	General Population	1 in 500	10%	1 in 555
Osteogenesis imperfecta, type xiv	TMEM38B	X	1	c.454+279_543-5092delinsAATTAAGGTATA	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Osteopetrosis, autosomal recessive 1	TCIRG1	X	2	c.117+4A>T; (c.1331G>T)	Ashkenazi Jewish	1 in 354	99%	1 in 35301
					General Population	1 in 354	10%	1 in 393
Osteopetrosis, autosomal recessive 8	SNX10	No	1	(c.152G>A)	Bedouin Arab	1 in 211	99%	1 in 21001
					General Population	1 in 250	10%	1 in 278
Otospondylomegaepiphyseal dysplasia	COL11A2	No	1	(c.3991C>T)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Pendred syndrome	SLC26A4	X	11	(c.1001G>T); (c.1151A>G); (c.1198delT); c.1246A>C; (c.1341+1delG); (c.2000T>G); c.2168A>G; c.349C>T; c.707T>C; (c.716T>A); c.919-2A>G	Asian	1 in 74	53%	1 in 156
					Bedouin Arab (Galilee)	N/A	99%	1 in 256
					Caucasian	1 in 88	69%	1 in 282
					General Population	1 in 80	69%	1 in 256
Peroxisome biogenesis disorder 1A (Zellweger)	PEX1	No	4	c.2097dupT; c.2528G>A; c.475G>A; c.475G>C	Caucasian	1 in 147	68%	1 in 457
					General Population	1 in 500	68%	1 in 1560
Peroxisome biogenesis disorder 4A (Zellweger)	PEX6	No	1	(c.1715C>T)	General Population	1 in 500	10%	1 in 555
					Yemenite Jewish	N/A	99%	1 in 555
Peroxisome biogenesis disorder 5A (Zellweger)	PEX2	No	2	c.355C>T; c.550delC	Ashkenazi Jewish	1 in 123	99%	1 in 12201
					General Population	1 in 158	23%	1 in 205
					Karaite Jewish	1 in 111	99%	1 in 11001
Peroxisome biogenesis disorder 9B/Rhizomelic chondrodysplasia punctata, type 1	PEX7	No	2	c.653C>T; c.875T>A	General Population	1 in 500	10%	1 in 555
Phenylalanine hydroxylase deficiency (including phenylketonuria)	PAH	X	25	c.1042C>G; (c.1045T>C); c.1066-11G>A; c.117C>G; c.1208C>T; c.1222C>T; c.1223G>A; c.1241A>G; c.1315+1G>A; c.143T>C; c.165delT; (c.165T>G); c.194T>C; (c.441+5G>T); c.473G>A; c.533A>G; c.689T>C; c.722G>A; c.727C>T; (c.754C>T); c.755G>A; c.782G>A; c.842C>T; c.896T>G; c.898G>T	Ashkenazi Jewish	1 in 225	33%	1 in 337
					Bukharian Jewish	1 in 18	74%	1 in 66
					General Population	1 in 65	43%	1 in 113
					Iranian Jewish	1 in 18	74%	1 in 66
					Iraqi Jewish	1 in 18	74%	1 in 66
					Kavkazi (Caucasus) Jewish	1 in 18	74%	1 in 66
					Moroccan Jewish	1 in 18	74%	1 in 66
					Tunisian Jewish	1 in 18	74%	1 in 66
Polycystic kidney disease, autosomal recessive	PKHD1	No	5	c.107C>T; c.1342G>C; c.3761_3762delCCinsG; c.4870C>T; c.664A>G	Ashkenazi Jewish	1 in 182	99%	1 in 18101
					General Population	1 in 500	10%	1 in 555
Polymicrogyria, bilateral frontoparietal	GPR56	No	4	(c.1046G>C); (c.1167+3G>C); c.1693C>T; c.739_745delCAGGACC	General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Pontocerebellar hypoplasia, type 1A	VRK1	No	1	c.1072C>T	Ashkenazi Jewish	1 in 225	99%	1 in 22401
					General Population	1 in 500	10%	1 in 555
Pontocerebellar hypoplasia, type 2D (Progressive cerebello-cerebral atrophy, type 2D)	SEPSECS	X	2	c.1001A>G; c.715G>A	General Population	1 in 500	10%	1 in 555
					Iraqi Jewish	1 in 42	99%	1 in 4101
					Moroccan Jewish	1 in 42	99%	1 in 4101
Pontocerebellar hypoplasia, type 2E	VPS53	X	2	c.1556+5G>A; c.2084A>G	General Population	1 in 500	10%	1 in 555
					Moroccan Jewish	1 in 37	99%	1 in 3851
Primary ciliary dyskinesia-12 (RSPH9-related)	RSPH9	No	1	(c.804_806delGAA)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 86	10%	1 in 95
Primary ciliary dyskinesia-9 (DNAI2-related)	DNAI2	No	2	c.1304G>A; c.1494+1G>A	Ashkenazi Jewish	1 in 200	99%	1 in 19901
					General Population	1 in 500	10%	1 in 555
Prolidase deficiency	PEPD	X	3	(c.1103T>G); c.605C>T; (c.634G>C)	Druze Arab (Yarka)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Propionicacidemia	PCCA	No	3	c.1676G>T; c.184-618_300+3930del; (c.1850T>C)	General Population	1 in 137	10%	1 in 152
	PCCB	No	2	c.1218_1231delGGGCATCATCCGGCinsTAGAGCACAGGA; c.553C>T	General Population	1 in 500	10%	1 in 555
Proximal myopathy and ophthalmoplegia	MYH2	No	1	(c.2400delG)	Arab Muslim (East Jerusalem)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Pycnodysostosis	CTSK	X	1	(c.990A>G)	Arab Muslim (Bu'eine Nujeidat)	N/A	99%	1 in 304
					General Population	1 in 274	10%	1 in 304
Pyridoxamine 5'-phosphate oxidase deficiency	PNPO	No	1	(c.284G>A)	General Population	1 in 500	10%	1 in 555
Pyruvate carboxylase deficiency	PC	No	2	c.1892G>A; c.2540C>T	General Population	1 in 500	10%	1 in 555
Renal tubular acidosis; proximal; with ocular abnormalities	SLC4A4	X	1	(c.2321G>A)	Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Retinitis pigmentosa 12	CRB1	X	2	c.2843G>A; (c.4121_4130delCAACTCAGGG)	Arab Muslim (Mashhad)	N/A	99%	1 in 304
					General Population	1 in 274	10%	1 in 304
					Hispanic	1 in 107	10%	1 in 119
Retinitis pigmentosa 14	TULP1	No	1	(c.1495+2dupT)	General Population	1 in 274	10%	1 in 304
Retinitis pigmentosa 25	EYS	No	3	(c.1211dupA); (c.3715G>T); c.8155_8156delCA	General Population	1 in 274	10%	1 in 304
					Iraqi Jewish	1 in 94	10%	1 in 104
					Moroccan Jewish	1 in 94	10%	1 in 104
Retinitis pigmentosa 26	CERKL	No	2	c.238+1G>A; c.769C>T	General Population	1 in 274	10%	1 in 304
					Yemenite Jewish	1 in 22	33%	1 in 33
Retinitis pigmentosa 28	FAM161A	No	4	c.1309A>T; c.1355_1356delCA; c.1567C>T; (c.1618C>T)	Bulgarian Jewish	1 in 32	86%	1 in 220
					General Population	1 in 274	10%	1 in 304
					Libyan Jewish	1 in 32	86%	1 in 220
					Moroccan Jewish	1 in 32	86%	1 in 220
					Syrian Jewish	1 in 32	86%	1 in 220
					Tunisian Jewish	1 in 32	86%	1 in 220
Retinitis pigmentosa 36	PRCD	X	1	c.64C>T	General Population	1 in 274	10%	1 in 304
Retinitis pigmentosa 57	PDE6G	X	1	(c.187+1G>T)	General Population	1 in 274	10%	1 in 304
Retinitis pigmentosa 59	DHDDS	No	1	c.124A>G	Ashkenazi Jewish	1 in 118	99%	1 in 11800
					General Population	1 in 274	10%	1 in 304
Retinitis pigmentosa 64	C8ORF37	X	3	(c.497T>A); (c.529C>T); (c.545A>G)	General Population	1 in 274	10%	1 in 304
Rickets, vitamin D-resistant, type IIA	VDR	X	1	(c.885C>A)	General Population	1 in 500	10%	1 in 555
Roberts syndrome/SC phocomelia syndrome	ESCO2	No	1	c.1111dupA	General Population	1 in 500	10%	1 in 555
Sandhoff disease, infantile, juvenile, and adult forms	HEXB	X	4	(c.1082+5G>A); c.171delG; c.76delA; Exon1-5	Arab Christian (Maghar)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Schimke immunoosseous dysplasia	SMARCAL1	No	1	c.836T>C	General Population	1 in 500	10%	1 in 555
Severe combined immunodeficiency due to ADA deficiency	ADA	No	11	c.301C>T; c.302G>A; c.302G>T; c.320T>C; c.631C>T; c.632G>A; (c.703C>T); (c.792G>A); c.821C>T; c.872C>T; c.986C>T	General Population	1 in 500	10%	1 in 555
Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis	POC1A	X	1	(c.398T>C)	General Population	1 in 500	10%	1 in 555
Sialic acid storage disorder, infantile	SLC17A5	X	1	(c.983G>A)	General Population	1 in 500	47%	1 in 943
Sjogren-Larsson syndrome	ALDH3A2	No	2	c.1297_1298delGA; c.943C>T	General Population	1 in 500	10%	1 in 555
Smith-Lemli-Opitz syndrome	DHCR7	X	14	(c.1054C>T); c.1139G>A; c.1210C>T; c.1228G>A; c.278C>T; c.452G>A; (c.453G>A); c.470T>C; c.724C>T; c.725G>A; (c.755A>G); c.906C>G; c.964-1G>C; c.976G>T	Arab Muslim (Kfar Kanna)	N/A	99%	1 in 98
					Ashkenazi Jewish	1 in 36	75%	1 in 141
					General Population	1 in 71	50%	1 in 564
Spastic paraparesis 49, autosomal recessive	TECPR2	X	1	c.3416delT	Bukharian Jewish	1 in 38	99%	1 in 3701
					General Population	1 in 500	10%	1 in 555
Spastic paraplegia 53, autosomal recessive	VPS37A	X	1	(c.1146A>T)	Arab Muslim (Nachef)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Spinal Muscular Atrophy	SMN1	X	4	c.835G>T; Exon7; Exon7-8; g.27134T>G	African American	1 in 66	71%	1 in 225
					Arab Christian	1 in 29	90%	1 in 281
					Arab Muslim	1 in 73	90%	1 in 721
					Ashkenazi Jewish	1 in 41	94%	1 in 668
					Asian	1 in 53	93%	1 in 744
					Caucasian	1 in 35	95%	1 in 681
					Druze	1 in 47	90%	1 in 461

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
					General population	1 in 50	90%	1 in 491
Spondylometaepiphyseal dysplasia, short limb-hand type	DDR2	No	1	(c.2254C>T)	General Population	1 in 500	10%	1 in 555
Stargardt disease 1 including Cone-rod dystrophy 3	ABCA4	X	10	c.1648G>A; c.2588G>C; c.3113C>T; c.3607G>A; c.3608G>A; (c.4254-15delCCTGCTCTGTCCCAGTCACATGT); c.4539+1G>T; c.5018+2T>C; (c.5460+1G>A); (c.834delT)	Arab Christian (Deir Hanna)	N/A	99%	1 in 57
					General Population	1 in 51	10%	1 in 57
Striatonigral degeneration, infantile	NUP62	X	1	(c.1172A>C)	Bedouin Arab (Kafr Qasim)	N/A	99%	1 in 555
					Bedouin Arab (Negev)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome	LIFR	X	2	(c.1601-1G>A); (c.2472_2476delTATGT)	Arab Muslim (Jabel Mukaber)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Surfactant metabolism dysfunction, pulmonary, 3	ABCA3	No	1	c.316C>T	General Population	1 in 500	10%	1 in 555
Tay-Sachs disease	HEXA	X	23	c.1073+1G>A; c.1274_1277dupTATC; (c.1351C>G); c.1421+1G>C; (c.1444G>A); c.1496G>A; c.1510C>T; c.-2564_253+5128del7945insG; (c.459+2dupT); (c.496delC); (c.508C>T); c.509G>A; (c.532C>T); c.533G>A; c.533G>T; (c.540C>G); (c.571-2A>G); (c.749G>A); c.749G>T; (c.805+1G>A); c.805G>A; (c.835T>C); c.915_917delCTT	Ashkenazi Jewish	1 in 27	98%	1 in 1301
					Bedouin Arab (Negev)	N/A	99%	1 in 701
					Druze Arab (Abu Snan)	N/A	99%	1 in 701
					Druze Arab (Peki'in/Buqei'a)	N/A	99%	1 in 701
					French Canadian/Cajun	1 in 53	82%	1 in 290
					General Population	1 in 288	59%	1 in 701
					Iraqi Jewish	1 in 125	82%	1 in 695
					Moroccan Jewish	1 in 125	82%	1 in 695
Thiamine-responsive megaloblastic anemia syndrome	SLC19A2	No	1	(c.725delC)	General Population	1 in 500	10%	1 in 555
Thyroid dyshormonogenesis 5	DUOXA2	No	1	c.205+2T>C	General Population	1 in 500	10%	1 in 555
Trichohepatoenteric syndrome 1	TTC37	No	1	c.2808G>A	General Population	1 in 500	10%	1 in 555
Tumoral calcinosis, familial, hyperphosphatemic	GALNT3	No	1	(c.1524+5G>A)	General Population	1 in 500	10%	1 in 555
Tumoral calcinosis, familial, normophosphatemic	SAMD9	No	2	c.1030C>T; (c.4483A>G)	General Population	1 in 500	10%	1 in 555
					Yemenite Jewish	1 in 25	99%	1 in 2401
Tyrosinemia, type I	FAH	No	8	c.1009G>A; c.1062+5G>A; c.1069G>T; c.192G>T; (c.554-1G>T); (c.707-1G>C); c.782C>T; (c.786G>A)	Ashkenazi Jewish	1 in 150	99%	1 in 14901
					French Canadian	1 in 66	88%	1 in 543
					French Canadian (Saguenay-Lac Saint-Jean region)	1 in 22	88%	1 in 176
					General Population	1 in 158	26%	1 in 213
					Norwegian	1 in 123	38%	1 in 198
Tyrosinemia, type III	HPD	X	1	(c.415-1G>A)	Druze Arab (Julis)	N/A	99%	1 in 701
					General Population	1 in 500	10%	1 in 701
Usher syndrome, type 1B	MYO7A	No	9	(c.1190C>A); (c.1996C>T); c.2187+1G>A; (c.2476G>A); c.470+1G>A; (c.5581C>T); (c.6196delC); c.640G>A; c.93C>A	Algerian Jewish	1 in 50	99%	1 in 4901
					General Population	1 in 143	10%	1 in 159
					Moroccan Jewish	1 in 50	99%	1 in 4901
Usher syndrome, type 1C	USH1C	No	2	(c.1220delG); c.216G>A	General Population	1 in 141	10%	1 in 157
					Yemenite Jewish	1 in 119	99%	1 in 11801
Usher syndrome, type 1F	PCDH15	X	2	c.3717+1G>A; c.733C>T	Ashkenazi Jewish	1 in 147	75%	1 in 585
					General Population	1 in 237	33%	1 in 353
Usher syndrome, type 2A	USH2A	X	8	(c.1000C>T); c.12067-2A>G; (c.2209C>T); c.2276G>T; c.2299delG; c.236_239dupGTAC; c.4544C>T; (c.5519G>T)	Algerian Jewish	1 in 139	61%	1 in 357
					Bukharian Jewish	1 in 139	61%	1 in 357
					General Population	1 in 126	28%	1 in 175
					Iranian Jewish	1 in 139	61%	1 in 357
					Iraqi Jewish	1 in 139	61%	1 in 357
					Libyan Jewish	1 in 139	61%	1 in 357
					Moroccan Jewish	1 in 139	61%	1 in 357
					Syrian Jewish	1 in 139	61%	1 in 357
					Tunisian Jewish	1 in 139	61%	1 in 357
					Yemenite Jewish	1 in 139	61%	1 in 357
Usher syndrome, type 3A	CLRN1	X	2	c.144T>G; c.528T>G	Ashkenazi Jewish	1 in 120	95%	1 in 2381
					General Population	1 in 500	10%	1 in 555
Ventricular tachycardia, catecholaminergic polymorphic, 2	CASQ2	X	1	(c.919G>C)	Bedouin Arab (Kammana Sallama)	N/A	99%	1 in 555
					Bedouin Arab (Ras al-Ein)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	No	10	(c.1096C>T); c.1226C>T; c.1405C>T; c.1679-6G>A; c.388_390delGAG; (c.637G>A); (c.65C>A); (c.779C>T); c.799_802delGTTA; c.848T>C	Bedouin Arab (Negev)	N/A	99%	1 in 97
					General Population	1 in 87	10%	1 in 97
Vitamin D-dependent rickets, type I	CYP27B1	No	2	c.1166G>A; c.262delG	General Population	1 in 500	10%	1 in 555
Werner syndrome	WRN	No	2	c.1105C>T; c.3590delA	General Population	1 in 500	10%	1 in 555

Disease Name	Gene	IMOH/ ISMG	# of Variations	List of Genetic Variations	Ethnicity	Carrier	Detection Rate	Residual Risk
Wilson disease	ATP7B	X	15	(c.1340_1343delAAAC); (c.1544G>A); (c.1639delC); c.1934T>G; (c.2293G>A); c.2333G>A; c.2333G>T; (c.2337G>A); c.2906G>A; c.2972C>T; c.3191A>C; c.3207C>A; c.3402delC; (c.3649_3654delGTTCTG); c.845delT	Algerian Jewish	1 in 65	99%	1 in 6425
					Ashkenazi Jewish	1 in 100	88%	1 in 826
					Asian	1 in 50	57%	1 in 115
					Bukharian Jewish	1 in 65	99%	1 in 6425
					General Population	1 in 90	50%	1 in 179
					Iranian Jewish	1 in 65	99%	1 in 6425
					Iraqi Jewish	1 in 65	99%	1 in 6425
					Libyan Jewish	1 in 65	99%	1 in 6425
					Moroccan Jewish	1 in 65	99%	1 in 6425
					Tunisian Jewish	1 in 65	99%	1 in 6425
					Yemenite Jewish	1 in 65	99%	1 in 6425
Wiskott-Aldrich syndrome	WAS	No	1	(c.119G>T)	General Population	1 in 500	10%	1 in 555
Wolman disease	LIPA	X	4	c.1024G>A; c.260G>T; c.398delC; c.419G>A	Arab Muslim (Bi'ina)	N/A	99%	1 in 329
					Arab Muslim (Deir al-Asad)	N/A	99%	1 in 329
					General Population	1 in 296	10%	1 in 329
					Iranian Jewish	1 in 32	99%	1 in 3140
Woodhouse-Sakati syndrome	DCAF17	No	1	(c.436delC)	Bedouin Arab	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Xeroderma pigmentosum, group C	XPC	No	1	c.566_567delAT	General Population	1 in 500	10%	1 in 555
Xeroderma pigmentosum, group G/Cockayne syndrome	ERCC5	X	1	(c.205C>T)	Arab Muslim (Fureidis)	N/A	99%	1 in 555
					General Population	1 in 500	10%	1 in 555
Xeroderma pigmentosum, variant type	POLH	No	1	c.522G>T	General Population	1 in 500	10%	1 in 555
Grand Total	**363**		**1696**					