Supplementary Table A: All Non-Mild Disease Statistics.

Disease (OMIM Number)	Total Patients Screened		Carrier Frequency				
						Wilson 95% CI	
	n	%[a]	n	%	1 in __	Lower %	Upper %
TOTAL	23453	100.0%	5633	24.0%	4.2	24.0%	24.0%
alpha-1 antitrypsin deficiency (613490)	15484	66.0%	1178	7.6%	13.1	7.2%	8.0%
cystic fibrosis (602421)	23369	99.6%	842	3.6%	27.8	3.4%	3.8%
GJB2-related DFNB1 nonsyndromic hearing loss and deafness (220290)	15799	67.4%	371	2.3%	42.6	2.1%	2.6%
spinal muscular atrophy (253300)	23127	98.6%	405	1.8%	57.1	1.6%	1.9%
familial Mediterranean fever (249100)	15854	67.6%	247	1.6%	64.2	1.4%	1.8%
Smith-Lemli-Opitz syndrome (270400)	15825	67.5%	232	1.5%	68.2	1.3%	1.7%
sickle cell disease (603903)	21360	91.1%	307	1.4%	69.6	1.3%	1.6%
Gaucher disease (230800)	21473	91.6%	280	1.3%	76.7	1.2%	1.5%
factor XI deficiency (612416)	15724	67.0%	171	1.1%	92.0	0.9%	1.3%
achromatopsia (262300)	15798	67.4%	162	1.0%	97.5	0.9%	1.2%
hereditary thymine-uraciluria (274270)	15719	67.0%	146	0.9%	107.7	0.8%	1.1%
medium chain acyl-CoA dehydrogenase deficiency (607008)	15831	67.5%	140	0.9%	113.1	0.7%	1.0%
hereditary fructose intolerance (229600)	15814	67.4%	130	0.8%	121.6	0.7%	1.0%
congenital disorder of glycosylation type 1a (212065)	15812	67.4%	128	0.8%	123.5	0.7%	1.0%
Pompe disease (232300)	15462	65.9%	117	0.8%	132.2	0.6%	0.9%
beta-thalassemia (613985)	21096	90.0%	158	0.7%	133.5	0.6%	0.9%
phenylalanine hydroxylase deficiency (261600)	15852	67.6%	112	0.7%	141.5	0.6%	0.8%
hexosaminidase A deficiency (272800)	21958	93.6%	141	0.6%	155.7	0.5%	0.8%
Tay-Sachs disease (272800)	21937	93.5%	124	0.6%	176.9	0.5%	0.7%
carnitine palmitoyltransferase II deficiency (600650)	15812	67.4%	87	0.6%	181.7	0.4%	0.7%
short chain acyl-CoA dehydrogenase deficiency (201470)	15810	67.4%	69	0.4%	229.1	0.3%	0.6%
glycogen storage disease type V (232600)	15796	67.4%	65	0.4%	243.0	0.3%	0.5%
fragile X syndrome (300624)	3885	16.6%	15	0.4%	259.0	0.2%	0.6%
glycogen storage disease type Ia (232200)	17986	76.7%	69	0.4%	260.7	0.3%	0.5%
familial dysautonomia (223900)	21592	92.1%	77	0.4%	280.4	0.3%	0.4%
Bardet-Biedl syndrome, BBS1-related (209900)	15727	67.1%	56	0.4%	280.8	0.3%	0.5%
Wilson disease (277900)	15830	67.5%	56	0.4%	282.7	0.3%	0.5%
galactosemia (230400)	15813	67.4%	55	0.3%	287.5	0.3%	0.5%
fumarase deficiency (606812)	3034	12.9%	10	0.3%	303.4	0.2%	0.6%
Canavan disease (271900)	21581	92.0%	71	0.3%	304.0	0.3%	0.4%
sulfate transporter-related osteochondrodysplasia (606718)	15813	67.4%	48	0.3%	329.4	0.2%	0.4%
MYH-associated polyposis (608456)	3034	12.9%	9	0.3%	337.1	0.2%	0.6%
biotinidase deficiency (253260)	15811	67.4%	38	0.2%	416.1	0.2%	0.3%
maple syrup urine disease type 1B (248600)	17367	74.1%	40	0.2%	434.2	0.2%	0.3%
CLN3-related neuronal ceroid lipofuscinosis (204200)	12747	54.4%	28	0.2%	455.3	0.2%	0.3%
Pendred syndrome (274600)	15810	67.4%	34	0.2%	465.0	0.2%	0.3%
tyrosinemia type 1 (276700)	15814	67.4%	34	0.2%	465.1	0.2%	0.3%
Fanconi anemia type C (227645)	21476	91.6%	44	0.2%	488.1	0.2%	0.3%
Long chain 3-hydroxyacyl-CoA	15810	67.4%	31	0.2%	510.0	0.1%	0.3%

dehydrogenase deficiency (609016)							
metachromatic leukodystrophy (250100)	15810	67.4%	30	0.2%	527.0	0.1%	0.3%
Niemann-Pick disease, SMPD1-associated (607608)	17773	75.8%	32	0.2%	555.4	0.1%	0.3%
very long chain acyl-CoA dehydrogenase deficiency (201475)	12665	54.0%	22	0.2%	575.7	0.1%	0.3%
PEX1-related Zellweger syndrome spectrum (602136)	12777	54.5%	22	0.2%	580.8	0.1%	0.3%
Krabbe disease (245200)	15810	67.4%	27	0.2%	585.6	0.1%	0.2%
mucolipidosis IV (252600)	21476	91.6%	36	0.2%	596.6	0.1%	0.2%
cystinosis (219800)	15797	67.4%	26	0.2%	607.6	0.1%	0.2%
Bloom syndrome (210900)	21466	91.5%	35	0.2%	613.3	0.1%	0.2%
ABCC8-related hyperinsulinism (256450)	17176	73.2%	28	0.2%	613.4	0.1%	0.2%
Niemann Pick disease type A (257200)	3703	15.8%	6	0.2%	617.2	0.1%	0.4%
alkaptonuria (203500)	15797	67.4%	25	0.2%	631.9	0.1%	0.2%
TPP1-related neuronal ceroid lipofuscinosis (204500)	15810	67.4%	25	0.2%	632.4	0.1%	0.2%
Usher syndrome type 3 (276902)	17777	75.8%	27	0.2%	658.4	0.1%	0.2%
Usher syndrome type 1F (602083)	17778	75.8%	27	0.2%	658.4	0.1%	0.2%
lipoamide dehydrogenase deficiency (248600)	17184	73.3%	25	0.1%	687.4	0.1%	0.2%
rhizomelic chondrodysplasia punctata type 1 (215100)	15811	67.4%	23	0.1%	687.4	0.1%	0.2%
Joubert syndrome (608091)	14186	60.5%	20	0.1%	709.3	0.1%	0.2%
Hurler syndrome (607014)	15810	67.4%	21	0.1%	752.9	0.1%	0.2%
primary hyperoxaluria type 1 (259900)	15714	67.0%	20	0.1%	785.7	0.1%	0.2%
autosomal recessive polycystic kidney disease (263200)	16644	71.0%	21	0.1%	792.6	0.1%	0.2%
isovaleric academia (243500)	15810	67.4%	19	0.1%	832.1	0.1%	0.2%
hypophosphatasia, autosomal recessive (241500)	15810	67.4%	18	0.1%	878.3	0.1%	0.2%
Bardet-Biedl syndrome, BBS10-related (209900)	15810	67.4%	16	0.1%	988.1	0.1%	0.2%
NEB-related nemaline myopathy (256030)	14188	60.5%	13	0.1%	1,091.4	0.1%	0.2%
Herlitz junctional epidermolysis bullosa, LAMB3-related (226700)	15727	67.1%	14	0.1%	1,123.4	0.1%	0.1%
homocystinuria caused by cystathionine beta-synthase deficiency (236200)	15728	67.1%	13	0.1%	1,209.8	0.0%	0.1%
citrullinemia type 1 (215700)	12664	54.0%	10	0.1%	1,266.4	0.0%	0.1%
PPT1-related neuronal ceroid lipofuscinosis (256730)	15813	67.4%	12	0.1%	1,317.8	0.0%	0.1%
infantile Refsum disease (266510)	3034	12.9%	2	0.1%	1,517.0	0.0%	0.2%
D-bifunctional protein deficiency (261515)	12664	54.0%	8	0.1%	1,583.0	0.0%	0.1%
polyglandular autoimmune syndrome type 1 (240300)	15811	67.4%	9	0.1%	1,756.8	0.0%	0.1%
cartilage-hair hypoplasia (250250)	15813	67.4%	9	0.1%	1,757.0	0.0%	0.1%
Nijmegen breakage syndrome (251260)	15810	67.4%	8	0.1%	1,976.3	0.0%	0.1%
steroid resistant nephrotic syndrome (600995)	12664	54.0%	6	0.0%	2,110.7	0.0%	0.1%
muscle-eye-brain disease (253280)	15727	67.1%	7	0.0%	2,246.7	0.0%	0.1%
glutaric acidemia type 1 (231670)	15727	67.1%	7	0.0%	2,246.7	0.0%	0.1%
ataxia telangiectasia (208900)	15758	67.2%	7	0.0%	2,251.1	0.0%	0.1%
megalencephalic leukoencephalopathy with subcortical cysts (604004)	12665	54.0%	5	0.0%	2,533.0	0.0%	0.1%
PROP1-related combined pituitary hormone deficiency (262600)	12747	54.4%	5	0.0%	2,549.4	0.0%	0.1%
primary hyperoxaluria type 2 (260000)	15714	67.0%	6	0.0%	2,619.0	0.0%	0.1%
limb-girdle muscular dystrophy type 2E (604286)	15728	67.1%	6	0.0%	2,621.3	0.0%	0.1%
Leigh syndrome, French-Canadian type	3035	12.9%	1	0.0%	3,035.0	0.0%	0.2%

(256000)							
inclusion body myopathy 2 (600737)	15728	67.1%	5	0.0%	3,145.6	0.0%	0.1%
Niemann Pick disease type C (257220)	15755	67.2%	5	0.0%	3,151.0	0.0%	0.1%
alpha-mannosidosis (248500)	12664	54.0%	4	0.0%	3,166.0	0.0%	0.1%
limb-girdle muscular dystrophy type 2D (608099)	12665	54.0%	4	0.0%	3,166.3	0.0%	0.1%
Segawa syndrome (605407)	15727	67.1%	4	0.0%	3,931.8	0.0%	0.1%
glycogen storage disease type 1b (232200)	15730	67.1%	3	0.0%	5,243.3	0.0%	0.1%
ataxia with vitamin E deficiency (277460)	15797	67.4%	3	0.0%	5,265.7	0.0%	0.1%
carnitine palmitoyltransferase 1a deficiency (600528)	15727	67.1%	2	0.0%	7,863.5	0.0%	0.0%
Salla disease (604639)	15800	67.4%	2	0.0%	7,900.0	0.0%	0.0%
GRACILE syndrome (603358)	15810	67.4%	2	0.0%	7,905.0	0.0%	0.0%
congenital disorder of glycosylation type 1b (602579)	15810	67.4%	2	0.0%	7,905.0	0.0%	0.0%
congenital Finnish nephrosis (256300)	15813	67.4%	2	0.0%	7,906.5	0.0%	0.0%
Herlitz junctional epidermolysis bullosa, LAMC2-related (226700)	15727	67.1%	1	0.0%	15,727.0	0.0%	0.0%
glycogen storage disease type III (232400)	15729	67.1%	1	0.0%	15,729.0	0.0%	0.0%
Andermann syndrome (218000)	15800	67.4%	1	0.0%	15,800.0	0.0%	0.0%
hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (238970)	3034	12.9%	0	0.0%	N/A	0.0%	0.1%
ARSACS (270550)	15800	67.4%	0	0.0%	N/A	0.0%	0.0%
aspartylglycosaminuria (208400)	15800	67.4%	0	0.0%	N/A	0.0%	0.0%
CLN5-related neuronal ceroid lipofuscinosis (256731)	15813	67.4%	0	0.0%	N/A	0.0%	0.0%
choroideremia (303100)	15797	67.4%	0	0.0%	N/A	0.0%	0.0%
Northern epilepsy (610003)	15810	67.4%	0	0.0%	N/A	0.0%	0.0%
pycnodysostosis (265800)	15714	67.0%	0	0.0%	N/A	0.0%	0.0%
Sjogren-Larsson syndrome (270200)	15728	67.1%	0	0.0%	N/A	0.0%	0.0%
x-linked juvenile retinoschisis (312700)	15797	67.4%	0	0.0%	N/A	0.0%	0.0%

[a]Percentage of 23,453 individuals screened for a given disease.

Supplemental Table B: Variants Tested for Each Disease in Table 5

Disease	Gene Name	Variant Common Name
achromatopsia	*CNGB3*	819_826del8, 886-896del11insT, E336X, IVS8-3T>G, R403Q, T383fs
alpha-1 antitrypsin deficiency	*SERPINA1*	S allele, Z allele
biotinidase deficiency	*BTD*	A171T, D252G, D444H, F403V, *G98*:d7i3, Q456H, R538C
carnitine palmitoyltransferase II deficiency	*CPT2*	G549D, Leu178_Ile186delinsPhe, P227L, P50H, P604S, Q413fs, Q550R, R124X, R503C, R631C, S113L, S38fs, Y628S
citrullinemia type 1	*ASS1*	G390R, IVS6-2A>G
congenital disorder of glycosylation type Ia	*PMM2*	F119L, P113L, R141H, V231M
cystic fibrosis	*CFTR*	1078delT, 1161delC, 1288insTA, 1609delCA, 1677delTA, 1717-1G>A, 1811+1.6kbA>G, 1812-1G>A, 1898+1G>A, 1898+1G>T, 1898+5G>T, 1949del84, 2043delG, 2055del9>A, 2105-2117del13insAGAAA, 2108delA, 2143delT, 2183AA>G, 2184delA, 2184insA, 2307insA, 2789+5G>A, 2869insG, 296+12T>C, 3120+1G>A, 3120G>A, 3171delC, 3199del6, 3272-26A>G, 3659delC, 3667del4, 3791delC, 3821delT, 3849+10kbC>T, 3849+4A>G, 3876delA, 3905insT, 394delTT, 405+1G>A, 405+3A>C, 406-1G>A, 444delA, 457TAT>G, 574delA, 621+1G>T, 663delT, 711+1G>T, 711+5G>A, 712-1G>T, 935delA, 936delTA, A455E, A559T, C524X, D1152H, E60X, E92X, F311del, F508C, F508del, G178R, G330X, G480C, G542X, G551D, G622D, G85E, G91R, I148T, I506V, I507del, IVS8-5T, K710X, L206W, M1101K, N1303K, P574H, Q1238X, Q359K/T360K, Q493X, Q552X, Q890X, R1066C, R1070Q, R1158X, R1162X, R117C, R117H, R1283M, R334W, R347H, R347P, R352Q, R553X, R560T, R709X, R75X, R764X, S1196X, S1235R, S1251N, S1255X, S364P, S549I, S549N, S549R(A>C), S549R(T>G), T338I, V520F, W1089X, W1204X(c.3611G>A), W1204X(c.3612G>A), W1282X, Y1092X, Y122X, dele2-3 21kb
factor XI deficiency	*F11*	E117X, F283L, IVS14+1G>A, IVS14del14
familial dysautonomia	*IKBKAP*	IVS20+6T>C, P914L, R696P
familial Mediterranean fever	*MEFV*	A744S, F479L, I692del, K695R, M680I, M694I, M694V, P369S, R408Q, R653H, R761H, T267I, V726A
galactosemia	*GALT*	F171S, IVS2-2A>G, K285N, L195P, Q169K, Q188R, S135L, T138M, X380R, Y209C
Gaucher disease	*GBA*	1035insG, D409H, D409V, IVS2+1G>A, L444P, N370S, R463C, R463H, R496H, V394L
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	*GJB2*	167delT, 235delC, 313del14, 35delG, E120del, L90P, M34T, Q124X, R184P, V37I, W24X, W77R, W77X
hereditary fructose intolerance	*ALDOB*	A149P, A174D, Delta4E4, N334K, Y204X
hereditary thymine-uraciluria	*DPYD*	IVS14+1G>A

hexosaminidase A deficiency	*HEXA*	1278insTATC, 7.6kb del, G250D, G269S, IVS12+1G>C, IVS7+1G>A, IVS9+1G>A, R170W, R178C, R178H, R247W
inclusion body myopathy 2	*GNE*	M712T, V572L
medium chain acyl-CoA dehydrogenase deficiency	*ACADM*	G170R, G242R, K304E, L59F, R181C, R181H, Y42H
megalencephalic leukoencephalopathy with subcortical cysts	*MLC1*	135insC, IVS2-10T>A, c.176G>A, c.278C>T
Pendred syndrome	*SLC26A4*	E384G, H723R, IVS8+1G>A, L236P, T416P
phenylalanine hydroxylase deficiency	*PAH*	E280K, G272X, I65T, IVS-10int-546, IVS12+1G>A, L48S, P281L, R158Q, R252W, R261Q, R408Q, R408W, S349P, Y414C
Pompe disease	*GAA*	525delT, D645E, IVS1-13T>G, R854X
short chain acyl-CoA dehydrogenase deficiency	*ACADS*	G185S, R107C
sickle cell disease/beta thalassemia	*HBB*	-28A>G, -29A>G, -30T>A, -87C>G, -88C>T, 619 bp deletion, CAP+1 A>C, Glu6fs, Gly16fs, Gly24 T>A, Hb C, Hb D-Punjab, Hb E, Hb O-Arab, Hb S, IVS-I-1(G>A), IVS-I-1(G>T), IVS-I-110, IVS-I-5, IVS-I-6, IVS-II-654, IVS-II-705, IVS-II-745, IVS-II-844, IVS-II-849(A>C), IVS-II-849(A>G), IVS-II-850, K17X, Lys8fs, Phe41fs, Phe71fs, Poly A: AATAAA->AATAAG, Poly A: AATAAA->AATGAA, Pro5fs, Q39X, Ser9fs, W15X
Smith-Lemli-Opitz syndrome	*DHCR7*	C380Y, E448L, F302L, G410S, IVS8-1G>C, L109P, L157P, R242C, R242H, R352Q, R352W, R404C, S169L, T93M, V326L, W151X(c.452G>A), W151X(c.453G>A)
spinal muscular atrophy	*SMN1*	Exon 7 deletion
tyrosinemia type I	*FAH*	E357X, IVS12+5G>A, IVS6-1G>T, IVS8-1G>C, P261L, Q64H, W262X

Supplementary Table C: References for Literature Estimates of Carrier Frequencies

Disease	Literature Frequency (1 in)	Citation
All Populations (N=23453)		
alpha-1 antitrypsin deficiency	11.5	de Serres, FJ, Blanco, I, Fernández-Bustillo, E(2010). Ethnic differences in alpha-1 antitrypsin deficiency in the United States of America. Ther Adv Respir Dis, 4, 2:63-70.
cystic fibrosis	31.7	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	42.8	Smith RJH, Van Camp G. Nonsyndromic Hearing Loss and Deafness, DFNB1. 1998 Sep 28 [Updated 2011 Jul 14]. In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews (TM) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1272/
spinal muscular atrophy	54	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
familial Mediterranean fever	unknown	Mikula, M, Buller, A, Sun, W, Strom, CM(2008). Prevalence of known mutations in the familial Mediterranean fever gene (MEFV) in various carrier screening populations. Genet. Med. , 10, 5:349-52.
Smith-Lemli-Opitz syndrome	123	Kelley, RI, Hennekam, RC(2000). The Smith-Lemli-Opitz syndrome. J. Med. Genet. , 37, 5:321-35.
sickle cell disease/beta thalassemia	158	Galanello, R, Origa, R(2010). Beta-thalassemia. Orphanet J Rare Dis, 5:11.
Gaucher disease	200	Horowitz, M, Pasmanik-Chor, M, Borochowitz, Z, Falik-Zaccai, T, Heldmann, K, Carmi, R, Parvari, R, Beit-Or, H, Goldman, B, Peleg, L, Levy-Lahad, E, Renbaum, P, Legum, S, Shomrat, R, Yeger, H, Benbenisti, D, Navon, R, Dror, V, Shohat, M, Magal, N, Navot, N, Eyal, N(1998). Prevalence of glucocerebrosidase mutations in the Israeli Ashkenazi Jewish population. Hum. Mutat. , 12, 4:240-4.
factor XI deficiency	rare	Duga, S, Salomon, O(2009). Factor XI Deficiency. Semin. Thromb. Hemost. , 35, 4:416-25.
achromatopsia	123	Sharpe LT, Stockman A, Jagle H, Nathans . Opsin genes, cone photopigments, color vision, and color blindness. In: Gegenfurtner K, Sharpe LT, eds. Color Vision: from Genes to Perception. Cambridge: Cambridge University Press; 1999:3-52.

Northwestern Europe (N=12915)		
alpha-1 antitrypsin deficiency	11.4	de Serres, FJ, Blanco, I, Fernández-Bustillo, E(2010). Ethnic differences in alpha-1 antitrypsin deficiency in the United States of America. Ther Adv Respir Dis, 4, 2:63-70.
cystic fibrosis	28	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	33.2	Smith RJH, Van Camp G. Nonsyndromic Hearing Loss and Deafness, DFNB1. 1998 Sep 28 [Updated 2011 Jul 14]. In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews (TM) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1272/
spinal muscular atrophy	47	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
Smith-Lemli-Opitz syndrome	123.9	Nowaczyk, MJ, Zeesman, S, Waye, JS, Douketis, JD(2004). Incidence of Smith-Lemli-Opitz syndrome in Canada: results of three-year population surveillance. J. Pediatr. , 145, 4:530-5.
medium chain acyl-CoA dehydrogenase deficiency	61.7	Grosse, SD, Khoury, MJ, Greene, CL, Crider, KS, Pollitt, RJ(2006). The epidemiology of medium chain acyl-CoA dehydrogenase deficiency: an update. Genet. Med. , 8, 4:205-12.
hereditary fructose intolerance	81.1	Santer, R, Rischewski, J, von Weihe, M, Niederhaus, M, Schneppenheim, S, Baerlocher, K, Kohlschütter, A, Muntau, A, Posselt, HG, Steinmann, B, Schneppenheim, R(2005). The spectrum of aldolase B (ALDOB) mutations and the prevalence of hereditary fructose intolerance in Central Europe. Hum. Mutat. , 25, 6:594.
achromatopsia	123	Sharpe LT, Stockman A, Jagle H, Nathans . Opsin genes, cone photopigments, color vision, and color blindness. In: Gegenfurtner K, Sharpe LT, eds. Color Vision: from Genes to Perception. Cambridge: Cambridge University Press; 1999:3-52.
hereditary thymine-uraciluria	33	Eidens M, Prause S, Weise A, Klemm M, Weber MM, Pfuetzner A. Dihydropyrimidine Dehydrogenase Genotyping and Phenotyping for 5-Fluorouracil Dysmetabolism: Moving Towards Personalized Chemotherapy in Patients with Cancer. Curr Pharmacogenomics Person Med. 2009, 7(4), 275-283.
familial Mediterranean fever	unknown, assumed rare	Ben-Chetrit, E, Touitou, I(2009). Familial mediterranean Fever in the world. Arthritis Rheum. , 61, 10:1447-53.

Ashkenazi Jewish (N=2410)		
factor XI deficiency	8	Gomez, K, Bolton-Maggs, P(2008). Factor XI deficiency. Haemophilia, 14, 6:1183-9.
familial Mediterranean fever	10.5	Aksentijevich, I, Torosyan, Y, Samuels, J, Centola, M, Pras, E, Chae, JJ, Oddoux, C, Wood, G, Azzaro, MP, Palumbo, G, Giustolisi, R, Pras, M, Ostrer, H, Kastner, DL(1999). Mutation and haplotype studies of familial Mediterranean fever reveal new ancestral relationships and evidence for a high carrier frequency with reduced penetrance in the Ashkenazi Jewish population. Am. J. Hum. Genet. , 64, 4:949-62.
Gaucher disease	17	Horowitz, M, Pasmanik-Chor, M, Borochowitz, Z, Falik-Zaccai, T, Heldmann, K, Carmi, R, Parvari, R, Beit-Or, H, Goldman, B, Peleg, L, Levy-Lahad, E, Renbaum, P, Legum, S, Shomrat, R, Yeger, H, Benbenisti, D, Navon, R, Dror, V, Shohat, M, Magal, N, Navot, N, Eyal, N(1998). Prevalence of glucocerebrosidase mutations in the Israeli Ashkenazi Jewish population. Hum. Mutat. , 12, 4:240-4.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	21	Morell, RJ, Kim, HJ, Hood, LJ, Goforth, L, Friderici, K, Fisher, R, Van Camp, G, Berlin, CI, Oddoux, C, Ostrer, H, Keats, B, Friedman, TB(1998). Mutations in the connexin 26 gene (GJB2) among Ashkenazi Jews with nonsyndromic recessive deafness. N. Engl. J. Med. , 339, 21:1500-5.
cystic fibrosis	29	Moskowitz SM, Chmiel JF, Sternen DL, et al. CFTR-Related Disorders. 2001 Mar 26 [Updated 2008 Feb 19]. In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews (TM) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1250/
alpha-1 antitrypsin deficiency	16	de Serres, FJ(2002). Worldwide racial and ethnic distribution of alpha1-antitrypsin deficiency: summary of an analysis of published genetic epidemiologic surveys. Chest, 122, 5:1818-29.
hexosaminidase A deficiency	27.4	Scott, SA, Edelmann, L, Liu, L, Luo, M, Desnick, RJ, Kornreich, R(2010). Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. Hum. Mutat. , 31, 11:1240-50.
short chain acyl-CoA dehydrogenase deficiency	unknown/ underdiagnosed	Jethva, R, Bennett, MJ, Vockley, J(2008). Short-chain acyl-coenzyme A dehydrogenase deficiency. Mol. Genet. Metab. , 95, 4:195-200.
familial dysautonomia	31	Scott, SA, Edelmann, L, Liu, L, Luo, M, Desnick, RJ, Kornreich, R(2010). Experience with carrier screening and prenatal diagnosis for 16 Ashkenazi Jewish genetic diseases. Hum. Mutat. , 31, 11:1240-50.

carnitine palmitoyltransferase II deficiency	rare/too few patients worldwide	Taroni, F, Verderio, E, Dworzak, F, Willems, PJ, Cavadini, P, DiDonato, S(1993). Identification of a common mutation in the carnitine palmitoyltransferase II gene in familial recurrent myoglobinuria patients. Nat. Genet. , 4, 3:314-20.
Hispanic (N=2302)		
alpha-1 antitrypsin deficiency	9.2	de Serres, FJ, Blanco, I, Fernández-Bustillo, E(2010). Ethnic differences in alpha-1 antitrypsin deficiency in the United States of America. Ther Adv Respir Dis, 4, 2:63-70.
cystic fibrosis	59	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	100	Shan, J, Chobot-Rodd, J, Castellanos, R, Babcock, M, Shanske, A, Parikh, SR, Morrow, BE, Samanich, J(2010). GJB2 mutation spectrum in 209 hearing impaired individuals of predominantly Caribbean Hispanic and African descent. Int. J. Pediatr. Otorhinolaryngol. , 74, 6:611-8. Samanich, J, Lowes, C, Burk, R, Shanske, S, Lu, J, Shanske, A, Morrow, BE(2007). Mutations in GJB2, GJB6, and mitochondrial DNA are rare in African American and Caribbean Hispanic individuals with hearing impairment. Am. J. Med. Genet. A, 143A, 8:830-8.
spinal muscular atrophy	68	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
sickle cell disease/beta thalassemia	128	Michlitsch, J, Azimi, M, Hoppe, C, Walters, MC, Lubin, B, Lorey, F, Vichinsky, E(2009). Newborn screening for hemoglobinopathies in California. Pediatr Blood Cancer, 52, 4:486-90.
Pompe disease	100	Taglia, A, Picillo, E, D'Ambrosio, P, Cecio, MR, Viggiano, E, Politano, L(2011). Genetic counseling in Pompe disease. Acta Myol, 30, 3:179-81.
congenital disorder of glycosylation type Ia	unknown	Schollen, E, Kjaergaard, S, Legius, E, Schwartz, M, Matthijs, G(2000). Lack of Hardy-Weinberg equilibrium for the most prevalent PMM2 mutation in CDG-Ia (congenital disorders of glycosylation type Ia). Eur. J. Hum. Genet. , 8, 5:367-71.

familial Mediterranean fever	unknown	Vergara, C, Borzutzky, A, Gutierrez, MA, Iacobelli, S, Talesnik, E, Martinez, ME, Stange, L, Basualdo, J, Maluje, V, Jimenez, R, Wiener, R, Tinoco, J, Jarpa, E, Aróstegui, JI, Yagüe, J, Alvarez-Lobos, M(2012). Clinical and genetic features of hereditary periodic fever syndromes in Hispanic patients: the Chilean experience. Clin. Rheumatol. , 31, 5:829-34.
African American (N=1193)		
sickle cell disease/beta thalassemia	10	Norman, BJ, Miller, SD(2011). Human genome project and sickle cell disease. Soc Work Public Health, 26, 4:405-16.
alpha-1 antitrypsin deficiency	37.3	de Serres, FJ, Blanco, I, Fernández-Bustillo, E(2010). Ethnic differences in alpha-1 antitrypsin deficiency in the United States of America. Ther Adv Respir Dis, 4, 2:63-70.
cystic fibrosis	84	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
Pompe disease	59.7	Hirschhorn R, Reuser AJ. Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. In: Scriver CR, Beaudet A, Sly WS, Valle D, eds. The Metabolic and Molecular Bases of Inherited Disease. New York: McGraw-Hill; 2001:3389-420.
spinal muscular atrophy	72	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
galactosemia	94	Suzuki, M, West, C, Beutler, E(2001). Large-scale molecular screening for galactosemia alleles in a pan-ethnic population. Hum. Genet. , 109, 2:210-5.
Gaucher disease	34.6	Landgren, O, Turesson, I, Gridley, G, Caporaso, NE(2007). Risk of malignant disease among 1525 adult male US Veterans with Gaucher disease. Arch. Intern. Med. , 167, 11:1189-94.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	25.3	Ross, SA, Novak, Z, Kumbla, RA, Zhang, K, Fowler, KB, Boppana, S(2007). GJB2 and GJB6 mutations in children with congenital cytomegalovirus infection. Pediatr. Res. , 61, 6:687-91.
Smith-Lemli-Opitz syndrome	137.8	Wright, BS, Nwokoro, NA, Wassif, CA, Porter, FD, Waye, JS, Eng, B, Nowaczyk, MJ(2003). Carrier frequency of the RSH/Smith-Lemli-Opitz IVS8-1G >C mutation in African Americans. Am. J. Med. Genet. A, 120A, 1:139-41.
South Asia (N=1123)		

achromatopsia	123	Sharpe LT, Stockman A, Jagle H, Nathans . Opsin genes, cone photopigments, color vision, and color blindness. In: Gegenfurtner K, Sharpe LT, eds. Color Vision: from Genes to Perception. Cambridge: Cambridge University Press; 1999:3-52.
sickle cell disease/beta thalassemia	25	Madan, N, Sharma, S, Sood, SK, Colah, R, Bhatia, LH(2010). Frequency of -thalassemia trait and other hemoglobinopathies in northern and western India. Indian J Hum Genet, 16, 1:16-25.
cystic fibrosis	118	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
spinal muscular atrophy	52	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
hereditary thymine-uraciluria	33	Morsman, JM, Sludden, J, Ameyaw, MM, Githang'A, J, Indalo, A, Ofori-Adjei, D, McLeod, HL(2000). Evaluation of dihydropyrimidine dehydrogenase activity in South-west Asian, Kenyan and Ghanaian populations. Br J Clin Pharmacol, 50, 3:269-72.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	148	Mahdieh, N, Rabbani, B(2009). Statistical study of 35delG mutation of GJB2 gene: a meta-analysis of carrier frequency. Int J Audiol, 48, 6:363-70.
megalencephalic leukoencephalopathy with subcortical cysts	unknown, founder effect	Gorospe, JR, Singhal, BS, Kainu, T, Wu, F, Stephan, D, Trent, J, Hoffman, EP, Naidu, S(2004). Indian Agarwal megalencephalic leukodystrophy with cysts is caused by a common MLC1 mutation. Neurology, 62, 6:878-82.
biotinidase deficiency	123	Wolf, B(1991). Worldwide survey of neonatal screening for biotinidase deficiency. J. Inherit. Metab. Dis. , 14, 6:923-7.
tyrosinemia type I	173	Mitchell GA, Grompe M, Lambert M, Tanguay RM. Hypertyrosinemia. In: Scriver CR, Beaudet AL, Sly WS, Valle D, eds. The Metabolic and Molecular Bases of Inherited Disease. New York: McGraw Hill; 2001:1777-806.
Eastern Asia (N=1121)		
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	33 - 200	Liu, Y, Ke, X, Qi, Y, Li, W, Zhu, P(2002). Connexin26 gene (GJB2): prevalence of mutations in the Chinese population. J. Hum. Genet. , 47, 12:688-90.

		Dai, P, Yu, F, Han, B, Liu, X, Wang, G, Li, Q, Yuan, Y, Liu, X, Huang, D, Kang, D, Zhang, X, Yuan, H, Yao, K, Hao, J, He, J, He, Y, Wang, Y, Ye, Q, Yu, Y, Lin, H, Liu, L, Deng, W, Zhu, X, You, Y, Cui, J, Hou, N, Xu, X, Zhang, J, Tang, L, Song, R, Lin, Y, Sun, S, Zhang, R, Wu, H, Ma, Y, Zhu, S, Wu, BL, Han, D, Wong, LJ(2009). GJB2 mutation spectrum in 2,063 Chinese patients with nonsyndromic hearing impairment. J Transl Med, 7:26.
sickle cell disease/beta thalassemia	50	Li, DZ(2009). Premarital screening for thalassemia in mainland China. Prenat. Diagn. , 29, 6:637-8.
		Cheng, TC, Orkin, SH, Antonarakis, SE, Potter, MJ, Sexton, JP, Markham, AF, Giardina, PJ, Li, A, Kazazian, HH(1984). beta-Thalassemia in Chinese: use of in vivo RNA analysis and oligonucleotide hybridization in systematic characterization of molecular defects. Proc. Natl. Acad. Sci. U. S. A. , 81, 9:2821-5.
spinal muscular atrophy	59	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
achromatopsia	123	Sharpe LT, Stockman A, Jagle H, Nathans . Opsin genes, cone photopigments, color vision, and color blindness. In: Gegenfurtner K, Sharpe LT, eds. Color Vision: from Genes to Perception. Cambridge: Cambridge University Press; 1999:3-52.
cystic fibrosis	242	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
alpha-1 antitrypsin deficiency	450	de Serres, FJ(2002). Worldwide racial and ethnic distribution of alpha1-antitrypsin deficiency: summary of an analysis of published genetic epidemiologic surveys. Chest, 122, 5:1818-29.
Pendred syndrome	51	Park, HJ, Shaukat, S, Liu, XZ, Hahn, SH, Naz, S, Ghosh, M, Kim, HN, Moon, SK, Abe, S, Tukamoto, K, Riazuddin, S, Kabra, M, Erdenetungalag, R, Radnaabazar, J, Khan, S, Pandya, A, Usami, SI, Nance, WE, Wilcox, ER, Riazuddin, S, Griffith, AJ(2003). Origins and frequencies of SLC26A4 (PDS) mutations in east and south Asians: global implications for the epidemiology of deafness. J. Med. Genet. , 40, 4:242-8.
Pompe disease	112	Lin, CY, Hwang, B, Hsiao, KJ, Jin, YR(1987). Pompe's disease in Chinese and prenatal diagnosis by determination of alpha-glucosidase activity. J. Inherit. Metab. Dis. , 10, 1:11-7.

carnitine palmitoyltransferase II deficiency	rare/too few patients worldwide	Taroni, F, Verderio, E, Dworzak, F, Willems, PJ, Cavadini, P, DiDonato, S(1993). Identification of a common mutation in the carnitine palmitoyltransferase II gene in familial recurrent myoglobinuria patients. Nat. Genet. , 4, 3:314-20.
Southern Europe (N=1063)		
alpha-1 antitrypsin deficiency	13.1	de Serres, FJ, Blanco, I, Fernández-Bustillo, E(2010). Ethnic differences in alpha-1 antitrypsin deficiency in the United States of America. Ther Adv Respir Dis, 4, 2:63-70.
cystic fibrosis	28	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	33.2	Smith RJH, Van Camp G. Nonsyndromic Hearing Loss and Deafness, DFNB1. 1998 Sep 28 [Updated 2011 Jul 14]. In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews (TM) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1272/
spinal muscular atrophy	47	Sugarman, EA, Nagan, N, Zhu, H, Akmaev, VR, Zhou, Z, Rohlfs, EM, Flynn, K, Hendrickson, BC, Scholl, T, Sirko-Osadsa, DA, Allitto, BA(2012). Pan-ethnic carrier screening and prenatal diagnosis for spinal muscular atrophy: clinical laboratory analysis of >72,400 specimens. Eur. J. Hum. Genet. , 20, 1:27-32.
sickle cell disease/beta thalassemia	50	Galanello, R, Origa, R(2010). Beta-thalassemia. Orphanet J Rare Dis, 5:11.
familial Mediterranean fever	unknown	Ben-Chetrit, E, Touitou, I(2009). Familial mediterranean Fever in the world. Arthritis Rheum. , 61, 10:1447-53.
phenylalanine hydroxylase deficiency	50.5	Scriver CR, Kaufman S (2001) The hyperphenylalaninemias. In: Scriver CR, Beaudet AL, Sly SW, Valle D (eds) Childs B, Kinzler KW, Vogelstein B (assoc eds) The Metabolic and Molecular Bases of Inherited Disease, 8 ed. McGraw-Hill, New York, Ch. 77 2001.
Pompe disease	100	Martiniuk, F, Chen, A, Mack, A, Arvanitopoulos, E, Chen, Y, Rom, WN, Codd, WJ, Hanna, B, Alcabes, P, Raben, N, Plotz, P(1998). Carrier frequency for glycogen storage disease type II in New York and estimates of affected individuals born with the disease. Am. J. Med. Genet. , 79, 1:69-72.
Smith-Lemli-Opitz syndrome	unknown	Nowaczyk, MJ, Zeesman, S, Waye, JS, Douketis, JD(2004). Incidence of Smith-Lemli-Opitz syndrome in Canada: results of three-year population surveillance. J. Pediatr. , 145, 4:530-5.

hereditary thymine-uraciluria	33	Eidens M, Prause S, Weise A, Klemm M, Weber MM, Pfuetzner A. Dihydropyrimidine Dehydrogenase Genotyping and Phenotyping for 5-Fluorouracil Dysmetabolism: Moving Towards Personalized Chemotherapy in Patients with Cancer. Curr Pharmacogenomics Person Med. 2009, 7(4), 275-283.
Middle East (N=512)		
familial Mediterranean fever	variable (1/10 - 1/20)	Shohat M, Halpern GJ. Familial Mediterranean Fever. 2000 Aug 8 [Updated 2012 Apr 26]. In: Pagon RA, Bird TD, Dolan CR, et al., editors. GeneReviews (TM) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-. Available from: http://www.ncbi.nlm.nih.gov/books/NBK1227/
sickle cell disease/beta thalassemia	30 (but variable)	Al-Riyami, A, Ebrahim, GJ(2003). Genetic Blood Disorders Survey in the Sultanate of Oman. J. Trop. Pediatr. , 49 Suppl 1:i1-20.
hereditary thymine-uraciluria	33	Morsman, JM, Sludden, J, Ameyaw, MM, Githang'A, J, Indalo, A, Ofori-Adjei, D, McLeod, HL(2000). Evaluation of dihydropyrimidine dehydrogenase activity in Southwest Asian, Kenyan and Ghanaian populations. Br J Clin Pharmacol, 50, 3:269-72.
achromatopsia	123	Sharpe LT, Stockman A, Jagle H, Nathans . Opsin genes, cone photopigments, color vision, and color blindness. In: Gegenfurtner K, Sharpe LT, eds. Color Vision: from Genes to Perception. Cambridge: Cambridge University Press; 1999:3-52.
cystic fibrosis	91	Rohlfs, EM, Zhou, Z, Heim, RA, Nagan, N, Rosenblum, LS, Flynn, K, Scholl, T, Akmaev, VR, Sirko-Osadsa, DA, Allitto, BA, Sugarman, EA(2011). Cystic fibrosis carrier testing in an ethnically diverse US population. Clin. Chem. , 57, 6:841-8.
spinal muscular atrophy	25	Lyahyai, J, Sbiti, A, Barkat, A, Ratbi, I, Sefiani, A(2012). Spinal muscular atrophy carrier frequency and estimated prevalence of the disease in Moroccan newborns. Genet Test Mol Biomarkers, 16, 3:215-8.
GJB2-related DFNB 1 nonsyndromic hearing loss and deafness	83	Najmabadi, H, Cucci, RA, Sahebjam, S, Kouchakian, N, Farhadi, M, Kahrizi, K, Arzhangi, S, Daneshmandan, N, Javan, K, Smith, RJ(2002). GJB2 mutations in Iranians with autosomal recessive non-syndromic sensorineural hearing loss. Hum. Mutat. , 19, 5:572.

inclusion body myopathy 2	15 (Iranian Jews), unknown in others	Eisenberg, I, Avidan, N, Potikha, T, Hochner, H, Chen, M, Olender, T, Barash, M, Shemesh, M, Sadeh, M, Grabov-Nardini, G, Shmilevich, I, Friedmann, A, Karpati, G, Bradley, WG, Baumbach, L, Lancet, D, Asher, EB, Beckmann, JS, Argov, Z, Mitrani-Rosenbaum, S(2001). The UDP-N-acetylglucosamine 2-epimerase/N-acetylmannosamine kinase gene is mutated in recessive hereditary inclusion body myopathy. Nat. Genet. , 29, 1:83-7.
hereditary fructose intolerance	unknown	James, CL, Rellos, P, Ali, M, Heeley, AF, Cox, TM(1996). Neonatal screening for hereditary fructose intolerance: frequency of the most common mutant aldolase B allele (A149P) in the British population. J. Med. Genet. , 33, 10:837-41.
Smith-Lemli-Opitz syndrome	rare	Al-Owain, M, Imtiaz, F, Shuaib, T, Edrees, A, Al-Amoudi, M, Sakati, N, Al-Hassnan, Z, Bamashmous, H, Rahbeeni, Z, Al-Ameer, S, Faqeih, E, Meyer, B, Al-Hashem, A, Garout, W, Al-Odaib, A, Rashed, M, Al-Aama, J(2012). Smith-Lemli-Opitz syndrome among Arabs. Clin Genet, 82, 2:165-172.