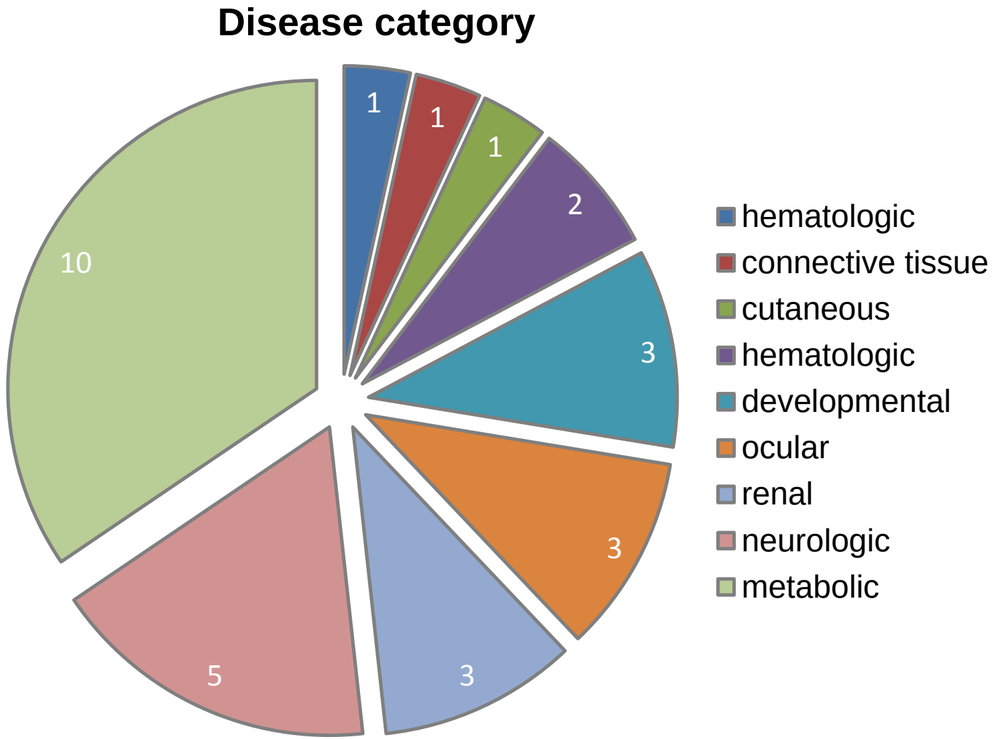
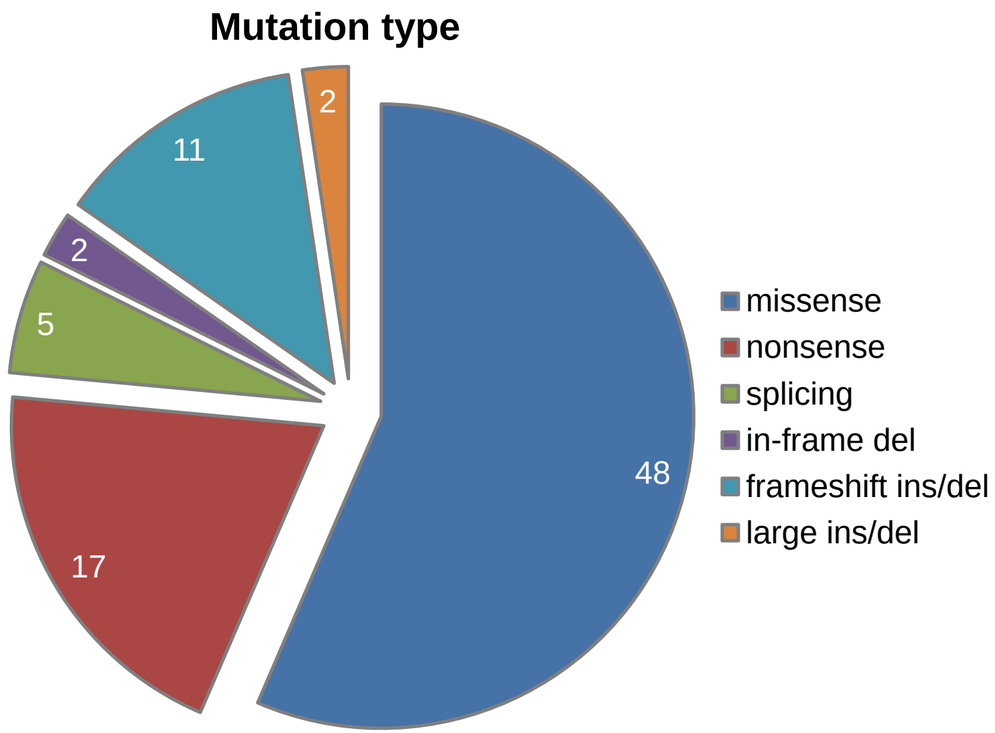
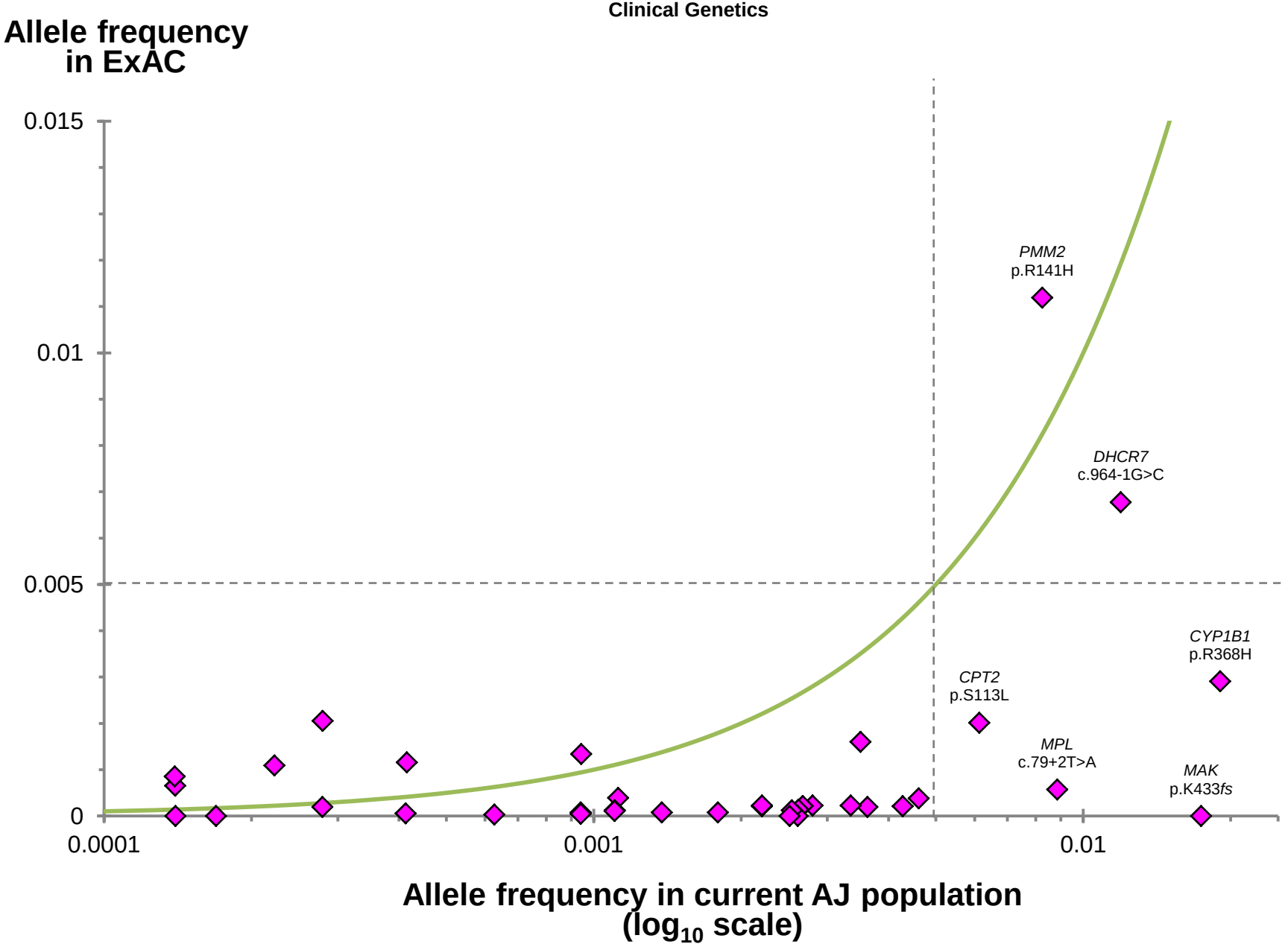
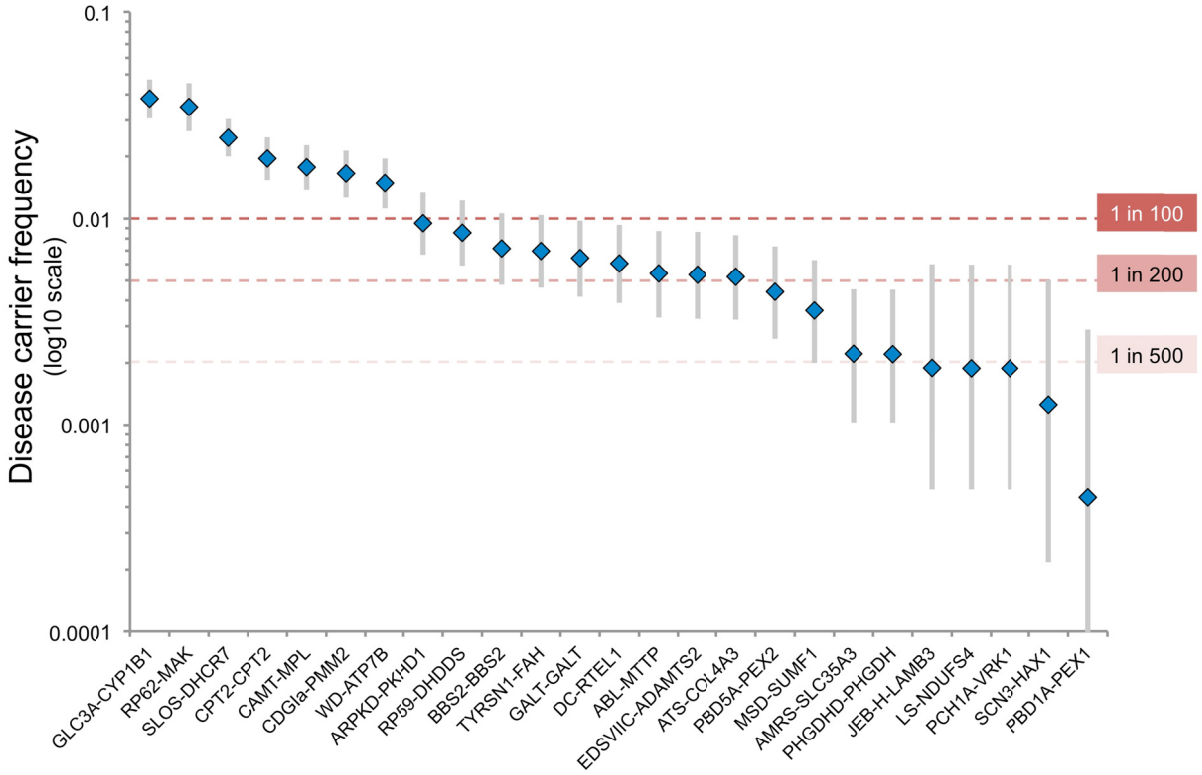
a


Disease category
1
1
1
2
3
3
3
5
10
hematologic
connective tissue
cutaneous
hematologic
developmental
ocular
renal
neurologic
metabolic

b


Mutation type
2
11
2
5
17
48
missense
nonsense
splicing
in-frame del
frameshift ins/del
large ins/del


Allele frequency in ExAC
0.015
0.01
0.005
0
0.0001
0.001
0.01
PMM2
p.R141H
DHCR7
c.964-1G>C
CYP1B1
p.R368H
CPT2
p.S113L
MPL
c.79+2T>A
MAK
p.K433fs
Allele frequency in current AJ population
(log10 scale)


Disease carrier frequency
(log10 scale)
0.1
0.01
0.001
0.0001
1 in 100
1 in 200
1 in 500
GLC3A-CYP1B1
RP62-MAK
SLOS-DHCR7
CPT2-CPT2
CAMT-MPL
CDG1a-PMM2
WD-ATP7B
ARPKD-PKHD1
RP59-DHDDS
BBS2-BBS2
TYRSN1-FAH
GALT-GALT
DC-RTEL1
ABL-MTTP
EDSVIIC-ADAMTS2
ATS-CCL4A3
PBD5A-PEX2
MSD-SUMF1
AMRS-SLC35A3
PHGDHD-PHGDH
JEB-H-LAMB3
LS-NDUFS4
PCH1A-VRK1
SCN3-HAX1
PBD1A-PEX1

Supplementary Table S1: Brief description for 29 conditions that have been tested in current study

Disease Name (abbrevation)	OMIM#	Gene	Inheritance	Disease category	Disease discription
Abetalipoproteinemia (ABL)	200100	*MTTP*	AR	hematologic	severe malabsorption of dietary fats and fat-soluble vitamins causing failure to thrive, diarrhea, blood abnormalities (acanthocytosis), and stool abnormalities (steatorrhea). Later in childhood symptoms include poor muscle coordination, ataxia, and retinitis pigmentosa.
Alport syndrome, autosomal recessive (ATS)	203780	*COL4A3*	AR	renal	progressive loss of kidney function (hematuria, proteinuria) resulting in end-stage renal disease, sensorineural hearing loss, and eye abnormalilties such as anterior lenticonus.
Alport syndrome, X-linked (ATS)	301050	*COL4A5*	XL-R	renal	
Arthrogryposis, mental retardation, and seizures (AMRS)	615553	*SLC35A3*	AR	neurologic	arthrogryposis, mental retardation, autism spectrum disorder, epilepsy, microcephaly, and hypotonia.
Bardet-Biedl syndrome 2 (BBS2)	209900	*BBS2*	AR	developmental	features include retinitis pigmentosa, obesity polydactyly, intellectual disability/ developmental delay, renal problems, anosmia, genital abnormalities, and male infertility. Other affected organs include the heart, liver and digestive system. There is variable age of onset and severity of symptoms.
Carnitine palmitoyltransferase II deficiency (CPT2)	255110;608836	*CPT2*	AR	metabolic	characterized by recurrent episodes of myalgia and rhabdomyolysis causing myoglobinuria which may be triggered by exercise, stress, exposure to extreme temperatures, infections, or fasting. The first episode usually occurs during childhood or adolescence. This can damage the kidneys, in some cases leading to life-threatening kidney failure.
Congenital amegakaryocytic thrombocytopenia (CAMT)	604498	*MPL*	AR	hematologic	pancytopenia, decreased bone marrow activity, and very low platelet counts.
Congenital disorder of glycosylation Ia (CDG1A)	212065	*PMM2*	AR	metabolic	hypotonia, abnormal fat distribution, strabismus, developmental delay, and failure to thrive appear in infancy. Other symptoms include elevated liver function tests, seizures, and pericardial effusion that could lead to death under 1 year of life due to multiple organ failure. Affected individuals who survive infancy may have intellectual disability, lethargy, temporary paralysis, neuropathy, kyphoscoliosis, ataxia, contractures and retinitis pigmentosa.
Congenital hydrocephalus (HYC1)	236600	*CCDC88C*	AR	neurologic	enlarged ventricles due to a disturbance of cerebrospinal fluid accumulation, onset in utero, moderate to severe intellectual disability
Dyskeratosis congenita, autosomal recessive (DC)	615190	*RTEL1*	AR	developmental	abnormally growing and poorly growing fingernails and toenails, pigmentary changes on neck and chest. Symptoms include bone marrow failure, aplastic anemia and increased risk for leukemia. Increased risk for cancers of the head, neck, anus, or genitals. Other features include pulmonary fibrosis, hair loss, osteoporosis, avascular necrosis of the joints, liver disease and short stature.
Ehlers-Danlos syndrome, type VIIC (EDSVIIC)	225410	*ADAMTS2*	AR	connective tissue	hypermobility, easy bruising, fragile skin, and blue sclera.
Galactosemia (GALT)	230400	*GALT*	AR	metabolic	feeding difficulties, lethargy, failure to thrive, jaundice, and bleeding within a few days after birth. Increased risk for sepsis and shock, developmental delay/intellectual disability, and cataracts. Managed by dietary restrictions.
Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (HHH)	238970	*SLC25A15*	AR	metabolic	neonatal to infantile onset, manifestations result from hyperammonemia, including chronic neurocognitive deficits, acute encephalopathy and livver dysfunction
Junctional epidermolysis bullosa, Herlitz type (JEB-H)	226700	*LAMB3*	AR	cutaneous	severe blistering presented at birth and may lead to significant granulation tissue. Mucosal involvement of the mouth, upper respiratory tract, esophagus, bladder, urethra, and corneas is also common. Renal and ureteral anomalies that can be seen.
Leigh syndrome (LS)	256000	*NDUFS4*	AR	neurologic	characterized by onset of symptoms typically between age three and 12 months, often following a viral infection. Decompensation during an intercurrent illness is typically associated with psychomotor retardation or regression. Neurologic features include hypotonia, spasticity, movement disorders, cerebellar ataxia, and peripheral neuropathy. Extraneurologic manifestations may include hypertrophic cardiomyopathy. High mortality rate by age three years, most often as a result of respiratory or cardiac failure.
Leigh syndrome (LS)	252010	*NDUFAF5*	AR	neurologic	
Multiple sulfatase deficiency (MSD)	272200	*SUMF1*	AR	metabolic	accumulation of sulfatides, sulfateglycosaminoglycans, sphingolipids, and steroid sulfates causing neurologic deterioration with mental retardation, skeletal anomalies, organomegaly, and ichthyosis
Severe congenital neutropenia, autosomal recessive (SCN3)	610738	*HAX1*	AR	hematologic	neutrophils deficiency at birth or soon after, recurrent infections, fevers, gingivitis. The disease is also associated with osteopenia and increased risk for leukemia or myelodysplastic syndrom.
3-Phosphoglycerate dehydrogenase deficiency (PHGDHD)	601815	*PHGDH*	AR	metabolic	microcephaly, psychomotor retardation, and seizures
Polycystic kidney disease, autosomal recessive (ARPKD)	263200	*PKHD1*	AR	renal	cyst development in the kidneys causes kidney enlargement and can lead to kidney failure. Symptoms include cysts in the liver, hypertension, hematuria, recurrent urinary tract infections, kidney stones, and an increased risk for aneurysms. This condition is often lethal early in life.
Pontocerebellar hypoplasia type 1A (PCH1A)	607596	*VRK1*	AR	neurologic	severe neurodegenerative disorders affecting growth and function of the brainstem and cerebellum, resulting in little or no development, central and peripheral motor dysfunction associated with anterior horn cell degeneration resembling infantile spinal muscular atrophy
Primary Congenital Glaucoma (GLC3A)	231300	*CYP1B1*	AR	ocular	characterized by elevated intraocular pressure , enlargement of the globe, edema, and opacification of the cornea with rupture of Descemet's membrane , thinning of the anterior sclera and iris atrophy, anomalously deep anterior chamber, and structurally normal posterior segment except for progressive glaucomatous optic atrophy. Symptoms usually occur in the first year of life.
Retinitis Pigmentosa (RP59)	613861	*DHDDS*	AR	ocular	childhood loss of night vision developing into peripheral blind spots and, later, leading to tunnel vision and blindness.
Retinitis Pigmentosa (RP62)	614181	*MAK*	AR	ocular	late-onset progressive, visual acuity is preserved but visual fields is severely affected.

Clinical Genetics

Smith-Lemli-Opitz syndrome (SLOS)	270400	*DHCR7*	AR	developmental	characteristic facial features, microcephaly, intellectual disability, and behavioral problems (e.g. autism). Abnormalities of the heart, lungs, kidneys, gastrointestinal tract, fingers/toes and genitalia are also common. Variable severity of symptoms.
Tyrosinemia type 1 (TYRSN1)	276700	*FAH*	AR	metabolic	tyrosine aminotransferase deficiency that can affect the eyes, skin, and mental development. Symptoms include photophobia, painful skin lesions on the palms and soles, and intellectual disability.
Wilson Disease (WD)	277900	*ATP7B*	AR	metabolic	copper accumulation in the liver (causing jaundice, fatigue, loss of appetite, and abdominal swelling), brain (causing nervous system and psychiatric problems), and eyes (causing Kayser-Fleischer rings and restricted ability to gaze upwards) with variable age of onset.
Peroxisome biogenesis disorder 1A (PBD1A)	214100	*PEX1*	AR	metabolic	demyelination of white matter causing hypotonia, feeding problems, hearing loss, vision loss, and seizures. Other affected organs include the liver, heart, kidneys, and bones and there is a shortened life expectancy.
Peroxisome biogenesis disorder 5A (PBD5A)	614866	*PEX2*	AR	metabolic	

Supplementary Table S2: 85 Candidate variants screened in current study and inclusion reasons

Disease[a]-gene	Chromosome location	Mutation nomenclature		Evidence[b]	Reference (PMID)
		cDNA	Protein		
ABL-*MTTP*	4q24	c.2593G>T	p.G865*	AJ;FD	17275380
		c.2212delT	p.S738*fs*	AJ	17275380
ATS-*COL4A3*	2q36.3	c.40_63del	p.L14_L21del	AJ;FQ;FD	23927549
ATS-*COL4A5*	Xq22	c.5030G>A	p.R1677Q	AJ;FD	9150741
AMRS-*SLC35A3*	1p21	c.886A>G	p.S296G	AJ;FD;FQ	24031089
		c.514C>T	p.Q172*	AJ;FD	24031089
BBS2-*BBS2*	16q12.2	c.311A>C	p.D104A	AJ;FQ	23829372
		c.1895G>C	p.R632P	AJ;FQ	23829372
CPT2-*CPT2*	1p32	c.338C>T	p.S113L	AJ	10090476
		c.1239_1240delGA	p.Q413*fs*	AJ	10090476;11477613
		c.370C>T	p.R124*	RC	9562964
		c.110_111dupGC	p.S38*fs*	AJ	12410208
CAMT-*MPL*	1p34.2	c.79+2T>A		AJ;FQ;FD	21489838
CDG1A-*PMM2*	16p13.2	c.338C>T	p.P113L	RC	9140401
		c.357C>A	p.F119L	RC	9140401
		c.422G>A	p.R141H	RC	9140401
		c.691G>A	p.V231M	RC	9140401
HYC1-*CCDC88C*	14q32.1	c.934C>T	p.R312*	AJ	23042809
DC-*RTEL1*	20q13.3	c.3791G>A (uc021wge.1)[c]	p.R1264H (uc021wge.1)[c]	AJ;FQ;FD	24009516
		c.1548G>T	p.M516I	AJ	23959892
		c.2941C>T	p.R981W	RC	23453664
		c.2992C>T	p.R998*	AJ	23959892
		c.2288G>T	p.G763V	AJ	23453664
EDSVIIC-*ADAMTS2*	5q35.3	c.673C>T	p.Q225*	AJ	18973246
		c.2384G>A	p.W795*	AJ	10417273
GALT-*GALT*	9p13.3	c.-1039_+789del5573ins129		AJ;FQ	21059483
		c.253-2A>G		RC	11754113
		c.404C>T	p.S135L	RC	11754113
		c.563A>G	p.Q188R	RC	11754113

		c.413C>T	p.T138M	RC	11754113
		c.512T>C	p.F171S	RC	9635294
		c.584T>C	p.L195P	RC	1373122
		c.626A>G	p.Y209C	RC	10408771
		c.855G>T	p.K285N	RC	11754113
HHH-*SLC25A15*	13q14.1	c.562_564delTTC	p.F188del	AJ,RC	2013 ACMG meeting
JEB-H-*LAMB3*	1q32.2	c.2528delA	p.N843*fs*	AJ	16439963
		c.124C>T	p.R42*	AJ	16439963
		c.1903C>T	p.R635*	RC	8824879
		c.727C>T	p.Q243*	RC	11023379
LS-*NDUFS4*	5q11.2	c.462delA	p.K154*fs*	AJ;FQ	19107570
		c.355G>C	p.D119H	AJ	19364667
LS-*NDUFAF5*	20p12.1	c.749G>T	p.G250V	AJ	21607760
MSD-*SUMF1*	3p26.1	c.463T>C	p.S155P	AJ;RC	unpublished data
SCN3-*HAX1*	1q21.3	c.125dupG	p.S43*fs*	AJ;FD	19036076
PHGDHD-*PHGDH*	1p12	c.1468G>A	p.V490M	AJ;FQ	11751922
ARPKD-*PKHD1*	6p12.2	c.3761_3762delCCinsG	p.A1254*fs*	AJ	unpublished data
		c.107C>T	p.T36M	RC	12506140
		c.1486C>T	p.R496*	RC	12506140
		c.5895dupA	p.L1966*fs*	RC	19914852
		c.10412T>G	p.V3471G	RC	12506140
		c.9689delA	p.D3230*fs*	RC	19940839
PCH1A-*VRK1*	14q32.2	c.1072C>T	p.R358*	AJ;FD;FQ	19646678
GLC3A-*CYP1B1*	2p22.2	c.1103G>A	p.R368H	AJ	21168818
RP59-*DHDDS*	1p36.11	c.124A>G	p.K42E	AJ;FD;FQ	21295282
RP62-*MAK*	6p24	c.1297ins(Alu)	p.K433fs	AJ;FD;FQ	22110072
		c.497G>A	p.R166H	AJ	21835304
SLOS-*DHCR7*	11q13.4	c.964-1G>C		AJ;RC	15776424
		c.452G>A	p.W151*	RC	9653161
		c.453G>A	p.W151*	RC	17965227
		c.1A>G	p.M1V	AJ	15776424
		c.976G>T	p.V326L	RC	17965227

		c.278C>T	p.T93M	RC	17965227
		c.1055G>A	p.R352Q	RC	10677299
		c.1054C>T	p.R352W	RC	17965227
		c.1210C>T	p.R404C	RC	10677299
		c.506C>T	p.S169L	RC	10677299
		c.724C>T	p.R242C	RC	10677299
		c.725G>A	p.R242H	RC	17965227
		c.906C>G	p.F302L	RC	17965227
		c.1228G>A	p.G410S	RC	17965227
		c.1342G>A	p.E448K	RC	10602371
TYRSN1-*FAH*	15q25.1	c.782C>T	p.P261L	AJ	11754109
		c.554-1G>T		RC	11754109
		c.1062+5G>A		RC	8829657
		c.192G>T	p.Q64H	RC	7942842
		c.786G>A	p.W262*	RC	8829657
		c.1069G>T	p.E357*	RC	8829657
WD-*ATP7B*	13q14.3	c.3191A>C	p.E1064A	AJ	9482578
		c.3207C>A	p.H1069Q	AJ	9482578
		c.2333G>T	p.R778L	RC	7626145
		c.1934T>G	p.M645R	AJ	9482578
PBD1A-*PEX1*	7q21.2	c.2528G>A	p.G843D	RC	11389485
	7q21.2	c.2097dupT	p.I700*fs*	RC	11389485
	7q21.2	c.2916delA	p.G983*fs*	RC	21031596
PBD5A-*PEX2*	8q21.11	c.355C>T	p.R119*	AJ;FQ	23590336

[a]See Supplementary Table S1 for disease abbreviations. [b]Evidence code: AJ, reported in Ashkenazi Jewish individual(s); FD, founder effected suggested; FQ, population frequency available; RC, recurrent in other ethnic group(s). [c]cDNA and protein coding number based on the transcript isoform uc021wge.1.

Supplementary Table S3: PCR primer information for mutations screened by Luminex assay

Disease-gene	Mutation	PCR primer sequence (5' to 3')		Primer working concentration (μM)	Amplicon size (bp)
		Forward	Reverse		
ABL-*MTTP*	p.G865*	AGCCTTTGACACTCATATCCTTTCTCA	CAGAGAGCAAGTACTTAGTCATGCCACA	1.5	384
	p.S738*fs*	TTTTCAACGGATACAGTGATTTGAT	CTCCAATTAAAGGCACTTATTAATCATC	1.5	319
AS-*COL4A5*	p.R1677Q	GGATCTGATTGTCTTATTTCTTATTTCC	AGGTACATCTGAATCTTCATGGTTACTA	1.5	282
CPT2-*CPT2*	p.S113L	TTTTCCTAATCATTCATTTTAAGAAGGT	AGGAGAGAATATGCCTTACTTTTCATT	5.0	389
	p.Q413*fs*	GCAGTGTTCTGTCTCTGCCTAGAT	AGTCAATAGTGAGGGTTTTCATGGT	1.5	373
CAMT-*MPL*	c.79+2T>A	CCTGAAGGGAGGATGGGCTA	GAGCCTCTTACTCTACCTCCCC	1.5	221
EDSVIIC-*ADAMTS2*	p.Q225*	TCATCGAACCCTTGGAGAAGGGGCTGG	GGAGAGCTGCCTGAGTCTCTGGGATGC	1.5	210
	p.W795*	AGCCGTCAAGAACCTGGAGACAGGCA	CAGCCCTGCTTGGACTCACCAGAACG	1.5	188
JEB-H-*LAMB3*	p.N843*fs*	ACTTTAGTGTAAAATGGGAACTCTGACT	CTTTGGGTAGCCTCTTTAACACC	1.5	329
	p.R42*	CCAAGTACATTCTAAGTATTCAGTGGAC	AAGGATATAATCTTGCTAATGTCAGCTT	1.5	349
	p.R635*	TAATGCACTGTACAAATGTAAGGAAGGA	ACCAGTGCCAGCGAGGCTAC	1.5	380
	p.Q243*	TTAAATACCCAAGTCTTAGATTTAGTGC	CTAACTTCCTTTCTTTATTTTCCATTCT	1.5	373
LS-*NDUFS4*	p.K154*fs*	CTTTTCAGGTATCCTCTTTAATTCTGTT	TATTCACAGTCAAGCAGAGATATAGTCA	1.5	200
	p.D119H	ATTTAACACAGAAAAAGGTATTCCAACT	AAACCACTGGATTAGATACAATGAAAC	4.0	377
LS-*NDUFAF5*	p.G250V	CACAATGCAGGATATTTAATCAGC	CTACCTTTCCCAATGGAGTCTAAA	2.0	210
SCN3-*HAX1*	p.S43*fs*	AGCCATTGTATTAATGTTTTGATGTG	TACTAGGTCATCAAAGCCGAAGTTAT	1.5	358
PHGDHD-*PHGDH*	p.V490M	ATCACAAAAGCTTTGAACTTCTGATT	GATTTTCTCTCCCTATTGATCACAGT	5.0	327
ARPKD-*PKHD1*	p.A1254*fs*	GCTATTTGTGATTTCTCCTTGCAT	AATATTTTGCTTTCCGTATTCATACTTT	1.5	341
	p.T36M	ACTATGCTACACACTTCCATATCATCTC	AAATATTGCTTAAAATATTGCAGAAGGT	4.0	442
	p.R496*	AATCTCAGGAATAAGACATCACATTTTT	TACTACCTGGAAGCAGAGCATCAT	1.5	346
	p.L1966*fs*	CTGAGGATAAATTGTCAACTAAGTTCTG	ATACTTATACTATCCCGCCCAAAAAC	4.0	403
	p.D3230*fs*	AGAAATTATAAACTACCATTGTGGCTATC	TGGAGATAGAGAACATTACTCTGGTAGA	1.5	498
	p.V3471G	AAGAGGTTATCCATGATGTTGAAATAGT	CTTGTTTTGCTTCTTTTCTTTTATAGGT	1.5	498
PCH1A-*VRK1*	p.R358*	CATCCATTCCTTTAGGTAACTACTATCC	CCTCCTCTGTCTGTGTGTTTGA	1.5	251
RP59-*DHDDS*	p.K42E	TTACTGAGTTAAAAGCACAAAGTACCC	GGTTTTCTAGCTATAATCTACCTCTCCA	1.5	327
RP62-*MAK*	p.R166H	AAAAGTTATAGCAACTTAGGGACAAGAG	GGGAACATGTGTTGTGTCCTATTAT	1.5	389
SLOS-*DHCR7*	c.964-1G>C	GATGTGTAGGAGCACTCGATGAC	AGGTAGAAGGCAGGTAGAGTTACTCATT	1.5	393
	p.W151* (c.452G>A)	GTGAAGTTTGCACTTTCTACATCAG	TATAGGGGCCTCAGAGATGGT	1.5	424
	p.W151* (c.453G>A)	GTGAAGTTTGCACTTTCTACATCAG	TATAGGGGCCTCAGAGATGGT	1.5	424
	p.M1V	CTCTGAGACCACACTTTACTTTCTAGC	CAAGGAAGAGAGTCTAAGACAAGATGAT	1.5	340
	p.V326L	GATGTGTAGGAGCACTCGATGAC	AGGTAGAAGGCAGGTAGAGTTACTCATT	1.5	393
	p.T93M	CTTACAGCTGAGGATGAATCTGAAC	GGAGGTGGACTGGTTTTCACT	1.5	401
TYRSN1-*FAH*	p.P261L	CTGATCAGCCTTTGTAAGTCCTG	AGCTTCCCTCCTGATGGTCT	1.5	281

Supplementary Table S4: ASPE primer information for mutations screened by Luminex assay

Disease-gene	Mutation	ASPE primer (5' to 3')	Bead Tag ID	Primer working concentration (μM)
ABL-*MTTP*	p.G865*	WT: GATGGAGCGGGAATTCACATCC	LUA_10	0.25
		MUT: GATGGAGCGGGAATTCACATCA	LUA_28	0.25
	p.S738*fs*	WT: TGAAAGGACTTATTCTGCTAATAGATCATT	MTAG-B049	0.25
		MUT: TGAAAGGACTTATTCTGCTAATAGATCATC	MTAG-A026	0.25
AS-*COL4A5*	p.R1677Q	WT: ACTTGAGGACACGAATTAGCCG	MTAG-B098	0.25
		MUT: ACTTGAGGACACGAATTAGCCA	MTAG-A019	0.25
CPT2-*CPT2*	p.S113L	WT: AGCCCAGCCTACCTACCCG	MTAG-A063	0.25
		MUT: AGCCCAGCCTACCTACCCA	MTAG-A039	0.25
	p.Q413*fs*	WT: TCAGTCAGCTCGAAGTTGAGTTTCT	MTAG-B095	0.25
		MUT: TCAGTCAGCTCGAAGTTGAGTTTGC	MTAG-A054	0.25
CAMT-*MPL*	c.79+2T>A	WT: CCAAGTCAGCAGCCAAGGT	MTAG-A037	0.25
		MUT: CCAAGTCAGCAGCCAAGGA	MTAG-A053	0.25
EDSVIIC-*ADAMTS2*	p.Q225*	WT: TCACCTGTGTCCAGGGCCTG	LUA_24	0.25
		MUT: TCACCTGTGTCCAGGGCCTA	LUA_37	0.25
	p.W795*	WT: ATTGCCATGGGCGTGGAGTG	LUA_29	0.25
		MUT: ATTGCCATGGGCGTGGAGTA	LUA_59	0.25
JEB-H-*LAMB3*	p.N843*fs*	WT: CTGAGCAGCTGCGGGGCTTCAA	MTAG-A039	0.25
		MUT: CTGAGCAGCTGCGGGGCTTCAT	MTAG-A074	0.25
	p.R42*	WT: TTGGGAGGACCCGGTTTCTCC	MTAG-A028	0.25
		MUT: TTGGGAGGACCCGGTTTCTCT	MTAG-A027	0.25
	p.R635*	WT: GGCTGCTGAGAACTGCTCG	MTAG-A012	0.25
		MUT: GGCTGCTGAGAACTGCTCA	MTAG-A057	0.25
	p.Q243*	WT: AGCGCCTACTATGCTGTGTCCC	MTAG-A054	0.25
		MUT: AGCGCCTACTATGCTGTGTCCT	MTAG-A072	0.25
LS-*NDUFS4*	p.K154*fs*	WT: GACATTGAAGAGAGGAAGGTTCCAAAA	MTAG-A048	0.25
		MUT: GACATTGAAGAGAGGAAGGTTCCAAAC	MTAG-A045	0.25
	p.D119H	WT: GGTTAGAACCATGTTGGATAAGGGATC	MTAG-B080	0.25
		MUT: GGTTAGAACCATGTTGGATAAGGGATG	MTAG-B097	0.25
	p.G250V	WT: CACTGATGAAATTCAAGTTAACTATCCTGG	MTAG-A078	0.25
		MUT: CACTGATGAAATTCAAGTTAACTATCCTGT	MTAG-A075	0.25
SCN3-*HAX1*	p.S43*fs*	WT: AGGAAGAAGAAGAAGAAGGGGGC	MTAG-A077	0.25
		MUT: AAGATGATGATGAGGAAGAAGAAGAAGAAGGGGGG	MTAG-A019	0.25
PHGDHD-*PHGDH*	p.V490M	WT: TCCTGGCAGAGGCAGGCG	MTAG-A064	0.25
		MUT: TCCTGGCAGAGGCAGGCA	MTAG-A073	0.25
ARPKD-*PKHD1*	p.A1254*fs*	WT: GCATCGGGTATCTGGGGGG	MTAG-A051	0.25
		MUT: GCATCGGGTATCTGGGGCC	MTAG-A052	0.25
	p.T36M	WT: CCATCAAAAATGACTGTGATCCACG	MTAG-A025	0.25
		MUT: CCATCAAAAATGACTGTGATCCACA	MTAG-A015	0.25
	p.R496*	WT: AGAAGCACCAGATCCGAGTCC	MTAG-A022	0.25
		MUT: AGAAGCACCAGATCCGAGTCT	MTAG-A062	0.25
	p.L1966*fs*	WT: CTTCACACACCTTTAATGTGCAGTA	MTAG-A055	0.25
		MUT: CTTCACACACCTTTAATGTGCAGTT	MTAG-A066	0.25
	p.D3230*fs*	WT: GCACTCAGCCAACTTGACATCAACAGA	MTAG-A053	0.25
		MUT: GCACTCAGCCAACTTGACATCAACAGT	MTAG-A037	0.25
	p.V3471G	WT: CCCATCAGGCAAATCACCAAAGT	MTAG-A067	0.25
		MUT: CCCATCAGGCAAATCACCAAAGG	MTAG-A065	0.25
PCH1A-*VRK1*	p.R358*	WT: GGTTCCTTGCTTTCTTCAATTTCTTTCTTTCG	MTAG-A043	0.25
		MUT: GGTTCCTTGCTTTCTTCAATTTCTTTCTTTCA	MTAG-A044	0.25
RP59-*DHDDS*	p.K42E	WT: GGGAACCGTCGCTATGCCAAGA	MTAG-B097	0.25
		MUT: GGGAACCGTCGCTATGCCAAGG	MTAG-A078	0.25
RP62-*MAK*	p.R166H	WT: CTGAAGATCTCAGTAAAACTTCAGGGGCAC	MTAG-A067	0.25
		MUT: CTGAAGATCTCAGTAAAACTTCAGGGGCAT	MTAG-A063	0.25
SLOS-*DHCR7*	c.964-1G>C	WT: TTCCCCCTCGCCCCCCAG	MTAG-A044	0.25
		MUT: TTCCCCCTCGCCCCCCAC	MTAG-A043	0.25
	p.W151* (c.452G>A)	WT: TATCAGATCAACGGCCTGCAAGCCTG	MTAG-A048	0.25
		MUT: TATCAGATCAACGGCCTGCAAGCCTA	MTAG-A036	0.25
	p.W151* (c.453G>A)	WT: AGAGCAGGTGCGTGAGGAGC	MTAG-B087	0.25
		MUT: GCAAACCAGAGCAGGTGCGTGAGGAGT	MTAG-A022	0.25
	p.M1V	WT: GTTCTCTTTCTTGCAGGGCCCAA	MTAG-A051	0.25
		MUT: GTTCTCTTTCTTGCAGGGCCCAG	MTAG-B095	0.25
	p.V326L	WT: ACAGCTGCACGGGGTGGTACAC	MTAG-A075	0.25
		MUT: ACAGCTGCACGGGGTGGTACAA	MTAG-A061	0.25
	p.T93M	WT: ATCTGGGCCAAGACTCCACCTATAAC	MTAG-A077	0.25
		MUT: ATCTGGGCCAAGACTCCACCTATAAT	MTAG-B082	0.25
TYRSN1-*FAH*	p.P261L	WT: AAGAGTTTTGGGACCACTGTCTCTCC	MTAG-A025	0.25
		MUT: AAGAGTTTTGGGACCACTGTCTCTCT	MTAG-A015	0.25

Supplementary Table S5: PCR primer and extension probe information for mutations screened by Agena Bioscience™ assay

Disease-gene	Mutation	PCR primer (5' to 3')		Primer working concentration (μM)	Amplicon size (bp)	Universal extension probe (5' to 3')	Probe working concentration (μM)
		Forward	Reverse				
ATS-*COL4A3*	p.L14_L21del	ACGTTGGATGGACCGAGCCCTACAAAACC	ACGTTGGATGGTGGAGGAGGGATGGAAGTG	1.0	302	CCCAGGCCGCAGGTGCTCCTGG	15.0
AMRS-*SLC35A3*	p.S296G	ACGTTGGATGCTCCTATTTTTGGCTTCAAG	ACGTTGGATGATCAGATCACCTTACAAAG	0.5	133	GGCTTCAAGATTTTGTGCCAACC	15.0
	p.Q172*	ACGTTGGATGCCTCAGATTCTCAGCTTGAT	ACGTTGGATGAAAACATGCTGTGAGAACTG	0.5	103	ACTTTCAGCTGGTTCT	7.0
BBS2-*BBS2*	p.D104A	ACGTTGGATGCTATGATGCCCTTTTAGTGG	ACGTTGGATGCTCTCTGTAGAACAAATCCG	0.5	102	GGGACACAGACTAATCTTTTGGCTTATG	15.0
	p.R632P	ACGTTGGATGGCACCTGTACTAACCATGAC	ACGTTGGATGTGATCCGAAGTTTGCTGGTC	0.5	106	TCACATGTCCCTCATCAGA	5.0
CPT2-*CPT2*	p.S113L	ACGTTGGATGGGAGAAATCATACCCACAGC	ACGTTGGATGGCTGGTTGCTCTGGACAAAC	0.5	100	AGCCCAGCCTACCTACCC	5.0
	p.Q413*fs*	ACGTTGGATGTAGCAGCTGTGATGCCAGTC	ACGTTGGATGACCACTGACTCTACTGTCAC	0.5	99	CAGTCAGCTCGAAGTTGAGTTT	10.0
	p.R124*	ACGTTGGATGGGGTCAGGATTGAAAGCCAT	ACGTTGGATGTAGGACCCTGGTTTGATATG	0.5	105	AAAGTTCAGAACAACGGAGTCTC	10.0
	p.S38*fs*	ACGTTGGATGTGTCCTGGTAGTGCATGGTG	ACGTTGGATGTTGGTCCGGGAGCCCCCAGT	0.5	116	GTGGGCACGATGCTGCGC	7.0
CAMT-*MPL*	c.79+2T>A	ACGTTGGATGCTCAAAACCTGGCCCAAGTC	ACGTTGGATGcTCCCTCTTCCTGGGCATAG	0.5	97	ACCTGGCCCAAGTCAGCAGCCAAGG	15.0
CDG1A-*PMM2*	p.P113L	ACGTTGGATGAGCCCACCATGTGACACTAC	ACGTTGGATGGTGAGGCCCTAATCCAAGAT	0.5	154	AAACCCACCTCTTCTTC	5.0
	p.F119L	ACGTTGGATGCGTACTTTATCGAGTTCGTAG	ACGTTGGATGTGTCACCCTTTCATTCCCAG	1.0	145	CATCCCATTTCGGAATTCAAT	10.0
	p.R141H	ACGTTGGATGCGTACTTTATCGAGTTCGTAG	ACGTTGGATGTGTCACCCTTTCATTCCCAG	1.0	145	TTTATCGAGTTCGTAGAACTCAATG	15.0
	p.V231M	ACGTTGGATGATCTTCACAGACCCCAGAAC	ACGTTGGATGGAACAGCAGTTCACAGATCC	0.5	98	ACCCCAGAACCATGGGCTACTCC	15.0
HYC1-*CCDC88C*	p.R312*	ACGTTGGATGTTCTCCCGCAGGGAATCCAG	ACGTTGGATGTGTCCACCTTGCAGAACATC	0.5	105	CCCGCAGGGAATCCAGCTCGTCTC	15.0
DC-*RTEL1*	p.R1264H	ACGTTGGATGTTCCAGTGCCCTGCCTGTGA	ACGTTGGATGACCCAGCAGGAACTGTAGTG	0.5	113	GCCTGTGACTTCCAGC	5.0
	p.M516I	ACGTTGGATGTGATGTCAGAGGTGTCACCG	ACGTTGGATGCGGTGTCCTCCTTTGCTCT	0.5	102	AGGGGTGGCCCGTACATCTG	10.0
	p.R981W	ACGTTGGATGTTCCTCTACAGGCTTCTACC	ACGTTGGATGCGGCTGTGCCCGCTGCCTT	1.0	156	AGGCTTCTACCAGTTTGTG	10.0
	p.R998*	ACGTTGGATGTTCCTCTACAGGCTTCTACC	ACGTTGGATGCGGCTGTGCCCGCTGCCTT	1.0	156	TGTATCCAGCTGACAGGA	5.0
	p.G763V	ACGTTGGATGCAGCAGATGAGGGTCTTCACTT	ACGTTGGATGTCGGCAACACGGAAGAACTG	0.5	220	ACTGGGCCACGTCTCGGATGACATGG	10.0
GALT-*GALT*	5kb del	ACGTTGGATGTGGAGCTTCTGATAGTCCTG	ACGTTGGATGACAGAGCAGACTCCGTCACA	0.5	105	CTGATAGTCCTGTACTCTCCAC	10.0
	c.253-2A>G	ACGTTGGATGGTGAGTGCTTCTAGCCTATC	ACGTTGGATGCTGGGAAGTCGTTGTCAAAC	0.5	100	AGCCTATCCTTGTCGGT	5.0
	p.S135L	ACGTTGGATGCCAGTAAGGTCATGTGCTTC	ACGTTGGATGAACAACAGCCCGGATCTCAG	0.5	100	AGTAAGGTCATGTGCTTCCACCCCTGGT	15.0
	p.Q188R	ACGTTGGATGcCACTGGAGCCCCTGACACC	ACGTTGGATGCTTGGTGTCTTTTGGCTAAC	0.5	147	CCCCTGACACCCTTACC	3.5
	p.T138M	ACGTTGGATGCCAGTAAGGTCATGTGCTTC	ACGTTGGATGAACAACAGCCCGGATCTCAG	0.5	100	GACATGAGTGGCAGC	5.0
	p.F171S	ACGTTGGATGcCACTGGAGCCCCTGACACC	ACGTTGGATGCTTGGTGTCTTTTGGCTAAC	0.5	147	TCATGGCACCTTTGTTTTCA	5.0
	p.L195P	ACGTTGGATGcTTGACAGGTATGGGCCAGC	ACGTTGGATGCTGGCGGCTGTACTCCATTA	0.5	136	ATGGGCCAGCAGTTTCC	5.0
	p.Y209C	ACGTTGGATGcTTGACAGGTATGGGCCAGC	ACGTTGGATGCTGGCGGCTGTACTCCATTA	0.5	136	ctctCGATCTCAGCAGGCCT	10.0
	p.K285N	ACGTTGGATGCCTCCATCATGAAGAAGCTC	ACGTTGGATGTTGAAAAGCCTCACCATGCC	0.5	112	GAAGAAGCTCTTGACCAA	5.0
HHH-*SLC25A15*	p.F188del	ACGTTGGATGTACCATGGACTCTCAAGCAC	ACGTTGGATGAAAAAGGACCGGCTCAGTTC	0.5	106	AGTACCAGGCTATTTCTTCTTC	15.0
MSD-*SUMF1*	p.S155P	ACGTTGGATGCCTACCTTCCTTATAGGCTG	ACGTTGGATGTGGTCTTCACTTGCTCACTC	0.5	97	TAGGCTGAGAAGTTTGGCGAC	10.0
PHGDH-*PHGDH*	p.V490M	ACGTTGGATGACACCAGTGAAGTCTGGTAG	ACGTTGGATGTGCCAACCAGGAGTTTCTTC	0.5	103	GTAGGACAGCAGCCGCA	5.0
ARPKD-*PKHD1*	p.A1254*fs*	ACGTTGGATGAAGAACCTGTTGCCAGCCCA	ACGTTGGATGGAGCATCTGGTGTGAAACCC	0.5	119	GCATCGGGTATCTGGGG	5.0
	p.T36M	ACGTTGGATGTGTGCACTTGGTAAAACCCC	ACGTTGGATGACCTGAAGAAGGTAGCCTTG	2.0	100	ACCATCAAAAATGACTGTGATCCAC	15.0
	p.R496*	ACGTTGGATGTGTGGTCACCACTTACCTAC	ACGTTGGATGACACAGACCTGTACTTCTGG	0.5	98	CGGGAGAAGCACCAGATCCGAGTC	10.0
	p.L1966*fs*	ACGTTGGATGATGGCCAATTGCTTCTGCTG	ACGTTGGATGACCTACAAACTTCACACACC	0.5	99	GACACTAACACAAGCATCCTCAACTTA	10.0
	p.V3471G	ACGTTGGATGCCCCAATAGAAAAAAGCGCA	ACGTTGGATGTTCTATCTTACCCATCAGGC	0.5	102	TGAGGAGTTTGATCCATGAAGCAG	10.0
	p.D3230*fs*	ACGTTGGATGTTCAGGACAAAGTGAAGCCG	ACGTTGGATGTTCGACCTCCTCTTGGATTG	0.5	98	CAGCCAACTTGACATCAACAG	7.0
GLC3A-*CYP1B1*	p.R368H	ACGTTGGATGAGAATTGGATCAGGTCGTGG	ACGTTGGATGAAAGGAAGGCCAGGACATAG	0.5	103	GGATCAGGTCGTGGGGAGGGACC	10.0
RP59-*DHDDS*	p.K42E	ACGTTGGATGCATTCATAATGGACGGGAAC	ACGTTGGATGTTGTTGAAGCCCTGTGAGTG	0.5	102	TGGACGGGAACCGTCGCTATGCCAAG	15.0
SLOS-*DHCR7*	c.964-1G>C	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	0.5	520	TTCCCCCTCGCCCCCCA	5.0
	p.W151* (c.452G>A)	ACGTTGGATGTCCAGTTGTCGAAGATGATG	ACGTTGGATGtGTAACAAGTATCAGATCA	0.5	136	AGAGCAGGTGCGTGAGGAGC	7.5
	p.W151* (c.453G>A)	ACGTTGGATGTCCAGTTGTCGAAGATGATG	ACGTTGGATGtGTAACAAGTATCAGATCA	0.5	136	tgcacACCAGAGCAGGTGCGTGAGGAG	15.0
	p.M1V	ACGTTGGATGTGACGCCATCTAGACTCTTG	ACGTTGGATGCAAGGTTCTCTTTCTTGCAG	0.5	101	GGGTTGCGATTTTGCAGCCA	5.0
	p.V326L	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	2.0	520	ggcatCTGCACGGGGTGGTACA	10.0
	p.T93M	ACGTTGGATGgGACATGCTCGGCTCTCGGA	ACGTTGGATGACCCACAAGGTATAGAGCTG	0.5	100	CCAAGACTCCACCTATAA	3.5
	p.R352Q	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	0.5	520	TGGGCTACTACATCTTCC	5.0
	p.R352W	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	0.5	520	GGTGGGCTACTACATCTTC	5.0
	p.R404C	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	2.0	520	GGGCTTCTGGGGCGTGGCC	7.5
	p.S169L	ACGTTGGATGtGTGAACAAGTATCAGATCA	ACGTTGGATGTCCAGTTGTCGAAGATGATG	0.5	136	TCATCTCCTGTCCTGGTTCT	5.0
	p.R242C	ACGTTGGATGGAAGTGGTTTGACTTCAAGC	ACGTTGGATGCTGCGAAGGACAGGTTGATG	0.5	100	GACTTCAAGCTGTTCTTCAATGGG	10.0
	p.R242H	ACGTTGGATGGAAGTGGTTTGACTTCAAGC	ACGTTGGATGCTGCGAAGGACAGGTTGATG	0.5	100	ggccGGCGACGATCCCGGGG	10.0
	p.F302L	ACGTTGGATGGGTACCTGAAGACCATTGAC	ACGTTGGATGAAGATAAGGCAGCCAGACAC	2.0	103	CCATTGACATCTGCCATGACCACTT	15.0
	p.G410S	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	2.0	520	GGCCAGGCTGCCCATCAGGTCGC	10.0
	p.E448K	ACGTTGGATGTAGAAGATTCCAGGCAGCAG	ACGTTGGATGTACATGGCCATCCTGCTGAC	0.5	145	GGCCGTACTTGCTGGCGCAGCGGTGCT	20.0
TYRSN1-*FAH*	p.P261L	ACGTTGGATGTCCTTGGGAAGAGTTTTGGG	ACGTTGGATGCACAGCAAAGGGCATGAGAG	0.5	94	TGGGACCACTGTCTCTC	5.0
	c.554-1G>T	ACGTTGGATGAGCTTGCAGGCACCATATAC	ACGTTGGATGTGTTCCCACACAGAGAGTTC	0.5	122	CACCATATACGGGAGGCTTAG	10.0
	c.1062+5G>A	ACGTTGGATGGAAATCTCTTCTCGAGGCTG	ACGTTGGATGTCCTGGCTTCTGGGACCATC	0.5	145	GCCCTCAGTGCAGCCAGATA	5.0
	p.Q64H	ACGTTGGATGTCAGGGACAAGTCGTTTCAC	ACGTTGGATGACCTCTTTACTGGTCCTGTC	0.5	100	CAAGTCGTTTCACAATGTCCTAC	10.0
	p.W262*	ACGTTGGATGTCCTTGGGAAGAGTTTTGGG	ACGTTGGATGCACAGCAAAGGGCATGAGAG	0.5	94	GGCATGAGAGCATCCATGGGCACCAC	15.0
	p.E357*	ACGTTGGATGTATGGGCTTCGTTCCCTTCC	ACGTTGGATGTTCTTCCCTTTCCTGTGATG	0.5	100	GACAGTTCCAACATGGAGCCGAAGTTTT	15.0
WD-*ATP7B*	p.E1064A	ACGTTGGATGAGTATTTGGTGACTGCCACG	ACGTTGGATGTCAGGAAGGTTCTGGCTGTG	2.0	98	GTTCTGGCTGTGGTGGGGACTGCGG	25.0
	p.H1069Q	ACGTTGGATGAGTATTTGGTGACTGCCACG	ACGTTGGATGTCAGGAAGGTTCTGGCTGTG	0.5	98	GACTGCCACGCCCAAGGG	5.0
	p.R778L	ACGTTGGATGGCAGCTCTTTTCTGAACCTG	ACGTTGGATGGCTCTTTGTGTTCATTGCCC	0.5	102	TTACCTTTGCCAAGTGTTCCAGCCAC	15.0
	p.M645R	ACGTTGGATGAGAGTTGGGCCCAGGTAGAG	ACGTTGGATGCCCACAAAGTCTACTGAGGCA	0.5	296	gGTTCTACCTACTGCTTTATTTCC	10.0
PBD1A-*PEX1*	p.G843D	ACGTTGGATGCCTGCATAAACCTAGAGACC	ACGTTGGATGAGTATCCATGAGTATCTGCC	0.5	96	AGACCTGGGTTGGGACAAGATTG	10.0
	p.I700*fs*	ACGTTGGATGTTGTGGCAATCAGTGCAACC	ACGTTGGATGGCTTAATTCCAGCTTTGAATG	0.5	95	CAAACTTCCCATGGAGATAAA	10.0
	p.G983*fs*	ACGTTGGATGGATGACATGATGACATTTTG	ACGTTGGATGCCGAGTAGTTAACCAGTTGC	0.5	128	ATAATTATTACCCTGTAAGCCT	10.0
PBD5A-*PEX2*	p.R119*	ACGTTGGATGGTACAATTGGTGGCAGGTGG	ACGTTGGATGACACACTGCTTGACTTTCCC	0.5	111	GCAGGTGGTTAGAAGAA	5.0

Supplementary Table S6: Agena Bioscience™ PCR primer and extension probe information for the clinical panel of 18 new AJ diseases

Disease-gene	Mutation	PCR primer (5' to 3')		Primer working concentration (μM)	Amplicon size (bp)	Universal extension probe (5' to 3')	Probe working concentration (μM)
		Forward	Reverse				
ABL-*MTTP*	p.G865*	ACGTTGGATGTGTCCACAGGCAGAGGTTATG	ACGTTGGATGCCTTTCGGGGAGACAAATTCAA	0.5	209	TGAGGGAGCGGGAATTCACATC	10.0
	p.S738*fs*	ACGTTGGATGTCAGTGTGGTGAAAGGACTT	ACGTTGGATGCCAAGATGGGCACAATTTTG	0.5	172	AGGACTTATTCTGCTAATAGATCAT	15.0
ATS-*COL4A3*	p.L14_L21del	ACGTTGGATGGACCGAGCCCTACAAAACC	ACGTTGGATGGTGGAGGAGGGATGGAAGTG	1.0	302	CCCAGGCCGCAGGTGCTCCTGG	15.0
AMRS-*SLC35A3*	p.S296G	ACGTTGGATGCTCCTATTTTTGGCTTCAAG	ACGTTGGATGATCAGATCACCTTACAAAG	0.5	133	GGCTTCAAGATTTTGTGCCAACC	15.0
	p.Q172*	ACGTTGGATGCCTCAGATTCTCAGCTTGAT	ACGTTGGATGAAAACATGCTGTGAGAACTG	0.5	103	ACTTTCAGCTGGTTCT	7.0
BBS2-*BBS2*	p.D104A	ACGTTGGATGCTATGATGCCCTTTTAGTGG	ACGTTGGATGCTCTCTGTAGAACAAATCCG	0.5	102	GGGACACAGACTAATCTTTTGGCTTATG	15.0
	p.R632P	ACGTTGGATGGCACCTGTACTAACCATGAC	ACGTTGGATGTGATCCGAAGTTTGCTGGTC	0.5	106	TCACATGTCCCTCATCAGA	5.0
CPT2-*CPT2*	p.S113L	ACGTTGGATGGGAGAAATCATACCCACAGC	ACGTTGGATGGCTGGTTGCTCTGGACAAAC	0.5	100	AGCCCAGCCTACCTACCC	5.0
	p.Q413*fs*	ACGTTGGATGTAGCAGCTGTGATGCCAGTC	ACGTTGGATGACCACTGACTCTACTGTCAC	0.5	99	CAGTCAGCTCGAAGTTGAGTTT	10.0
	p.R124*	ACGTTGGATGGGGTCAGGATTGAAAGCCAT	ACGTTGGATGTAGGACCCTGGTTTGATATG	0.5	105	AAAGTTCAGAACAACGGAGTCTC	10.0
	p.S38*fs*	ACGTTGGATGTGTCCTGGTAGTGCATGGTG	ACGTTGGATGTTGGTCCGGGAGCCCCCAGT	0.5	116	GTGGGCACGATGCTGCGC	7.0
CAMT-*MPL*	c.79+2T>A	ACGTTGGATGCTCAAAACCTGGCCCAAGTC	ACGTTGGATGcTCCCTCTTCCTGGGCATAG	0.5	97	ACCTGGCCCAAGTCAGCAGCCAAGG	15.0
CDG1A-*PMM2*	p.P113L	ACGTTGGATGAGCCCACCATGTGACACTAC	ACGTTGGATGGTGAGGCCCTAATCCAAGAT	0.5	154	AAACCCACCTCTTCTTC	5.0
	p.F119L	ACGTTGGATGCGTACTTTATCGAGTTCGTAG	ACGTTGGATGTGTCACCCTTTCATTCCCAG	1.0	145	CATCCCATTTCGGAATTCAAT	10.0
	p.R141H	ACGTTGGATGCGTACTTTATCGAGTTCGTAG	ACGTTGGATGTGTCACCCTTTCATTCCCAG	1.0	145	TTTATCGAGTTCGTAGAACTCAATG	15.0
	p.V231M	ACGTTGGATGATCTTCACAGACCCCAGAAC	ACGTTGGATGGAACAGCAGTTCACAGATCC	0.5	98	ACCCCAGAACCATGGGCTACTCC	15.0
DC-*RTEL1*	p.R1264H	ACGTTGGATGTTCCAGTGCCCTGCCTGTGA	ACGTTGGATGACCCAGCAGGAACTGTAGTG	0.5	113	GCCTGTGACTTCCAGC	5.0
	p.M516I	ACGTTGGATGTGATGTCAGAGGTGTCACCG	ACGTTGGATGCGGTGTCCTCCTTTGCTCT	0.5	102	AGGGGTGGCCCGTACATCTG	10.0
	p.R981W	ACGTTGGATGTTCCTCTACAGGCTTCTACC	ACGTTGGATGCGGCTGTGCCCGCTGCCTT	1.0	156	AGGCTTCTACCAGTTTGTG	10.0
	p.R998*	ACGTTGGATGTTCCTCTACAGGCTTCTACC	ACGTTGGATGCGGCTGTGCCCGCTGCCTT	1.0	156	TGTATCCAGCTGACAGGA	5.0
	p.G763V	ACGTTGGATGCAGCAGATGAGGGTCTTCACTT	ACGTTGGATGTCGGCAACACGGAAGAACTG	0.5	220	ACTGGGCCACGTCTCGGATGACATGG	10.0
EDSVIIC-*ADAMTS2*	p.Q225*	ACGTTGGATGCCAAGCTGGTCTGTGGAGAG	ACGTTGGATGCCGTGTGCATGTGGTGTATC	0.5	195	GGAATCACCTGTGTCCAGGGCCT	15.0
	p.W795*	ACGTTGGATGTGAAGGGTCAGGAGGCTCA	ACGTTGGATGTCCAAAACCTTCATTGCCATGG	0.5	219	CTCCTCGTCTCTGTACTCC	5.0
GALT-*GALT*	5kb del	ACGTTGGATGTGGAGCTTCTGATAGTCCTG	ACGTTGGATGACAGAGCAGACTCCGTCACA	0.5	105	CTGATAGTCCTGTACTCTCCAC	10.0
	c.253-2A>G	ACGTTGGATGGTGAGTGCTTCTAGCCTATC	ACGTTGGATGCTGGGAAGTCGTTGTCAAAC	0.5	100	AGCCTATCCTTGTCGGT	5.0
	p.S135L	ACGTTGGATGCCAGTAAGGTCATGTGCTTC	ACGTTGGATGAACAACAGCCCGGATCTCAG	0.5	100	AGTAAGGTCATGTGCTTCCACCCCTGGT	15.0
	p.Q188R	ACGTTGGATGcCACTGGAGCCCCTGACACC	ACGTTGGATGCTTGGTGTCTTTTGGCTAAC	0.5	147	CCCCTGACACCCTTACC	3.5
	p.T138M	ACGTTGGATGCCAGTAAGGTCATGTGCTTC	ACGTTGGATGAACAACAGCCCGGATCTCAG	0.5	100	GACATGAGTGGCAGC	5.0
	p.F171S	ACGTTGGATGcCACTGGAGCCCCTGACACC	ACGTTGGATGCTTGGTGTCTTTTGGCTAAC	0.5	147	TCATGGCACCTTTGTTTTCA	5.0
	p.L195P	ACGTTGGATGcTTGACAGGTATGGGCCAGC	ACGTTGGATGCTGGCGGCTGTACTCCATTA	0.5	136	ATGGGCCAGCAGTTTCC	5.0
	p.Y209C	ACGTTGGATGcTTGACAGGTATGGGCCAGC	ACGTTGGATGCTGGCGGCTGTACTCCATTA	0.5	136	ctctCGATCTCAGCAGGCCT	10.0
	p.K285N	ACGTTGGATGCCTCCATCATGAAGAAGCTC	ACGTTGGATGTTGAAAAGCCTCACCATGCC	0.5	112	GAAGAAGCTCTTGACCAA	5.0
MSD-*SUMF1*	p.S155P	ACGTTGGATGCCTACCTTCCTTATAGGCTG	ACGTTGGATGTGGTCTTCACTTGCTCACTC	0.5	97	TAGGCTGAGAAGTTTGGCGAC	10.0
PHGDHD-*PHGDH*	p.V490M	ACGTTGGATGACACCAGTGAAGTCTGGTAG	ACGTTGGATGTGCCAACCAGGAGTTTCTTC	0.5	103	GTAGGACAGCAGCCGCA	5.0
ARPKD-*PKHD1*	p.A1254*fs*	ACGTTGGATGAAGAACCTGTTGCCAGCCCA	ACGTTGGATGGAGCATCTGGTGTGAAACCC	0.5	119	GCATCGGGTATCTGGGG	5.0
	p.T36M	ACGTTGGATGTGTGCACTTGGTAAAACCCC	ACGTTGGATGACCTGAAGAAGGTAGCCTTG	2.0	100	ACCATCAAAAATGACTGTGATCCAC	15.0
	p.R496*	ACGTTGGATGTGTGGTCACCACTTACCTAC	ACGTTGGATGACACAGACCTGTACTTCTGG	0.5	98	CGGGAGAAGCACCAGATCCGAGTC	10.0
	p.L1966*fs*	ACGTTGGATGATGGCCAATTGCTTCTGCTG	ACGTTGGATGACCTACAAACTTCACACACC	0.5	99	GACACTAACACAAGCATCCTCAACTTA	10.0
	p.V3471G	ACGTTGGATGCCCCAATAGAAAAAAGCGCA	ACGTTGGATGTTCTATCTTACCCATCAGGC	0.5	102	TGAGGAGTTTGATCCATGAAGCAG	10.0
	p.D3230*fs*	ACGTTGGATGTTCAGGACAAAGTGAAGCCG	ACGTTGGATGTTCGACCTCCTCTTGGATTG	0.5	98	CAGCCAACTTGACATCAACAG	7.0
RP59-*DHDDS*	p.K42E	ACGTTGGATGCATTCATAATGGACGGGAAC	ACGTTGGATGTTGTTGAAGCCCTGTGAGTG	0.5	102	TGGACGGGAACCGTCGCTATGCCAAG	15.0
SLOS-*DHCR7*	c.964-1G>C	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	0.5	520	TTCCCCCTCGCCCCCCA	5.0
	p.W151* (c.452G>A)	ACGTTGGATGTCCAGTTGTCGAAGATGATG	ACGTTGGATGtGTGAACAAGTATCAGATCA	0.5	136	AGAGCAGGTGCGTGAGGAGC	7.5
	p.W151* (c.453G>A)	ACGTTGGATGTCCAGTTGTCGAAGATGATG	ACGTTGGATGtGTGAACAAGTATCAGATCA	0.5	136	tgcacACCAGAGCAGGTGCGTGAGGAG	15.0
	p.M1V	ACGTTGGATGTGACGCCATCTAGACTCTTG	ACGTTGGATGCAAGGTTCTCTTTCTTGCAG	0.5	101	GGGTTGCGATTTTGCAGCCA	5.0
	p.V326L	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	2.0	520	ggcatCTGCACGGGGTGGTACA	10.0
	p.T93M	ACGTTGGATGgGACATGCTCGGCTCTCGGA	ACGTTGGATGACCCACAAGGTATAGAGCTG	0.5	100	CCAAGACTCCACCTATAA	3.5
	p.R352Q	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	0.5	520	TGGGCTACTACATCTTCC	5.0
	p.R352W	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	0.5	520	GGTGGGCTACTACATCTTC	5.0
	p.R404C	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	2.0	520	GGGCTTCTGGGGCGTGGCC	7.5
	p.S169L	ACGTTGGATGtGTGAACAAGTATCAGATCA	ACGTTGGATGTCCAGTTGTCGAAGATGATG	0.5	136	TCATCTCCTGTCCTGGTTCT	5.0
	p.R242C	ACGTTGGATGGAAGTGGTTTGACTTCAAGC	ACGTTGGATGCTGCGAAGGACAGGTTGATG	0.5	100	GACTTCAAGCTGTTCTTCAATGGG	10.0
	p.R242H	ACGTTGGATGGAAGTGGTTTGACTTCAAGC	ACGTTGGATGCTGCGAAGGACAGGTTGATG	0.5	100	ggccGGCGACGATCCCGGGG	10.0
	p.F302L	ACGTTGGATGGGTACCTGAAGACCATTGAC	ACGTTGGATGAAGATAAGGCAGCCAGACAC	2.0	103	CCATTGACATCTGCCATGACCACTT	15.0
	p.G410S	ACGTTGGATGTGGCCATGTAGATGATGTAGAA	ACGTTGGATGGGTGCTTTTATTCACAGAAGAGA	2.0	520	GGCCAGGCTGCCCATCAGGTCGC	10.0
	p.E448K	ACGTTGGATGTAGAAGATTCCAGGCAGCAG	ACGTTGGATGTACATGGCCATCCTGCTGAC	0.5	145	GGCCGTACTTGCTGGCGCAGCGGTGCT	20.0
TYRSN1-*FAH*	p.P261L	ACGTTGGATGTCCTTGGGAAGAGTTTTGGG	ACGTTGGATGCACAGCAAAGGGCATGAGAG	0.5	94	TGGGACCACTGTCTCTC	5.0
	c.554-1G>T	ACGTTGGATGAGCTTGCAGGCACCATATAC	ACGTTGGATGTGTTCCCACACAGAGAGTTC	0.5	122	CACCATATACGGGAGGCTTAG	10.0
	c.1062+5G>A	ACGTTGGATGGAAATCTCTTCTCGAGGCTG	ACGTTGGATGTCCTGGCTTCTGGGACCATC	0.5	145	GCCCTCAGTGCAGCCAGATA	5.0
	p.Q64H	ACGTTGGATGTCAGGGACAAGTCGTTTCAC	ACGTTGGATGACCTCTTTACTGGTCCTGTC	0.5	100	CAAGTCGTTTCACAATGTCCTAC	10.0
	p.W262*	ACGTTGGATGTCCTTGGGAAGAGTTTTGGG	ACGTTGGATGCACAGCAAAGGGCATGAGAG	0.5	94	GGCATGAGAGCATCCATGGGCACCAC	15.0
	p.E357*	ACGTTGGATGTATGGGCTTCGTTCCCTTCC	ACGTTGGATGTTCTTCCCTTTCCTGTGATG	0.5	100	GACAGTTCCAACATGGAGCCGAAGTTTT	15.0
WD-*ATP7B*	p.E1064A	ACGTTGGATGAGTATTTGGTGACTGCCACG	ACGTTGGATGTCAGGAAGGTTCTGGCTGTG	2.0	98	GTTCTGGCTGTGGTGGGGACTGCGG	25.0
	p.H1069Q	ACGTTGGATGAGTATTTGGTGACTGCCACG	ACGTTGGATGTCAGGAAGGTTCTGGCTGTG	0.5	98	GACTGCCACGCCCAAGGG	5.0
	p.R778L	ACGTTGGATGGCAGCTCTTTTCTGAACCTG	ACGTTGGATGGCTCTTTGTGTTCATTGCCC	0.5	102	TTACCTTTGCCAAGTGTTCCAGCCAC	15.0
	p.M645R	ACGTTGGATGAGAGTTGGGCCCAGGTAGAG	ACGTTGGATGCCCACAAAGTCTACTGAGGCA	0.5	296	gGTTCTACCTACTGCTTTATTTCC	10.0
PBD5A-*PEX2*	p.R119*	ACGTTGGATGGTACAATTGGTGGCAGGTGG	ACGTTGGATGACACACTGCTTGACTTTCCC	0.5	111	GCAGGTGGTTAGAAGAA	5.0

Supplementary Table S7: Screening results for 85 variants among over 3,000 AJ and over 6,000 non-AJ individuals

Disease[a]-gene	Mutation	AJ population					Non-AJ population				
		Sample size	HOM	HET	WT	HWE p	Sample size	HOM	HET	WT	HWE p
Diseases that are included in AJ carrier screening panel											
ABL-*MTTP*	p.G865*	3347	1	15	3331	<0.001	7071	0	5	7066	0.98
	p.S738fs	2954	0	1	2953	0.99	7071	0	0	7071	N/A
ATS-*COL4A3*	p.L14_L21del	3639	0	19	3620	0.87	7240	0	2	7238	0.99
AMRS-*SLC35A3*	p.S296G	3622	0	8	3614	0.95	7239	0	2	7237	0.99
	p.Q172*	3617	0	0	3617	N/A	7239	0	0	7239	N/A
BBS2-*BBS2*	p.D104A	3627	0	10	3617	0.93	7232	0	10	7222	0.95
	p.R632P	3626	0	16	3610	0.89	7232	0	0	7232	N/A
CPT2-*CPT2*	p.S113L	3587	0	44	3543	0.71	6815	0	52	6763	0.75
	p.Q413fs	3586	0	26	3560	0.83	6815	0	0	6815	N/A
	p.R124*	3590	0	0	3590	N/A	6815	0	0	6815	N/A
	p.S38fs	3573	0	0	3573	N/A	6815	0	0	6815	N/A
CAMT-*MPL*	c.79+2T>A	3617	0	64	3553	0.59	7069	0	14	7055	0.93
CDG1A-*PMM2*	p.P113L	3575	0	0	3575	N/A	6813	0	64	6749	0.70
	p.F119L	3579	0	0	3579	N/A	6813	0	0	6813	N/A
	p.R141H	3578	0	59	3519	0.62	6813	0	0	6813	N/A
	p.V231M	3575	0	0	3575	N/A	6813	0	0	6813	N/A
DC-*RTEL1*	p.R1264H	3627	0	19	3608	0.87	7239	0	14	7225	0.93
	p.M516I	3630	0	3	3627	0.98	7239	0	0	7239	N/A
	p.R981W	3627	0	0	3627	N/A	7239	0	0	7239	N/A
	p.R998*	3627	0	0	3627	N/A	7239	0	0	7239	N/A
	p.G763V	3619	0	0	3619	N/A	7239	0	0	7239	N/A
EDSVIIC-*ADAMTS2*	p.Q225*	3367	0	18	3349	0.88	7235	0	7	7228	0.97
	p.W795*	3367	0	0	3367	N/A	7235	0	0	7235	N/A
GALT-*GALT*	c.-1039_+789del5573ins129	3582	0	18	3564	0.88	6815	0	42	6773	0.80
	c.253-2A>G	3581	0	0	3581	N/A	6815	0	0	6815	N/A
	p.S135L	3576	0	1	3575	0.99	6815	0	0	6815	N/A
	p.Q188R	3576	0	2	3574	0.99	6815	0	0	6815	N/A
	p.T138M	3575	0	0	3575	N/A	6815	0	0	6815	N/A
	p.F171S	3582	0	0	3582	N/A	6815	0	0	6815	N/A
	p.L195P	3575	0	0	3575	N/A	6815	0	0	6815	N/A
	p.Y209C	3573	0	0	3573	N/A	6815	0	0	6815	N/A

	p.K285N	3581	0	2	3579	0.99	6815	0	0	6815	N/A
MSD-*SUMF1*	p.S155P	3625	0	13	3612	0.91	7238	0	8	7230	0.96
PHGDHD-*PHGDH*	p.V490M	3625	0	8	3617	0.95	7242	0	3	7239	0.99
ARPKD-*PKHD1*	p.A1254fs	3578	0	33	3545	0.78	6815	0	30	6785	0.86
	p.T36M	3587	0	1	3586	0.99	6815	0	0	6815	N/A
	p.R496*	3581	0	0	3581	N/A	6815	0	0	6815	N/A
	p.L1966fs	3581	0	0	3581	N/A	6815	0	0	6815	N/A
	p.V3471G	3583	0	0	3583	N/A	6815	0	0	6815	N/A
	p.D3230fs	3581	0	0	3581	N/A	6815	0	0	6815	N/A
RP59-*DHDDS*	p.K42E	3621	0	31	3590	0.8	7238	0	19	7219	0.91
SLOS-*DHCR7*	c.964-1G>C	3604	0	86	3518	0.47	15369	0	126	15243	0.61
	p.W151* (c.452G>A)	3612	0	3	3609	0.98	15369	0	0	15369	N/A
	p.W151* (c.453G>A)	3612	0	0	3614	N/A	15369	0	0	15369	N/A
	p.M1V	3615	0	0	3615	N/A	15369	0	0	15369	N/A
	p.V326L	3602	0	0	3602	N/A	15369	0	0	15369	N/A
	p.T93M	3612	0	0	3612	N/A	15369	0	0	15369	N/A
	p.R352Q	3601	0	0	3601	N/A	15369	0	0	15369	N/A
	p.R352W	3602	0	0	3602	N/A	15369	0	0	15369	N/A
	p.R404C	3602	0	0	3602	N/A	15369	0	0	15369	N/A
	p.S169L	3615	0	0	3615	N/A	15369	0	0	15369	N/A
	p.R242C	3614	0	0	3614	N/A	15369	0	0	15369	N/A
	p.R242H	3613	0	0	3613	N/A	15369	0	0	15369	N/A
	p.F302L	3613	0	0	3613	N/A	15369	0	0	15369	N/A
	p.G410S	3602	0	0	3602	N/A	15369	0	0	15369	N/A
	p.E448K	3606	0	0	3606	N/A	15369	0	0	15369	N/A
TYRSN1-*FAH*	p.P261L	3581	0	24	3557	0.84	6813	0	11	6802	0.95
	c.554-1G>T	3571	0	0	3571	N/A	6813	0	0	6813	N/A
	c.1062+5G>A	3578	0	1	3577	0.99	6813	0	0	6813	N/A
	p.Q64H	3577	0	0	3577	N/A	6813	0	0	6813	N/A
	p.W262*	3574	0	0	3574	N/A	6813	0	0	6813	N/A
	p.E357*	3566	0	0	3566	N/A	6813	0	0	6813	N/A
WD-*ATP7B*	p.E1064A	3572	0	20	3552	0.87	6813	0	36	6777	0.83
	p.H1069Q	3565	0	25	3540	0.83	6813	0	0	6813	N/A
	p.R778L	3578	0	0	3578	N/A	6813	0	0	6813	N/A
	p.M645R	3563	0	8	3555	0.95	6813	0	0	6813	N/A
PBD5A-*PEX2*	p.R119*	3626	0	16	3610	0.89	7232	0	5	7227	0.98

Diseases that are not appropriate for AJ carrier screening

ATS-*COL4A5*	p.R1677Q	1598[b]	0	0	1598[b]	N/A
HYC1-*CCDC88C*	p.R312*	2238	0	0	2238	N/A
HHH-*SLC25A15*	p.F188del	2242	0	0	2242	N/A
JEB-H-*LAMB3*	p.N843fs	1590	0	0	1590	N/A
	p.R42*	1590	0	0	1590	N/A
	p.R635*	1590	0	3	1587	0.97
	p.Q243*	1586	0	0	1586	N/A
LS-*NDUFS4*	p.K154fs	1594	0	3	1591	0.97
	p.D119H	1596	0	0	1596	N/A
LS-*NDUFAF5*	p.G250V	1597	0	0	1597	N/A
SCN3-*HAX1*	p.S43fs	1596	0	2	1594	0.98
PCH1A-*VRK1*	p.R358*	1595	0	3	1592	0.97
GLC3A-*CYP1B1*	p.R368H	2231	2	81	2148	0.18
RP62-*MAK*	p.K433fs	1494	0	52	1442	0.49
	p.R166H	1591	0	0	1591	N/A
PBD1A-*PEX1*	p.G843D	2240	0	0	2240	N/A
	p.I700fs	2243	0	1	2242	0.99
	p.G983fs	2236	0	0	2236	N/A

HOM, homozygote; HET, heterozygote; WT, wild-type; CI, confidence interval; HWE, Hardy-Weinberg equilibrium.

[a]See Table 1 for disease abbreviations. [b]899 females and 699 males

Supplementary Table S8: Cumulative risks for AJ panels

ACMG/ACOG recommendation		Commercially available additional AJ-9		Additional AJ-18 in current study	
Disease name [a]	AJ carrier frequency	Disease name	AJ carrier frequency [b]	Disease name	AJ carrier frequency [b]
GD	1 in 15	GSD1A	1 in 64	SLOS	1 in 40
CF	1 in 23	FH	1 in 68	CPT2	1 in 51
TSD	1 in 27	WWS	1 in 90	CAMT	1 in 57
FD	1 in 31	MSUD Ib	1 in 97	CDG1A	1 in 61
CD	1 in 55	E3	1 in 108	WD	1 in 67
MLIV	1 in 88	JBS2	1 in 110	ARPKD	1 in 105
FANCC	1 in 100	NM	1 in 114	RP59	1 in 117
NPD	1 in 115	USH3A	1 in 120	BBS2	1 in 139
BS	1 in 133	USH1F	1 in 147	TYRSN1	1 in 143
				GALT	1 in 156
				DC	1 in 165
				ABL	1 in 185
				EDSVIIC	1 in 187
				ATS	1 in 192

		PBD5A	1 in 227
		MSD	1 in 279
		ARMS	1 in 453
		PHGDHD	1 in 453
Combined carrier frequency [c]			
1 in 4.3	1 in 10.6		1 in 6
Cumulative carrier frequency [c]			
1 in 4.3	**1 in 3**		**1 in 2**

AJ, Ashkenazi Jewish; [a]Additional disease abbreviations: GD, Gaucher Disease (MIM#230800); CF, cystic fibrosis ((MIM#219700); TSD, Tay-Sachs disease (MIM#272800); FD, familial dysautonomia (MIM#223900); CD, Canavan disease (MIM#271900); MLIV, mucolipidosis IV (MIM#252650); FANCC, Fanconi anemia, complementation group C (MIM#227645); NPD, Niemann-Pick disease type A (MIM#257200),; BS, Bloom syndrome (MIM#210900); GSD1A, glycogen storage disease type 1A (MIM#232200); FH, familial hyperinsulinism (MIM#256450); WWS, Walker-Warburg syndrome (MIM#253800); MSUD, maple syrup urine disease (MIM#248600); E3, dihydrolipoamide dehydrogenase deficiency (MIM#246900); JBS, Joubert syndrome (MIM#608091); NM, Nemaline myopathy (MIM#256030); USH3A, Usher syndrome, Type IIIA (MIM#276902); USH1F, Usher syndrome, Type IF (MIM#602083). [b]AJ carrier frequencies for the first two panels were from Scott *et al.* 2010 except for WWS and JBS2, whose AJ carrier frequencies were from unpublished data. [c]Refers to the simple mathematical sum of all disease frequencies in the panel. Proportions of multi-mutation carrier were not considered.

Supplementary Table S9: Population frequency comparison between current AJ cohort and literature report

			Current AJ cohort		Literature				
Disease	**Gene**	**Mutation**	**Carrier frequency**	**Sample size**	**Carrier frequency**	**Sample size**	**Population Ethnicity**	**Literature (PMID)**	**p-value**
ABL	*MTTP*	p.G865*	1 in 394	3347	1 in 113	393	AJ	17275380	0.65
ABL	*MTTP*	p.S738*fs*	1 in 2954	2954	0	430	AJ	17275380	0.70
AS	*COL4A3*	p.L14_L21del	1 in 192	3639	1 in 183	2017	AJ	23927549	0.91
AMRS	*SLC35A3*	p.S296G	1 in 453	3622	1 in 205	2045	AJ	24031089	0.08
AMRS	*SLC35A3*	p.Q172*	0	3617	0	2045	AJ	24031089	1.00
BBS2	*BBS2*	p.D104A	1 in 363	3627	1 in 211	19010	AJ	23829372	0.13
BBS2	*BBS2*	p.R632P	1 in 227	3626	1 in 383	18025	AJ	23829372	0.09
CPT2	*CPT2*	p.S113L	1 in 82	3587	1 in 35	280	AJ	2013 ACMG meeting abstract 522	0.05
CAMT	*MPL*	c.79+2T>A	1 in 57	3617	1 in 75	2018	AJ	21489838	0.23
CDG1A	*PMM2*	p.R141H	1 in 61	3578	1 in 40	280	AJ	2013 ACMG meeting abstract 522	0.33
GALT	*GALT*	c.-1039_+789del5573ins129	1 in 199	3582	1 in 127	760	AJ	21059483	0.29
HHH	*SLC25A15*	p.F188del	0	2242	1 in 94	280	AJ	2013 ACMG meeting abstract 522	**<0.01**
LS	*NDUFS4*	p.K154*fs*	1 in 531	1594	1 in 1000	5000	AJ	19107570	0.41
LS	*NDUFAF5*	p.G250V	0	1597	1 in 290	869	AJ	21607760	**0.04**
PHGDHD	*PHGDH*	p.V490M	1 in 453	3625	1 in 400	400	AJ	11751922	0.61
PCH1A	*VRK1*	p.R358*	1 in 532	1595	1 in 225	449	AJ	19646678	0.30
GLC3A	*CYP1B1*	p.R368H	1 in 26	2231	1 in 31	280	AJ	2013 ACMG meeting abstract 522	0.83
					1 in 322	322	AJ	21295282	0.51
					1 in 90	717	AJ	21295283	0.51
RP59	*DHDDS*	p.K42E	1 in 117	3621	1 in 120	1207	AJ	22110072	1.00
RP62	*MAK*	p.K433fs	1 in 29	1494	1 in 55	1207	AJ	22110072	**<0.01**
RP62	*MAK*	p.R166H	0	1591	1 in 217	217	Israel oriental Jews	21835304	0.12
SLOS	*DHCR7*	c.964-1G>C	1 in 42	3604	1 in 35	280	AJ	2013 ACMG meeting abstract 522	0.55
SLOS	*DHCR7*	p.W151* (c.452G>A)	1 in 1204	3612	1 in 338	7774	European Caucasians	17965227(Euro Caucasians)	**0.03**
WD	*ATP7B*	p.E1064A	1 in 179	3572	1 in 100	100	AJ	9482578	0.44
PBD5A	*PEX2*	p.R119*	1 in 227	3626	1 in 123	2091	AJ	23590336	0.10