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שרותי בריאות הציבור
גנטיקה קהילתית
Department of Community Genetics

משרד הבריאות
לחיים בריאים יותר

Department of Community Genetics
Public Health Services
Ministry of Health
Israel

MENDELIAN DISORDERS AMONG JEWS

TABLES

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ASHKENAZI JEWS; Autosomal recessive

1. Abetalipoproteinemia.	1:131	MTP; **[G865X]**; [2212 delT]
2. Acyl-CoA dehydrogenase deficiency Short chain	1:15	SCAD; **[319C>T]**; [625G>A], [511C>T]
3. Aicardi-Goutieres syndrome	1:110	SAMHD1 **[del exon 1]**; [c.676C>G]; [c.649_650insG]
4. Alport	1:183	COL4A3 **[c.40_63del]**
5. Amegakaryocytic thrombocytopenia	1:75	MPL **[c.79+2T>A]**
6. Arthrogryposis autism and epilepsy	1:204	SLC35A3 **[c.886A>G]**; [c.514C>T]
7. Adrenal hyperplasia, non classical 21 OH deficiency	1:6	CPY21 **[VAL281LEU]**
8. Bardet Biedl	1:143	BBS2 **[c.311A>C]**; [c.1895G>C]
10 Bloom syndrome	1:134*	BLM **[6-BP DEL/7-BP INS]**
10. Canavan disease	1: 62*	ASPA **[GLU285ALA]**; [TYR231TER]
11. Carnitine plamoyltransferase II deficiency	?	CPTII **[1237delAG]**; **[F448 L]**; [S113L]
12. Choreoacanthocytosis	?	VPS13A **[6059delC]**
13. Cystic fibrosis	1:26*	CFTR [PHE508DEL]; [TRP1282TER]; [GLY542TER]; [ASN1303LYS]; [IVS19, C-T, +10]
14. Cystinuria	?	SLC3A1 **[C808T]**
15. Deafness	1:25 1:180 ?	GJB2 **[1-BP DEL, 167T]**;[1-BP DEL,30G] GJB6 [deletion,140 kb] LOXHD1 **[R1572X]** STRC [c.4171CC>G]; **[deletion]**
16. Dysautonomia, familial	1:37*	IKBKAP **[IVS20+6T>C];** [R696P]
17. Ehlers-Danlos syndrome type VII-C	?	ADAMTS2 **[Q225X]**
18. Enhanced S-cone	?	NR2E3 **[c.923G>A]**
19. Factor XI (PTA) deficiency	1:12	F11 **[p.Glu117stop]**; **[p.phe283leu]**; [c.1716+1G>A]
20. Familial Mediterranean Fever	1:10	MEFV **[E148Q]**; [P369S]; [K395R]; [V726A]
21. Fanconi anemia	1:99*	FACC **[IVS4+4, A-to-T]**
22. Galactosemia	1:120	GALT **[5bp deletion]**
23. Gaucher disease type I	1:17*	GBA **[ASN370SER]**; [84GG]; [LEU444PRO]; [IVS2DS, G-A, +1]
24. Glycogenosis Ia	1:82*	G6PC **[R83C]**
25. Glycogenosis IV (adult)	1:34	GBE **[c.1076A>C]**
26. GlycogenosisVII Phosphokinase deficiency, late onset	Rare	PFKM [IVS5DS, G-A, +1]; [ARG95TER] [ARG39LEU];
27. Hermansky-Pudlak syndrome	1:235	HSP3 **[1303+1G→A]**
28. Hoyerall-Hreidarsson syndrome	1:220	RTEL1 **[c.3791G>A]**; [c.C2920T]; [c.G1476T]
29. Hyperoxaluria III	Rare	DHDPSL **[GLU315DEL]**; [GLY287VAL]
30. Joubert syndrome 2	1:102*	TMEM216 **[c.35G>T]**
31. Leber amaurosis	1:100	LCA5 **[c. 835C>T]**
32. Leigh syndrome	1:290	C20ORF7 **[G250V]**
33. Lipoamide dehydrogenase deficiency	1:99*	LAD **[GLY229CYS]**; [1-bp ins Tyr35Ter]
34. Microcephaly with complex motor and sensory axonal neuropathy	1:225	VRK1 **[R358X]**
35. MSUD	1:120*	E1β **[R183P]**
36. Mucolipidosis IV	1:102*	MCOLN1 **[IVS3-1A-G]**; [del (EX1- EX7)]
37. Nemaline myopathy	1:140*	NEB **[R2478_D2521del]**
38. Niemann-Pick disease A	1:119*	SMPD1 [ARG496LEU]; [LEU302PRO]; [PRO330FS]
39. Osteopetrosis	1:350	TCIRG1**[c.117+4A>T]**
40. Pentosuria	1:29	DCXR **[c.598delC]**; [55+1G>A]
41. Persistent hyperinsulinemic hypoglycemia of infancy	1:61*	ABCC8 **[c.3989-9G>A],** [p.F1387del]

42. Primary ciliary dyskinesia	1:200 1:344 1:357 1:1000 1:172 1:526 1:208	DNAI2 [c.1304G>A] CCDC65 [c.877_878delAT] DNAL1 [c.1490G>A] DNAH11 [c.11929G>T] DNAH5 [c.7502G>C] CCDC114 [c.939delT] C21orf59 [c.735C>G]
43. Retinitis pigmentosa	1:115 1:120	DHDDS **[c.124A>G]** MAK **[353-bp Alu ins Ex9]**
44. Retinitis pigmentosa non syndromic	1:143	BBS2 [**c.311A>C**]; [c.1895G>C]
45. Tay Sachs disease	1:27*	HEXA **[4-BP INS, EX11]** ; [IVS12DS, G-C, +1]; [GLY269SER]
46. Thalassemia, alpha	1:17	HBA **[alpha 3.7]**
47. Tyrosinemia	?	FAH **[P261L]**
48. Usher syndrome I	1:131*	PCDH15 **[R245X]**
49. Usher syndrome III	1:109*	USH3 **[N48K]**
50. Walker Warburg	1:81*	FCMD **[c.1167_1168insA]**
51. Zelweger syndrome	1:120	PEX2 **[c.355C>T]**

*carrier frequency as determined in large population carrier screening

ASHKENAZI JEWS; Autosomal dominant

1. Breast/ovarian cancer	1:100 1:75	BRCA1 [185delAG]; BRCA1[5382insC]; BRCA2 [6174delT]
2. Cerebral cavernous malformations	?	CCM2 **[c.30+5_6delinsTT]**
3. Hypercholesterolemia, familial (Lithuanian Jews)	1:69	LDLR **[GLY197DEL]**
4. MODY	?	GK **[T206P]**
5. Lynch syndrome		MSH2 **[1906C>G]**; MSH6 [c.3984_3987dupGTCA]; MSH6 [c.3939_3962delCAAG]
6. Torsion Dystonia (idiopathic)	1:4,000	DYT1 **[3-BP DEL, GLU DEL]**
7. Zinc deficiency, transient neonatal	?	SLC30A2 **[G78R]**

ASHKENAZI JEWS; X linked

Alport syndrome	?	COL4A5 **[R1677Q]**

ASHKENAZI JEWS; complex inheritance

Crohn disease		NOD2 [p.R702W]; [pG908R; [p.L1007fs]
Colon cancer	1:20	APC **[ILE1307LYS]**
Parkinson disease		LRRK2 [G2019S]
Prostate cancer		RASEL

ASHKENAZI JEWS; Autosomal recessive, one family

Deafness	MYO15A [c.373delCG], [p.R2728H] STRC [c.4171C]
Citrin deficiency	SLC25A13 [T446P]
Comel-Netherton syndrome	SPINK5[c.C649T; c.691delC]
Erythropoietic protoporphyria	FECH [IVS3-48]
Foveal hypoplasia	SLC38A8 [c.848A>C]
Hereditary sensory autonomic neuropathy	DST [c.14865delA
Leigh syndrome	NDUFS4 [462delA]
Nephrotic syndrome steroid resistant	ARHGDIA [c.518G>T]
Niemann Pick type C	NPC1 [c.2792-3delAG]
Renal hypouricemia	SLC2A9 [36kb deletion]

Retinitis pigmentosa	FAM161A [c.1355_6delCA]
Peeling skin syndrome	CDSN [c164_167dupGCCT]
Pendred syndrome	SLC26A4 [c.349C>T]
Smith-Lemli-Opitz	DHCR7 [c.964-1G>C]

JEWS FROM NORTH AFRICA

Ataxia telangiectasia	>1:80	ATM [ARG35TER]
Familial Mediterranean Fever	1:7	MEFV 70% [M694VAL]
Chronic hemolysis and childhood relapsing immune mediated polyneuropathy.	1:66	CD59 [p.Cys89Tyr]
Leber congenital amaurosis	1:90	RPE65 [c.95-2A>T]
Leber congenital amaurosis	4 families	GUCY2D [pc.389delC]
Leber congenital amaurosis	4 families	AIPL1 [p.Val71Phe]
Retinitis pigmentosa	1:94	EYS [c.1211_1212insA]
Retinitis pigmentosa*	3 families	FAM161A [c.C.1567C>T]
Tay Sachs disease	>1:110	HEXA [several mutations]
Usher syndrome 1B	Several families	USH1B [Ala214Arg]

*Also in two Syrian families and a Bulgarian individual

JEWS FROM ALGERIA

Distal spinal muscular atrophy	4 families	GARS [G526R]
Frontotemporal dementia	1 family	MAP Tau gene [P301S]

JEWS FROM LIBYA

LIBYA - TUNISIA

Creuzfeld-Jakob disease	1:24,000	PRNP [E200K]

LIBYA

Cystinuria	1:25	SLC7A9 [V170M]
Limb girdle muscular dystrophy	1:10	DYSF [deletion]
Megaencephalic leukoencephalopathy	1:50	MLC [G59E]

LIBYA, one or 2 families

Usher syndrome 2A	USH2A
Multiple mitochondrial deletions	unknown

JEWS FROM TUNISIA

TUNISIA - LIBYA

Creuzfeld-Jakob disease	1:24,000	PRNP [E200K]

TUNISIA - MOROCCO

Melanoma, cutaneous	unknown	CDKN2A [V59G]

TUNISIA

Brittle cornea syndrome	ZNF469 [5943 delA]
Factor V and VIII deficiency	LMAN1 [IVS383 DS, T-C, +2]
Fanconi anemia	FANCA [890-893del]
Fragile X syndrome	FMR1 unique haplotype
Properdin deficiency	PCF
Selective intestinal malabsorption B12	AMN [IVS3, A-G, -2]
Mucolipidosis III	GNPTAG [500 insC]
Penylketonuria	PHA [L48S] [1]

[1]also found in Caucasian Jews and Jews from Bukhara

TUNISIA, one or 2 families

Abetalipoproteinemia	1 family	MTP [K03X][1]
3 methyl crotonyl CoA carboxylase	1 family	unknown
Usher syndrome type 2A	1 family	USH2A [c.1000C>T] [2]
Osteoporosis-pseudoglioma syndrome	1 family	LRP5 [R353Q]
Wilson disease	?	ATP7B [R969Q]

[1]one parent from Algeria

[2] found also in one Moroccan Jewish family

JEWS FROM MOROCCO

MOROCCO - ALGERIA

Usher syndrome	Several families	USH1B [Ala214Arg], [Gly214Arg], [2065del]
Deafness	1 family	CDH23 [c.7903G>T]

MOROCCO - LIBYA

Retinitis pigmentosa*	1:32	FAM161A [c.1355_6delCA]

*Also in two Ashkenazi and on Bulgarian individual

MOROCCO - TUNISIA

Melanoma, cutaneous	unknown	CDKN2A [V59G]

MOROCCO - IRAQ - IRAN

Factor VII	1:42	F7 [ALA244VAL]
Myasthenia gravis, infantile	unknown	RAPSN [-38A-->G] [3]
Pituitary dwarfism II (Laron)	unknown	GHR [ex 5,6 DEL] [3]

MOROCCO

Adrenal hyperplasia, 11 beta hydroxylase	1:30-1:128	CYP11B1 [R448H], [R448C]
Adrenoleukodystrophy	3 families	ALD [L229P]
Albinism, oculocutaneous	1:30	TYR [G47D]
Cerebrotendinous xanthomatosis	1:70	CYP27 [IVS4DS, G-A, +1]; [1-BP DEL, FS]
Chronic granulomatous disease	3 families	CYBA insertion G after G171 frameshift
Complement C7 deficiency	1:100	C7 [G357R]
Congenital dyserythropoietic anemia type II	1:100	SEC23B [E109K], [T710M]
Congenital insensivity to pain with anhidrosis	rare	NTRK1[c.207-208 delTG]
Cystinosis	1:100	CTNS [G339]
Deafness	1:50	TMC1 **[c.1939T>C];** [c.1810C>T] [c.1165C>T]; [c.1210T>C]
Dubin Johnson syndrome	1:100	MRP2 [R1150H]
Fanconi disease	1:100	FANCA [2172-2173insG]; [4275delT]
Glycogen storage disease III	1:35	AGL [FS1502TER]
GM1 gangliosidosis	3 families	GAA, [D441]
Monilethrix	1:100	DSG4 [R289X]
Progressive cerebello-cerebral atrophy type1	1:40	SepSecS [c.715G>A]
Progressive cerebello-cerebral atrophy type 2	1:37	VPS53 [c.2084A>G; c.1556+5G>A]
Wilson disease	3 families	ATP7B [H1069Q]

MOROCCO, one or 2 families

Acyl CoA dehydrogenase deficiency, short/branched chains	1 family	ACADSB [443C>T]
Afibrinogenemia congenital	1 family	unknown
Atrichia	1 family	HR [Q478X]
Brugada syndrome	1 family	SCN5A [G35S]; [R104Q]
Combined pituitary hormone deficiencies	1 family	PROP1 [R120C]
Dyggve-Melchior-Clausen syndrome	1 family	unknown
Enhanced S-cone[2]	Unknown	NR2E3 [c.923G>A]
Epilepsy and mental retardation limited to females	1 family	unknown
Familial Medullary thyroid carcinoma	2 families	RET [C618R]
Heriditrary motor neuropathy, distal	1 family	HSJ1 [c.352+1G>A]
Leber congenital amaurosis	1 family	CRB1 [620dlC]
Renal tubular acidosis, progressive sensorineural deafness	1 family	ATP6V1B1 [1155-1156 insC]
Seborrhea-like dermatitis with psoriasiform elements	1 family	ZNF750 [56_57 dupCC]
Usher syndrome type 1B		USH1B [2065del]
Usher syndrome type 2A	2 families	USH2A [c.1000C>T] [1], [c.5519G>T]

[1] found also in one Tunisian Jewish family, [2] founder Ashkenazi mutation

JEWS FROM IRAN AND IRAQ

Color blindness, total (achromatopsia)	unknown	GNGA3 [V529M] [1]
G6PD deficiency	1:4 males	G6PD
Growth hormone deficiency	unknown	GH [7.5 kb DEL] [2]
Microphthalmia/ anophthalmia	unknown	unknown
Myasthenia gravis, infantile	unknown	RAPSN [-38A-->G] [3]
Usher syndrome type 2A	1:25-1:50	USH2A [c.236-239dupGTAC] [4]
Pituitary dwarfism II (Laron)	unknown	GHR [ex 5,6 DEL][3]
Pseudocholinesterase deficiency (E1)	1:11	CHE1
Reticulosis, familial hystiocytic	unknown	unknown
Thalassemia, beta	1:6	HBB, many
Wilson disease	unknown	ATP7B [845delT]

[1] also found in Jews from Bukhara, and Afghanistan,
[2] also found in Jews from Morocco,
[3] also found in Jews from Yemen,
[4] also found in Jews from Afghanistan and Yemen and two non-Jewish Spanish families

JEWS FROM IRAQ

IRAQ - ASHKENAZI

Factor XI deficiency (PTA deficiency)	1:30	F11 [GLU117TER]

IRAQ

Deafness, high frequency	1:40	SYNE4 [c.228delAT]
Diarrhea intractable	5 families	unknown
Factor V and VIII deficiency	rare	LMAN1
Familial Mediterranean Fever	1:15	MEFV 40% [MET694VAL] ; 20% [VAL726ALA]
Monilethrix	1:25	DSG4 [P267R]
Pituitary dwarfism II (Laron)	unknown	GHR [W15X], [R11H]
Spondyloepimetaphyseal dysplasia	4 families	unknown
Progressive cerebellocerebral atrophy	1:40	SepSecS [c.715G>A]
Retinitis pigmentosa	Several	EYS [8218_8219delCA], [p.His2740TyrfsX27]
Thrombasthenia (Glanzmann)	1:40	GPIIIa [11 –BP DEL, EX 12], [11.2 Kb DEL]
Type III 3-Methyl glutaconic aciduria	1:10	OPA3 [G→C intron1 acceptor]
Usher syndrome type 2A	unknown	USH2A [c.2209C>T]
Xeroderma pigmentosum, variant	several	POLH [p.Trp174Cys]

IRAQ, one or 2 families

Acyl CoA dehydrogenase deficiency very long chains	1 family	VLCD [G637A]
Afibrinogenemia congenital	1 family	unknown
Atrichia, congenital	1 family	HR [c.1557-1G>T][1]
Camurati-Engelmann disease	1 family	TGFβ1
Cataracts	1 family	LIM2 [Phe105Val]
Choreoacanthocytosis	1 family	VPS13A [EX23del]
Comel-Netherton syndrome	1 family	SPINK5[c.691delC]
Deafness, autosomal recessive	1 family	GJB2 [L90P]
Dyskeratosis congenital	1 family	TERT [c.1892G>A]
Factor VII	1 family	F7 [ALA244VAL] [2]
Fechtner syndrome	1 family	unknown
Glutaric acidemia I	1 family	GCDH [G110R]
Myasthenia gravis, congenital Ic.	1 family	COLQ [G240X]
Niemann Pick type C	1 family	NPC1 [c.3673T>G], c.1241_2delTC]
Ornithine aminotransferase deficiency	1 family	OAT [deletion]
Peripheral sensory neuropathy	1 family	unknown
Pontocerebellar hypoplasia[3]	1 family	RARS [IVS2+5A>G]
APECED[4]	1 family	AIRE [Y85C]

[1] one of three alleles, the other two were of Iranian origin,
[2] frequent among Jews from Morocco and Iran,
[3] the parents were Iraqi/Syrian/Turkish
[4] Frequent among Iranian Jews

JEWS FROM IRAN

IRAN - MOROCCO

Factor VII	1:40	F7 [ALA244VAL] [1]

[1] also found in a Jewish family from Iraq

IRAN

Corticosterone methyl oxydase II deficiency	1:30	CYP11B2 [R181W;V386A]
Dubin Johnson syndrome	1:20	ABCC2 [p.I1173]
Factor V and VIII deficiency	unknown	LMAN1 [1-BP INS, 86G]
Familial Mediterranean Fever		MEFV [G632S] 10% alleles
Lu6 blood group	Unknown	Anti Lu
Mitochondrial myopathy, sideroblastic anemia	3 families	PUS1 [p.R144W]
Monilethrix	1:100	DSG4 [763delT]
Multiple cutaneous and uterine leiomyomata	4 families	FH [905-1G>A]
Myoneurogastrointestinal encephalopathy	Several families	TYMP [E298A]
Inclusion Body Myopathy	1:12	GNE [p.M7432T]
APCED	1:50	AIRE [Y85C]

IRAN one or 2 families

Abetalipoproteinemia	1 family	MTP [148-2A>G]
Adrenal hyperplasia	1 family	CYP11B1 [insGTG467]
Asparagine synthetase deficiency	2 families	ASNS [p.F362V]
Atrichia, congenital	1 family	HR [557-1G>T] [1]
Disphosphoglycerate mutase deficiency	1 family	BPGM [Arg62Gln]
Bloom syndrome	1 family	unknown
Cataracts, autosomal recessive	1 family	RYAA [W9X]
Dyskeratosis congenita	1 family	TERT [R901W]
Dystonia primary torsion, AR	1 family	unknown
Glycogen storage disease type II (Pompe disease)	1 family	GAA [G648P]
Myosin myopathy with external ophthalmoplegia	1 family	MYH2 [c.706G>A]
Peroxisomal biogenesis disorder	1 family	PEX12[c.102A>T]
Primary carnitine deficiency	1 family	SLC22A5 [R399Q]
Primary ciliary deficiency	1 kindred	MCIDAS c.1142G>A
Primary ciliary deficiency	1 kindred	DNAI2 [IVS11+1G>A]
Truncus arteriosus	1 family	PLXD1 [Arg1299Cys]
Wilson disease		ATP7B [845delT]
Xanthinuria	1 family	XDH [1658insC]

[1] two of three alleles, the other one was of Iraqi origin

JEWS FROM SYRIA

Renal tubular acidosis and deafness	1 family	ATP6V1B1 [1037C>G]
Retinitis pigmentosa*	2 families	FAM161A [c.1567C>T]
Proximal renal tubular acidosis	1 family	

*Also in individuals from North Africa

KURDISH JEWS

COXPD3	1 family	TSFM [C997T]
Erythrokeratodermia variabilis	1 family	CON30.3 [F137L]
Familial Mediterranean Fever	1:15	MEFV 40% [MET694VAL] ; 20% [VAL726ALA]
G6PD		
Glutaric academia I	1 family	GCDH [1173delG]
Thalassemia, beta	1:6	HBB, many

JEWS FROM DAGHASTAN (Caucasian)

Chronic granulomatous disease	Several families	NCF1 [579 G>A] [1]
Huntington disease	Several families	HD [expansion]
Infantile cerebral cerebellar atrophy	1:20	MED17 [L371P]
Juvenile open angle glaucoma	1 family	MYOC [Y371D]
Limb girdle muscular dystrophy	1:25	DYSF [2779delG]
McArdle disease	Several families	PYGM [c.632delG]
Mitochondrial complex 1 deficiency	1:25	NDUFS6 [c.344G>A]
Phenyl ketonuria	Unknown	PHA [E178G], [P281L], [L48S] [2]

[1] also found in Jews from Georgia and Azerbaijan, [2] also found in Tunisian Jews.

JEWS FROM UZBEKISTAN (Bukhara)

Acyl CoA dehydrogenase deficiency very long chains	3 families	VLCD [fst nt 826]
Bartter syndrome, infantile variant with sensorineuronal deafness		BSND [c.167ins6 [TTTCCC]]
Bernard Soulier syndrome	1 family	GPIalpha [Trp207Gly]
Color blindness, total (achromatopsia)	unknown	GNGA3 [V529M] [1]
Deafness, AR	unknown	GJB2 [51del112insA], [W24X]
Hereditary spastic paraparesis type 49	3 families	TECPR2 [c.3416delT]
Methylenetetrahydrofolate reductase deficiency	1:39	MTHFR [c.474A > T]
Oculopharyngeal Muscular Dystrophy	1:700	PABP2, expansion
Phenyl ketonuria	Unknown	PHA [E178G], [P281L], [L48S]
TRMT10A dysfunction	1 family	TRMT10A [Gly206Arg]
Usher syndrome type 2A	3 families	USH2A [c.12067-2A>G]
Wolman disease	1 family	LIPA [G60V]

[1] also found in Jews from Iran, Iraq and Afghanistan.

GEORGIAN JEWS

Acyl CoA dehydrogenase deficiency, short/branched chains	1 family	ACADSB [443C>T]
Albinism	1 family	TYR [c.74_75 insT]
Chronic granulomatous disease	2 families	NCF1 [c.579 G>A] [1]
Comel-Netherton syndrome	3 families	SPINK5[c.2557T; c.C649T]
Congenital disorder of glycolysation	2 families	TMEM165 [c.792+182G>A]
Polyarteritis nodosa	1:10	ADA2 [Gly47Arg]

[1] also found in Jews from Dagestan and Azerbaijan

JEWS FROM YEMEN

IRAQ – IRAN- YEMEN

Pituitary dwarfism II (Laron)	unknown	GHR [ex 5,6 Del]
Myasthenia gravis, infantile	unknown	RAPSN [-38A-->G]

YEMEN

Acute infantile liver failure	1:40	TRMU 75% [Y77H]
Breast/ovarian cancer (predisposition)	1:150	BRCA2 [8765delAG]
Calcinosis, tumoral normophosphatemic	1:1:27	SAMD9 [K1495E], [R344X]
Neutropenia, chronic familial	1:4	unknown
Machado-Joseph	unknown	SCA3, expansion
Metachromatic leukodystrophy	1:50	ARSA [PRO377LEU]
Myotonic dystrophy	1:6,000	DMPK, expansion
Phenylketonuria	1:35	PHA [EX3DEL]
Retinitis pigmentosa macular involvement	1:22	CERKL [c.238+1G>A]
Thalassemia, alpha	1:8	HBA [deletion]
Usher syndrome	1:119	USH1C [c.1220DelG]

YEMEN one or 2 families

AGAT deficiency	1 family	GAMT [c.1111_1112 insA]
Ataxia Telangiectasia	2 families	ATM [delA368]
Chronic granulomatous disease	1 family	CYBA [G71A]
Familial amyloid polyneuropathy	1 family	TTR [TYR 77]
Glycogenosis V, myophosphorylase deficiency, McArdle	2 families	PYGM [R270X]
Limb girdle muscular dystrophy LGMD2	1 family	DYSF [fst 5711]
Pseudohypoaldosteronism	1 family	SCNN1B [c.1612C>T]
Peroxisome biogenesis disorders	1 family	PEX6 [1715C-T]
Parkinson disease, juvenile	1 family	PARK2 [Ex 3 del]
Pituitary dwarfism II (Laron)	2 families	GHR [R271X]
Wilson disease	1 family	ATP7B [D765N]

BULGARIAN JEWS

Dysautonomia, familial		IKBKAP [IVS20+6T>C]; [R696P]
Oculopharyngeal Muscular Dystrophy	5 families	PABP2, expansion

JEWS FROM AZERBAIJAN

Comel-Netherton syndrome	1 family	SPINK5[c.2240+5G>A]]

INDIAN JEWS

ACTH deficiency	1 family	Tbox19 [IVS4+1G>A]
Factor XIII deficiency	1 family	FXIII [IVS11+1]
Fanconi anemia	2 families	FANCA [S858R]
Foveal hypoplasia	1:10	SLC38A8 [c.95T>G]
Papillon Lefevre syndrome (Cochin)	Several families	CTSC [2127A>G]

ETHIOPIAN JEWS

Factor XIII deficiency	1 family	FXIII [10bp deletion 1562-1661]
Familial cold autoinflamatory syndrome	1 family	CIAS1 [F525C]
Glucocorticoid deficiency	1 family	MRAP [L31X]

KARAITE JEWS

Color blindness, total (achromatopsia)	unknown	GNE [M712T] [1]
Huntington disease	unknown	HD [expansion]
San Filippo IIIA	1 family	SGSH [T271M]
Spinal Muscular Atrophy I	Several families	SMN [deletion]
Frontoparietal polymicrogyria	2 families	GPR56 [W349S]
Zelweger syndrome	Several families	PEX2 [550delC]

[1] mutation frequent among Jews from Iran