

**Supplemental Table 1.** PCR primers and conditions.

Gene	Exon	Primer Sequences 5' to 3'; Forward and Reverse Primers	Annealing Temp	Amplimer Size (nt)	Notes
<b>CA4 (RP17)</b>	1	GGAGAGCGGAGGATTTTCAGATG – TATCAGGGACTCCCAACTCG	56° C	555	nested sequencing; HotStarTaq and Q-Solution*
	1 seq	TGGAGAGGGGAGGGCAGGC – AGTCCTCCCGCACCCCTCGC	---	---	sequencing primers
	2	TCCTCCTGTGTGTGTGAGC – TCCATTTCCGGCTTGCGTGG	56° C	185	
	3	TGAAGTTGTCCCTACCCCG – GTGCTCTCTCCTCCTGGAAG	52° C	618	
	4-5,7	GAGCTCATGAAGGTTGGGAGGCAGG – ACAGCTCAGGCTTTTCTTGC	64° C	1307	nested sequencing; HotStarTaq without Q-Solution
	4 seq-5 seq	TTGGGAGGCAGGAGACAATG – AGTCCCAGGTCTCTGAATCC	---	---	sequencing primers
	7s	TCTCCCACCCCTCACTGAC – CTCCCCACCTGTATCCTG	---	---	sequencing primers
	6	TCCAACCTCCGCCTCTGTTTC – CACCCACCCCTTCCTTTGTG	60° C	306	
<b>CRX</b>	8	GGTCACACGGCAGGGAGTG – GGGACAGGGTGGGGTAGTTG	60° C	429	
	1	CTGCACGTCACCCCATGGTGAGTAAC – CAGAGGTCTCCAAGAGATGAGGCC	64° C	257	
	2	GGATGGAATTCTTGGTCATCCCAG – CTCTTTGTTCCGGGCAGGCCTC	64° C	314	
	3a	GGATGCAAAGTAGACAGATG – GGCGTAGGTCATGGCATAGGG	60° C	463	
	3b	CCTCCACAGATGTGTGTCCAGAC – TGGGAGAAAGGTAGGGGTCTAGG	64° C	294	
3c	GCCTCCGCTTTCTGCTCTTC – GCCCGATGGAGAGAGATGGAGACTGC	60° C	340		

<b>FSCN2</b>	1	AGGGGGTTCGTGACGCC – TCGGTGCCTCCGAAGA	60° C	991	
	1 seq	TCCGCAGCAGCCACCT – TCGGTGCCTCCGAAGAA	---	---	sequencing primers
	2	GGTCTCTGAGAGGTGCCTTC – ATGGGGATGATGGAGGAGG	60° C	347	
	3	GATTGCCGTAGCAGCTCAGT – TCCAGCTCTTGGTGGAGATG	60° C	399	
	4a	CACATGAGGCAATGGCA – CAGGTGCAAGACGTCGTAGA	60° C	263	
	4b	AACCAGCTGGACACCAA – ACTCGAAGACGAAGTCCTCG	58° C	245	
	5	ACGTCTTCCACCTGAGCTTC – GAGAGACACGGTTTTAATGCG	60° C	381	
<b>IMPDH1 (RP10)</b>	1	GGCGCGCGTCAGCAGTAGCAG – TCGCCGTGCCACGTCCGTCTGCTC	56° C	373	
	2	ACCCAGTAGACCTTTCGCT – ATGCCCTGCCCTGAGCAAG	56° C	364	
	3-4	TGGGTGATAAACTCTTTACGTGG – GCCTCTGAGGTGGGGACT	56° C	399	
	5-6	GCTTTCTTCCAGCCTTTTCCT – AACAACGGGACTGTGGAC	56° C	548	
	7	AGTGAATCTCTGGAGTGGTC – CCTGGGTCCCTATAAACCTC	56° C	377	nested sequencing
	7 seq	CATCTTCACCCTCCTAAACATC – AACCTCCACTCTGCTGAACCAC	---	---	sequencing primers
	8-9	TTCATCCACTCAGGCTCTCC – CTGGGGAACAAAGGCGAGG	56° C	561	
	10-11	ACACTCATCCTGGTGGTATTTG – CCATCTGGGGAAGTCGGTG	56° C	644	
	12-13	AAGAGGTGGGGTGGGGT – ACCTGCCCAACCCACTGC	64° C	438	HotStarTaq without Q-Solution

	14	GGGAAAGGGTTTTGGGAAG – TGTGCCCAAAGTGGACAC	56° C	154	
<b>NRL</b>	1	CTCAGAGAGCTGGCCCTTTA - AGAGGGGGTTCTAGGTGAGC	53° C	217	
	2	CCATGTGCTCCAGACCTCTC – CTCTCTTGGGCAGTCCTCCT	64° C	504	
	3	GGGGATCCCAGAGACGAG – AAGGCGCTCTGGTAACGAT	64° C	727	
	3 seq	GCGGAGCCAGGTAGCGTC – AAGGCGCTCTGGTAAGCAT	---	---	sequencing primers
<b>PRPF3 (RP18)</b>	11	GCAAATGCTGAAGTGATTGG – AAGGTACTTGCCCGAGGTTT	56° C	317	
<b>PRPF8 (RP13)</b>	42	CATCTTGCTGTGAACCGC – TGGCTGGCTTGGAGGTGG	65° C	499	nested sequencing
	42 seq	TGGCTGGCTTGGAGGTGG – ACAAGCCATCAGGAGGTC	---	---	sequencing primers
<b>PRPF31 (RP11)</b>	2-3	CTGGGGGAGAATCATCGCTC – AAGGCTCTGGAAAAGGCT	56° C	557	
	4	CGAGAGGGGGTAGGGATTTAGATAC – ACCTCGATCTGAGCTTGGGCTTAG	52° C	252	
	5	ACAACCTCCTGTCCCGTTTACC – GCACCCACCTTCTCTGCG	56° C	148	
	6-7	CTTCCTGAGTTCCCGAGCC – AGCTTTCCCAAGGTCACAGTG	56° C	574	nested sequencing due to CA repeat
	6 seq-7 seq	TCCAGCCTAATCCCCAATC – AACCGAGAGGGCTGGGGC	---	---	sequencing primers
	8	TACTCACCCCCACCTCTCTG – CCCATTTGATAGGGGAGGAG	56° C	467	HotStarTaq without Q-Solution
	9	AAAGCCCCCTTCCAGGACC – GGTAGGACAGTGCTCGCTG	62° C	315	
	10-11	AGGCAGCATTAGGTGCTGAT – CAGCGCCCTAGACATGAG	64° C	540	HotStarTaq without Q-Solution

	12-13	GGAGGGGGTGCCTCGGTGG – TGCCAGGAGCTGTCTACGTC	62° C	524	HotStarTaq without Q-Solution
	14	ACCCATCATCCTCTCTCCCTCAC – CCAGAACCCGATCCTAGCC	56° C	239	
<b>RDS</b>	1a-1b	AGCTGTGCTGTGGGAAGCAA – CTTCAGCCAGGGCTTCCATC	62° C	338	
	1c-1d	TGGACCCAGCCAAGTATGCC – CTGACCCCAGGACTGGAAGC	62° C	352	
	2	AAGCCCATCTCCAGCTGTCT – TTACCCTTACCCCCAGCTGGCCCAG	62° C	311	
	3	TGCCTCTAAATCTCCTCTCCCAC – TGCACTATTTCTCAGTGTTCCGGG	62° C	286	
<b>RHO</b>	1	GTCAGCCACAAGGGCCACAG – CACACCCACCCACACCC	62° C	410	
	2	TGCACCCTCCTTAGGCAGTG – TGACTGGAGGACCCTACAAAGA	62° C	284	
	3-4	GTCTCCCTACCTGCCTGTCC – TAGCTTGTCCTTGGCAGGCAGC	62° C	613	
	5	CCCTGACTCAAGCCTCTTGC – GTCGGCCACAGAGTCCTAGG	62° C	164	
<b>ROM1</b>	1	GGGGCTGACAGGGGGGCG – GGATTCAAGGCAGCAGGAGG	64° C	829	
	1a seq	CTGACTCAGCATCCTGCC – GGGGCAGGACAGGGAAGT	---	---	sequencing primers
	1b seq	CCTGTCAGTTCCTGTCCCTGC – GAGGAAGGGAGACGCAAATCAC	---	---	sequencing primers
	2	AGGCCTCTATCTCCAGACAT – AGTCCCGGGTGGGAGGAGGT	61° C	337	
	3	CAACCCTGGGCCTCTTGGAA – GCCTTGGTAAGGAGTTGTGA	56° C	353	
<b>RP1</b>	4a	TGGGGAAAACAAGTCTGAGT – TGCTGGCAACAGATGACAAA	56° C	539	

	4b	GCTCAGTGTGGTTTAAACAAAAC – ACCGATTTCTCTTTTTTTAGGTG	54° C	441	
	4c	ATTCCAAGGTTCAAGGAC – CAATTTACCACACTCGTTTCATTTC	56° C	541	
	4d	GCAGAACATAAATCCATATCCAAC – ATCTACTTGAATGGCAGC	56° C	457	
<b>RP9 (PAP1)</b>	1	GTTGCCCGAGCGGCGCT – TGGCCGCGCGCGGACGGCA	62° C	213	
	2	AAATCTCTGATTA AAAATCCTATAGCC – AAAAGGAGATTTAACATCATGCAA	62° C	230	
	3	CAGGAAAAGCCAGGCAAG – GAGGGCTGTGATGAGAACAAG	56° C	320	
	4	TGCTGATTCTTTATCTTGAGTAGGTG – TGGTGACTTTCTGCTTCACTG	54° C	231	
	5	GGTTTTTCATAACATAGGCATTTCA – TGTTTACTGCACCATTCTCT	53° C	245	
	6	CATCCTATACTGCTTTTGAATGACA – TGCATCTTCTCTGTTCCCTG	64° C	357	hybridizes to both orthologs
	6 alt	AGTAGGCATTCAGGTGGA – TGCATCTTCTCTGTTCCCTG	64° C	432	hybridizes to PAP1 only
<b>RPGR</b>	ORF 15a	CGGTATGGCAGGAAATTGATTGA – CCATCCTCTGCTTCTCCAC	61° C	299	
	ORF15b	AGGAAGGAGCAGAGGATTCA – CCTTCTCTGCTAGTTCCTTCTCTCCC	56° C	330	
	ORF 15c	GGGAGAAGGGGGAGAAAGACAAGG – TTCCCTCCTCTACTT	56° C	448	reverse direction only; 2 x 50 ul PCR reactions, gel purified
	ORF15d	TGTCTGACTGGCCATAATCG – TGTCTGACTGGCCATAATCG	60-58° C	1630	nested sequencing; 3 x 50 ul PCR reactions, gel purified; HotStarTaq and Q-Solution
	15d nest 1	GTTTGCCATATTTACAGATCC	58° C	---	sequencing primers 1, one direction only
	15d nest 2	GAAGTAGAGGGAGGGGAAGTAG	58° C	---	sequencing primers 2, one direction only

15d nest 3	TCCTTCCTCCTCTTCCCCCTCCC	63° C	---	sequencing primers 3, one direction only
ORF15e	GCAGGATGGAGAGGAGTACA – GAGAGAGGCCAAAATTTACCA	56° C	415	

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\*HotStarTaq (Qiagen) and Q-Solution (Qiagen)

**Supplemental Table 2.** Cohort family data

Family ID	Ethnicity	Mutation	Gen	Aff	M-M	Skip	Lods d:r	Lods d:x	Prior Publication
BCMAD013	Caucasian	RP1 Leu762fs/ter777 (2285_2289del)	3	5	y	0	10	7	
BCMAD014	Caucasian		6	25	y	2	40	9	
BCMAD015	Caucasian		5	9	n	0	13	4	
BCMAD032	Caucasian	RHO Pro23His (68 C>A)	4	4	n	0	11	1	1
BCMAD033	Caucasian	RDS Tyr141Cys (422 A>G)	3	5	n	0	11	4	1
BCMAD900	Caucasian		6	18	n	2	20	-1	
RFS001	Caucasian	RHO Pro23His (68 C>A)	4	6	n	0	15	1	1
RFS003	Caucasian	RHO Asp190Asn (568 G>A)	3	6	n	0	10	3	1
RFS004	Caucasian	RHO Leu46Arg (137 T>G)	3	4	n	0	11	1	1,2
RFS009	Caucasian		4	6	n	0	13	4	
RFS010	Caucasian	RHO Pro23His (68 C>A)	5	14	y	0	21	5	1
RFS012	Caucasian	RHO IVS4/exon5junction (IVS4-1 G>A)	4	10	y	0	11	9	
RFS015	Caucasian	IMPDH1 Asp226Asn (676 G>A)	6	29	y	0	50	20	3
RFS016	African Amer.		5	9	y	1	14	1	
RFS019	Caucasian	PRPF8 Glu2331fs/ter2358 (6991delG)	3	5	y	0	11	8	
RFS020	Caucasian	RHO Pro23His (68 C>A)	5	15	y	0	16	7	
RFS021	Caucasian		4	10	n	1	14	-1	
RFS023	Caucasian	RHO Pro23His (68 C>A)	4	10	n	0	20	4	1
RFS024	Caucasian	PRPF31 Cys299Arg (895 T>C)	4	13	y	0	11	9	
RFS025	Caucasian	RHO Pro23His (68 C>A)	3	3	y	0	10	5	1
RFS026	Caucasian		3	2	n	1	6	2	
RFS027	Caucasian	RDS exon2/IVS2junction (IVS2+3 A>T)	6	40	y	0	23	8	1
RFS029	Caucasian		3	5	n	0	10	3	
RFS030	Caucasian	PRPF8 Arg2310Gly (6928 A>G)	4	4	n	0	14	2	
RFS031	Caucasian	PRPF8 Glu2331fs/ter2358 (6991delG)	5	33	y	0	86	53	
RFS033	Caucasian	PRPF31 Gln74ter (220 C>T)	3	2	n	1	5	0	
RFS034	Caucasian		4	10	y	0	34	14	
RFS035	Caucasian		3	4	y	0	6	4	
RFS036	Caucasian	RHO Cys110Phe (329 G>T)	3	4	n	0	11	3	1
RFS037	Caucasian	RHO Pro23His (68 C>A)	3	4	y	0	11	5	1
RFS038	Caucasian		4	8	y	0	8	4	
RFS046	Caucasian		2	2	y	0	7	4	
RFS048	Caucasian		4	6	n	0	15	0	

RFS049	Caucasian	RHO Pro171Ser (511 C>T)	2	2	y	0	7	5	1
RFS053	Caucasian	PRPF8 Phe2304Leu (6912 C>G)	2	2	n	0	7	2	
RFS061	unknown		3	5	y	0	11	5	
RFS066	Caucasian		5	15	y	0	21	8	
RFS069	Caucasian	RHO Pro23His (68 C>A)	4	7	y	0	12	4	1
RFS070	Caucasian	RDS Pro216Leu (647 C>T)	2	6	n	0	9	2	1
RFS071	unknown	RHO Pro23His (68 C>A)	2	3	y	0	23	12	1
RFS072	Hispanic	RHO Gly106Arg (318 A>G)	3	15	y	0	7	6	1
RFS073	Caucasian		3	4	y	0	11	4	
RFS076	Caucasian		2	4	n	1	2	1	
RFS078	Caucasian	PRPF31 Asn131fs/ter197 (390delC)	2	3	y	0	7	4	
RFS079	Caucasian	RDS Pro210Arg (629 C>G)	3	6	n	0	10	1	1
RFS083	Caucasian	RDS exon2/IVS2junction (IVS2+3 A>T)	5	9	y	0	9	3	1
RFS084	Caucasian	PRPF3 Pro493Ser (1477 C>T)	3	5	n	0	11	3	
RFS085	African Amer.	PRPF8 Ala2328Val (6983 C>T)	3	3	n	0	10	3	
RFS087	Caucasian	CRX Arg41Gln (122 G>A)	4	4	y	0	14	5	1,4
RFS092	Caucasian		4	10	y	2	14	8	
RFS094	Hispanic		5	4	n	1	5	-1	
RFS096	Caucasian	RHO Arg135Trp (403 C>T)	3	6	y	0	14	5	1
RFS097	Caucasian		3	4	y	0	7	4	
RFS099	Caucasian		4	6	n	0	7	0	
RFS101	Caucasian	PRPF31 exon10/IVS10junction (IVS10+1 G>A)	4	7	n	0	13	4	
RFS118	Caucasian	RDS Arg172Gln (516 G>T)	4	7	n	0	14	4	
RFS119	Caucasian		4	4	n	5	3	-7	
RFS132	African Amer.		6	13	y	0	35	10	
RFS136	Caucasian	RHO Pro23His (68 C>A)	5	12	n	0	18	6	1
RFS137	Caucasian	RP1 Arg677ter (2029 C>T)	6	22	y	0	23	18	1,5
RFS138	Hispanic		4	14	y	0	15	12	
RFS146	Caucasian		4	10	y	0	8	7	
RFS153	Caucasian	RHO Pro170Arg (509 C>G)	3	7	n	0	9	3	1
RFS156	unknown	RDS exon2/IVS2junction (IVS2+3 A>T)	3	5	y	0	12	6	1
RFS163	African Amer.	RHO Ala164Val (491 C>T)	3	6	n	0	9	2	1
RFS164	Caucasian		2	4	y	0	8	4	
RFS169	Caucasian		5	13	y	0	18	6	
RFS170	Caucasian		3	12	y	0	9	3	
RFS173	Caucasian	RHO Gly106Trp (316 G>T)	4	7	n	1	10	1	



RFS175	Caucasian		3	3	n	0	10	2	
RFS178	Caucasian		4	12	y	2	17	-1	
RFS179	Caucasian		3	3	n	0	11	1	
RFS182	Caucasian	RHO Thr58Arg (173 C>G)	4	7	y	0	14	6	
RFS183	Caucasian		3	2	y	1	5	3	
RFS184	Caucasian	RDS Pro216Leu (647 C>T)	3	3	n	0	5	-1	
RFS191	Caucasian		3	7	n	0	11	0	
RFS202	Caucasian	PRPF8 IVS41/exon42junction (IVS41-4 G>A)	3	3	y	0	11	3	
RFS204	Caucasian	RHO Pro23His (68 C>A)	4	19	y	0	27	5	
RFS207	Caucasian	PRPF31 Gly253fs/ter317 (758_767del)	4	5	y	0	13	7	
RFS222	Caucasian		3	5	y	0	6	3	
RFS268	Caucasian		4	13	y	0	23	7	
RFS275	Caucasian	RDS exon2/IVS2junction (IVS2+3 A>T)	3	5	n	0	15	4	
RFS900	Caucasian	CRX Leu146_Pro149del (436_447del)	6	12	y	0	56	17	1, 4
RFS910	Caucasian		4	9	y	0	15	8	
RP01	Caucasian	RP1 Arg677ter (2029 C>T)	8	200+	y	2	206	78	1,5,6
RP01B	Caucasian	RHO Arg135Leu (404 G>T + 405 G>T)	5	29	y	0	21	8	
UTAD002	Caucasian		3	3	n	0	11	-1	
UTAD003	Caucasian		6	17	y	0	44	18	
UTAD005	Caucasian	RPGR Glu256fs/ter492 (ORF15 + 764_765del)	4	6	n	2	17	3	
UTAD007	Hispanic		3	6	n	0	10	2	
UTAD008	unknown		3	4	n	0	11	2	
UTAD010	Caucasian	RHO Pro23His (68 C>A)	3	3	n	0	11	1	1
UTAD013	Caucasian	RHO Gly106Trp (316 G>T)	3	5	y	0	13	5	1
UTAD017	Caucasian		3	4	n	0	9	0	
UTAD018	unknown	RHO Pro23His (68 C>A)	5	7	y	1	16	14	1
UTAD022	Caucasian	RHO His211Arg (632 A>G)	4	4	n	1	8	0	
UTAD023	Caucasian	RDS Val206_Val209del (616_627del)	3	5	y	0	14	8	1
UTAD028	Caucasian		4	4	n	0	14	2	
UTAD029	Caucasian	RP1 Leu762fs/ter777 (2285_2289del)	3	4	y	0	7	4	1, 5
UTAD030	unknown	IMPDH1 Asp226Asn (676 G>A)	5	10	n	0	14	3	7
UTAD034	unknown		2	3	y	0	6	3	
UTAD035	African Amer.		4	12	y	0	24	20	
UTAD036	Caucasian		5	10	n	2	26	1	
UTAD039	Caucasian	RP1 Leu762fs/ter777 (2285_2289del)	5	15	y	0	26	10	1, 5
UTAD040	Caucasian		3	2	n	1	5	-1	

UTAD042	unknown	RHO Ser270Arg (810 C>A)	3	3	n	0	11	2	1
UTAD045	unknown	IMPDH1 Asp226Asn (676 G>A)	6	38	y	0	40	15	3,8
UTAD049	Caucasian		3	6	y	0	7	4	
UTAD055	Caucasian		5	10	y	2	17	6	
UTAD056	Caucasian	RDS Gly266Asp (797 G>A)	5	13	y	1	22	6	1
UTAD058	other	RDS Gly266Asp (797 G>A)	3	5	y	0	14	12	1
UTAD069	Caucasian		4	4	n	0	14	3	
UTAD070	Caucasian	RHO Leu57Arg (170 C>T)	4	5	n	0	9	1	1
UTAD075	Caucasian		4	4	y	1	8	8	
UTAD076	Caucasian		4	18	n	1	24	0	
UTAD079	Caucasian	PRPF31 exon10/IVS10junction (1049_IVS10+20del/insCCCCT)	4	5	n	1	6	-1	
UTAD080	Hispanic		4	8	y	0	13	7	
UTAD081	Caucasian	RHO Pro347Thr 1039 C>A	3	10	y	0	15	6	1
UTAD082	Caucasian		4	9	n	1	7	3	
UTAD084	Caucasian	RHO Pro23His (68 C>A)	5	12	y	0	20	7	1
UTAD085	Caucasian	RHO Pro23His (68 C>A)	4	5	y	1	12	-3	1
UTAD088	Caucasian	RDS Pro210Leu (629 C>T)	3	3	y	0	10	5	1
UTAD089	other	RHO Glu181Lys (541 G>A)	5	17	y	0	26	12	1
UTAD094	Caucasian		3	4	n	0	6	0	
UTAD095	Asian	RDS Pro216Ser (646 C>T)	4	20	y	0	31	15	1
UTAD096	African Amer.	RHO Pro171Gln (512 C>A)	4	6	n	0	14	3	1
UTAD099	Caucasian		5	6	n	0	11	0	
UTAD102	Hispanic		3	4	y	0	11	8	
UTAD117	Caucasian		5	4	n	2	11	2	
UTAD119	Caucasian		6	12	n	2	24	5	
UTAD120	unknown	RHO Arg135Trp (403 C>T)	5	25	y	0	25	14	1
UTAD140	Caucasian	RHO Pro347Leu (1040 C>T)	3	6	?	?	15	5	1
UTAD147	Caucasian		5	8	n	2	5	1	
UTAD159	unknown	RHO Arg135Trp (403 C>T)	4	5	n	0	11	4	1
UTAD168	Caucasian		3	3	n	0	10	2	
UTAD170	unknown		4	7	n	0	14	2	
UTAD177	Caucasian	IMPDH1 Asp226Asn (676 G>A)	3	11	y	0	11	8	3,7
UTAD178	Hispanic		5	19	n	0	10	6	
UTAD181	Hispanic		3	5	n	1	13	1	
UTAD184	Caucasian	PRPF31 Glu325ter (973 G>T)	4	3	n	1	9	4	
UTAD188	Caucasian	RHO Pro347Leu (1040 C>T)	3	7	n	0	11	6	1

UTAD198	Hispanic		3	5	y	0	11	8	
UTAD2006	Caucasian		5	12	y	3	31	5	
UTAD201	Hispanic		4	10	n	0	11	4	
UTAD215	Caucasian	RDS Leu185Pro (554 T>C) + ROM1 Leu114fs/ter131 (339insG)	4	12	y	0	21	6	1
UTAD230	unknown	RHO Pro23His (68 C>A)	3	3	n	0	11	2	1
UTAD242	unknown		3	3	n	1	8	-1	
UTAD246	Asian		3	2	n	1	3	0	
UTAD247	Hispanic		4	7	n	1	10	-1	
UTAD259	unknown		3	2	n	1	6	0	
UTAD280	Caucasian	RDS Leu126Arg (377 T>G)	5	9	y	2	22	7	1
UTAD305	Caucasian		4	15	n	1	12	6	
UTAD352	Caucasian		2	2	y	0	6	3	
UTAD388	Asian		4	21	y	0	26	11	
UTAD392	Caucasian		4	9	y	0	22	13	
UTAD395	Caucasian	RHO Arg135Leu (404 G>T + 405 G>T)	3	5	y	0	11	6	
UTAD399	unknown	RDS Arg46ter (136 C>T)	2	4	y	0	5	4	1
UTAD401	other	PRPF31 exon2/IVS2junction (IVS2+1 G>A)	4	12	y	2	19	15	
UTAD404	Caucasian		4	10	n	0	17	8	
UTAD407	unknown	RHO Pro347Ala (1039 C>G)	6	33	y	1	41	21	1
UTAD410	Caucasian	PRPF3 Thr494Met (1481 C>T)	4	10	y	0	14	9	
UTAD414	unknown	RHO Pro23His (68 C>A)	2	4	y	0	8	5	1
UTAD437	Caucasian	RHO Pro23His (68 C>A)	5	4	n	1	12	2	1
UTAD441	Caucasian	RP1 Gly723ter 2167 G>T	3	6	y	2	17	10	1 (UTAD411)*
UTAD442	unknown	RPGR Arg195fs/ter229 (ORF15 + 558del)	5	18	n	0	38	5	
UTAD449	Hispanic		8	3	y	0	8	5	
UTAD472	Caucasian		5	13	n	0	21	7	
UTAD476	unknown		5	7	y	0	17	6	
UTAD479	Caucasian	PRPF31 Ala291Pro (871 G>C)	4	10	y	0	14	2	
UTAD481	Caucasian	PRPF31 Glu141ter (421 G>T)	3	6	y	0	9	7	
UTAD482	unknown		5	5	n	0	16	4	
UTAD483	Caucasian	RHO Cys187Tyr (560 G > A)	2	4	y	0	6	5	
UTAD488	Caucasian		5	13	n	0	21	1	
UTAD493	Caucasian	RHO Arg135Trp (403 C>T)	3	4	y	0	11	5	
UTAD497	Caucasian	RHO Val345Met (1033 G>A)	5	20	n	0	35	10	
UTAD498	Caucasian		3	3	n	0	10	1	
UTAD503	Caucasian	PRPF31 Met212fs/ter238 (636delG)	3	5	y	1	14	12	

UTAD506	Hispanic		3	6	y	0	7	4	
UTAD513	Hispanic	RHO Thr17Met (50 C>T)	3	4	y	0	11	9	
UTAD514	Caucasian		3	4	n	0	10	3	
UTAD519	Asian	RDS Ser198Arg (594 C>G)	3	7	y	0	9	4	
UTAD522	Caucasian		4	4	y	2	9	5	
UTAD526	Caucasian	RP1 Arg677ter (2029 C>T)	3	4	n	0	10	1	
UTAD529	Caucasian	RHO ter349Gln (1045 T>C)	4	10	y	0	15	11	
UTAD530	Caucasian		6	18	n	0	30	10	
UTAD531	unknown	RHO Asp190Asn (568 G>A)	4	5	n	0	13	4	
UTAD533	Caucasian	RHO Pro23His (68 C>A)	4	6	y	0	10	7	
UTAD537	Caucasian		4	4	n	1	9	3	
UTAD541	Asian	RDS Pro216Arg (647 C>G)	4	7	y	0	17	12	
UTAD543	Asian		5	8	n	0	17	5	
UTAD544	Asian		3	3	n	0	11	1	
UTAD545	unknown	RHO Pro267Leu (800 C>T)	3	8	n	0	17	5	
UTAD549	Caucasian		4	5	y	0	11	5	
UTAD551	Caucasian		4	3	y	1	9	3	
UTAD553	Caucasian	RHO Arg135Leu (404 G>T + 405 G>T)	5	8	y	0	13	5	
UTAD554	Caucasian	RHO Pro23His (68 C>A)	3	7	y	0	14	6	
UTAD555	African Carrib.		4	8	n	1	13	-2	
UTAD557	Caucasian	IMPDH1 Asp226Asn (676 G>A)	4	12	y	0	19	15	7
UTAD562	Caucasian		5	13	y	2	24	8	
UTAD565	Caucasian		4	8	y	1	21	3	

\*Incorrectly listed as UTAD411 in Sohocki 2001

## Supplemental Table 2 - References

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