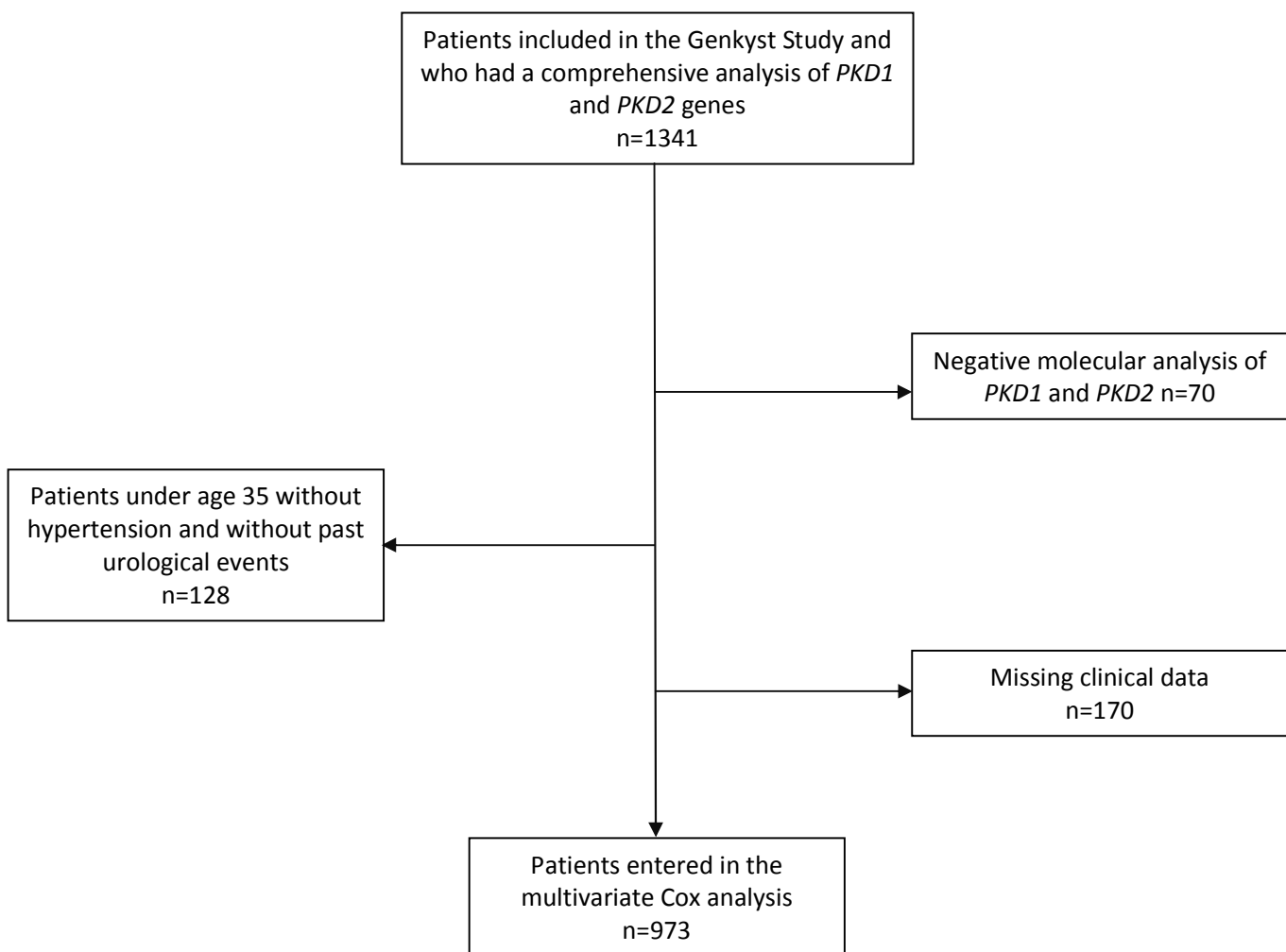
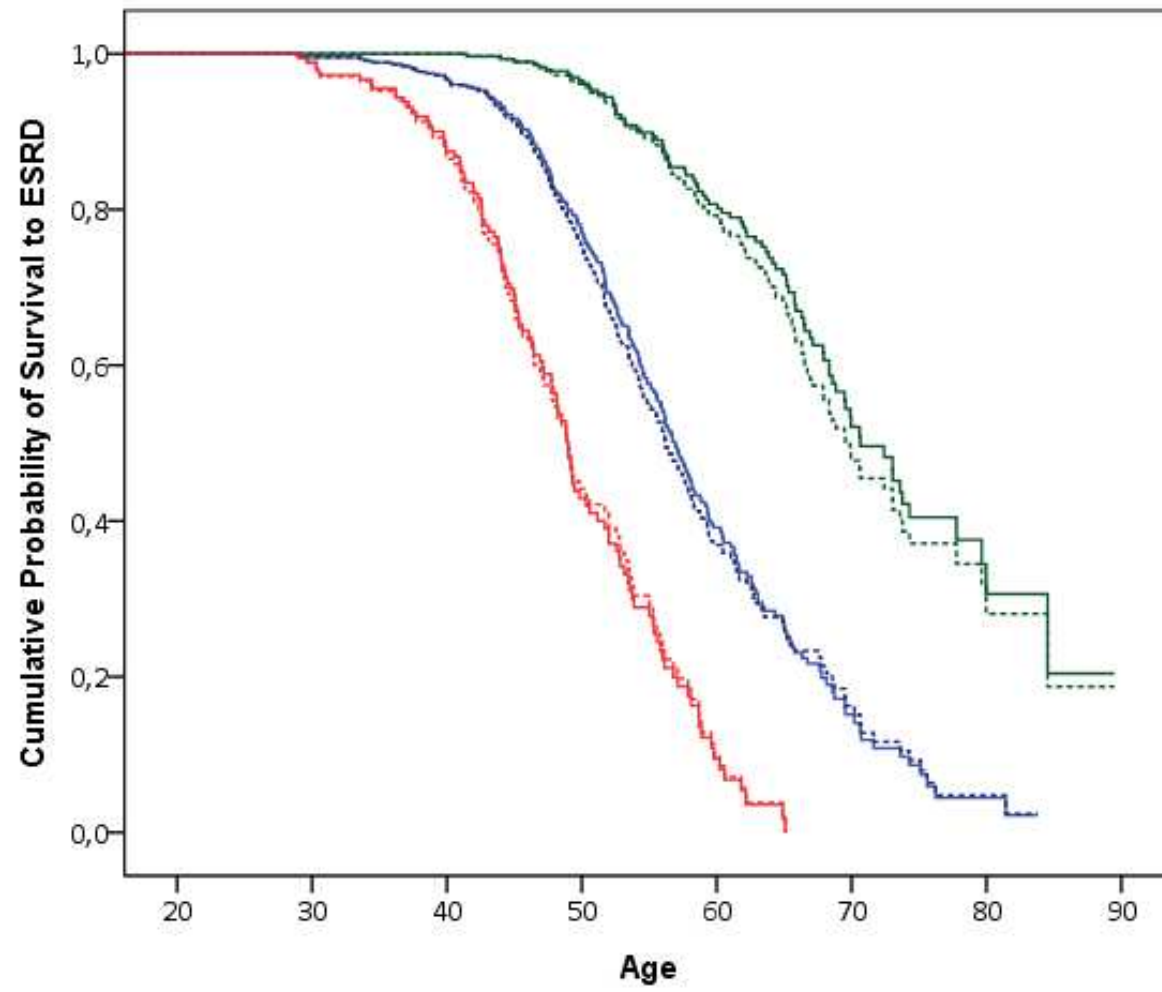


Supplemental figure 1

Flow chart of the study design: 1341 patients were included in Genkyst study between 2010 and January 2015, from which 973 were finally entered in the multivariate analysis. In addition to the 70 patients in whom the mutation was not identified, 298 patients were not entered in the final multivariate analysis either because they were under age 35 and had not developed hypertension and urological complications (n=128), either because of one or both of these clinical data were missing (n=170).



Supplemental figure 2



Comparison of renal survival curves in the three prognostic groups, low (green), intermediate (blue) and high (red) risk of progression to ESRD, in the developmental cohort (solid curves) and the 10 training data-set pooled together (dotted curves). Renal survival in the developmental cohort and the 10 training data-set pooled together did not differ.

Supplemental Table 2

Performance characteristics of the PROPKD score for the additional cut-off points (or thresholds) of 1, 2, 4, 5, 7 and 8 (in complement to the main data presented in table 5) and for different censored ages. The test is positive when the score is greater than the cut-off point and negative when the score is less than or equal to the cut-off point (n=973 patients).

Cut-off point	Censored age, years	Se (SE_Se) %	Sp (SE_Sp) %	PPV (SE_PPV)%	NPV (SE_NPV) %
1	40	100.0 (0.0)	16.9 (1.3)	4.5 (0.8)	100.0 (0.0)
	45	10.00 (0.0)	18.4 (1.5)	11.8 (1.2)	100.0 (0.0)
	50	100.0 (0.0)	22.7 (1.8)	26.6 (1.8)	100.0 (0.0)
	55	97.7 (1.0)	27.0 (2.3)	42.7 (2.1)	95.5 (2.0)
	60	96.4 (1.1)	34.6 (3.1)	60.2 (2.3)	90.6 (2.9)
	65	95.0 (1.3)	44.5 (4.3)	72.6 (2.4)	85.2 (3.7)
2	40	100.0 (0.0)	28.2 (1.5)	5.2 (0.9)	100.0 (0)
	45	97.5 (1.7)	29.9 (1.7)	13.2 (1.4)	99.1 (0.6)
	50	96.3 (1.5)	34.1 (2.0)	29.0 (1.9)	97.1 (1.2)
	55	92.4 (1.7)	39.6 (2.6)	46.0 (2.3)	90.3 (2.2)
	60	90.8 (1.7)	50.6 (3.3)	65.4 (2.4)	84.3 (2.8)
	65	87.4 (1.9)	58.4 (4.2)	76.5 (2.5)	75.0 (3.6)
4	40	94.4 (3.8)	56.2 (1.7)	7.9 (1.3)	99.6 (0.3)
	45	87.8 (3.6)	59.5 (1.8)	19.1 (2.0)	97.8 (0.67)
	50	78.2 (3.2)	65.5 (2.1)	38.8 (2.7)	91.5 (1.4)
	55	70.7 (2.9)	71.3(2.4)	58.9 (2.9)	81.3 (2.0)
	60	65.0 (2.7)	80.1 (2.6)	77.0 (2.8)	69.1 (2.5)
	65	60.4 (2.4)	85.4 (3.0)	86.5 (2.7)	58.2 (2.9)
5	40	82.6 (6.4)	71.7 (1.5)	10.4 (1.8)	99.1 (0.4)
	45	72.0 (5.0)	74.9 (1.6)	23.8 (2.7)	96.1 (0.8)
	50	60.4 (3.8)	80.6 (1.7)	46.6 (3.4)	87.9 (1.4)
	55	48.4 (3.1)	83.3 (2.0)	61.8 (3.6)	71.4 (2.0)
	60	44.5 (2.7)	90.5 (1.9)	82.7 (3.2)	61.4 (2.4)
	65	41.0 (2.6)	94.9 (1.9)	92.6 (2.6)	50.9 (2.6)

7	40	28.3 (7.6)	94.4 (0.8)	16.7 (4.8)	97.1 (0.6)
	45	22.2 (4.5)	95.0 (0.8)	32.8 (6.3)	91.8 (1.0)
	50	16.2 (2.8)	96.5 (0.8)	56.1 (7.3)	80.5 (1.5)
	55	13.0 (2.0)	97.0 (0.9)	70.7 (7.1)	66.7 (1.9)
	60	11.5 (1.7)	98.3 (0.9)	87.2 (5.8)	52.0 (2.1)
	65	10.4 (1.5)	99.3 (0.7)	95.6 (4.2)	41.7 (2.2)
8	40	14.0 (5.8)	97.6 (0.5)	18.9 (7.7)	96.6 (0.6)
	45	10.5 (3.3)	97.7 (0.6)	33.5 (9.3)	90.9 (1.0)
	50	7.2 (1.9)	98.3 (0.5)	54.7 (10.8)	79.1 (1.5)
	55	6.6 (1.5)	98.9 (0.5)	77.2 (9.8)	65.5 (1.8)
	60	5.5 (1.2)	99.6 (0.4)	92.8 (6.8)	50.7 (2.1)
	65	4.9 (1.1)	100.0 (0.0)	100.0 (0.0)	40.4 (2.2)

Definitively pathogenic mutations

Genkyst ID	Family ID	Gene	Exon/Intron	Nucleotide change	Effect on Protein	Mutation type	Reference
020108	PK11122	PKD1	1	c.(?_209)_(*1017_?)*del		Large rearrangement	This study
080009	PK10111	PKD1	1	c.(?_209)_(*1017_?)*del		Large rearrangement	Audrezet, 2012
010883	PK10111	PKD1	1	c.(?_209)_(*1017_?)*del		Large rearrangement	Audrezet, 2012
031371	PK10111	PKD1	1	c.(?_209)_(*1017_?)*del		Large rearrangement	Audrezet, 2012
131045	PK11248	PKD1	1	c.(?_209)_(*1201+?)*del		Large rearrangement	This study
021266	PK11032	PKD1	1	c.(?_209)_(*1201+?)*del		Large rearrangement	This study
021367	PK11032	PKD1	1	c.(?_209)_(*1201+?)*del		Large rearrangement	This study
020138	PK10350	PKD1	1	c.(?_209)_(*215+?)*del		Large rearrangement	Audrezet, 2012
020061	PK10132	PKD1	1	c.107_108dup	p.Cys37Profs*37	Frameshift	Rossetti, 2007
191709	PK11379	PKD1	1	c.114del	p.Cys39Alafs*34	Frameshift	This study
020083	PK10412	PKD1	1	c.158_179del	p.Gly53Valfs*13	Frameshift	Audrezet, 2012
020086	PK10412	PKD1	1	c.158_179del	p.Gly53Valfs*13	Frameshift	Audrezet, 2012
050019	PK10975	PKD1	1	c.160_166dup	p.Leu56Profs*60	Frameshift	This study
010176	PK10164	PKD1	1	c.165_171del	p.Leu56Argfs*15	Frameshift	Rossetti, 2007
060038	PK11121	PKD1	1	c.-1933_215+3456del		Large rearrangement	This study
021104	PK11121	PKD1	1	c.-1933_215+3456del		Large rearrangement	This study
030053	PK10384	PKD1	1	c.2161G>C		Splicing	Audrezet, 2012
091245	PK11160	PKD1	1	c.74_75delinsT	p.Gly25Valfs*48	Frameshift	This study
191555	PK11315	PKD1	1	c.74del	p.Gly25Alafs*48	Frameshift	Hoefle, 2010
130044	PK10592	PKD1	2	c.288-2A>G		Splicing	Rossetti, 2001
131043	PK10592	PKD1	2	c.288-2A>G		Splicing	Rossetti, 2001
131048	PK10592	PKD1	2	c.288-2A>G		Splicing	Rossetti, 2001
021320	PK10592	PKD1	2	c.288-2A>G		Splicing	Rossetti, 2001
131398	PK10592	PKD1	2	c.288-2A>G		Splicing	Rossetti, 2001
131876	PK10592	PKD1	2	c.288-2A>G		Splicing	Rossetti, 2001
021201	PK11126	PKD1	3	c.(?_288)_(*7863+?)*del		Large rearrangement	This study
021119	PK10856	PKD1	3	c.348_352del	p.Asn116Lysfs*2	Frameshift	Garcia-Gonzales, 2007
191500	PK11303	PKD1	3	c.359-2T>G		Splicing	This study
030058	PK11035	PKD1	4	c.360-131_4395del		Large rearrangement	This study
021206	PK10928	PKD1	4	c.371_386dup	p.Cys129Trpfs*5	Frameshift	This study
030040	PK10322	PKD1	4	c.393_394del	p.Cys131Trpfs*47	Frameshift	Audrezet, 2012
031300	PK10322	PKD1	4	c.393_394del	p.Cys131Trpfs*47	Frameshift	Audrezet, 2012
031026	PK10767	PKD1	4	c.412C>T	p.Arg138*	nonsense	Rossetti, 2007
031075	PK10792	PKD1	4	c.412C>T	p.Arg138*	nonsense	Rossetti, 2007
191725	PK11387	PKD1	5	c.1029del	p.Ala344Profs*121	Frameshift	This study
130043	PK10591	PKD1	5	c.1111G>T	p.Glu371*	nonsense	Audrezet, 2012
191716	PK11386	PKD1	5	c.1202-1G>C		Splicing	This study
100001	PK10291	PKD1	5	c.617dup	p.Pro207Alafs*54	Frameshift	Audrezet, 2012
040902	PK10670	PKD1	5	c.618dup	p.Pro207Alafs*54	Frameshift	This study
041528	PK10670	PKD1	5	c.618dup	p.Pro207Alafs*54	Frameshift	This study
030086	PK10622	PKD1	5	c.661_701del	p.Leu221Glyfs*26	Frameshift	Audrezet, 2012
031172	PK10622	PKD1	5	c.661_701del	p.Leu221Glyfs*26	Frameshift	Audrezet, 2012
021366	PK11208	PKD1	5	c.696_697del	p.Cys232Trpfs*28	Frameshift	This study
030803	PK10590	PKD1	5	c.726dup	p.Ala243Cysfs*18	Frameshift	This study
091019	PK10967	PKD1	5	c.730dup	p.Cys244Leufs*17	Frameshift	This study

211818	PK11483	PKD1	5	c.755dup	p-Pro253Alafs*8	Frameshift	Petola, 2005
211831	PK11483	PKD1	5	c.755dup	p-Pro253Alafs*8	Frameshift	Petola, 2005
030078	PK10463	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
060006	PK10588	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
130013	PK10643	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
031177	PK10892	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
060024	PK10588	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
091414	PK11207	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
091595	PK11390	PKD1	5	c.856_862del	p.Gly287*	Frameshift	Audrezet, 2012
031085	PK10807	PKD1	5	c.891dup	p.Leu298Alafs*73	Frameshift	This study
030974	PK10901	PKD1	5	c.935_938dup	p.Glu313Aspfs*59	Frameshift	This study
090831	PK10683	PKD1	6	c.1291C>T	p.Gln431*	nonsense	This study
191542	PK11311	PKD1	7	c.1445del	p.Pro482Argfs*76	Frameshift	This study
191776	PK11440	PKD1	7	c.1495G>T	p.Glu499*	nonsense	This study
020042	PK10386	PKD1	7	c.1516G>T	p.Glu506*	nonsense	Audrezet, 2012
020167	PK10052	PKD1	7	c.1516G>T	p.Glu506*	nonsense	Audrezet, 2012
021313	PK10052	PKD1	7	c.1516G>T	p.Glu506*	nonsense	Audrezet, 2012
021198	PK10936	PKD1	7	c.1522_1541del	p.Cys508Argfs*4	Frameshift	This study
020099	PK10472	PKD1	7	c.1524C>A	p.Cys508*	nonsense	Audrezet, 2012
021192	PK10937	PKD1	7	c.1591G>T	p.Glu531*	nonsense	This study
030033	PK10287	PKD1	7	c.1606+2T>C		Splicing	Audrezet, 2012
030057	PK10407	PKD1	8	c.1609_1723-44del	p.Pro537_Glu574del	In Frame deletion	Audrezet, 2012
060956	PK10938	PKD1	8	c.1615C>T	p.Gln539*	nonsense	This study
030034	PK10290	PKD1	8	c.1670del	p.Leu557Argfs*27	Frameshift	Audrezet, 2012
030087	PK10482	PKD1	8	c.1670del	p.Leu557Argfs*27	Frameshift	Audrezet, 2012
031082	PK10803	PKD1	8	c.1670del	p.Leu557Argfs*27	Frameshift	Audrezet, 2012
031088	PK10361	PKD1	8	c.1670del	p.Leu557Argfs*27	Frameshift	Audrezet, 2012
031294	PK10482	PKD1	8	c.1670del	p.Leu557Argfs*27	Frameshift	Audrezet, 2012
030013	PK10361	PKD1	8	c.1670del	p.Leu557Argfs*27	Frameshift	Audrezet, 2012
021735	PK11235	PKD1	8	c.1684C>T	p.Gln562*	nonsense	Rossetti, 2007
010863	PK10777	PKD1	8	c.1712_1722+17del	p.Gln571Glyfs*12	Frameshift	This study
191481	PK11264	PKD1	8	c.1722+1_1722-3delinsAGT		Splicing	This study
191546	PK11264	PKD1	8	c.1722+1_1722+3delinsAGT		Splicing	This study
070912	PK10229	PKD1	8	c.1722+1G>A		Splicing	Audrezet, 2012
191768	PK11451	PKD1	9	c.1751_1757del	p.Ser584Lysfs*199	Frameshift	This study
020015	PK10321	PKD1	9	c.1772del	p.Thr591Argfs*194	Frameshift	Audrezet, 2012
131160	PK11038	PKD1	9	c.1830C>G	p.Tyr510*	nonsense	This study
031231	PK11033	PKD1	10	c.1849-59_2805del		Large rearrangement	This study
020068	PK10391	PKD1	10	c.1930G>T	p.Gly644*	nonsense	Audrezet, 2012
021202	PK10391	PKD1	10	c.1930G>T	p.Gly644*	nonsense	Audrezet, 2012
021207	PK10391	PKD1	10	c.1930G>T	p.Gly644*	nonsense	Audrezet, 2012
020034	PK10372	PKD1	10	c.2038del	p.Tyr680Metfs*105	Frameshift	Audrezet, 2012
020126	PK10522	PKD1	10	c.2089C>T	p.Gln697*	nonsense	Audrezet, 2012
021114	PK10908	PKD1	10	c.2097+2T>G		Splicing	This study
020076	PK10405	PKD1	11	c.2152C>T	p.Gln718*	nonsense	Audrezet, 2012
030975	PK10737	PKD1	11	c.2152C>T	p.Gln718*	nonsense	Audrezet, 2012
021442	PK11246	PKD1	11	c.2152C>T	p.Gln718*	nonsense	Audrezet, 2012
191586	PK10737	PKD1	11	c.2152C>T	p.Gln718*	nonsense	Audrezet, 2012
191587	PK10737	PKD1	11	c.2152C>T	p.Gln718*	nonsense	Audrezet, 2012
070006	PK10402	PKD1	11	c.2298_2308del	p.Cys767Alafs*27	Frameshift	Hoelle, 2010
091418	PK11297	PKD1	11	c.2326G>T	p.Glu776*	nonsense	This study

191756	PK11415	PKD1	11	c.2444_2445del	p.Phe815*	Frameshift	This study
030806	PK10602	PKD1	11	c.2487dup	p.Ala830Cysfs*42	Frameshift	Audrezet, 2012
191581	PK11346	PKD1	11	c.2494dup	p.Arg852Profs*40	Frameshift	Rossetti, 2012
010070	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010025	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010062	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010101	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010003	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010063	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010045	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010092	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010061	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010134	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
021056	PK10761	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
021063	PK10761	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
010899	PK10230	PKD1	11	c.2636_2655dup	p.Trp887Profs*18	Frameshift	Audrezet, 2012
090028	PK10507	PKD1	11	c.2684_2685del	p.Ser895Cysfs*9	Frameshift	Audrezet, 2012
020078	PK10406	PKD1	11	c.2839C>T	p.Gln947*	nonsense	Audrezet, 2012
131303	PK11137	PKD1	11	c.2853+1G>C		Splicing	This study
090012	PK10375	PKD1	11	c.2854-1G>A		Splicing	Audrezet, 2012
090023	PK10375	PKD1	11	c.2854-1G>A		Splicing	Audrezet, 2012
120001	PK10563	PKD1	11	c.2854-1G>A		Splicing	Audrezet, 2012
150002	PK10563	PKD1	11	c.2854-1G>A		Splicing	Audrezet, 2012
091348	PK11219	PKD1	11	c.2854-1G>A		Splicing	Audrezet, 2012
130029	PK10491	PKD1	12	c.2962_2963del	p.Ser988Argfs*112	Frameshift	Audrezet, 2012
010054	PK10028	PKD1	12	c.2968_2969delinsT	p.Ala990Trpfs*7	Frameshift	Audrezet, 2012
010017	PK10028	PKD1	12	c.2968_2969delinsT	p.Ala990Trpfs*7	Frameshift	Audrezet, 2012
010066	PK10028	PKD1	12	c.2968_2969delinsT	p.Ala990Trpfs*7	Frameshift	Audrezet, 2012
010889	PK10028	PKD1	12	c.2968_2969delinsT	p.Ala990Trpfs*7	Frameshift	Audrezet, 2012
010019	PK10035	PKD1	12	c.2985+1G>A		Splicing	Audrezet, 2012
010838	PK10035	PKD1	12	c.2985+1G>A		Splicing	Audrezet, 2012
010842	PK10035	PKD1	12	c.2985+1G>A		Splicing	Audrezet, 2012
010891	PK10035	PKD1	12	c.2985+1G>A		Splicing	Audrezet, 2012
010893	PK10035	PKD1	12	c.2985+1G>A		Splicing	Audrezet, 2012
010026	PK10359	PKD1	13	c.2985+21_*1007delinsCACAGCTC		Large rearrangement	Audrezet, 2012
010095	PK10359	PKD1	13	c.2985+21_*1007delinsCACAGCTC		Large rearrangement	Audrezet, 2012
031077	PK11128	PKD1	13	c.2985+26_8161+37del		Large rearrangement	This study
010124	PK10158	PKD1	13	c.3058C>T	p.Gln1020*	nonsense	Pelola, 2005
020009	PK10309	PKD1	13	c.3058C>T	p.Gln1020*	nonsense	Pelola, 2005
020082	PK10309	PKD1	13	c.3058C>T	p.Gln1020*	nonsense	Pelola, 2005
021197	PK11006	PKD1	13	c.3058C>T	p.Gln1020*	nonsense	Pelola, 2005
130003	PK10445	PKD1	14	c.3212_3245del	p.Tyr1071Trpfs*72	Frameshift	Audrezet, 2012
030031	PK10305	PKD1	14	c.3213C>G	p.Tyr1071*	nonsense	Audrezet, 2012
021205	PK10305	PKD1	14	c.3213C>G	p.Tyr1071*	nonsense	Audrezet, 2012
021611	PK11389	PKD1	14	c.3247dup	p.Ala1083Glnfs*18	Frameshift	This study
021319	PK11091	PKD1	14	c.3285C>A	p.Tyr1095*	nonsense	Rossetti, 2007
020145	PK10684	PKD1	14	c.3293del	p.Pro1098Glnfs*6	Frameshift	This study
021601	PK10684	PKD1	14	c.3293del	p.Pro1098Glnfs*6	Frameshift	This study
010898	PK11388	PKD1	14	c.3296-1G>T		Splicing	This study
031296	PK11138	PKD1	14	c.3296-2A>C		Splicing	Tan, 2008
060952	PK10672	PKD1	15	c.3319del	p.Ala1107Hisfs*8	Frameshift	This study

060954	PK10672	PKD1	15	c.3319del	p.Ala1107Hisfs*8	Frameshift	This study
191504	PK11304	PKD1	15	c.3423_3484del	p.Arg1142Leufs*48	Frameshift	This study
100009	PK10560	PKD1	15	c.3441C>G	p.Tyr1147*	nonsense	Audrezet, 2012
031087	PK10809	PKD1	15	c.3441C>G	p.Tyr1147*	nonsense	Audrezet, 2012
030067	PK10481	PKD1	15	c.3493del	p.Asp1165Thrfs*6	Frameshift	Audrezet, 2012
031173	PK10877	PKD1	15	c.3503del	p.Pro1168Leufs*3	Frameshift	This study
031223	PK10877	PKD1	15	c.3503del	p.Pro1168Leufs*3	Frameshift	This study
091459	PK11351	PKD1	15	c.3607C>T	p.Gln1203*	nonsense	This study
030001	PK10231	PKD1	15	c.3624_3645delinsT	p.Phe1209_Leu1215del	In Frame deletion	Audrezet, 2012
040009	PK10429	PKD1	15	c.3729G>A	p.Trp1243*	nonsense	Audrezet, 2012
031089	PK10810	PKD1	15	c.3761C>A	p.Ser1254*	nonsense	This study
191517	PK11287	PKD1	15	c.3812_3813del	p.Thr1271Serfs*29	Frameshift	This study
191493	PK11287	PKD1	15	c.3812_3813del	p.Thr1271Serfs*29	Frameshift	This study
020103	PK10479	PKD1	15	c.3907_3908del	p.Ala1303Leufs*7	Frameshift	Audrezet, 2012
021199	PK10479	PKD1	15	c.3907_3908del	p.Ala1303Leufs*7	Frameshift	Audrezet, 2012
191571	PK10595	PKD1	15	c.3920dup	p.Gln1308Alafs*3	Frameshift	This study
031179	PK10917	PKD1	15	c.3922C>T	p.Gln1308*	nonsense	This study
090026	PK10454	PKD1	15	c.3942_3969del	p.Ala1315Thrfs*22	Frameshift	Audrezet, 2012
010059	PK10029	PKD1	15	c.4135G>T	p.Glu1379*	nonsense	Audrezet, 2012
010854	PK10029	PKD1	15	c.4135G>T	p.Glu1379*	nonsense	Audrezet, 2012
130036	PK10547	PKD1	15	c.4198_4201del	p.Leu1400Trpfs*31	Frameshift	Audrezet, 2012
020058	PK10428	PKD1	15	c.4247_4248del	p.Phe1416Trpfs*14	Frameshift	Audrezet, 2012
020107	PK10483	PKD1	15	c.4255_4274delinsTGAGG	p.Glu1419*	In Frame deletion	Audrezet, 2012
030062	PK10452	PKD1	15	c.4297_4298insCACGT	p.Phe1433Serfs*13	Frameshift	Audrezet, 2012
030809	PK10582	PKD1	15	c.4297_4298insCACGT	p.Phe1433Serfs*13	Frameshift	Audrezet, 2012
031072	PK10582	PKD1	15	c.4297_4298insCACGT	p.Phe1433Serfs*13	Frameshift	Audrezet, 2012
030100	PK10545	PKD1	15	c.4306C>T	p.Arg1436*	nonsense	Garcia-Gonzales, 2007
021619	PK11370	PKD1	15	c.4306C>T	p.Arg1436*	nonsense	Garcia-Gonzalez, 2007
060007	PK10142	PKD1	15	c.4331_4332del	p.Thr1444Serfs*12	Frameshift	Audrezet, 2012
060009	PK10685	PKD1	15	c.4331_4332del	p.Thr1444Serfs*12	Frameshift	Audrezet, 2012
020017	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
020157	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
021360	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
021450	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
021609	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
021613	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
021741	PK10310	PKD1	15	c.4442dup	p.Gln1482Alafs*41	Frameshift	Audrezet, 2012
010015	PK10033	PKD1	15	c.4444C>T	p.Gln1482*	nonsense	Audrezet, 2012
010078	PK10208	PKD1	15	c.4444C>T	p.Gln1482*	nonsense	Audrezet, 2012
010135	PK10033	PKD1	15	c.4444C>T	p.Gln1482*	nonsense	Audrezet, 2012
010136	PK10033	PKD1	15	c.4444C>T	p.Gln1482*	nonsense	Audrezet, 2012
010148	PK10438	PKD1	15	c.4447C>T	p.Gln1483*	nonsense	Hoefle, 2010
091411	PK10438	PKD1	15	c.4447C>T	p.Gln1483*	nonsense	Hoefle, 2010
010114	PK10299	PKD1	15	c.4460_4461insA	p.Phe1487Leufs*36	Frameshift	Audrezet, 2012
050008	PK10456	PKD1	15	c.4551C>G	p.Tyr1517*	nonsense	Audrezet, 2012
020140	PK10721	PKD1	15	c.4551C>G	p.Tyr1517*	nonsense	Audrezet, 2012
050011	PK10790	PKD1	15	c.4551C>G	p.Tyr1517*	nonsense	Audrezet, 2012
191720	PK11369	PKD1	15	c.4580del	p.Val1527Glyfs*7	Frameshift	This study
030003	PK10295	PKD1	15	c.4617G>A	p.Trp1539*	nonsense	Audrezet, 2012
030027	PK10109	PKD1	15	c.4617G>A	p.Trp1539*	nonsense	Audrezet, 2012
030037	PK10121	PKD1	15	c.4617G>A	p.Trp1539*	nonsense	Audrezet, 2012

030050	PK10295	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
030070	PK10109	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
020120	PK10121	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
030105	PK10561	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
030111	PK10573	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
020154	PK10573	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
030978	PK10573	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
031385	PK10573	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
030022	PK10121	PKD1	15	c.4617G>A	p-Trp1539*	noise	Audrezet, 2012
021054	PK10113	PKD1	15	c.4880_4881del	p-Tyr1627Cysfs*30	Frameshift	Audrezet, 2012
021058	PK10178	PKD1	15	c.4881T>A	p-Tyr1627*	noise	This study
010129	PK10202	PKD1	15	c.4906C>T	p-Gln1636*	noise	Rossetti, 2002
060011	PK10141	PKD1	15	c.4906C>T	p-Gln1636*	noise	Rossetti, 2002
060017	PK10141	PKD1	15	c.4906C>T	p-Gln1636*	noise	Rossetti, 2002
060937	PK10141	PKD1	15	c.4906C>T	p-Gln1636*	noise	Rossetti, 2002
091012	PK10958	PKD1	15	c.4906C>T	p-Gln1636*	noise	Rossetti, 2002
191754	PK11368	PKD1	15	c.4924del	p-Arg1642Alafs*80	Frameshift	This study
031090	PK10839	PKD1	15	c.4929C>G	p-Tyr1643*	noise	This study
020133	PK10552	PKD1	15	c.4957C>T	p-Gln1653*	noise	Phakdeekitcharoen, 2000
020134	PK10552	PKD1	15	c.4957C>T	p-Gln1653*	noise	Phakdeekitcharoen, 2000
010083	PK10271	PKD1	15	c.4973_5006dup	p-Trp1669Tyrfs*12	Frameshift	Audrezet, 2012
010846	PK10157	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
010097	PK10037	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
010055	PK10195	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
010120	PK10037	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
010028	PK10215	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
020014	PK10388	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
020050	PK10426	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
020069	PK10426	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
031022	PK10765	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
021112	PK10665	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
021259	PK10426	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
021267	PK10426	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
021357	PK11175	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
031380	PK11212	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
191600	PK11385	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
191791	PK11385	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
191792	PK11436	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
191859	PK11490	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 1999
192083	PK11385	PKD1	15	c.5014_5015del	p-Arg1672Glyfs*98	Frameshift	Wamtek, 2003
021015	PK10959	PKD1	15	c.5058dup	p-Val1687Argfs*84	Frameshift	This study
021327	PK11139	PKD1	15	c.5079C>A	p-Tyr1693*	noise	Bataille, 2011
191714	PK11383	PKD1	15	c.5079C>A	p-Tyr1693*	noise	Bataille, 2011
040006	PK10599	PKD1	15	c.5080_5103del	p-His1694_Asn1701del	In Frame deletion	Audrezet, 2012
040999	PK10599	PKD1	15	c.5080_5103del	p-His1694_Asn1701del	In Frame deletion	Audrezet, 2012
031165	PK10599	PKD1	15	c.5080_5103del	p-His1694_Asn1701del	In Frame deletion	Audrezet, 2012
031237	PK10599	PKD1	15	c.5080_5103del	p-His1694_Asn1701del	In Frame deletion	Audrezet, 2012
041001	PK10599	PKD1	15	c.5080_5103del	p-His1694_Asn1701del	In Frame deletion	Audrezet, 2012
010022	PK10124	PKD1	15	c.5186del	p-Pro1729Glnfs*30	Frameshift	Audrezet, 2012
010173	PK10124	PKD1	15	c.5186del	p-Pro1729Glnfs*30	Frameshift	Audrezet, 2012
010067	PK10124	PKD1	15	c.5186del	p-Pro1729Glnfs*30	Frameshift	Audrezet, 2012

010069	PK10124	PKD1	15	c.5186del	p.Pro1729Glnfs*30	Frameshift	Audrezeit, 2012
100008	PK10124	PKD1	15	c.5186del	p.Pro1729Glnfs*30	Frameshift	Audrezeit, 2012
010089	PK10118	PKD1	15	c.5242_5257del	p.Val1748Profs*6	Frameshift	Audrezeit, 2012
020129	PK10550	PKD1	15	c.5479_5480dup	p.Gln1828Glyfs*9	Frameshift	Audrezeit, 2012
021430	PK10550	PKD1	15	c.5479_5480dup	p.Gln1828Glyfs*9	Frameshift	Audrezeit, 2012
010127	PK10027	PKD1	15	c.5482C>T	p.Gln1828*	nonsense	Rossetti, 2002
120010	PK10515	PKD1	15	c.5515del	p.Trp1839Glyfs*10	Frameshift	Audrezeit, 2012
021311	PK11056	PKD1	15	c.5558_5559insT	p.Met1854Hisfs*16	Frameshift	This study
191549	PK11310	PKD1	15	c.5599_5600del	p.Asn1867Cysfs*122	Frameshift	This study
010165	PK10498	PKD1	15	c.5628_5629delinsC	p.Ala1877Profs*72	Frameshift	Audrezeit, 2012
010103	PK10148	PKD1	15	c.5758del	p.Arg1920Alafs*29	Frameshift	Audrezeit, 2012
010056	PK10148	PKD1	15	c.5758del	p.Arg1920Alafs*29	Frameshift	Audrezeit, 2012
021356	PK11172	PKD1	15	c.5764C>T	p.Gln1922*	nonsense	Thomas, 1999
020045	PK10424	PKD1	15	c.5861dup	p.Asn1954Lysfs*36	Frameshift	Audrezeit, 2012
131155	PK10865	PKD1	15	c.5861dup	p.Asn1954Lysfs*36	Frameshift	Audrezeit, 2012
021602	PK11283	PKD1	15	c.5878C>T	p.Gln1960*	nonsense	Phakdeekitchareon, 2000
191707	PK11380	PKD1	15	c.5923C>T	p.Gln1975*	nonsense	Bataille, 2011
191778	PK11439	PKD1	15	c.5923C>T	p.Gln1975*	nonsense	Bataille, 2011
021603	PK11292	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191506	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191584	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191578	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191757	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191781	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191841	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191848	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
191894	PK11291	PKD1	15	c.5968_5969del	p.Arg1990Glufs*59	Frameshift	This study
060032	PK10520	PKD1	15	c.5969_6016del	p.Asn1991_Trp2006del	In Frame deletion	Audrezeit, 2012
191866	PK11486	PKD1	15	c.5982_5994del	p.Arg1995Alafs*117	Frameshift	This study
191502	PK10480	PKD1	15	c.6031C>T	p.Gln2011*	nonsense	Audrezeit, 2012
010068	PK10200	PKD1	15	c.6090del	p.Val2031Trpfs*85	Frameshift	Audrezeit, 2012
090001	PK10207	PKD1	15	c.6090del	p.Val2031Trpfs*85	Frameshift	Audrezeit, 2012
040013	PK10207	PKD1	15	c.6090del	p.Val2031Trpfs*85	Frameshift	Audrezeit, 2012
030977	PK10207	PKD1	15	c.6090del	p.Val2031Trpfs*85	Frameshift	Audrezeit, 2012
111091	PK10783	PKD1	15	c.6090del	p.Val2031Trpfs*85	Frameshift	Audrezeit, 2012
010104	PK10201	PKD1	15	c.6115C>T	p.Gln2039*	nonsense	Phakdeekitchareon, 2000
091342	PK10201	PKD1	15	c.6115C>T	p.Gln2039*	nonsense	Phakdeekitchareon, 2000
010872	PK11163	PKD1	15	c.6232C>T	p.Gln2078*	nonsense	This study
191568	PK11289	PKD1	15	c.6247_6259dup	p.Pro2087Hisfs*25	Frameshift	This study
030092	PK10601	PKD1	15	c.6307C>T	p.Gln2103*	nonsense	Audrezeit, 2012
091452	PK11245	PKD1	15	c.6319G>T	p.Gln2107*	nonsense	This study
031168	PK10867	PKD1	15	c.6344dup	p.Arg2116Glufs*59	Frameshift	This study
191763	PK11403	PKD1	15	c.6472C>T	p.Gln2158*	nonsense	Rossetti, 2002
191851	PK11488	PKD1	15	c.6472C>T	p.Gln2158*	nonsense	Rossetti, 2002
010126	PK10199	PKD1	15	c.6475_6476insCCCC	p.Val2159Alafs*17	Frameshift	Audrezeit, 2012
010007	PK10199	PKD1	15	c.6475_6476insCCCC	p.Val2159Alafs*17	Frameshift	Audrezeit, 2012
010142	PK10199	PKD1	15	c.6475_6476insCCCC	p.Val2159Alafs*17	Frameshift	Audrezeit, 2012
100005	PK10199	PKD1	15	c.6475_6476insCCCC	p.Val2159Alafs*17	Frameshift	Audrezeit, 2012
100011	PK10199	PKD1	15	c.6475_6476insCCCC	p.Val2159Alafs*17	Frameshift	Audrezeit, 2012
100003	PK10199	PKD1	15	c.6475_6476insCCCC	p.Val2159Alafs*17	Frameshift	Audrezeit, 2012

041339	PK11271	PKD1	15	c.6483dup	p.Arg2162Alafs*13	Frameshift	This study
010169	PK10513	PKD1	15	c.6487C>T	p.Arg2163*	nonsense	Rossetti, 2001
020143	PK10666	PKD1	15	c.6487C>T	p.Arg2163*	nonsense	Rossetti, 2001
021261	PK10666	PKD1	15	c.6487C>T	p.Arg2163*	nonsense	Rossetti, 2001
060020	PK10453	PKD1	15	c.6543C>G	p.Tyr2181*	nonsense	Audrezet, 2012
021189	PK10872	PKD1	15	c.6574_6580del	p.Thr2192Alafs*18	Frameshift	Roelfsema, 1997
021111	PK10769	PKD1	15	c.6577_6583del	p.Cys2195Glyfs*15	Frameshift	Audrezet, 2012
191503	PK10769	PKD1	15	c.6577_6583del	p.Cys2195Glyfs*15	Frameshift	Audrezet, 2012
191721	PK11366	PKD1	15	c.6720_6721del	p.Leu2241Aspfs*20	Frameshift	This study
191810	PK11366	PKD1	15	c.6720_6721del	p.Leu2241Aspfs*20	Frameshift	This study
030009	PK10270	PKD1	15	c.6726_6727del	p.Gln2243Glufs*18	Frameshift	Rossetti, 2001
030076	PK10270	PKD1	15	c.6726_6727del	p.Gln2243Glufs*18	Frameshift	Rossetti, 2001
030059	PK10514	PKD1	15	c.6730_6731del	p.Ser2244Hisfs*17	Frameshift	Yu, 2011
030074	PK10516	PKD1	15	c.6730_6731del	p.Ser2244Hisfs*17	Frameshift	Yu, 2011
130049	PK10514	PKD1	15	c.6730_6731del	p.Ser2244Hisfs*17	Frameshift	Yu, 2011
130045	PK10598	PKD1	15	c.6799_6803dup	p.Ser2269Cysfs*47	Frameshift	Audrezet, 2012
021052	PK10789	PKD1	15	c.6799_6803dup	p.Ser2269Cysfs*47	Frameshift	Audrezet, 2012
140010	PK10985	PKD1	15	c.6799_6803dup	p.Ser2269Cysfs*47	Frameshift	Audrezet, 2012
050004	PK10822	PKD1	15	c.6803G>A	p.Trp2268*	nonsense	This study
010864	PK10798	PKD1	15	c.6829_6845del	p.Asp2277Argfs*137	Frameshift	This study
010882	PK10798	PKD1	15	c.6829_6845del	p.Asp2277Argfs*137	Frameshift	This study
030810	PK10788	PKD1	15	c.6831del	p.Asp2277Glufs*37	Frameshift	This study
191511	PK11288	PKD1	15	c.6831del	p.Asp2277Glufs*37	Frameshift	This study
061127	PK11161	PKD1	15	c.6913C>T	p.Gln2305*	nonsense	This study
191710	PK11381	PKD1	15	c.6915+2T>C		Splicing	This study
010110	PK10112	PKD1	15	c.6915+2T>G		Splicing	Perrichot, 2000
010099	PK10112	PKD1	15	c.6915+2T>G		Splicing	Perrichot, 2000
030971	PK10112	PKD1	15	c.6915+2T>G		Splicing	Perrichot, 2000
010858	PK10112	PKD1	15	c.6915+2T>G		Splicing	Perrichot, 2000
020098	PK10478	PKD1	16	c.6994_7000del	p.Ala2332Trpfs*7	Frameshift	Phakdeekitchareon, 2000
020169	PK10770	PKD1	16	c.6994_7000del	p.Ala2332Trpfs*7	Frameshift	Phakdeekitchareon, 2000
191501	PK11290	PKD1	16	c.6994_7000del	p.Ala2332Trpfs*7	Frameshift	Phakdeekitchareon, 2000
091773	PK11406	PKD1	16	c.6994_7000dup	p.Val2334Glyfs*88	Frameshift	Rossetti, 2001
010128	PK10032	PKD1	16	c.7008C>A	p.Tyr2336*	nonsense	Perrichot, 2000
120002	PK10032	PKD1	16	c.7008C>A	p.Tyr2336*	nonsense	Perrichot, 2000
021186	PK10897	PKD1	16	c.7065+2_7065+4del		Splicing	This study
080001	PK10346	PKD1	16	c.7066-2A>G		Splicing	Audrezet, 2012
060938	PK10878	PKD1	17	c.7112_7113insATA	p.Ser2372*	nonsense	This study
191793	PK11435	PKD1	17	c.7113_7114del	p.Ser2372Leufs*47	Frameshift	Rossetti, 2001
191547	PK11309	PKD1	17	c.7126C>T	p.Gln2376*	nonsense	This study
030063	PK10411	PKD1	17	c.7137C>G	p.Tyr2379*	nonsense	Audrezet, 2012
040016	PK10411	PKD1	17	c.7137C>G	p.Tyr2379*	nonsense	Audrezet, 2012
040018	PK10411	PKD1	17	c.7137C>G	p.Tyr2379*	nonsense	Audrezet, 2012
030075	PK10411	PKD1	17	c.7137C>G	p.Tyr2379*	nonsense	Audrezet, 2012
031029	PK10411	PKD1	17	c.7137C>G	p.Tyr2379*	nonsense	Audrezet, 2012
030108	PK10562	PKD1	17	c.7158C>G	p.Tyr2386*	nonsense	Audrezet, 2012
191509	PK11312	PKD1	17	c.7204C>T	p.Arg2402*	nonsense	Phakdeekitchareon, 2000
211834	PK11470	PKD1	17	c.7209+1G>A		Splicing	This study
020109	PK10494	PKD1	18	c.7250del	p.Leu2417Argfs*203	Frameshift	Audrezet, 2012
030041	PK10341	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
020088	PK10433	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000

060027	PK10484	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
130015	PK10493	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
141037	PK10823	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
021053	PK10814	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
060002	PK10816	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
131041	PK10493	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
031288	PK11063	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
091420	PK11234	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
191715	PK11384	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
191803	PK10823	PKD1	18	c.7288C>T	p.Arg2430*	nonsense	Phakdeekitchareon, 2000
021351	PK11167	PKD1	18	c.7318del	p.Asp2440TTris*180	Frameshift	This study
010853	PK10743	PKD1	18	c.7324_7342del	p.Gln2442Seris*172	Frameshift	This study
021113	PK10866	PKD1	18	c.7324_7344dup	p.Gln2442_Leu2448dup	In Frame insertion	This study
191769	PK11404	PKD1	18	c.7390del	p.Arg2464Alafs*156	Frameshift	This study
031235	PK11030	PKD1	19	c.7642G>T	p.Gln2548*	nonsense	This study
021358	PK11174	PKD1	19	c.7648_7649del	p.Gly2550Profs*44	Frameshift	This study
020039	PK10381	PKD1	19	c.7672dup	p.Gln2558Profs*37	Frameshift	Audrezet, 2012
020081	PK10381	PKD1	19	c.7672dup	p.Gln2558Profs*37	Frameshift	Audrezet, 2012
030006	PK10272	PKD1	19	c.7703+1G>A		Splicing	Audrezet, 2012
020041	PK10387	PKD1	20	c.7730_7737del	p.Pro2577Glnfs*15	Frameshift	Audrezet, 2012
020084	PK10387	PKD1	20	c.7730_7737del	p.Pro2577Glnfs*15	Frameshift	Audrezet, 2012
191808	PK11468	PKD1	20	c.7804C>T	p.Gln2602*	nonsense	Audrezet, 2012
020146	PK10732	PKD1	20	c.7816C>T	p.Gln2606*	nonsense	This study
191758	PK11453	PKD1	20	c.7816C>T	p.Gln2606*	nonsense	This study
010140	PK10108	PKD1	20	c.7821_7825del	p.Val2608Argfs*51	Frameshift	Audrezet, 2012
090049	PK10108	PKD1	20	c.7821_7825del	p.Val2608Argfs*51	Frameshift	Audrezet, 2012
010138	PK10475	PKD1	20	c.7864+1G>T		Splicing	Audrezet, 2012
020150	PK11128	PKD1	21	c.7864-1J_18791+7del		Large rearrangement	Audrezet, 2012
060015	PK10422	PKD1	21	c.7866C>A	p.Tyr2622*	nonsense	Audrezet, 2012
191565	PK10422	PKD1	21	c.7866C>A	p.Tyr2622*	nonsense	Audrezet, 2012
021324	PK11090	PKD1	21	c.7903G>T	p.Gln2635*	nonsense	This study
130008	PK10432	PKD1	21	c.7915C>T	p.Arg2639*	nonsense	Perrichot, 2000
140019	PK10432	PKD1	21	c.7915C>T	p.Arg2639*	nonsense	Perrichot, 2000
130028	PK10432	PKD1	21	c.7915C>T	p.Arg2639*	nonsense	Perrichot, 2000
020144	PK10731	PKD1	21	c.7915C>T	p.Arg2639*	nonsense	Perrichot, 2000
131399	PK10432	PKD1	21	c.7915C>T	p.Arg2639*	nonsense	Perrichot, 2000
191556	PK11402	PKD1	21	c.7921C>T	p.Gln2641*	nonsense	This study
021069	PK10817	PKD1	21	c.7975del	p.Asp2659Metfs*26	Frameshift	This study
030942	PK10667	PKD1	21	c.8008C>T	p.Gln2670*	nonsense	Audrezet, 2012
021263	PK10667	PKD1	21	c.8008C>T	p.Gln2670*	nonsense	Audrezet, 2012
031374	PK10667	PKD1	21	c.8008C>T	p.Gln2670*	nonsense	Audrezet, 2012
031475	PK10667	PKD1	21	c.8008C>T	p.Gln2670*	nonsense	Audrezet, 2012
010073	PK10240	PKD1	21	c.8017-2_8017-1del		Splicing	Rossetti, 2001
120006	PK10240	PKD1	21	c.8017-2_8017-1del		Splicing	Rossetti, 2001
100018	PK10240	PKD1	21	c.8017-2_8017-1del		Splicing	Rossetti, 2001
020132	PK10549	PKD1	21	c.8017-2A>G		Splicing	Rossetti, 2007
021426	PK11236	PKD1	21	c.8017-2A>G		Splicing	Rossetti, 2007
191548	PK11308	PKD1	21	c.8017-2A>G		Splicing	Rossetti, 2007
091884	PK10089	PKD1	22	c.8056C>T	p.Gln2686*	nonsense	Garcia-Gonzales, 2007
191717	PK10089	PKD1	22	c.8070del	p.Lys2690Asnfs*5	Frameshift	Audrezet, 2012
021610	PK10966	PKD1	23	c.8196_8197dup	p Ala2733Glyfs*24	Frameshift	This study

010859	PK10766	PKD1	23	c.8219del	p.Gly2740Glufs*16	Frameshift	This study
010851	PK10025	PKD1	23	c.8257_8260del	p.Tyr2753Thrfs*2	Frameshift	Audrezet, 2012
010008	PK10025	PKD1	23	c.8257_8260del	p.Tyr2753Thrfs*2	Frameshift	Audrezet, 2012
010158	PK10025	PKD1	23	c.8257_8260del	p.Tyr2753Thrfs*2	Frameshift	Audrezet, 2012
010852	PK10025	PKD1	23	c.8257_8260del	p.Tyr2753Thrfs*2	Frameshift	Audrezet, 2012
010862	PK10025	PKD1	23	c.8257_8260del	p.Tyr2753Thrfs*2	Frameshift	Audrezet, 2012
110021	PK10855	PKD1	23	c.8284_8295del	p.Ile2762_Arg2765del	In Frame deletion	This study
110024	PK10858	PKD1	23	c.8284_8295del	p.Ile2762_Arg2765del	In Frame deletion	This study
191801	PK11419	PKD1	23	c.8284_8295del	p.Ile2762_Arg2765del	In Frame deletion	This study
061130	PK10845	PKD1	23	c.8284_8295dup	p.Ile2762_Arg2765dup	In Frame insertion	This study
021193	PK10845	PKD1	23	c.8284_8295dup	p.Ile2762_Arg2765dup	In Frame insertion	This study
021194	PK10845	PKD1	23	c.8284_8295dup	p.Ile2762_Arg2765dup	In Frame insertion	This study
130025	PK10466	PKD1	23	c.8308_8311delinsCTCGTT	p.Asn2770Leufs*6	Frameshift	Audrezet, 2012
130005	PK10467	PKD1	23	c.8308_8311delinsCTCGTT	p.Asn2770Leufs*6	Frameshift	Audrezet, 2012
021190	PK10875	PKD1	23	c.8308_8311delinsCTCGTT	p.Asn2770Leufs*6	Frameshift	Audrezet, 2012
091250	PK10131	PKD1	23	c.8308_8311delinsCTCGTT	p.Asn2770Leufs*6	Frameshift	Audrezet, 2012
040022	PK10500	PKD1	23	c.8320_8321del	p.Leu2774Aspfs*47	Frameshift	Audrezet, 2012
030801	PK10589	PKD1	23	c.8320_8321del	p.Leu2774Aspfs*47	Frameshift	Audrezet, 2012
031281	PK10589	PKD1	23	c.8320_8321del	p.Leu2774Aspfs*47	Frameshift	Audrezet, 2012
041335	PK10589	PKD1	23	c.8320_8321del	p.Leu2774Aspfs*47	Frameshift	Audrezet, 2012
020071	PK10423	PKD1	23	c.8350C>T	p.Gln2784*	nonsense	Audrezet, 2012
111092	PK10791	PKD1	23	c.8385C>A	p.Cys2795*	nonsense	This study
010002	PK10183	PKD1	23	c.8560C>T	p.Gln2884*	nonsense	Audrezet, 2012
080003	PK10030	PKD1	23	c.8560C>T	p.Gln2884*	nonsense	Audrezet, 2012
021057	PK10183	PKD1	23	c.8560C>T	p.Gln2884*	nonsense	Audrezet, 2012
031083	PK10808	PKD1	23	c.8560C>T	p.Gln2884*	nonsense	Audrezet, 2012
010049	PK10190	PKD1	23	c.8586del	p.Ile2863Serfs*12	Frameshift	Audrezet, 2012
030107	PK10190	PKD1	23	c.8586del	p.Ile2863Serfs*12	Frameshift	Audrezet, 2012
060033	PK10190	PKD1	23	c.8586del	p.Ile2863Serfs*12	Frameshift	Audrezet, 2012
010894	PK10190	PKD1	23	c.8586del	p.Ile2863Serfs*12	Frameshift	Audrezet, 2012
010004	PK10186	PKD1	23	c.8603C>A	p.Ser2868*	nonsense	Audrezet, 2012
010843	PK10186	PKD1	23	c.8603C>A	p.Ser2868*	nonsense	Audrezet, 2012
131309	PK11194	PKD1	23	c.8709_8713dup	p.Val2905Alafs*91	Frameshift	This study
020139	PK11125	PKD1	24	c.8791+89_9397+743del	Large rearrangement	Large rearrangement	This study
021200	PK11125	PKD1	24	c.8791+89_9397+743del	Large rearrangement	Large rearrangement	This study
020079	PK10431	PKD1	24	c.8799C>G	p.Tyr2933*	nonsense	Audrezet, 2012
091349	PK11230	PKD1	24	c.8830_8834del	p.Val2944Profs*9	Frameshift	This study
030008	PK10274	PKD1	24	c.8905C>T	p.Gln2969*	nonsense	Inoue, 2002
030819	PK10274	PKD1	24	c.8905C>T	p.Gln2969*	nonsense	Inoue, 2002
021214	PK10979	PKD1	24	c.8906del	p.Gln2969Argfs*25	Frameshift	This study
090963	PK10153	PKD1	25	c.8972dup	p.Tyr2991*	Frameshift	Audrezet, 2012
191558	PK11382	PKD1	25	c.8973C>G	p.Tyr2991*	nonsense	Rossetti, 2002
020096	PK10473	PKD1	25	c.9002G>A	p.Trp3001*	nonsense	Phakdeekitchareon, 2000
020077	PK10430	PKD1	25	c.9018_9035dup	p.Val3008_Ser3013dup	In Frame insertion	This study
010098	PK10031	PKD1	25	c.9127del	p.Cys3043Alafs*31	Frameshift	Audrezet, 2012
010088	PK10031	PKD1	25	c.9127del	p.Cys3043Alafs*31	Frameshift	Audrezet, 2012
010855	PK10031	PKD1	25	c.9127del	p.Cys3043Alafs*31	Frameshift	Audrezet, 2012
010856	PK10031	PKD1	25	c.9127del	p.Cys3043Alafs*31	Frameshift	Audrezet, 2012
030980	PK10031	PKD1	25	c.9127del	p.Cys3043Alafs*31	Frameshift	Audrezet, 2012
031174	PK10876	PKD1	25	c.9148dup	p.Ala3050Glyfs*19	Frameshift	Baraille, 2011
020043	PK10374	PKD1	25	c.9201+1G>C	Splicing	Splicing	Audrezet, 2012

020137	PK10587	PKD1	26	c.9214del	p.Asp3072Metfs*2	Frameshift	Audrezet, 2012
010847	PK10012	PKD1	26	c.9240_9241del	p.Ala3082Cysfs*96	Frameshift	Thongrooprakun, 2004
020090	PK10476	PKD1	26	c.9240_9241del	p.Ala3082Cysfs*96	Frameshift	Thongrooprakun, 2004
120015	PK10012	PKD1	26	c.9240_9241del	p.Ala3082Cysfs*96	Frameshift	Thongrooprakun, 2004
021055	PK10768	PKD1	26	c.9240_9241del	p.Ala3082Cysfs*96	Frameshift	Thongrooprakun, 2004
021215	PK10768	PKD1	26	c.9240_9241del	p.Ala3082Cysfs*96	Frameshift	Thongrooprakun, 2004
141278	PK11190	PKD1	26	c.9240_9241del	p.Ala3082Cysfs*96	Frameshift	Thongrooprakun, 2004
030004	PK10276	PKD1	26	c.9283_9293del	p.Leu3095Glyfs*80	Frameshift	Audrezet, 2012
030068	PK10443	PKD1	26	c.9317dup	p.Arg3107Profs*72	Frameshift	Audrezet, 2012
030069	PK10443	PKD1	26	c.9317dup	p.Arg3107Profs*72	Frameshift	Audrezet, 2012
031071	PK10443	PKD1	26	c.9317dup	p.Arg3107Profs*72	Frameshift	Audrezet, 2012
021353	PK11141	PKD1	26	c.9398-1G>C		Splicing	This study
010085	PK10224	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
060019	PK10224	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
060013	PK10224	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
020106	PK10527	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
060022	PK10224	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
050001	PK10224	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
021361	PK11176	PKD1	27	c.9547C>T	p.Arg3183*	nonsense	Audrezet, 2012
020060	PK10414	PKD1	27	c.9547del	p.Arg3183Glnfs*133	Frameshift	Audrezet, 2012
010038	PK10140	PKD1	27	c.9555G>A	p.Trp3185*	nonsense	Audrezet, 2012
010895	PK10140	PKD1	27	c.9555G>A	p.Trp3185*	nonsense	Audrezet, 2012
010034	PK10239	PKD1	27	c.9555G>A	p.Trp3185*	nonsense	Audrezet, 2012
010032	PK10140	PKD1	27	c.9555G>A	p.Trp3185*	nonsense	Audrezet, 2012
010016	PK10140	PKD1	27	c.9555G>A	p.Trp3185*	nonsense	Audrezet, 2012
030065	PK10143	PKD1	28	c.9616C>T	p.Gln3206*	nonsense	Perrichot, 2000
130020	PK10143	PKD1	28	c.9616C>T	p.Gln3206*	nonsense	Perrichot, 2000
030024	PK10143	PKD1	28	c.9616C>T	p.Gln3206*	nonsense	Perrichot, 2000
130039	PK10143	PKD1	28	c.9616C>T	p.Gln3206*	nonsense	Perrichot, 2000
030813	PK10143	PKD1	28	c.9616C>T	p.Gln3206*	nonsense	Perrichot, 2000
131880	PK10143	PKD1	28	c.9616C>T	p.Gln3206*	nonsense	Perrichot, 2000
050002	PK10503	PKD1	28	c.9668del	p.Ser320Argfs*96	Frameshift	Perrichot, 2000
191770	PK11450	PKD1	29	c.9788G>A	p.Trp3263*	nonsense	This study
191588	PK11378	PKD1	31	c.10073del	p.Thr3358Asnfs*39	Frameshift	This study
030946	PK10722	PKD1	31	c.10151C>G	p.Ser3384*	nonsense	Petola, 2005
140006	PK10523	PKD1	31	c.10167+25_10167+43del		Splicing	Peral, 1997
021317	PK11253	PKD1	31	c.10167+25_10167+43del		Splicing	Peral, 1997
091413	PK11252	PKD1	31	c.10167+25_10167+43del		Splicing	Peral, 1997
021424	PK11253	PKD1	31	c.10167+25_10167+43del		Splicing	Peral, 1997
021184	PK10910	PKD1	32	c.10189_10190del	p.Lys3397Glnfs*2	Frameshift	This study
191564	PK11313	PKD1	32	c.10215dup	p.Lys3406*	Frameshift	This study
191580	PK11313	PKD1	32	c.10215dup	p.Lys3406*	Frameshift	This study
031295	PK11116	PKD1	33	c.10232G>A	p.Trp3411*	nonsense	This study
020070	PK10415	PKD1	33	c.10335del	p.Leu3446Trpfs*27	Frameshift	Audrezet, 2012
020119	PK10541	PKD1	33	c.10345_10406-24del	p.Gln3449Metfs*4	Frameshift	Audrezet, 2012
020162	PK10541	PKD1	33	c.10345_10406-24del	p.Gln3449Metfs*4	Frameshift	Audrezet, 2012
020005	PK10306	PKD1	33	c.10374dup	p.Tyr3459Leufs*12	Frameshift	Audrezet, 2012
020100	PK10306	PKD1	33	c.10374dup	p.Tyr3459Leufs*12	Frameshift	Audrezet, 2012
030093	PK10605	PKD1	33	c.10405+1G>T		Splicing	Audrezet, 2012
040905	PK10147	PKD1	34	c.10423C>T	p.Gln3475*	nonsense	Perrichot, 1999
041000	PK10147	PKD1	34	c.10423C>T	p.Gln3475*	nonsense	Perrichot, 1999

020075	PK10450	PKD1	34	c.10440_10441dup	p.Val3481Glyfs*47	Frameshift	Audrezeit, 2012
021196	PK10450	PKD1	34	c.10440_10441dup	p.Val3481Glyfs*47	Frameshift	Audrezeit, 2012
021329	PK10450	PKD1	34	c.10440_10441dup	p.Val3481Glyfs*47	Frameshift	Audrezeit, 2012
060041	PK10644	PKD1	34	c.10441dup	p.Val3481Glyfs*146	Frameshift	Audrezeit, 2012
020156	PK11018	PKD1	35	c.10519A>T	p.Lys3507*	nonsense	This study
020166	PK11018	PKD1	35	c.10519A>T	p.Lys3507*	nonsense	This study
021253	PK11018	PKD1	35	c.10519A>T	p.Lys3507*	nonsense	This study
040014	PK10355	PKD1	35	c.10540C>T	p.Gln3514*	nonsense	Peral, 1996
021606	PK11317	PKD1	35	c.10563_10570del	p.Pro3522Argfs*102	Frameshift	This study
020104	PK10504	PKD1	35	c.10618+2T>C		Splicing	Audrezeit, 2012
020142	PK10504	PKD1	35	c.10618+2T>C		Splicing	Audrezeit, 2012
211812	PK11434	PKD1	35	c.10618+2T>C		Splicing	Audrezeit, 2012
020089	PK10455	PKD1	36	c.10701_10720del	p.Ala3569Glyfs*51	Frameshift	Audrezeit, 2012
010145	PK10409	PKD1	36	c.10722_10732del	p.Trp3575Glnfs*48	Frameshift	Audrezeit, 2012
020170	PK10813	PKD1	36	c.10725G>A	p.Trp3575*	nonsense	Zang, 2005
010850	PK10686	PKD1	36	c.10728_10731dup	p.Ala3578Glyfs*50	Frameshift	This study
021257	PK11037	PKD1	36	c.10730del	p.Gly3577Valfs*8	Frameshift	This study
191566	PK11280	PKD1	36	c.10739del	p.Phe3580Serfs*5	Frameshift	This study
020013	PK10323	PKD1	36	c.10745dup	p.Val3584Argfs*43	Frameshift	Audrezeit, 2012
020122	PK10323	PKD1	36	c.10745dup	p.Val3584Argfs*43	Frameshift	Audrezeit, 2012
030090	PK10659	PKD1	36	c.10745dup	p.Val3584Argfs*43	Frameshift	Audrezeit, 2012
141038	PK10835	PKD1	36	c.10745dup	p.Val3584Argfs*43	Frameshift	Audrezeit, 2012
010036	PK10078	PKD1	36	c.10789del	p.Leu3597Trpfs*10	Frameshift	Audrezeit, 2012
010115	PK10078	PKD1	36	c.10789del	p.Leu3597Trpfs*10	Frameshift	Audrezeit, 2012
031178	PK10921	PKD1	36	c.10821+IG>A		Splicing	This study
191543	PK11281	PKD1	36	c.10821+IG>A		Splicing	This study
191569	PK11281	PKD1	36	c.10821+IG>A		Splicing	This study
191755	PK11416	PKD1	36	c.10821+IG>A		Splicing	This study
020037	PK10340	PKD1	37	c.10833del	p.Ala3612Profs*20	Frameshift	Audrezeit, 2012
020124	PK10340	PKD1	37	c.10833del	p.Ala3612Profs*20	Frameshift	Audrezeit, 2012
010125	PK10162	PKD1	37	c.10897_10898del	p.Ser3633Profs*88	Frameshift	Audrezeit, 2012
191572	PK11318	PKD1	37	c.10949_10950dup	p.Gly3651Thrfs*34	Frameshift	This study
100992	PK11024	PKD1	37	c.10964_10965delinsAA	p.Phe3655*	nonsense	This study
010023	PK10034	PKD1	37	c.10966del	p.Leu3656Trpfs*28	Frameshift	Audrezeit, 2012
010075	PK10155	PKD1	37	c.11004dup	p.Gly3669Trpfs*53	Frameshift	Audrezeit, 2012
030061	PK10005	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
030071	PK10497	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
030095	PK10005	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
031162	PK10836	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
031180	PK10836	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
031383	PK10836	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
031384	PK10005	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
091453	PK10836	PKD1	37	c.11016+IG>A		Splicing	Audrezeit, 2012
191513	PK11265	PKD1	37	c.11017-IG>C		Splicing	This study
090967	PK10700	PKD1	38	c.11078C>A	p.Ser3693*	nonsense	This study
010060	PK10146	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010072	PK10146	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010077	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010018	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010011	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010040	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999

010030	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010081	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010074	PK10146	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010116	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010122	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010106	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010159	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
100015	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
100016	PK10059	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010870	PK10891	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010879	PK10891	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
010885	PK10891	PKD1	38	c.11078dup	p.Cys3694Metfs*28	Frameshift	Perrichot, 1999
030106	PK10618	PKD1	38	c.11091del	p.His697Glnfs*129	Frameshift	Audrezet, 2012
191577	PK11305	PKD1	39	c.11177G>A	p.Trp3726*	non sense	This study
030973	PK10729	PKD1	39	c.11178dup	p.Met3727Aspfs*89	Frameshift	This study
030023	PK11505	PKD1	39	c.11202>A	p.Tyr3734*	non sense	This study
191573	PK11306	PKD1	39	c.11270-1G>T		Splicing	Kumar, 2012
110014	PK10634	PKD1	40	c.11296delinsCG	p.Val3767Glyfs*49	Frameshift	Audrezet, 2012
191807	PK11545	PKD1	40	c.11314_11324del	p.Ala3772Leufs*40	Frameshift	This study
030043	PK10325	PKD1	40	c.11316_11317insA	p.Ala3773Serfs*43	Frameshift	Audrezet, 2012
010096	PK10311	PKD1	40	c.11327_11353del	p.Phe3776_Gly3784del	In Frame deletion	Audrezet, 2012
091412	PK10311	PKD1	40	c.11327_11353del	p.Phe3776_Gly3784del	In Frame deletion	Audrezet, 2012
030081	PK10510	PKD1	40	c.11340_11346dup	p.Val3783Leufs*35	Frameshift	Audrezet, 2012
030038	PK10369	PKD1	40	c.11344_11363dup	p.His3789Thrfs*44	Frameshift	Audrezet, 2012
031170	PK10869	PKD1	40	c.11354_11355ins14	p.Trp3785*	Frameshift	This study
070004	PK10645	PKD1	40	c.11379del	p.Thr3794Argfs*32	Frameshift	Bogdanova, 2000
031076	PK10781	PKD1	40	c.11379del	p.Thr3794Argfs*32	Frameshift	Bogdanova, 2000
191785	PK11316	PKD1	40	c.11386dup	p.Ala3796Glyfs*20	Frameshift	This study
030080	PK10509	PKD1	40	c.11397_11398del	p.Pro3800Glyfs*15	Frameshift	Audrezet, 2012
010027	PK10036	PKD1	41	c.11418dup	p.Ser3807Valfs*9	Frameshift	Audrezet, 2012
010065	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010051	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010021	PK10169	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010064	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010084	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010139	PK10170	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010079	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010013	PK10247	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010094	PK10247	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010109	PK10169	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
020033	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010132	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010147	PK10247	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
010188	PK10427	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
120985	PK10110	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
191505	PK11282	PKD1	41	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000
041003	PK10942	PKD1	41	c.11525G>A	p.Trp3842*	non sense	Rossetti, 2007
020056	PK10356	PKD1	41	c.11537+1G>T		Splicing	Audrezet, 2012
020057	PK10356	PKD1	41	c.11537+1G>T		Splicing	Audrezet, 2012
020110	PK10565	PKD1	41	c.11538-2A>G		Splicing	Audrezet, 2012
010143	PK10427	PKD1	42	c.11461C>T	p.Gln3821*	non sense	Perrichot, 2000

021449	PK11269	PKD1	42	c.11554del	p.Leu3852Trpfs*93	Frameshift	Rossetti, 2012
020035	PK10324	PKD1	42	c.11630del	p.Gly3877Alafs*68	Frameshift	Audrezet, 2012
021269	PK10324	PKD1	42	c.11630del	p.Gly3877Alafs*68	Frameshift	Audrezet, 2012
030056	PK10401	PKD1	42	c.11633_11660dup	p.Phe3888Argfs*82	Frameshift	Audrezet, 2012
030047	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
020054	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
070002	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
070005	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
060003	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
060953	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
070917	PK10348	PKD1	42	c.11648_11660dup	p.Phe3888Glnfs*77	Frameshift	Audrezet, 2012
020044	PK10349	PKD1	43	c.11745del	p.Val8916Trpfs*29	Frameshift	Audrezet, 2012
090008	PK10304	PKD1	43	c.11773del	p.Glu3925Lysfs*20	Frameshift	Audrezet, 2012
110010	PK10304	PKD1	43	c.11773del	p.Glu3925Lysfs*20	Frameshift	Audrezet, 2012
191712	PK11418	PKD1	43	c.11792_11793dup	p.Arg3932Cysfs*14	Frameshift	This study
010043	PK10267	PKD1	43	c.11802_11803del	p.Ala3935Leufs*25	Frameshift	Audrezet, 2012
191482	PK11296	PKD1	43	c.11817G>A	p.Trp3939*	nonsense	This study
030098	PK10690	PKD1	43	c.11863C>T	p.Gln3955*	nonsense	Rossetti, 2007
020153	PK10717	PKD1	43	c.11863C>T	p.Gln3955*	nonsense	Rossetti, 2007
021060	PK10717	PKD1	43	c.11863C>T	p.Gln3955*	nonsense	Rossetti, 2007
041008	PK11096	PKD1	43	c.11863C>T	p.Gln3955*	nonsense	Rossetti, 2007
130031	PK10567	PKD1	43	c.11909_11926del	p.Pro3971_Ser3976del	In Frame deletion	Audrezet, 2012
131305	PK10567	PKD1	43	c.11909_11926del	p.Pro3971_Ser3976del	In Frame deletion	Audrezet, 2012
191576	PK11410	PKD1	43	c.11923_11942del	p.Trp3975Alafs*175	Frameshift	This study
030025	PK10275	PKD1	43	c.11935C>T	p.Gln3979*	nonsense	Hoelele, 2010
030077	PK10538	PKD1	43	c.11935C>T	p.Gln3979*	nonsense	Hoelele, 2010
020127	PK10275	PKD1	43	c.11935C>T	p.Gln3979*	nonsense	Hoelele, 2010
021191	PK10911	PKD1	43	c.11935C>T	p.Gln3979*	nonsense	Hoelele, 2010
031381	PK10275	PKD1	43	c.11935C>T	p.Gln3979*	nonsense	Hoelele, 2010
091592	PK11413	PKD1	43	c.11935C>T	p.Gln3979*	nonsense	Hoelele, 2010
090045	PK10978	PKD1	43	c.11974del	p.Ala3992Profs*47	Frameshift	This study
010052	PK10255	PKD1	43	c.12004-2A>G		Splicing	Audrezet, 2012
010048	PK10255	PKD1	43	c.12004-2A>G		Splicing	Audrezet, 2012
120025	PK10255	PKD1	43	c.12004-2A>G		Splicing	Audrezet, 2012
191570	PK11452	PKD1	43	c.12004-2A>G		Splicing	Audrezet, 2012
191753	PK11452	PKD1	43	c.12004-2A>G		Splicing	Audrezet, 2012
191767	PK11452	PKD1	43	c.12004-2A>G		Splicing	Audrezet, 2012
010086	PK10258	PKD1	44	c.12010C>T	p.Gln4004*	nonsense	Gao, 2006
010118	PK10077	PKD1	44	c.12013C>T	p.Gln4005*	nonsense	Iglesias, 1999
020066	PK10408	PKD1	44	c.12013C>T	p.Gln4005*	nonsense	Iglesias, 1999
091455	PK11272	PKD1	44	c.12031C>T	p.Gln4011*	nonsense	Daniells, 1998
010010	PK10152	PKD1	44	c.12035G>A	p.Trp4012*	nonsense	Audrezet, 2012
020040	PK10354	PKD1	44	c.12035G>A	p.Trp4012*	nonsense	Audrezet, 2012
010091	PK10300	PKD1	44	c.12061C>T	p.Arg4021*	nonsense	Turco, 1996
110019	PK10300	PKD1	44	c.12061C>T	p.Arg4021*	nonsense	Turco, 1996
021446	PK11326	PKD1	44	c.12061C>T	p.Arg4021*	nonsense	Turco, 1996
010050	PK10174	PKD1	44	c.12085_12121del	p.Val4029Profs*157	Frameshift	Audrezet, 2012
020087	PK10460	PKD1	44	c.12085_12121del	p.Val4029Profs*157	Frameshift	Audrezet, 2012
020114	PK10174	PKD1	44	c.12085_12121del	p.Val4029Profs*157	Frameshift	Audrezet, 2012
080018	PK10460	PKD1	44	c.12085_12121del	p.Val4029Profs*157	Frameshift	Audrezet, 2012
021217	PK10981	PKD1	44	c.12124C>T	p.Gln4042*	nonsense	turco, 1996

191497	PK11267	PKD1	44	c.12124C>T	p.Gln4042*	nonsense	Turco, 1996
191554	PK11411	PKD1	44	c.12124C>T	p.Gln4042*	nonsense	Turco, 1996
090021	PK10461	PKD1	44	c.12138+2T>G		Splicing	Audrezet, 2012
020006	PK10307	PKD1	44	c.12139-1G>A		Splicing	Audrezet, 2012
130018	PK10525	PKD1	45	c.12215_12216del	p.Ser4072Tyrfs*84	Frameshift	Audrezet, 2012
020008	PK10308	PKD1	45	c.12224_12225del	p.Cys4075Serfs*81	Frameshift	Audrezet, 2012
021427	PK11197	PKD1	45	c.12244_12245del	p.Leu4082Valfs*74	Frameshift	This study
191561	PK11314	PKD1	45	c.12253del	p.Leu4085Cysfs*113	Frameshift	This study
031232	PK10980	PKD1	45	c.12288G>A	p.Trp4096*	nonsense	Audrezet, 2012
021736	PK11519	PKD1	45	c.12288G>A	p.Trp4096*	nonsense	Audrezet, 2012
021745	PK11519	PKD1	45	c.12288G>A	p.Trp4096*	nonsense	Audrezet, 2012
021744	PK11519	PKD1	45	c.12288G>A	p.Trp4096*	nonsense	Audrezet, 2012
060044	PK10647	PKD1	45	c.12310_12313del	p.Val4104Phefs*93	Frameshift	Audrezet, 2012
020064	PK11145	PKD1	45	c.12419G>A	p.Trp4140*	nonsense	Perrichot, 1999
090022	PK10119	PKD1	45	c.12420G>A	p.Trp4140*	nonsense	Perrichot, 1999
010137	PK10485	PKD1	45	c.12421dup	p.Met4141Asnfs*16	Frameshift	Audrezet, 2012
010866	PK10485	PKD1	45	c.12421dup	p.Met4141Asnfs*16	Frameshift	Audrezet, 2012
191864	PK11544	PKD1	45	c.12440dup	p.Gln4148Glyfs*9	Frameshift	This study
191809	PK11547	PKD1	45	c.12440dup	p.Gln4148Glyfs*9	Frameshift	This study
021209	PK10944	PKD1	45	c.12445-2A>T		Splicing	This study
110012	PK10607	PKD1	46	c.12445-5_12451dup	p.Arg4150_His4151insProGlnPheArg	In Frame insertion	Audrezet, 2012
110017	PK10607	PKD1	46	c.12445-5_12451dup	p.Arg4150_His4151insProGlnPheArg	In Frame insertion	Audrezet, 2012
030089	PK10539	PKD1	46	c.12530dup	p.Pro4178Thrfs*32	Frameshift	Audrezet, 2012
070013	PK10649	PKD1	46	c.12568dup	p.Ser4190Phefs*20	Frameshift	Audrezet, 2012
030073	PK10477	PKD1	46	c.12577_12580dup	p.Leu4194Profs*17	Frameshift	Audrezet, 2012
120011	PK10540	PKD1	46	c.12605_12632del	p.Arg4203Profs*146	Frameshift	Audrezet, 2012
191782	PK11467	PKD1	46	c.12608_12635del	p.Arg4203Profs*146	Frameshift	Audrezet, 2012
010046	PK10253	PKD1	46	c.12624_12625del	p.Cys4208*	Frameshift	Audrezet, 2012
120004	PK10253	PKD1	46	c.12624_12625del	p.Cys4208*	Frameshift	Audrezet, 2012
120003	PK10253	PKD1	46	c.12624_12625del	p.Cys4208*	Frameshift	Audrezet, 2012
021210	PK10961	PKD1	46	c.12640del	p.Arg4214Alafs*144	Frameshift	This study
021270	PK10961	PKD1	46	c.12640del	p.Arg4214Alafs*144	Frameshift	This study
020113	PK10566	PKD1	46	c.12691C>T	p.Gln4231*	nonsense	Stekrova, 2009
031163	PK10842	PKD1	46	c.12721C>T	p.Gln4241*	nonsense	This study
030812	PK10842	PKD1	46	c.12721C>T	p.Gln4241*	nonsense	This study
191723	PK10842	PKD1	46	c.12721C>T	p.Gln4241*	nonsense	This study
091014	PK10902	PKD2	1	c.12721C>T		Large rearrangement	This study
010901	PK10734	PKD2	1	c.169C>T	p.Gln57*	nonsense	This study
020121	PK10664	PKD2	1	c.200_213del	p.Pro67Argfs*20	Frameshift	Audrezet, 2012
020027	PK10418	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
090018	PK10420	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
090033	PK10569	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
050017	PK10753	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
030943	PK10968	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
040904	PK10771	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
040906	PK10771	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
040907	PK10771	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
030979	PK10837	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
090970	PK10420	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
010860	PK10420	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999
060957	PK11129	PKD2	1	c.203dup	p.Ala69Glyfs*23	Frameshift	Torra, 1999

010875	PK10420	PKD2	1	c.203dup	p.Ala69Glys*23	Frameshift	Torna, 1999
091451	PK11345	PKD2	1	c.203dup	p.Ala69Glys*23	Frameshift	Torna, 1999
021445	PK11364	PKD2	1	c.203dup	p.Ala69Glys*23	Frameshift	Torna, 1999
021605	PK11392	PKD2	1	c.203dup	p.Ala69Glys*23	Frameshift	Torna, 1999
021110	PK10963	PKD2	1	c.432C>G	p.Tyr144*	nonsense	This study
140009	PK10733	PKD2	1	c.478C>T	p.Gln160*	nonsense	Reiterova, 2002
020067	PK10499	PKD2	1	c.595+1G>A		Splicing	Magstrom, 2003
030048	PK10396	PKD2	1	c.595+1G>C		Splicing	Audrezet, 2012
031464	PK11457	PKD2	1	c.595+1G>C		Splicing	Audrezet, 2012
191575	PK11464	PKD2	2	c.619G>T	p.Glu207*	nonsense	This study
021616	PK11495	PKD2	2	c.704_709+2del	p.Cys235_1le236del	Frameshift	This study
020036	PK10399	PKD2	2	c.709+1G>A		Splicing	Veidhøisen, 1997
020051	PK10437	PKD2	3	c.1095-2A>G		Splicing	Audrezet, 2012
191563	PK11456	PKD2	3	c.817_818del	p.Leu273Valfs*29	Frameshift	Rossetti, 2003
030045	PK10377	PKD2	3	c.843+1G>A		Splicing	Audrezet, 2012
031166	PK10377	PKD2	3	c.843+1G>A		Splicing	Audrezet, 2012
010166	PK10669	PKD2	3	g.88952830_g.89050619	p.Arg561*	Large rearrangement	Audrezet, 2012
010844	PK11052	PKD2	3	g.88952830_g.89050619		Large rearrangement	Audrezet, 2012
041007	PK11293	PKD2	3	g.88952830_g.89050619		Large rearrangement	Audrezet, 2012
060932	PK10470	PKD2	4	c.1081C>T		nonsense	Chung, 2006
060936	PK10825	PKD2	4	c.1081C>T	p.Arg361*	nonsense	Chung, 2006
010837	PK10570	PKD2	4	c.1094+1G>A		Splicing	Chung, 2006
030817	PK10682	PKD2	4	c.1094+1G>C		Splicing	Yu, 2011
021258	PK11061	PKD2	4	c.12424G>A	p.Gly414Ser	nonsense	Rossetti, 2007
021608	PK11458	PKD2	4	c.866del	p.Asp289Valfs*28	Frameshift	This study
110008	PK10471	PKD2	4	c.958C>T	p.Arg320*	nonsense	Reynolds, 1999
040020	PK10554	PKD2	4	c.958C>T	p.Arg320*	nonsense	Reynolds, 1999
090024	PK10628	PKD2	4	c.958C>T	p.Arg320*	nonsense	Reynolds, 1999
030804	PK10680	PKD2	4	c.973C>T	p.Arg325*	nonsense	Rossetti, 2007
030947	PK10772	PKD2	4	c.973C>T	p.Arg325*	nonsense	Rossetti, 2007
031284	PK11131	PKD2	4	c.973C>T	p.Arg325*	nonsense	Rossetti, 2007
191492	PK11598	PKD2	4	c.973C>T	p.Arg325*	nonsense	Rossetti, 2007
040001	PK10371	PKD2	5	c.1140G>A	p.Trp380*	nonsense	Audrezet, 2012
040002	PK10371	PKD2	5	c.1140G>A	p.Trp380*	nonsense	Audrezet, 2012
040003	PK10371	PKD2	5	c.1140G>A	p.Trp380*	nonsense	Audrezet, 2012
030072	PK10371	PKD2	5	c.1140G>A	p.Trp380*	nonsense	Audrezet, 2012
041334	PK11321	PKD2	5	c.1140G>A	p.Trp380*	nonsense	Audrezet, 2012
041407	PK10371	PKD2	5	c.1140G>A	p.Trp380*	nonsense	Audrezet, 2012
030035	PK10376	PKD2	5	c.1249C>T	p.Arg417*	nonsense	Pei, 1998
030055	PK10435	PKD2	5	c.1249C>T	p.Arg417*	nonsense	Pei, 1998
030096	PK10376	PKD2	5	c.1249C>T	p.Arg417*	nonsense	Pei, 1998
110003	PK10353	PKD2	5	c.1320+1G>A		Splicing	Audrezet, 2012
110015	PK10353	PKD2	5	c.1320+1G>A		Splicing	Audrezet, 2012
161144	PK10934	PKD2	5	c.1320+1G>A		Splicing	Audrezet, 2012
180008	PK11009	PKD2	5	c.1320+1G>A		Splicing	Audrezet, 2012
141280	PK11227	PKD2	5	c.1320+1G>A		Splicing	Audrezet, 2012
030044	PK10416	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbøy, 1997
020062	PK10468	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbøy, 1997
130030	PK10557	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbøy, 1997
030816	PK10416	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbøy, 1997
021117	PK10991	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbøy, 1997

021208	PK10991	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbay, 1997
031239	PK10416	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbay, 1997
010876	PK10091	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbay, 1997
021447	PK11395	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbay, 1997
021615	PK11462	PKD2	6	c.1390C>T	p.Arg464*	nonsense	Virbay, 1997
100012	PK10486	PKD2	6	c.1391del	p.Arg464Hisfs*50	Frameshift	Audrezet, 2012
010888	PK11294	PKD2	6	c.1412dup	p.Phe472Leufs*6	Frameshift	This study
211813	PK11531	PKD2	6	c.1412dup	p.Phe472Leufs*6	Frameshift	This study
211827	PK11531	PKD2	6	c.1412dup	p.Phe472Leufs*6	Frameshift	This study
021185	PK10997	PKD2	6	c.1461T>A	p.Tyr487*	nonsense	This study
091247	PK11134	PKD2	7	c.1589C>A	p.Ser530*	nonsense	This study
091345	PK11134	PKD2	7	c.1589C>A	p.Ser530*	nonsense	This study
021330	PK11134	PKD2	7	c.1589C>A	p.Ser530*	nonsense	This study
091800	PK11134	PKD2	7	c.1589C>A	p.Ser530*	nonsense	This study
090042	PK10637	PKD2	7	c.1704dup	p.Val569Cysfs*4	Frameshift	Audrezet, 2012
130027	PK10556	PKD2	7	c.1717-2A>G		Splicing	Audrezet, 2012
130042	PK10658	PKD2	7	c.1717-2A>G		Splicing	Audrezet, 2012
080016	PK10658	PKD2	7	c.1717-2A>G		Splicing	Audrezet, 2012
131049	PK10556	PKD2	7	c.1717-2A>G		Splicing	Audrezet, 2012
010161	PK10099	PKD2	8	c.1774C>T	p.Arg592*	nonsense	Audrezet, 2012
130052	PK10712	PKD2	8	c.1774C>T	p.Arg592*	nonsense	Audrezet, 2012
020149	PK10726	PKD2	8	c.1811del	p.Phe604Serfs*6	Frameshift	This study
021107	PK10726	PKD2	8	c.1811del	p.Phe604Serfs*6	Frameshift	This study
010053	PK10234	PKD2	8	c.1837C>T	p.Chr613*	nonsense	Audrezet, 2012
020019	PK10370	PKD2	9	c.1960C>T	p.Arg654*	nonsense	Irazabal, 2011
040008	PK10394	PKD2	9	c.1960C>T	p.Arg654*	nonsense	Irazabal, 2011
191498	PK11399	PKD2	9	c.1960C>T	p.Arg654*	nonsense	Irazabal, 2011
020074	PK10464	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
130016	PK10639	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
110013	PK10640	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
030110	PK10641	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
030113	PK10676	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
130921	PK10714	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
131046	PK10932	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
131307	PK10676	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
131391	PK10464	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
131396	PK10639	PKD2	9	c.2019+2T>A		Splicing	Audrezet, 2012
060029	PK10154	PKD2	10	c.2059_2063del	p.Val687Ilefs*2	Frameshift	Audrezet, 2012
010887	PK10154	PKD2	10	c.2059_2063del	p.Val687Ilefs*2	Frameshift	Audrezet, 2012
010107	PK10378	PKD2	10	c.2077C>T	p.Gln693*	nonsense	Audrezet, 2012
131310	PK11241	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
030026	PK10301	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
030042	PK10532	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
020046	PK10531	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
030060	PK10532	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
130001	PK10536	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
130004	PK10610	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
020080	PK10531	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
130002	PK10611	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
130007	PK10612	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012
130010	PK10613	PKD2	10	g.88983145_g.89011203		Large rearrangement	Audrezet, 2012

I30024	PK10651	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30019	PK10652	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
030083	PK10652	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
030101	PK10662	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30033	PK10880	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30034	PK10739	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
030099	PK10881	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30041	PK10740	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30047	PK10613	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30048	PK10688	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30014	PK10611	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30923	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30925	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30924	PK10752	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30927	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I30930	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31042	PK10536	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31047	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I61142	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I60001	PK10905	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I80997	PK10947	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31152	PK10612	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
070915	PK10955	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
031221	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31158	PK10301	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
021251	PK10532	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31304	PK10880	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31306	PK11186	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
021328	PK11188	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I31394	PK11186	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I21532	PK11365	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
I21271	PK11365	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
051136	PK10752	PKD2	10	g-88983145.g-8901203	Large rearrangement	Audrezet, 2012
031175	PK11001	PKD2	11	c-2159del	Frameshift	Pei, 1998
031290	PK11136	PKD2	11	c-2159del	Frameshift	Pei, 1998
031291	PK10103	PKD2	11	c-2159del	Frameshift	Pei, 1998
091593	PK10103	PKD2	11	c-2159del	Frameshift	Pei, 1998
010020	PK10217	PKD2	11	c-2159dup	Frameshift	Pei, 1998
031171	PK10998	PKD2	11	c-2159dup	Frameshift	Pei, 1998
031236	PK10998	PKD2	11	c-2159dup	Frameshift	Pei, 1998
091457	PK11401	PKD2	11	c-2159dup	Frameshift	Pei, 1998
191582	PK11459	PKD2	11	c-2203A>T	noise	This study
021268	PK11133	PKD2	11	c-2224C>T	noise	Mochizuki, 1996
021325	PK11185	PKD2	11	c-2240+1G>A	Splicing	Tan, 2011
191708	PK11463	PKD2	11	c-2240+1G>A	Splicing	Tan, 2011
091347	PK11407	PKD2	11	c-2533C>T	noise	Torn, 2000
031287	PK11132	PKD2	12	c-2266del	Frameshift	This study
020026	PK10380	PKD2	12	c-2358+1G>A	Splicing	Rosetti, 2007
060028	PK10624	PKD2	13	c-2407C>T	noise	Deltas, 2001
031028	PK10815	PKD2	13	c-2407C>T	noise	Deltas, 2001
031388	PK10815	PKD2	13	c-2407C>T	noise	Deltas, 2001

091796	PK10624	PKD2	13	c.2407C>T	p.Arg803*	noise/ise	Deltas, 2001
050006	PK10351	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
050003	PK10351	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
050007	PK10352	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
010105	PK10398	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
130009	PK10553	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
010168	PK10351	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
130023	PK10625	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
130037	PK10636	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
030807	PK10677	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
090036	PK10668	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
090044	PK10668	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
060042	PK10711	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
020160	PK10774	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
120983	PK11082	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
131156	PK10711	PKD2	13	c.2419C>T	p.Arg807*	noise/ise	Virbø, 1997
030015	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
040004	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
090004	PK10382	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
040010	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
090006	PK10419	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
040015	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
040011	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
010163	PK10382	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
130050	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
040909	PK10382	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
041331	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
041408	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
041409	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
041522	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
041523	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
041526	PK10417	PKD2	13	c.2508C>G	p.Tyr836*	noise/ise	Audrezet, 2012
090965	PK10999	PKD2	14	c.2527del	p.Val843*	Frameshift	This study
031378	PK11319	PKD2	14	c.2527del	p.Val843*	Frameshift	This study
021733	PK11530	PKD2	14	c.2527del	p.Val843*	Frameshift	This study
010900	PK11319	PKD2	14	c.2527del	p.Val843*	Frameshift	This study
091343	PK11184	PKD2	14	c.2528_2529del	p.Val843Glnfs*18	Frameshift	This study
010121	PK10188	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
090020	PK10469	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
010144	PK10555	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
010174	PK10555	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
090027	PK10629	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
010180	PK10188	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
010181	PK10188	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
010849	PK10725	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
090964	PK11000	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
010884	PK11182	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
031297	PK11189	PKD2	14	c.2533C>T	p.Arg845*	noise/ise	Torn, 2000
021216	PK11083	PKD2	14	c.2564del	p.Ser855Thrfs*2	Frameshift	This study
191516	PK11393	PKD2	14	c.2568_2569del	p.Ser858Glnfs*3	Frameshift	This study
191764	PK11536	PKD2	14	c.2568_2569del	p.Ser858Glnfs*3	Frameshift	This study

010183	PK10298	PKD2	14	c.2614C>T	p.Arg872*	nonsense	Reynolds, 1999
040910	PK10993	PKD2	14	c.2614C>T	p.Arg872*	nonsense	Reynolds, 1999
041406	PK10993	PKD2	14	c.2614C>T	p.Arg872*	nonsense	Reynolds, 1999
030109	PK10638	PKD2	15	c.2845_2848del	p.Ser949Asnfs*49	Frameshift	Audrezet, 2012
040007	PK10393	PKD2	15	c.2905T>C	p.*969Arget*13	nonstop	Audrezet, 2012
021188	PK10393	PKD2	15	c.2905T>C	p.*969Arget*13	nonstop	Audrezet, 2012

Genkyst ID	Family ID	Gene	Exon/Intron	Nucleotide Change	Effect on Protein	Mutation type	Score	Reference
010111	PK10303	PKD1	1	c.153C>G	p.Cys51Trp	Missense	16	Audrezet, 2012
021737	PK11522	PKD1	1	c.153C>G	p.Cys51Trp	Missense	16	Audrezet, 2012
031240	PK11109	PKD1	1	c.216-8C>G		Splicing		This study
131400	PK11109	PKD1	1	c.216-8C>G		Splicing		This study
070008	PK10642	PKD1	3	c.359T>C	p.Ile120Thr	Missense	20	Audrezet, 2012
070911	PK10642	PKD1	3	c.359T>C	p.Ile120Thr	Missense	20	Audrezet, 2012
010039	PK10990	PKD1	5	c.548G>A	p.Cys183Tyr	Missense	20	Audrezet, 2012
031389	PK11257	PKD1	5	c.629G>A	p.Cys210Tyr	Missense	16	Rossetti, 2012
051138	PK11257	PKD1	5	c.629G>A	p.Cys210Tyr	Missense	16	rossetti, 2012
040025	PK10673	PKD1	5	c.695G>A	p.Cys232Tyr	Missense	14	Comtec-Le Gall, 2013
010047	PK10288	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
110001	PK10289	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
030046	PK10373	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
010149	PK10288	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
130026	PK10289	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
031023	PK10373	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
051131	PK10288	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
131301	PK10289	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
191596	PK11479	PKD1	5	c.974A>G	p.Tyr325Cys	Missense	14	Rossetti, 2007
021354	PK11202	PKD1	6	c.1295C>T	p.Ala432Val	Missense	19	Rossetti, 2007
021739	PK11202	PKD1	6	c.1295C>T	p.Ala432Val	Missense	19	Rossetti, 2007
120981	PK10719	PKD1	7	c.1544G>T	p.Gly515Trp	Missense	14	Comtec-Le Gall, 2013
030010	PK10939	PKD1	7	c.1605A>G		Splicing		This study
030094	PK10939	PKD1	7	c.1605A>G		Splicing		This study
030814	PK11067	PKD1	7	c.1605A>G		Splicing		This study
021181	PK10939	PKD1	7	c.1605A>G		Splicing		This study
031285	PK11067	PKD1	7	c.1605A>G		Splicing		This study
031933	PK11067	PKD1	7	c.1605A>G		Splicing		This study
010170	PK10138	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
020063	PK10284	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
030066	PK10404	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
130022	PK10530	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
090040	PK10586	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
010841	PK10623	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
020155	PK10742	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
021070	PK10854	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
111096	PK11302	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
191557	PK11427	PKD1	11	c.2180T>C	p.Leu727Pro	Missense	20	Rossetti, 2007
031230	PK11043	PKD1	11	c.2534T>C	p.Leu458Ser	Missense	19	Thomas, 1999
100004	PK10964	PKD1	12	c.2985+8G>A		Splicing		This study
021182	PK10898	PKD1	13	c.3161+5G>T		Splicing		This study
191553	PK10898	PKD1	13	c.3161+5G>T		Splicing		This study
030014	PK10273	PKD1	14	c.3296-3C>G		Splicing		Audrezet, 2012
031372	PK10273	PKD1	14	c.3296-3C>G		Splicing		Audrezet, 2012
031379	PK10273	PKD1	14	c.3296-3C>G		Splicing		Audrezet, 2012
020094	PK10457	PKD1	15	c.3719_3721del	p.Asn1240del	In Frame deletion		Rossetti, 2003
021204	PK10916	PKD1	15	c.3719_3721del	p.Asn1240del	In Frame deletion		Rossetti, 2003

010868	PK10114	PKD1	15	c.3719_3721del	p.Asn1240del	In Frame deletion	Rossetti, 2003
010880	PK10114	PKD1	15	c.3719_3721del	p.Asn1240del	In Frame deletion	Rossetti, 2003
091419	PK11258	PKD1	15	c.4115_4117del	p.Trp1372del	In Frame deletion	This study
021118	PK10851	PKD1	15	c.4349_4351del	p.Asn1450del	In Frame deletion	This study
031234	PK10982	PKD1	15	c.4349_4351del	p.Asn1450del	In Frame deletion	This study
021365	PK11213	PKD1	15	c.4981_4983del	p.Asn1661del	In Frame deletion	This study
021607	PK11424	PKD1	15	c.5995G>A	p.Gly1999Ser	Missense	Rossetti, 2007
020038	PK10389	PKD1	15	c.5976_5978del	p.Phe1992_Thr1993delinsLeu	In Frame deletion	Rossetti, 2001
020115	PK10389	PKD1	15	c.5976_5978del	p.Phe1992_Thr1993delinsLeu	In Frame deletion	Rossetti, 2001
030112	PK10614	PKD1	15	c.6845A>G	p.Tyr2282Cys	Missense	Audrezet, 2012
060016	PK10335	PKD1	15	c.6778_6780del	p.Ile2256del	In Frame deletion	Rossetti, 2001
030052	PK10425	PKD1	15	c.6778_6780del	p.Ile2256del	In Frame deletion	Rossetti, 2001
060025	PK10335	PKD1	15	c.6778_6780del	p.Ile2256del	In Frame deletion	Rossetti, 2001
060045	PK10335	PKD1	15	c.6778_6780del	p.Ile2256del	In Frame deletion	Rossetti, 2001
031373	PK11201	PKD1	15	c.6778_6780del	p.Ile2256del	In Frame deletion	Rossetti, 2001
010033	PK10256	PKD1	15	c.6892T>G	p.Trp2298Gly	Missense	Audrezet, 2012
010035	PK10286	PKD1	16	c.7066-17_7066-9del		Splicing	Audrezet, 2012
060039	PK10286	PKD1	16	c.7066-17_7066-9del		Splicing	Audrezet, 2012
010141	PK10136	PKD1	17	c.7136A>G	p.Tyr2379Cys	Missense	Perrichot, 2000
090009	PK10338	PKD1	17	c.7136A>G	p.Tyr2379Cys	Missense	Perrichot, 2000
090038	PK10338	PKD1	17	c.7136A>G	p.Tyr2379Cys	Missense	Perrichot, 2000
010840	PK10136	PKD1	17	c.7136A>G	p.Tyr2379Cys	Missense	Perrichot, 2000
090030	PK10338	PKD1	17	c.7136A>G	p.Tyr2379Cys	Missense	Perrichot, 2000
010076	PK10235	PKD1	17	c.7136A>G	p.Tyr2379Cys	Missense	Perrichot, 2000
031286	PK11088	PKD1	17	c.7209+4_7209+7del		Splicing	Rossetti, 2007
021102	PK10896	PKD1	18	c.7271C>T	p.Thr2424Met	Missense	Yu, 2011
030084	PK10505	PKD1	18	c.7300C>T	p.Arg2434Trp	Missense	Hoefele, 2010
030949	PK10706	PKD1	18	c.7300C>T	p.Arg2434Trp	Missense	Hoefele, 2010
031224	PK11017	PKD1	18	c.7300C>T	p.Arg2434Trp	Missense	Hoefele, 2010
020053	PK10441	PKD1	18	c.7426T>A	p.Cys2476Ser	Missense	Audrezet, 2012
180006	PK10922	PKD1	18	c.7426T>A	p.Cys2476Ser	Missense	Audrezet, 2012
021183	PK10922	PKD1	18	c.7426T>A	p.Cys2476Ser	Missense	Audrezet, 2012
021260	PK10922	PKD1	18	c.7426T>A	p.Cys2476Ser	Missense	Audrezet, 2012
021748	PK10441	PKD1	18	c.7426T>A	p.Cys2476Ser	Missense	Audrezet, 2012
010082	PK10134	PKD1	18	c.7483T>C	p.Cys2495Arg	Missense	Audrezet, 2012
010087	PK10296	PKD1	19	c.7546C>T	p.Arg2516Cys	Missense	Rossetti, 2007
130928	PK10831	PKD1	19	c.7546C>T	p.Arg2516Cys	Missense	Rossetti, 2007
030088	PK10474	PKD1	19	c.7543_7548del	p.Arg2515_Arg2516del	In Frame deletion	Audrezet, 2012
021443	PK11277	PKD1	19	c.7546C>T	p.Arg2516Cys	Missense	Rossetti, 2007
010179	PK10225	PKD1	23	c.8299C>T	p.Arg2767Cys	Missense	Garcia-Gonzales, 2007
010153	PK10442	PKD1	23	c.8299C>T	p.Arg2767Cys	Missense	Garcia-Gonzales, 2007
010131	PK10189	PKD1	23	c.8300G>C	p.Arg2767Pro	Missense	Audrezet, 2012
010154	PK10189	PKD1	23	c.8300G>C	p.Arg2767Pro	Missense	Audrezet, 2012
010157	PK10189	PKD1	23	c.8300G>C	p.Arg2767Pro	Missense	Audrezet, 2012
010881	PK10189	PKD1	23	c.8300G>C	p.Arg2767Pro	Missense	Audrezet, 2012
010896	PK10189	PKD1	23	c.8300G>C	p.Arg2767Pro	Missense	Audrezet, 2012
120007	PK10385	PKD1	23	c.8309A>T	p.Asn2770Ile	Missense	Audrezet, 2012
010150	PK10130	PKD1	23	c.8311G>A	p.Gln2771Lys	Missense	Rossetti, 2001
020048	PK10366	PKD1	23	c.8311G>A	p.Gln2771Lys	Missense	Rossetti, 2001
020095	PK10366	PKD1	23	c.8311G>A	p.Gln2771Lys	Missense	Rossetti, 2001
030103	PK10546	PKD1	23	c.8311G>A	p.Gln2771Lys	Missense	Rossetti, 2001

020135	PK10551	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
090050	PK10650	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
090962	PK10650	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
090961	PK10650	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
050016	PK10793	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
131050	PK10846	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
191711	PK11377	PKD1	23	c.831G>A	p.Gln277Lys	Missense	11	Rossetti, 2001
020105	PK10528	PKD1	23	c.844T>C	p.Leu2816Pro	Missense	18	Rossetti, 2001
030029	PK10292	PKD1	23	c.8557_8562del	p.Phe2853_Gln2854del	In Frame deletion		Audrezet, 2012
090015	PK10495	PKD1	23	c.8557_8562del	p.Phe2853_Gln2854del	In Frame deletion		Audrezet, 2012
191779	PK10495	PKD1	23	c.8557_8562del	p.Phe2853_Gln2854del	In Frame deletion		Audrezet, 2012
160004	PK10596	PKD1	23	c.8791G>A		Splicing		Garcia-Gonzales, 2007
010184	PK10596	PKD1	23	c.8791G>A		Splicing		Garcia-Gonzales, 2007
021066	PK10801	PKD1	25	c.9202-3C>G		Splicing	20	This study
030049	PK10496	PKD1	26	c.9377C>T	p.Thr3126Ile	Missense	20	Audrezet, 2012
120008	PK10524	PKD1	26	c.9377C>T	p.Thr3126Ile	Missense	20	Audrezet, 2012
060030	PK10524	PKD1	26	c.9377C>T	p.Thr3126Ile	Missense	20	Audrezet, 2012
021370	PK10496	PKD1	26	c.9377C>T	p.Thr3126Ile	Missense	20	Audrezet, 2012
191510	PK11342	PKD1	26	c.9377C>T	p.Thr3126Ile	Missense	20	Audrezet, 2012
130011	PK10542	PKD1	27	c.9564_9566del	p.Asn3188del	In Frame deletion		This study
030028	PK10302	PKD1	27	c.9430G>T	p.Gly3144Trp	Missense	20	Audrezet, 2012
130926	PK10302	PKD1	27	c.9430G>T	p.Gly3144Trp	Missense	20	Audrezet, 2012
191583	PK11426	PKD1	27	c.9431G>A	p.Gly3144Glu	Missense	20	Audrezet, 2012
191599	PK11426	PKD1	27	c.9431G>A	p.Gly3144Glu	Missense	20	Audrezet, 2012
020029	PK10413	PKD1	27	c.9485G>A	p.Arg3162Leu	Missense	20	Audrezet, 2012
021429	PK10413	PKD1	27	c.9485G>T	p.Arg3162Leu	Missense	20	Audrezet, 2012
021604	PK10413	PKD1	27	c.9485G>T	p.Arg3162Leu	Missense	20	Audrezet, 2012
191790	PK11457	PKD1	27	c.9564_9566del	p.Asn3188del	In Frame deletion		This study
020148	PK10694	PKD1	27	c.9568G>A		Splicing		This study
021106	PK10782	PKD1	28	c.9924-3C>G		Splicing		This study
021316	PK10782	PKD1	28	c.9924-3C>G		Splicing		This study
PK10782	PK10782	PKD1	28	c.9924-3C>G		Splicing		This study
031080	PK10802	PKD1	29	c.9859_9861del	p.Leu3287del	In Frame deletion		This study
021734	PK11508	PKD1	29	c.9859_9861del	p.Leu3287del	In Frame deletion		Thongnopplakun, 2004
191806	PK11546	PKD1	29	c.9859_9861del	p.Leu3287del	In Frame deletion		Thongnopplakun, 2004
020001	PK10278	PKD1	29	c.9924-3C>G		Splicing		Audrezet, 2012
020020	PK10278	PKD1	29	c.9924-3C>G		Splicing		Audrezet, 2012
020072	PK10278	PKD1	29	c.9924-3C>G		Splicing		Audrezet, 2012
021062	PK10782	PKD1	29	c.9924-3C>G		Splicing		Audrezet, 2012
031079	PK10787	PKD1	29	c.9924-3C>G		Splicing		Audrezet, 2012
021109	PK10278	PKD1	29	c.9924-3C>G		Splicing		Audrezet, 2012
010029	PK10173	PKD1	37	c.10951G>A	p.Gly3651Ser	Missense	17	Zhang, 2005
020024	PK10347	PKD1	37	c.10951G>A	p.Gly3651Ser	Missense	17	Zhang, 2005
100014	PK10578	PKD1	37	c.10951G>A	p.Gly3651Ser	Missense	17	Zhang, 2005
120019	PK10619	PKD1	37	c.10951G>A	p.Gly3651Ser	Missense	17	Zhang, 2005
021254	PK11025	PKD1	39	c.11176T>C	p.Trp3726Arg	Missense	18	Reed, 2008
021255	PK11025	PKD1	39	c.11176T>C	p.Trp3726Arg	Missense	18	Reed, 2008
020052	PK11050	PKD1	37	c.11007C>T		Splicing		Rossetti, 2012
010108	PK10329	PKD1	37	c.11017-10C>A		Splicing		Perrichot, 1999
020163	PK10751	PKD1	39	c.11157G>C		Splicing		This study
021369	PK10751	PKD1	39	c.11157G>C		Splicing		This study

020021	PK10584	PKD1	39	c.11204T>A	p.Val3735Asp	Missense	18	Comec-Le Gall, 2013
020025	PK10357	PKD1	39	c.11249_11250delinsAA	p.Arg3750Gln	Missense	20	Audrezet, 2012
030091	PK10512	PKD1	39	c.11204_11209del	p.Val3735_His3736del	In Frame deletion		Audrezet, 2012
030104	PK10512	PKD1	39	c.11204_11209del	p.Val3735_His3736del	In Frame deletion		Audrezet, 2012
030948	PK10696	PKD1	39	c.11204_11209del	p.Val3735_His3736del	In Frame deletion		Audrezet, 2012
031479	PK10512	PKD1	39	c.11204_11209del	p.Val3735_His3736del	In Frame deletion		Audrezet, 2012
021067	PK10800	PKD1	39	c.11249_11250delinsAA	p.Arg3750Gln	Missense	20	Audrezet, 2012
191512	PK11266	PKD1	39	c.11249_11250delinsAA	p.Arg3750Gln	Missense	20	Audrezet, 2012
191515	PK10363	PKD1	39	c.11249_11250delinsAA	p.Arg3750Gln	Missense	20	Audrezet, 2012
010177	PK10196	PKD1	39	c.11251C>A	p.Gln3751Lys	Missense	16	Audrezet, 2012
010041	PK10196	PKD1	39	c.11251C>A	p.Gln3751Lys	Missense	16	Audrezet, 2012
010031	PK10196	PKD1	39	c.11251C>A	p.Gln3751Lys	Missense	16	Audrezet, 2012
020031	PK10339	PKD1	39	c.11257C>T	p.Arg3753Trp	Missense	20	Tsuchiya, 2001
020032	PK10339	PKD1	39	c.11257C>T	p.Arg3753Trp	Missense	20	Tsuchiya, 2001
030054	PK11053	PKD1	39	c.11257C>T	p.Arg3753Trp	Missense	20	Tsuchiya, 2001
020131	PK10608	PKD1	39	c.11257C>T	p.Arg3753Trp	Missense	20	Tsuchiya, 2001
021314	PK10608	PKD1	39	c.11257C>T	p.Arg3753Trp	Missense	20	Tsuchiya, 2001
010886	PK10167	PKD1	39	c.11258G>A	p.Arg3753Gln	Missense	15	Rossetti, 2007
131044	PK10818	PKD1	39	c.11258G>A	p.Arg3753Gln	Missense	15	Rossetti, 2007
021218	PK10984	PKD1	41	c.11451_11453dup	p.Gly3818dup	In Frame insertion		This study
010071	PK10319	PKD1	42	c.11614C>C	p.Gln3872Gln	Missense	19	Audrezet, 2012
020002	PK10327	PKD1	42	c.11614G>C	p.Gln3872Gln	Missense	19	Audrezet, 2012
021364	PK10327	PKD1	42	c.11614G>C	p.Gln3872Gln	Missense	14	Audrezet, 2012
090041	PK10145	PKD1	43	c.11980_11988dup	p.Phe3996_Leu3998dup	In Frame insertion		This study
191562	PK10145	PKD1	43	c.11980_11988dup	p.Phe3996_Leu3998dup	In Frame insertion		This study
191765	PK10145	PKD1	43	c.11980_11988dup	p.Phe3996_Leu3998dup	In Frame insertion		This study
010001	PK10080	PKD1	45	c.12388_12390del	p.Val4130del	In Frame deletion		Garcia-Gonzales, 2007
091013	PK10776	PKD1	45	c.12391_12393del	p.Gln4131del	In Frame deletion		This study
010112	PK10285	PKD1	46	c.12448C>T	p.Arg4150Cys	Missense	20	Garcia-Gonzales, 2007
060008	PK10444	PKD1	46	c.12448C>T	p.Arg4150Cys	Missense	20	Garcia-Gonzales, 2007
020093	PK10575	PKD1	46	c.12448C>T	p.Arg4150Cys	Missense	20	Garcia-Gonzales, 2007
120012	PK10444	PKD1	46	c.12448C>T	p.Arg4150Cys	Missense	20	Garcia-Gonzales, 2007
020168	PK10575	PKD1	46	c.12448C>T	p.Arg4150Cys	Missense	20	Garcia-Gonzales, 2007
010892	PK11251	PKD1	46	c.12448C>T	p.Arg4150Cys	Missense	20	Garcia-Gonzalez, 2007
121531	PK11408	PKD2	4	c.1034A>G	p.Tyr345Cys	Missense	16	Yu, 2011
010185	PK10233	PKD2	4	c.1094+3_1094+6del		Splicing		Magistri, 2003
010857	PK10233	PKD2	4	c.1094+3_1094+6del		Splicing		Magistri, 2003
031074	PK10832	PKD2	4	c.1094+3_1094+6del		Splicing		Magistri, 2003
170003	PK11099	PKD2	4	c.965G>A	p.Arg322Gln	Missense	20	Peltola, 2005
070007	PK10935	PKD2	4	c.1031G>A	p.Cys344Tyr	Missense	20	This study
021262	PK11152	PKD2	4	c.964C>T	p.Arg322Trp	Missense	20	Reiterova, 2002
141276	PK11183	PKD2	4	c.964C>T	p.Arg322Trp	Missense	20	Reiterova, 2002
091454	PK11529	PKD2	4	c.964C>T	p.Arg322Trp	Missense	20	Reiterova, 2002
021108	PK10903	PKD2	4	c.965G>A	p.Arg322Gln	Missense	12	Peltola, 2005
021732	PK10903	PKD2	4	c.965G>A	p.Arg322Gln	Missense	12	Peltola, 2005
021265	PK11149	PKD2	5	c.1320+5T>G		Splicing		This study
130032	PK10630	PKD2	6	c.1548+3A>C		Splicing		Audrezet, 2012
090827	PK10715	PKD2	11	c.2208_2213del	p.Leu736_Asn737del	In Frame deletion		Stekrova, 2004
131871	PK11455	PKD2	14	c.2657A>G	p.Asp86Gly	Missense	18	Reynolds, 1999

Genkyst ID	Family ID	Gene	Exon/Intron	Nucleotide Change	Effect on Protein	Mutation type	Score	Reference
021195	PK11008	PKD1	1	c.116G>T	p.Cys59Phe	Missense	16	This study
021219	PK11008	PKD1	1	c.116G>T	p.Cys59Phe	Missense	16	This study
021252	PK11008	PKD1	1	c.116G>T	p.Cys59Phe	Missense	16	This study
191507	PK11420	PKD1	2	c.230A>G	p.Asn77Ser	Missense	12	This study
191713	PK11498	PKD1	2	c.230A>G	p.Asn77Ser	Missense	12	This study
010113	PK10242	PKD1	4	c.385T>C	p.Cys129Arg	Missense	20	Audrezet, 2012
070003	PK10526	PKD1	4	c.464G>A	p.Cys155Tyr	Missense	20	Audrezet, 2012
191706	PK11477	PKD1	4	c.464G>A	p.Cys155Tyr	Missense	20	Audrezet, 2012
010873	PK11016	PKD1	5	c.1088G>A	p.Cys363Tyr	Missense	14	This study
061124	PK11016	PKD1	5	c.1088G>A	p.Cys363Tyr	Missense	14	This study
120018	PK10635	PKD1	5	c.674C>T	p.Ser225Leu	Missense	6	Comes-Le Gall, 2013
021115	PK10953	PKD1	6	c.1379T>A	p.Val460Asp	Missense	15	This study
020125	PK10653	PKD1	7	c.1576C>G	p.His526Asp	Missense	20	Audrezet, 2012
010093	PK10243	PKD1	9	c.1730T>G	p.Val577Gly	Missense	17	Audrezet, 2012
031283	PK11062	PKD1	9	c.1826T>G	p.Val609Gly	Missense	17	Rossetti, 2007
010133	PK10915	PKD1	9	c.1831C>T	p.Arg611Trp	Missense	20	Garcia-Gonzales, 2007
020085	PK10508	PKD1	9	c.1831C>T	p.Arg611Trp	Missense	20	Garcia-Gonzales, 2007
080005	PK10534	PKD1	9	c.1831C>T	p.Arg611Trp	Missense	20	Garcia-Gonzales, 2007
020158	PK10508	PKD1	9	c.1831C>T	p.Arg611Trp	Missense	20	Garcia-Gonzales, 2007
010836	PK10616	PKD1	10	c.2053G>A	p.Glu685Lys	Missense	17	Audrezet, 2012
020018	PK10332	PKD1	10	c.2092T>G	p.Tyr698Asp	Missense	20	Audrezet, 2012
020055	PK10440	PKD1	10	c.2092T>G	p.Tyr698Asp	Missense	20	Audrezet, 2012
020301	PK10440	PKD1	10	c.2092T>G	p.Tyr698Asp	Missense	20	Audrezet, 2012
191751	PK10440	PKD1	10	c.2092T>G	p.Tyr698Asp	Missense	20	Audrezet, 2012
191865	PK10440	PKD1	10	c.2092T>G	p.Tyr698Asp	Missense	20	Audrezet, 2012
061121	PK10904	PKD1	11	c.2387A>G	p.Tyr796Cys	Missense	20	Rossetti, 2012
111093	PK10951	PKD1	11	c.2485C>T	p.Pro829Ser	Missense	10	This study
010005	PK10205	PKD1	15	c.3617T>G	p.Val1206Gly	Missense	10	Audrezet, 2012
010042	PK10448	PKD1	15	c.3617T>G	p.Val1206Gly	Missense	10	Audrezet, 2012
100013	PK10579	PKD1	15	c.3617T>G	p.Val1206Gly	Missense	10	Audrezet, 2012
010861	PK10830	PKD1	15	c.3617T>G	p.Val1206Gly	Missense	10	Audrezet, 2012
091243	PK11106	PKD1	15	c.3617T>G	p.Val1206Gly	Missense	10	Audrezet, 2012
010890	PK11299	PKD1	15	c.3617T>G	p.Val1206Gly	Missense	10	Audrezet, 2012
020116	PK10581	PKD1	15	c.3722T>A	p.Ile1241Asn	Missense	14	Audrezet, 2012
021362	PK11220	PKD1	15	c.3872T>A	p.Val1291Asp	Missense	18	This study
161146	PK11015	PKD1	15	c.4076T>C	p.Leu1359Pro	Missense	16	This study
030815	PK10656	PKD1	15	c.4379T>G	p.Val1460Gly	Missense	20	Audrezet, 2012
031030	PK10656	PKD1	15	c.4379T>G	p.Val1460Gly	Missense	20	Audrezet, 2012
010869	PK10976	PKD1	15	c.4379T>G	p.Val1460Gly	Missense	20	Audrezet, 2012
010871	PK10976	PKD1	15	c.4379T>G	p.Val1460Gly	Missense	20	Audrezet, 2012
100991	PK10976	PKD1	15	c.4379T>G	p.Val1460Gly	Missense	20	Audrezet, 2012
031086	PK10860	PKD1	15	c.4385T>G	p.Val1462Gly	Missense	20	This study
031376	PK11218	PKD1	15	c.4462T>C	p.Ser1488Pro	Missense	16	This study
021065	PK10859	PKD1	15	c.5093G>C	p.Arg1698Pro	Missense	6	This study
020118	PK10633	PKD1	15	c.5180C>T	p.Pro1727Leu	Missense	20	Audrezet, 2012
020123	PK10633	PKD1	15	c.5180C>T	p.Pro1727Leu	Missense	20	Audrezet, 2012
020164	PK10812	PKD1	15	c.5180C>T	p.Pro1727Leu	Missense	20	Audrezet, 2012

131154	PK11026	PKD1	15	c.5180C>T	p.Pro1727Leu	Missense	20	Audrezet, 2012
021321	PK11143	PKD1	15	c.5180C>T	p.Pro1727Leu	Missense	20	Audrezet, 2012
131397	PK11026	PKD1	15	c.5180C>T	p.Pro1727Leu	Missense	20	Audrezet, 2012
130929	PK10785	PKD1	15	c.5936G>A	p.Cys1979Trp	Missense	8	Cornec-Le Gall, 2013
030972	PK10747	PKD1	15	c.5960G>A	p.Gly1987Asp	Missense	14	Cornec-Le Gall, 2013
090966	PK10746	PKD1	15	c.5983C>T	p.Arg1995Cys	Missense	10	Cornec-Le Gall, 2013
010839	PK10707	PKD1	15	c.5998T>C	p.Ser2000Pro	Missense	10	Cornec-Le Gall, 2013
191589	PK11478	PKD1	15	c.6082T>G	p.Tyr2028Asp	Missense	14	This study
050005	PK10763	PKD1	15	c.6086C>T	p.Thr2029Met	Missense	14	This study
070001	PK10705	PKD1	15	c.6382A>C	p.Asn2128His	Missense	14	Cornec-Le Gall, 2013
020102	PK10576	PKD1	15	c.6496C>T	p.Arg2166Cys	Missense	14	Garcia-Gonzalez, 2007
010178	PK10661	PKD1	15	c.6643C>T	p.Arg2215Trp	Missense	19	Cornec-Le Gall, 2013
060939	PK11249	PKD1	15	c.6643C>T	p.Arg2215Trp	Missense	19	Cornec-Le Gall, 2013
031467	PK11450	PKD1	15	c.6643C>T	p.Arg2215Trp	Missense	19	Cornec-Le Gall, 2013
020151	PK10720	PKD1	15	c.6647T>A	p.Leu2216Gln	Missense	14	Cornec-Le Gall, 2013
140003	PK10574	PKD1	15	c.6658C>T	p.Arg2220Trp	Missense	10	Vujic, 2010
141405	PK10574	PKD1	15	c.6658C>T	p.Arg2220Trp	Missense	10	Vujic, 2010
010877	PK11058	PKD1	15	c.6855C>A	p.Asn2285Lys	Missense	14	This study
030945	PK10703	PKD1	15	c.6892T>C	p.Trp2298Arg	Missense	18	This study
031226	PK10703	PKD1	15	c.6892T>C	p.Trp2298Arg	Missense	18	This study
191520	PK11359	PKD1	15	c.6892T>C	p.Trp2298Arg	Missense	18	This study
060018	PK10583	PKD1	16	c.6985C>T	p.Arg2329Trp	Missense	10	Perrichot, 2000
010024	PK10316	PKD1	17	c.7115C>G	p.Ser2372Cys	Missense	20	Audrezet, 2012
010186	PK10316	PKD1	17	c.7115C>G	p.Ser2372Cys	Missense	20	Audrezet, 2012
010187	PK10316	PKD1	17	c.7115C>G	p.Ser2372Cys	Missense	20	Audrezet, 2012
010119	PK10400	PKD1	17	c.7172G>A	p.Gly2391Asp	Missense	14	Hoefele, 2010
191789	PK11510	PKD1	18	c.7264A>C	p.Thr2422Pro	Missense	17	This study
191844	PK11510	PKD1	18	c.7264A>C	p.Thr2422Pro	Missense	17	This study
191852	PK11510	PKD1	18	c.7264A>C	p.Thr2422Pro	Missense	17	This study
030097	PK10606	PKD1	18	c.7376G>A	p.Gly2459Asp	Missense	14	Audrezet, 2012
140008	PK10882	PKD1	18	c.7222C>T	p.Arg2408Cys	Missense	10	Phakdeekitcharoen, 2000
031078	PK10841	PKD1	19	c.7622C>T	p.Pro2541Leu	Missense	17	This study
130051	PK11166	PKD1	20	c.7844T>C	p.Leu2615Pro	Missense	14	This study
091242	PK11040	PKD1	23	c.8207C>G	p.Pro2736Arg	Missense	14	This study
031164	PK10857	PKD1	20	c.7837_7839del	p.Leu2615del	In Frame deletion		This study
020152	PK10709	PKD1	23	c.8363C>G	p.Ser2788Trp	Missense	14	This study
060934	PK10675	PKD1	23	c.8537C>T	p.Thr2846Ile	Missense	20	Cornec-Le Gall, 2013
061128	PK10675	PKD1	23	c.8537C>T	p.Thr2846Ile	Missense	20	Cornec-Le Gall, 2013
110006	PK10125	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
030011	PK10264	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110002	PK10264	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110004	PK10315	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
010130	PK10315	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110005	PK10365	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
090011	PK10315	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
090017	PK10315	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110007	PK10264	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110011	PK10315	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
020112	PK10501	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110016	PK10315	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012
110018	PK10156	PKD1	23	c.8475_8476msGCT	p.Leu2825_Ile2826msAla	In Frame insertion		Audrezet, 2012

050013	PK10125	PKD1	23	c.8475_8476msGCT	p.Leu2825_ile2826msAla	In Frame insertion	8	Audrezet, 2012
021312	PK11105	PKD1	23	c.8639C>T	p.Ser2880Leu	Missense	This study	
010100	PK10708	PKD1	24	c.8933T>G	p.Phe2978Cys	Missense	20	Cornec-Le Gall, 2013
010102	PK10241	PKD1	26	c.9361G>A	p.Glu3121Lys	Missense	13	Cornec-Le Gall, 2013
010172	PK10529	PKD1	26	c.9361G>A	p.Glu3121Lys	Missense	13	Cornec-Le Gall, 2013
141401	PK11501	PKD1	26	c.9377C>G	p.Thr3126Arg	Missense	20	This study
020165	PK10748	PKD1	26	c.9395C>T	p.Ser3132Leu	Missense	20	Cornec-Le Gall, 2013
191550	PK11360	PKD1	27	c.9404C>T	p.Thr3153Met	Missense	20	Bataille, 2011
191585	PK11425	PKD1	27	c.9430G>A	p.Gly3144Arg	Missense	20	This study
031081	PK11005	PKD1	27	c.9499A>T	p.Ile3167Phe	Missense	12	Rossetti, 2002
060037	PK10654	PKD1	27	c.9543G>C	p.Lys3181Asn	Missense	17	Audrezet, 2012
191761	PK11499	PKD1	27	c.9551T>G	p.Val3184Gly	Missense	17	This study
021212	PK11013	PKD1	28	c.9569G>A	p.Gly3190Glu	Missense	20	This study
031021	PK10828	PKD1	28	c.9577C>T	p.Pro3193Ser	Missense	20	Cornec-Le Gall, 2013
090014	PK10038	PKD1	28	c.9578C>T	p.Pro3193Leu	Missense	20	Perrichot, 2000
090034	PK10038	PKD1	28	c.9578C>T	p.Pro3193Leu	Missense	20	Perrichot, 2000
090010	PK10038	PKD1	28	c.9578C>T	p.Pro3193Leu	Missense	20	Perrichot, 2000
031084	PK10894	PKD1	28	c.9644T>G	p.Val3215Gly	Missense	14	This study
091020	PK10894	PKD1	28	c.9644T>G	p.Val3215Gly	Missense	14	This study
031238	PK10894	PKD1	28	c.9644T>G	p.Val3215Gly	Missense	14	This study
191862	PK11525	PKD1	28	c.9694A>G	p.Lys3232Glu	Missense	13	Rossetti, 2012
010090	PK10362	PKD1	29	c.9829C>T homo	p.Arg3277Cys homo	Missense	16	Rossetti, 2009
191794	PK11509	PKD1	29	c.9829C>T homo	p.Arg3277Cys homo	Missense	16	Rossetti, 2009
030802	PK10655	PKD1	29	c.9898G>A	p.Gly3300Arg	Missense	16	Cornec-Le Gall, 2013
130021	PK10580	PKD1	30	c.10043G>A	p.Arg3348Gln	Missense	12	non out
130053	PK10580	PKD1	30	c.10043G>A	p.Arg3348Gln	Missense	12	non out
020049	PK10893	PKD1	37	c.10835C>A	p.Ala3612Asp	Missense	20	This study
020065	PK10893	PKD1	37	c.10835C>A	p.Ala3612Asp	Missense	20	This study
021941	PK10893	PKD1	37	c.10835C>A	p.Ala3612Asp	Missense	20	This study
030007	PK10326	PKD1	37	c.10958C>T	p.Ala3653Val	Missense	20	Audrezet, 2012
030036	PK10337	PKD1	37	c.10958C>T	p.Ala3653Val	Missense	20	Audrezet, 2012
030808	PK10326	PKD1	37	c.10958C>T	p.Ala3653Val	Missense	20	Audrezet, 2012
090826	PK10663	PKD1	39	c.11249G>A	p.Arg3750Gln	Missense	13	Audrezet, 2012
090833	PK10663	PKD1	39	c.11249G>A	p.Arg3750Gln	Missense	13	Audrezet, 2012
020022	PK10462	PKD1	41	c.11468T>C	p.Leu3823Pro	Missense	20	Audrezet, 2012
031298	PK11155	PKD1	41	c.11524T>G	p.Trp3842Gly	Missense	20	This study
030941	PK10692	PKD1	42	c.11711C>G	p.Ser3904Trp	Missense	14	Cornec-Le Gall, 2013
020004	PK10811	PKD1	43	c.11969T>G	p.Leu3990Arg	Missense	19	This study
020012	PK10811	PKD1	43	c.11969T>G	p.Leu3990Arg	Missense	19	This study
021264	PK10811	PKD1	43	c.11969T>G	p.Leu3990Arg	Missense	19	This study
061123	PK11107	PKD1	44	c.12051G>C	p.Lys4017Asn	Missense	16	This study
191489	PK11240	PKD1	45	c.12439A>G	p.Lys4147Gly	Missense	17	This study
060023	PK10604	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
060036	PK10620	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
130012	PK10648	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
060931	PK10620	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
131157	PK10648	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
131302	PK10648	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
141277	PK11157	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999
191519	PK11538	PKD1	46	c.12826C>T	p.Arg4276Trp	Missense	16	Badenas, 1999

040023	PK10724	PKD2	3	c.818T>C	p.Leu273Pro	Missense	19	This study
040903	PK10724	PKD2	3	c.818T>C	p.Leu273Pro	Missense	19	This study
131151	PK11003	PKD2	5	c.1242G>T	p.Trp14Cys	Missense	20	This study
130040	PK10723	PKD2	6	c.1349G>A	p.Glu450Asp	Missense	18	This study
031282	PK11148	PKD2	8	c.1778G>A	p.Cys593Tyr	Missense	15	This study
030085	PK10577	PKD2	9	c.1991T>C	p.Phe664Ser	Missense	19	This study
091249	PK11151	PKD2	14	c.2546T>A	p.Met849Lys	Missense	20	This study