Supp. Table S1. Mutations in MKS genes in allelic disorders.

		Allele 1			Allele 2			
Family ID	**Syndrome**	**Exon/int.**	**Mutation 1**	**Effect on protein**	**Exon/int.**	**Mutation 2**	**Effect on protein**	**Origin**	**Reference**
MKS1									
PB215-04	BBS		n.d. in original article	p.Cys492Trp		n.d. in original article	p.Phe371del	Turkey	[Leitch et al., 2008]
TMEM67 (MKS3)									
JS-661 and 660	JS	ex15	c.1538A>G	p.Tyr513Cys	ex22	c.2315_2323+4del13insGG	missplicing	Europe	[Baala et al., 2007b]
JS-05	JS	int23	c.2439+5G>C	Ile775_Ala813del ^	int23	c.2439+5G>C	Ile775_Ala813del ^	Algeria	[Baala et al., 2007b]
JS-09[§]	JS	int6	c.651+2T>G	pot. missplicing	ex21	c.2341G>A [#]	p.Gln747fs [#]	Europe	[Baala et al., 2007b]
		ex16	c.1634G>A	p.Gly545Glu					
NPH-786	JS	ex6	c.637C>T	p.Arg213Cys	ex21	c.2132A>C	p.Asp711Ala	Europe	[Baala et al., 2007b]
COR09	COACH	ex17	c.1769T>C	p.Phe590Ser	int19	c.1961-2A>C	Ala621_Glu700del ^	Europe	[Brancati et al., 2008a]
COR20	COACH	ex6	c.579_580delAG	p.Gly195fs	ex17	c.1769T>C	p.Phe590Ser	Europe	[Brancati et al., 2008a]
COR32	COACH	ex11	c.1115C>A	p.Thr372Lys	ex23	c.2345A>G	p.His782Arg	Europe	[Brancati et al., 2008a]
COR71	COACH	ex3	c.389C>G	p.Pro130Arg	ex7	c.675G>A	p.Trp225X	Europe	[Brancati et al., 2008a]
COR94	COACH	ex13	c.1319G>A	p.Arg440Gln	ex21	c.2182A>G	p.Ser728Gly	Europe	[Brancati et al., 2008a]
COR190	COACH	int2	c.312+5G>A	pot. missplicing	ex24	c.2498T>C	p.Ile833Thr	Europe	[Brancati et al., 2008a]
MTI124	COACH	ex24	c.2498T>C	p.Ile833Thr	int 24	c.2556+1G>T	pot. missplicing	Europe	[Brancati et al., 2008a]
CEP290 (MKS4)									
Family 7	CORS	ex26	c.2906dupA	p.Tyr969X	ex31	c.3793C>T	p.Gln1265X	Europe	[Baala et al., 2007a]
Family 8	CORS	ex27	c.3043G>T	p.Glu1015X	int27	c.3104-1G>A	pot. missplicing	Palestina	[Baala et al., 2007a]
Family 9	CORS	ex41	c.5649dupA	p. Leu1884fs	ex42	c.5850delT	p. Phe1950fs	Europe	[Baala et al., 2007a]
Family 10	CORS	ex20	c.1984C>T	p.Gln662X	ex20	c.1984C>T	p.Gln662X	Europe	[Baala et al., 2007a]
F4 (II-1) and (II-2)	SLS	ex23	c.2218_2222delccagATAGA	pot. missplicing	ex23	c.2218_2222delccagATAGA	pot. missplicing	Turkey	[Sayer et al., 2006]
F63 (II-1)	JS	ex36	c.4656delA	p.Glu1553fs Lys1552fs	ex41	c.5668G>T	p.Gly1890X	Europe	[Sayer et al., 2006]
A197(II-1)	JS	ex29	c.3175dupA 3175_3176insA	p.Ile1059fs	ex55	c.7341dupA 7341_7342insA	p.Leu2448fs	Europe	[Sayer et al., 2006]
F89 (II-1)	JS	ex41	c.5515_5518delGAGA	p.Glu1839fs	ex42	c.5649_5650insA 5649insA	p. Leu1884fs	Europe	[Sayer et al., 2006]

		Allele 1			Allele 2				
Family ID	**Syndrome**	**Exon/int.**	**Mutation 1**	**Effect on protein**	**Exon/int.**	**Mutation 2**	**Effect on protein**	**Origin**	**Reference**
In 6 families	JS/JSRD-SLS	ex41	c.5668G>T	p.Gly1890X	ex41	c.5668G>T	p.Gly1890X	United Arab Emirates, Turkey	[Brancati et al., 2007; Sayer et al., 2006; Valente et al., 2006]
F9	JS	ex41	c.5649dupA 5649_5650insA	p. Leu1884fs	ex42	c.5850delT	p. Phe1950fs	Europe	[Tory et al., 2007]
F72	JS	ex37	c.4963_4964delAG	p.Arg1665fs	int2	c.103-13_18delGCTTTT	pot. missplicing	Europe	[Tory et al., 2007]
F99	JS	ex22	c.2251C>T	p.Arg751X	ex50	c.6869delA	p. Asn2290fs	Europe	[Tory et al., 2007]
F265[a]	JS	ex5	c.287delA	p.Asn96fs	ex5	c.287delA	p.Asn96fs	Europe	[Tory et al., 2007]
F375	JS	ex15	c.1361delG	p.Gly454fs	ex50	c.6869delA	p. Asn2290fs	Europe	[Tory et al., 2007]
F358	JS	ex17	c.1645C>T	p.Arg549X	ex41	c.5649_5650insA	p. Leu1884fs	Europe	[Tory et al., 2007]
F419	JS	int28	c.3310-1G>C	pot. missplicing	int45	c.6271-8T>G	pot. missplicing	Argentina	[Tory et al., 2007]
F858-2	JS	int32	c.4195-1G>A	pot. missplicing	ex41	c.5649_5650insA	p. Leu1884fs	Europe	[Tory et al., 2007]
COR27	JS	ex36	c.4732G>T	p.Glu1578X	ex36	c.4732G>T	p.Glu1578X	Europe	[Valente et al., 2006]
MTI133	JS	ex42	c.5824C>T	p.Gln1924X	ex42	c.5824C>T	p.Gln1924X	Palestina	[Valente et al., 2006]
COR22	JS	ex2	c.21G>T	p.Trp7Cys	ex2	c.21G>T	p.Trp7Cys	Pakistan	[Valente et al., 2006]
MK05	JS	ex28	c.3176delT	p. Ile1059fs	ex28	c.3176delT	p. Ile1059fs	Turkey	[Valente et al., 2006]
In 3 families	JS	ex31	c.3811C>T	p.Arg1271X	ex43	c.5734delT	p.Trp1912fs Trp1911fs	Europe	[Brancati et al., 2007; den Hollander et al., 2006; Helou et al., 2007; Perrault et al., 2007]
A989	JS	ex38	c.4882C>T	p.Gln1682X	ex44	c.5941G>T	p.Glu1981X	Europe	[Helou et al., 2007]
F101	JS	int12	c.1066-1G>A	pot. missplicing	ex39	c.5163delT	p.Thr1721fs	America	[Helou et al., 2007]
F57	SLS	ex2	c.1A>G	p.Met1? p.Met1Val	ex15	c.1419_1423delAATAA	p.Ile474fs Lys473fs	Europe	[Helou et al., 2007]
in 11 families	LCA	int26	c.2991+1655A>G	p.Cys998X ^	int26	c.2991+1655A	p.C998X ^	Europe	[Cideciyan et al., 2007; den Hollander et al., 2006; Perrault et al., 2007]
13168	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex22	c.2249T>G	p.Leu750X	Europe	[den Hollander et al., 2006]
14964	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex55	c.7341dupA	p.Leu2448fs	Europe	[den Hollander et al., 2006]
15103	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex21	c.2118_2122dupTCAGC	p.Thr709fs	Europe	[den Hollander et al., 2006]
15212	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex43	c.5866G>T	p.Glu1956X	Europe	[den Hollander et al., 2006]
17971	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex31	c.3814C>T	p.Arg1272X	Europe	[den Hollander et al., 2006]
20152	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex10	c.679_680delGA	p.Glu227fs	Europe	[den Hollander et al., 2006]

		Allele 1			Allele 2			
Family ID	**Syndrome**	**Exon/int.**	**Mutation 1**	**Effect on protein**	**Exon/int.**	**Mutation 2**	**Effect on protein**	**Origin**	**Reference**
21393	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex5	c.265dupA	p.Thr89fs	Europe	[den Hollander et al., 2006]
21918	LCA	int26	c.2991+1655A>G	p.Cys998X ^	int3	c.180+1G>T	pot. missplicing	Europe	[den Hollander et al., 2006]
27242	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex16	c.1550delT	p.Leu517X	Canada	[den Hollander et al., 2006]
27245	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex32	c.4115_4116delTA	p. Ile1372fs	Canada	[den Hollander et al., 2006]
27246	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex37	c.4966G>T	p.Glu1656X	Canada	[den Hollander et al., 2006]
27250	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex42	c.5813_5817delCTTTA	p.Thr1938fs	Europe	[den Hollander et al., 2006]
P3	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex37	c.4882C>T	p.Gln1628X	n.d.	[Cideciyan et al., 2007]
P4 and P5	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex42	c.5668G>T	p.Gly1890X	n.d.	[Cideciyan et al., 2007]
P7	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex14	c.1260_1264delTAAAG	p.Lys421fs Thr420fs	n.d.	[Cideciyan et al., 2007]
in 2 families	LCA	int26	c.2991+1655A>G	p.Cys998X ^	int40	c.5587-1G>C	pot. missplicing	Europe	[Perrault et al., 2007]
654	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex19	c.1855_1858delAAAG	p.Arg621fs	Europe	[Perrault et al., 2007]
726	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex28	c.3175dupA 3175insA	p.Ile1059fs	Europe	[Perrault et al., 2007]
798	LCA	int26	c.2991+1655A>G	p.Cys998X ^	int38	c.5226+1G>A	pot. missplicing	Europe	[Perrault et al., 2007]
in 3 families	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex38	c.5163delT	p.Thr1722fs Thr1721fs	Europe	[Perrault et al., 2007]
659	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex20	c.1992delT	p.Pro665fs	Europe	[Perrault et al., 2007]
552	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex14	c.1219_1220delAT	p.Met407fs	Europe	[Perrault et al., 2007]
105	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex32	c.5255_5256delGG	p.Ala1753fs Arg1752fs	Europe	[Perrault et al., 2007]
166	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex48	c.6604delA	p. Ile2202fs	Europe	[Perrault et al., 2007]
247	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex37	c.4882C>T	p.Gln1628X	Europe	[Perrault et al., 2007]
264	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex31	c.3922C>T	p.Gln1308X	Europe	[Perrault et al., 2007]
334	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex42	c.5850delT	p.Phe1950fs	Europe	[Perrault et al., 2007]
355	LCA	int26	c.2991+1655A>G	p.Cys998X ^	int19	c.1910-2A>G	pot. missplicing	Europe	[Perrault et al., 2007]
382	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex28	c.3292G>T	p.Glu1098X	Europe	[Perrault et al., 2007]
389	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex37	c.4962_4963delAA	p.Gln1654fs	Europe	[Perrault et al., 2007]
445	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex31	c.4028delA	p.Lys1343fs	Europe	[Perrault et al., 2007]
513	LCA	int26	c.2991+1655A>G	p.Cys998X ^	ex6	c.384_387delTAGA	p.Asp128fs	Europe	[Perrault et al., 2007]
416	LCA	ex16	c.1593C>A	p.Tyr531X	ex1	c.2T>A	p.Met1Lys	Europe	[Perrault et al., 2007]
in 6 families	LCA	ex36	c.4723A>T	p.Lys1575X	ex36	c.4723A>T	p.Lys1575X	Europe	[Perrault et al., 2007]
809	LCA	ex36	c.4723A>T	p.Lys1575X	ex17	c.1709C>G	p.Ser570X	Europe	[Perrault et al., 2007]
797	LCA	ex42	c.5850delT	p.Phe1950fs	int40	c.5587-1G>C	pot. missplicing	Europe	[Perrault et al., 2007]

		Allele 1			Allele 2				
Family ID	**Syndrome**	**Exon/int.**	**Mutation 1**	**Effect on protein**	**Exon/int.**	**Mutation 2**	**Effect on protein**	**Origin**	**Reference**
800	LCA	ex42	c.5850delT	p.Phe1950fs	ex35	c.4661_4663delAAG	p.Glu1554del	Europe	[Perrault et al., 2007]
543	LCA	int17	c.1711+5G>A	pot. missplicing	int40	c.5587-1G>C	pot. missplicing	Europe	[Perrault et al., 2007]
A3	LCA	ex20	c.1936C>T	p.Gln646X	ex48	c.6604delA	p.Ile2202fs Ile2203fs	Europe	[Perrault et al., 2007]
COR083	JSRD-SLS	ex39	c.5163delT	p.Thr1721fs	ex39	c.5163delT	p.Thr1722fs Thr1721fs	Europe	[Brancati et al., 2007]
COR145	JSRD-SLS	ex17	1657_1666delA ¤	p.Leu552fs	ex44	c.6031C>T	p.Arg2011X	Europe	[Brancati et al., 2007]
MTI333aa and b	JSRD-SLS	ex37	c.4882C>T	p.Gln1628X	ex41	c.5610_5614delCAAA c.5610delCAAA	p.Gln1871fs Lys1870fs	America	[Brancati et al., 2007]
COR109	JSRD-SLS	ex17	c.1683delA 1682_1683delA	p.Glu562fs Ala560fs	ex31	c.3814C>T	p.Arg1272X	Europe	[Brancati et al., 2007]
COR084	JSRD-SLS	ex37	c.4882C>T	p.Gln1628X	ex43	c.5941G>T	p.Glu1981X	Russia	[Brancati et al., 2007]
MTI154	JSRD-SLS	ex41	c.5668G>T	p.Gly1890X	ex41	c.5668G>T	p.Gly1890X	India	[Brancati et al., 2007]
MTI328	JSRD-SLS	ex20	c.1985A>T	p.Gln662X	ex46	c.6277delG	p.Val2093fs Val2092fs	America	[Brancati et al., 2007]
MTI273	JSRD-SLS	ex36	c.4791_4794delTAAA 4786_4790delTAAA	p.Lys1598fs Ser1595fs	ex36	c.4791_4794delTAAA 4786_4790delTAAA	Lys1598fs Ser1595fs	Turkey	[Brancati et al., 2007]
COR125	JSRD-SLS	ex40	c.5434_5435delGA 5431_5433delGA	p.Glu1812fs Asn1810fs	ex41	c.5668G>T	p.Gly1890X	Europe	[Brancati et al., 2007]
MTI487	JSRD-SLS	ex42	c.5722G>T	p.Glu1908X	ex42	c.5722G>T	p.Glu1908X	Turkey	[Brancati et al., 2007]
MTI118	JSRD-SLS	ex28	c.3175dupA 3167_3175insA	p.Ile1059fs Ile1055fs	ex41	c.5668G>T	p.Gly1890X	Europe	[Brancati et al., 2007]
MTI111a and b	JSRD-SLS	ex44	c.6072C>A	p.Tyr2024X	ex54	c.7317_7320dupCTCT 7321dupCTCT	p.Leu2441fs Leu2440fs	Laos	[Brancati et al., 2007]
COR031	JSRD-SLS	ex34	c.4393C>T	p.Arg1465X	ex36	c.4723A>T	p.Lys1575X	Europe	[Brancati et al., 2007]
MTI012 MTI587	JSRD	ex41	c.5668G>T	p.Gly1890X	ex41	c.5668G>T	p.Gly1890X	United Arab Emirates	[Brancati et al., 2007; Valente et al., 2006]
RPGRIP1L (MKS5)									
F222-1	CORS	ex6	c.697A>T	p.Lys233X	ex15	c.1843A>C	p.Thr615Pro	Europe	[Delous et al., 2007]
F58-1	CORS	ex6	c.757C>T	p.Gln253X	ex15	c.1843A>C	p.Thr615Pro	Europe	[Delous et al., 2007]
F42-1, F42-2	CORS	ex15	c.2083G>C	p.Ala695Pro	ex15	c.2083G>C	p.Ala695Pro	Europe	[Delous et al., 2007]
UW42	JS	int16	c.2305-1G>A	pot. missplicing	int16	c.2305-1G>A	pot. missplicing	Turkey	[Arts et al., 2007]

		Allele 1			Allele 2			
Family ID	**Syndrome**	**Exon/int.**	**Mutation 1**	**Effect on protein**	**Exon/int.**	**Mutation 2**	**Effect on protein**	**Origin**	**Reference**
UW43	JS	ex15	c.1721delA	p. Tyr574fs	ex15	c.1721delA	p. Tyr574fs	Turkey	[Arts et al., 2007]
UW15	JS	ex15	c.2050C>T	p.Gln684X	ex15	c.1843A>C	p.Thr615Pro	Europe	[Arts et al., 2007]
in 4 families	JS	ex15	c.1843A>C	p.Thr615Pro	ex 15	c.1843A>C	p.Thr615Pro	Europe	[Brancati et al., 2008b; Wolf et al., 2005]
Family B	JS	ex16	c.2269delA 2268_2269delA	p.Thr757fs Ile756fs	ex 16	c.2269delA 2268_2269delA	p.Thr757fs Ile756fs	Morocco	[Brancati et al., 2008b]
A762 II-3	CORS	ex15	c.1897T>C	p.Cys633Arg	ex15	c.1897T>C	p.Cys633Arg	Europe	[Wolf et al., 2005]
CC2D2A (MKS6)									
Mianwali fam	JS[x]	int18	c.2182-1G>C	p.Val728fs ^	int18	c.2182-1G>C	p.Val728fs ^	Pakistan	[Noor et al., 2008]
UW36-IV:4, UW48-IV:7	JS	ex27	c.3364C>T	p.Pro1122Ser	ex27	c.3364C>T	p.Pro1122Ser	Saudi Arabian	[Gorden et al., 2008]
UW47-II:1	JS	ex25	c.3055C>T	p.Arg1019X	ex25	c.3288G>C	p.Gln1096His	n.d.	[Gorden et al., 2008]
UW41-IV:1	JS	ex23	c.2848C>T	p.Arg950X	ex23	c.2848C>T	p.Arg950X	n.d.	[Gorden et al., 2008]
F871-II:1	JS	ex36	c.4652T>C	p.Leu1551Pro	ex36	c.4652T>C	p.Leu1551Pro	n.d.	[Gorden et al., 2008]
UW50-VI	JS	ex36	c.4582C>T	p.Arg1528Cys	ex36	c.4582C>T	p.Arg1528Cys	Levanten Arab	[Gorden et al., 2008]
UW49-II:1	JS	ex27	c.3289delG	p.Val1097fs	ex36	c.4582C>T	p.Arg1528Cys	n.d.	[Gorden et al., 2008]

Nucleotide numbering reflects cDNA numbering with +1 corresponding to the A of the ATG translation initiation codon in the reference sequence. The initiation codon is 1.
[#] Mutation marking not recognized by the Mutalyzer program.
[x] Original publication by Noor et al. (2008) reported patients with autosomal recessive mental retardation (ARMR), but Gorden et al. considered the phenotype to resemble more likely JS.
[§] The authors could not determine which of the three detected variants were the disease causing mutations.
[a] A homozygous deletion in the *NPHP1* gene is also observed in the family.
¤ Unclear mutation.
n.d. not determined.

SUPP. TABLE S1 REFERENCES

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Supp. Table S2. PolyPhen ja SIFT predictions of the new variants.

Gene	Amino acid substitution	PolyPhen	SIFT
MKS1	Arg166Trp	Probably damaging	Tolerated
MKS3	Tyr54Cys	Probably damaging	Not tolerated
MKS3	Ser245Phe	Possibly damaging	Not tolerated
MKS3	Trp296Cys	Probably damaging	Tolerated
MKS3	Cys615Arg	Probably damaging	Tolerated
MKS6	Thr1114Met	Possibly damaging	Tolerated