

Supplementary Table 1. Novel pathogenic or likely pathogenic variants identified in Taiwan PKD

Gene	Variant (c.)	Variant (p.)	ACMG Classification	gnomAD MAF	Variant (c.)	Variant (p.)	ACMG Classification	gnomAD MAF
PKD1 NM_001009944.3								
	c.350T>A	p.Leu117*	P		c.7460T>C	p.Leu2487Pro	LP	
	c.427C>T	p.Gln143*	LP		c.7624G>T	p.Gly2542Cys	LP	
	c.689G>T	p.Cys230Phe	LP		c.7694C>T	p.Ala2565Val	LP	
	c.829delC	p.Leu277fs	P		c.7829_7836delAGTACTCG	p.Glu2610fs	P	
	c.890dup	p.Leu298fs	P		c.7831T>C	p.Tyr2611His	LP	0.0000319 (1/31372)
	c.1013_1019delCCCTGGG	p.Ala338fs	P		c.7844T>G	p.Leu2615Arg	LP	
	c.1201+2T>C	p.?	P		c.7886_7887insC	p.Ala2630fs	P	
	c.1265T>A	p.Leu422Gln	LP		c.7969A>C	p.Thr2657Pro	LP	
	c.1336_1339delATGG	p.Met446fs	P		c.8291_8308delTGCGCTCCCGCGTGCTCA	p.Met2764_Leu2769del	LP	
	c.1364T>C	p.Phe455Ser	LP		c.8299C>G	p.Arg2767Gly	LP	
	c.1385+1G>T	p.?	P		c.8306_8310delTCAAC	p.Leu2769fs	P	
	c.1688delA	p.Gln563fs	LP		c.8327T>C	p.Leu2776Pro	LP	
	c.1765delC	p.Leu589fs	P		c.8359C>G	p.Arg2787Gly	LP	
	c.1796delT	p.Leu599fs	P		c.8362_8371delTCGGACCCGC	p.Ser2788fs	P	
	c.1849+2T>G	p.?	P	0.00562 (60/10680, ALFA)	c.8377C>G	p.Leu2793Val	LP	0.000011 (3/125568, TOPMed)
	c.2073delC	p.Ala692fs	P		c.8751_8761delGGCCGGGCTGC	p.Ala2918fs	P	
	c.2803C>T	p.Leu935Phe	LP		c.9033C>G	p.Tyr3011*	P	
	c.2884G>T	p.Asp962Tyr	LP		c.9074G>A	p.Trp3025*	P	
	c.2894delT	p.Phe965fs	P		c.9124_9130delGTCTGCC	p.Val3042fs	P	
	c.3132_3156dup	p.Phe1053fs	P		c.9129C>A	p.Cys3043*	P	
	c.3426delG	p.Val1144fs	LP		c.9158C>A	p.Ala3053Asp	LP	
	c.3524_3527dup	p.Ala1177fs	P		c.9173C>T	p.Pro3058Leu	LP	0.0000261 (5/191614)

Supplementary Table 1. Novel pathogenic or likely pathogenic variants identified in Taiwan PKD (continued)

c.3675_3676insC	p.Gly1226fs	P		c.9240delA	p.Cys3081fs	LP	
c.3741G>A	p.Met1247Ile	LP		c.9400A>G	p.Thr3134Ala	LP	0.000004 (1/246260)
c.3833_3845delGC CCCGCCGGCCA	p.Ser1278fs	P		c.9507delG	p.Ile3170fs	LP	
c.4209delT	p.Cys1403fs	LP		c.9541A>G	p.Lys3181Glu	LP	
c.4834dup	p.Thr1612fs	P		c.9585G>T	p.Trp3195Cys	LP	
c.4868_4869dup	p.Ile1624fs	P		c.9644T>C	p.Val3215Ala	LP	
c.5157delG	p.Trp1720fs	P		c.9827delC	p.Thr3276fs	P	
c.5207_5208delTC	p.Val1736fs	P		c.9835C>T	p.Gln3279*	P	
c.5436delC	p.Gly1813fs	LP		c.10188dup	p.Lys3397fs	LP	
c.5517G>A	p.Trp1839*	P		c.10220+1G>A	p.?	P	
c.5602G>T	p.Ala1868Ser	LP	0.000012 (3/246184)	c.10406-2A>G	p.?	P	
c.5601_5609delTGC CTCCAA	p.Ser1869_Ala187 1del	LP		c.10616C>T	p.Thr3539Ile	LP	
c.5669_5682delTG GTGCTGTGGGCC	p.Leu1890fs	P		c.10710_10712delGG C	p.Ala3571del	LP	
c.5803delC	p.Arg1935fs	P		c.10711delG	p.Ala3571fs	LP	
c.5842dup	p.Val1948fs	P		c.10764G>A	p.Trp3588*	P	
c.5970delG	p.Asn1991fs	LP		c.11256dup	p.Arg3753fs	P	
c.6102delG	p.Leu2035fs	P		c.11274_11275delCT	p.Tyr3759fs	P	
c.6109dup	p.Glu2037fs	P		c.11411+1dup	p.?	P	
c.6224G>C	p.Arg2075Pro	LP		c.11510T>C	p.Leu3837Pro	LP	
c.6293A>C	p.Asp2098Ala	LP		c.11526G>T	p.Trp3842Cys	LP	
c.6296_6319delGG TCGCCAGGGCAG GACACAGATG	p.Gly2099_Asp210 6del	LP		c.11687_11694dup	p.Leu3899fs	P	
c.6341_6344delACC T	p.Tyr2114fs	P		c.11725_11727delCTG	p.Leu3909del	VUS_P	
c.6371T>A	p.Val2124Glu	LP		c.11758dup	p.Arg3920fs	LP	
c.6497G>T	p.Arg2166Leu	LP		c.11782delT	p.Trp3928fs	LP	
c.6512C>G	p.Ala2171Gly	LP		c.11955dup	p.Ala3986fs	LP	
c.6534_6550delCGT CACCTACCAGACT G	p.Cys2178fs	P		c.12003G>C	p.Lys4001Asn	LP	0.000009 (2/226208)
c.6555C>G	p.Tyr2185*	P		c.12013_12016delCAG C	p.Gln4005fs	LP	
c.6647T>C	p.Leu2216Pro	LP		c.12123_12124dup	p.Gln4042fs	LP	

Supplementary Table 1. Novel pathogenic or likely pathogenic variants identified in Taiwan PKD (continued)

PKD2 NM_000297.4	c.6667C>G	p.Leu2223Val	LP		c.12138G>A	p.Leu4046=	LP(splicing effect)	0.0000144 (4/277686)
	c.6669dup	p.Pro2224fs	P		c.12139_12146delGCC TTCCCCGCAGCTCG TGTC	p.Leu4047fs	P	
	c.6781G>T	p.Glu2261*	P		c.12245delT	p.Leu4082fs	P	
	c.6921_6922insCAC ACCG	p.Ala2308fs	LP		c.12299_12300insTAG G	p.Leu4100fs	P	
	c.6989T>A	p.Leu2330Gln	LP		c.12395T>A	p.Leu4132*	P	
	c.6991_7009delGC GGCTGGCGTGGA GTACA	p.Ala2331fs	P		c.12424G>T	p.Gly4142Cys	LP	0.000004 (1/245366)
	c.7009A>C	p.Thr2337Pro	LP		c.12442delG	p.Glu4148fs	P	
	c.7025T>G	p.Val2342Gly	LP		c.12456delA	p.Val4153fs	P	
	c.7109G>A	p.Cys2370Tyr	LP		c.12471delG	p.Met4158fs	P	
					c.12751_12823delAGC AGCCGGGCGCCCGC CGGATCTTCCCGTG GCCCCATCCCCGGGC CTGCGGCCAGCACT GCCCAGCCGCCTTG	p.Ser4251fs	P	
	c.7328delG	p.Gly2443fs	P					
	c.224delC	p.Pro75fs	LP		c.1049delT	p.Val350fs	LP	
	c.449_450insCG	p.Ser151fs	P		c.1091delC	p.Ala365fs	P	
	c.647_648delAC	p.Tyr216fs	P		c.1969G>T	p.Gly657*	P	
	c.755dup	p.Met252fs	LP		c.2011_2014delATTC	p.Ile671fs	P	
	c.1030T>C	p.Cys344Arg	LP		c.2320G>T	p.Glu774*	P	
PKHD1 NM_138694.4	c.1042T>A	p.Tyr348Asn	LP		c.2380_2392delAGTT CTTTACCAC	p.Ser794fs	P	
	c.227C>A	p.Pro76His	LP	0.0000079 6 (2/251386)	c.3299T>C	p.Phe1100Ser	LP	
	c.439G>A	p.Gly147Ser	LP	0.000004 (1/251188)	c.4039_4091delGTGA GCCTGTCTGGATGC TCCATCCCTCTTCAC AGTCTGGAGGCTGG CATCTA	p.Val1347fs	P	
	c.448+1G>A	p.?	P		c.4197_4199delTTCins AAG	p.Ser1400Arg	LP	

Supplementary Table 1. Novel pathogenic or likely pathogenic variants identified in Taiwan PKD (continued)

GANAB NM_198335.4	c.704delG	p.Gly235fs	P	0.000008 (2/251292) 0.0000283 (8/282550)	c.4202delC	p.Ala1401fs	LP
	c.1046G>A	p.Gly349Glu	LP		c.8432T>C	p.Leu2811Pro	LP
	c.1124G>A	p.Arg375Gln	LP		c.10284T>G	p.Tyr3428*	P
	c.252+2T>C	p.?	P		c.1900+1G>C	p.?	P
	c.1389G>A	p.Trp463*	P		c.2116C>T	p.Arg706*	P
	c.1803+1G>T	p.?	P		c.2504G>A	p.Trp835*	P
ALG8 NM_024079.5	c.396delA	p.Val133fs	P		c.824delG	p.Gly275fs	P

ACMG: The American College of Medical Genetics and Genomics, ALFA: Allele Frequency Aggregator (<https://www.ncbi.nlm.nih.gov/snp/docs/gsr/alfa/>), gnomAD: The Genome Aggregation Database (<https://gnomad.broadinstitute.org/>), LP: likely pathogenic, P: pathogenic, MAF: minor allele frequency, TOPMed: The Trans-Omics for Precision Medicine (<https://topmed.nhlbi.nih.gov/>).

Supplementary Table 2. Mutation Landscape of Taiwan Autosomal Dominant Polycystic Kidney Disease.

Gene	Family	Coding Change (c.)	Amino Acid Change	Mutation type	Decision	ACMG (VarSome)	ClinVar	De	Mayo PKDB	HGMD (P)	LOVD	dbSNP	Genome database Frequency	Same amino acid change but different coding changes in references		
PKD1	DY2288	c.196C>A	p.Pro66Thr	Missense	VUS	Uncertain Significance	LP									
	DY1143	c.256C>T	p.Leu86Phe	Missense	VUS	Uncertain Significance	LP					rs1263493445	0.000004 (1/264690, TOPMED)			
	DY828, DY1349, DY1535, DY1661, DY1907	c.266A>C	p.Asn89Thr	Missense	VUS	Benign			Indeterminate			rs1434978033	0.000163 (27/165760, GnomAD)			
	DY1289, DY2243	c.348_352delTTTAA	p.Asn116fs	Frameshift Indel	P	Pathogenic	Pathogenic	Definitely Pathogenic								
	DY1402	c.350T>A	p.Leu117*	Truncating	P	Pathogenic								PMID: 31740684	c.350T>G	
	DY318	c.356A>G	p.Glu119Gly	Missense	VUS	Uncertain Significance						rs748839748	0.000007 (1/149554, GnomAD)			
	DY2007	c.412C>T	p.Arg138*	Truncating	P	Pathogenic	Likely path	Definitely Pathc	+			rs1596591955				
	DY1441	c.417G>A	p.Trp139*	Truncating	P	Pathogenic				+						
	DY1192	c.427C>T	p.Gln143*	Truncating	P	Likely Pathogenic										
	DY1169	c.430C>T	p.Gln144*	Truncating	P	Likely Pathogenic										
	DY2232	c.445C>T	p.Gln149*	Truncating	P	Pathogenic				+	Pathogenic					
	DY1925	c.689G>T	p.Cys230Phe	Missense	P	Likely Pathogenic										
	DY2170	c.692T>C	p.Leu231Pro	Missense	VUS	Uncertain Significance	P									
	DY95	c.776G>A	p.Cys259Tyr	Missense	P	Likely Benig	Uncertain	Highly Likely Pa	+			rs529066905	0.000165 (28/169848, GnomAD)			
	DY1736, DY1782 (in-cis)	c.812C>A	p.Ala271Asp	Missense	P	Likely Pathogenic			Likely Pathogeni	+	Likely Pathogenic					
	DY154	c.829delC	p.Leu277fs	Frameshift Indel	P	Pathogenic								PMID: 27499327	not existed in the supplemental data	
	DY125, DY2231	c.856_862delTCTGGC	p.Gly287*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathogenic				rs1555459108	0			
	DY1347	c.890dup	p.Leu298fs	Frameshift Indel	P	Pathogenic								PMID: 33462484	non-PKD related	
	DY705, DY1638, DY1661, DY1956	c.974A>G	p.Tyr325Cys	Missense	P	Likely Patho	Conflicting	Highly Likely Pa	+			rs1232180956	0			
	DY1381	c.1013_1019delCCCTG	p.Ala338fs	Frameshift Indel	P	Pathogenic								PMID: 27499327	c.1013_1039del127	
	DY1844	c.1021delG	p.Ala341fs	Frameshift Indel	P	Pathogenic								PMID: 27499327		
	DY1964	c.1198C>T	p.Arg400*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+			rs774453006	0.00001 (1/71508, GnomAD)			
	KMUH2349	c.1201+2T>C	p.?	Splicing site	P	Pathogenic								PMID: 27835667	c.1201+1G>A	
	1_A4	c.1202-9G>A	p.?	Splicing site	P	Uncertain Si	Likely path	Likely Pathogenic								
	DY20	c.1202-1G>T	p.?	Splicing site	P	Pathogenic										
	DY2025	c.1222delT	p.Ser408fs	Frameshift Indel	P	Likely Pathogenic										
	DY1426	c.1261C>T	p.Arg421Cys	Missense	P	Likely Patho	Likely pathogenic	(Last revi	+			rs1567216536	0			
	KMUH_1736, DY828	c.1265T>A	p.Leu422Gln	Missense	P	Likely Pathogenic										
	DY292, DY1825, DY1862, DY1937, DY2223	c.1295C>T	p.Ala432Val	Missense	P	Likely Patho	Likely path	Highly Likely Pa	+			rs1060499699	0			
	DY2181	c.1336_1339delATGG	p.Met446fs	Frameshift Indel	P	Pathogenic										
	DY2015	c.1364T>C	p.Phe455Ser	Missense	P	Likely Pathogenic										
	DY2041	c.1385+1G>T	p.?	Splicing site	P	Pathogenic										
	DY2172	c.1591G>A	p.Glu531Lys	Missense	P	Uncertain Si	Uncertain significance		+			rs1567215326	None			
	DY25, DY304	c.1597C>T	p.Gln533*	Truncating	P	Pathogenic			Definitely Pathc	+						
	DY1458	c.1688delA	p.Gln563fs	Frameshift Indel	P	Likely Pathogenic										
	DY2092	c.1765delC	p.Leu589fs	Frameshift Indel	P	Pathogenic										
	DY13	c.1781T>A	p.Phe594Tyr	Missense	VUS	Uncertain Significance			Indeterminate							
	DY748	c.1781T>C	p.Phe594Ser	Missense	VUS	Uncertain Si	Uncertain significance									
	DY1858	c.1796T>C	p.Leu599Pro	Missense	VUS	Uncertain Significance										
	DY2056	c.1796delT	p.Leu599fs	Frameshift Indel	P	Pathogenic								PMID: 29529603	c.1796_1808del113	
	DY2159	c.1831C>T	p.Arg611Trp	Missense	P	Likely Patho	Conflicting	Likely Pathogeni	+		likely path	rs1555458413	0.00000 (0/10680, ALFA)			
	DY814	c.1849+2T>G	p.?	Splicing site	P	Pathogenic						rs1596582514	0.0232 (67/2894, KOREAN);0.00562 (60/10680, ALFA Project)			
	DY1770, DY1748	c.1849+5_1849+6delG	p.?	Splicing site	VUS	Uncertain Significance			Indeterminate							
	DY1239, DY1423	c.2016dup	p.Pro673fs	Frameshift Indel	P	Likely Pathogenic					Pathogenic					
	DY1707, DY2283	c.2073delC	p.Ala692fs	Frameshift Indel	P	Pathogenic										
	DY1356	c.2097+5G>A	p.?	Splicing site	VUS	Uncertain Si	Uncertain significance	(Last reviewed: Feb 5, 2020)								

DY297	c.2180T>C	p.Leu727Pro	Missense	P	Likely Patho	Conflicting	Highly Likely Pat	+	rs1616940	0.001 (1/998, GoNL)			
DY1826 (<i>in-cis</i>)	c.2432T>C	p.Leu811Pro	Missense	VUS	Uncertain Significance								
DY2130	c.2489C>T	p.Ala830Val	Missense	VUS	Uncertain Significance_LP				rs775988021	0.000038 (10/264690, TOPMED)			
DY1362, KMH1619, KMH1825	c.2494dupC	p.Arg832fs	Frameshift Indel	P	Pathogenic	Pathogenic	Definitely Pathogenic		Pathogenic	rs1567210630	0		
DY1645, DY1647, DY1848	c.2534T>C	p.Leu845Ser	Missense	P	Pathogenic	Pathogenic	Highly Likely Pathogenic		Likely Patho	rs199476100	0.000004 (1/230128, GnomAD)		
DY2106	c.2671G>A	p.Asp891Asn	Missense	VUS	Uncertain Significance_LP				rs770952448	0.000004 (1/264690, TOPMED)			
DY1997	c.2803C>T	p.Leu935Phe	Missense	P	Likely Pathogenic								
DY1305	c.2884G>T	p.Asp962Tyr	Missense	P	Likely Pathogenic								
DY1413	c.2885A>G	p.Asp962Gly	Missense	P	Likely Benig	Uncertain	Likely Pathogenic						
DY2139	c.2894delT	p.Phe965fs	Frameshift Indel	P	Pathogenic								
DY2268	c.2972T>C	p.Val991Ala	Missense	VUS	Uncertain Significance_LP				p.Val991Gly (VUS)				
DY349	c.2985+6T>C	p.?	Splicing site	VUS	Uncertain Significance								
DY2145	c.2985+6_2985+52del	p.?	Splicing site	VUS	Uncertain Significance_LP								
DY130	c.2986-1G>A	p.?	Splicing site	P	Pathogenic	Pathogenic							
DY99, DY2282	c.3088G>A	p.Val1030Met	Missense	VUS	Uncertain Significance				rs747210535	0.000016 (4/246314, GnomAD)			
DY2203	c.3132_3156dup	p.Phe1053fs	Frameshift Indel	P	Pathogenic								
DY1685, DY1815	c.3161+1G>A	p.?	Splicing site	P	Pathogenic				Likely Pathogenic				
DY1386	c.3196C>T	p.Gln1066*	Truncating	P	Pathogenic		Definitely Pathogenic						
DY1404	c.3202C>T	p.Gln1068*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathogenic						
DY1114	c.3322T>C	p.Ser1108Pro	Missense	VUS	Uncertain Significance								
DY1088, DY1162	c.3349C>T	p.Gln1117*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogenic				
DY1707	c.3380C>A	p.Pro1127His	Missense	VUS	Uncertain Significance								
DY55	c.3386T>A	p.Val1129Glu	Missense	VUS	Uncertain Significance								
DY1826 (<i>in-cis</i>)	c.3392T>C	p.Val1131Ala	Missense	VUS	Uncertain Significance								
DY2208	c.3426delG	p.Val1144fs	Frameshift Indel	P	Likely Pathogenic						PMID: 27499327	c.3425_3428dupGGCC	
DY691, DY1996, DY2081, DY2218	c.3490G>C	p.Gly1164Arg	Missense	P	Likely Pathogenic		+						
DY699	c.3496G>A	p.Gly1166Ser	Missense	P	Uncertain Si	Conflicting	Likely Pathogen	+	rs573566419	0.000065 (12/184266, GnomAD)			
DY1968	c.3524_3527dup	p.Ala1177fs	Frameshift Indel	P	Pathogenic								
DY1573	c.3675_3676insC	p.Gly1226fs	Frameshift Indel	P	Pathogenic								
DY1860	c.3741G>A	p.Met1247Ile	Missense	P	Likely Pathogenic								
DY1330	c.3833_3845delGCCC	p.Ser1278fs	Frameshift Indel	P	Pathogenic						PMID: 30042192	c.3831_3847del	
DY2036	c.3955G>A	p.Gly1319Arg	Missense	P	Likely Patho	Likely pathogenic		+	Likely path	rs747362311	0.000004 (1/243930, GnomAD_exome)		
DY789	c.3983G>A	p.Trp1328*	Truncating	P	Pathogenic			+					
DY1100, DY1191, DY1363, DY1394, DY1446, DY1524, DY1573, DY1579, DY1822	c.4052G>A	p.Arg1351Gln	Missense	VUS	Likely Benig	Uncertain significance			rs374129201	0.000231 (29/125568, TOPMED)			
DY1730	c.4069delC	p.Leu1357fs	Frameshift Indel	P	Pathogenic	Pathogenic	Definitely Pathogenic						
DY751	c.4099_4100delAG	p.Arg1367fs	Frameshift Indel	P	Pathogenic	Pathogenic	Definitely Pathogenic						
DY2266	c.4099_4100delAG	p.Arg1367fs	Frameshift Indel	P	Pathogenic		Definitely Pathogenic		Pathogenic				
DY2201	c.4209delT	p.Cys1403fs	Frameshift Indel	P	Likely Pathogenic								
DY1109, DY1807, DY1992, DY2183	c.4306C>T	p.Arg1436*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogenic				
A6, DY95	c.4369_4370delTC	p.Ala1458fs	Frameshift Indel	P	Pathogenic								
DY1930	c.4387C>T	p.Gln1463*	Truncating	P	Pathogenic		Definitely Pathc	+					
DY1091, DY1270 (<i>in-trans</i>)	c.4455C>G	p.Tyr1485*	Truncating	P	Pathogenic		Definitely Pathogenic		Pathogenic				

	DY1350 (in-cis)	c.4460T>C	p.Phe1487Ser	Missense	VUS	Uncertain Significance								
	DY1069, DY1189, DY1416,	c.4797C>A	p.Tyr1599*	Truncating	P	Pathogenic	Pathogenic	+		Pathogenic				
	DY1404	c.4798A>G	p.Thr1600Ala	Missense	VUS	Uncertain Significance					rs776529978	0.000016 (4/249458, GnomAD)		
	DY1386	c.4834dup	p.Thr1612fs	Frameshift Indel	P	Pathogenic								
	DY1225	c.4868_4869dup	p.Ile1624fs	Frameshift Indel	P	Pathogenic								
	DY43, DY320, DY1216,	c.5014_5015delAG	p.Arg1672fs	Frameshift Indel	P	Pathogenic	Pathogenic	Definitely Pathogenic			rs1555455457	0		
	DY1156, DY1350, DY1764, DY1795, DY1813	c.5037C>A	p.Ser1679Arg	Missense	VUS	Likely Benig	Uncertain significance				rs144091742	0.000441 (120/272282, GnomAD)		
	DY92	c.5157delG	p.Trp1720fs	Frameshift Indel	P	Pathogenic							PMID: 11857740	c.5159delG
	DY2051	c.5207_5208delTC	p.Val1736fs	Frameshift Indel	P	Pathogenic								
	DY1407	c.5363delG	p.Gly1788fs	Frameshift Indel	P	Likely Pathogenic	Definitely Pathogenic							
	DY1870	c.5366C>A	p.Ser1789*	Truncating	P	Pathogenic	Pathogenic	(Last reviewed Jan 17, 2019) ?			-			
	DY1435	c.5436delC	p.Gly1813fs	Frameshift Indel	P	Likely Pathogenic							PMID: 24611717	c.5438delG
	DY851	c.5482C>T	p.Gln1828*	Truncating	P	Pathogenic		Definitely Patho	+					
	DY831, DY1135, DY1186,	c.5483A>G	p.Gln1828Arg	Missense	VUS	Likely Benign	Indeterminate				rs563854855	0.000243 (53/217696, GnomAD)		
	DY357, DY701, DY2078	c.5517G>A	p.Trp1839*	Truncating	P	Pathogenic								
	DY2077	c.5585_5587delTCT	p.Phe1862del	In-frame Indel	VUS	Uncertain Significance_P								
	DY1195, DY1788	c.5602G>T	p.Ala1868Ser	Missense	P	Likely Pathogenic					rs753398195	0.000012 (3/246184, GnomAD)		
	DY1253	c.5601_5609delTGCC	p.Ser1869_Ala187	In-frame Indel	P	Likely Pathogenic								
	DY1597	c.5669_5682delTGGTC	p.Leu1890fs	Frameshift Indel	P	Pathogenic								
	DY1348	c.5682_5699delCAGC	p.Ser1895_Ala190	In-frame Indel	VUS	Uncertain Significance								
	DY1439	c.5704G>T	p.Gly1902Trp	Missense	VUS	Uncertain Significance								
	DY728 (in-cis)	c.5705G>A	p.Gly1902Glu	Missense	VUS	Uncertain Significance								
	DY45	c.5803delC	p.Arg1935fs	Frameshift Indel	P	Pathogenic								
	DY1241	c.5819dup	p.Arg1942fs	Frameshift Indel	P	Pathogenic								
	DY1230	c.5842dup	p.Val1948fs	Frameshift Indel	P	Pathogenic								
	DY1832	c.5852G>A	p.Arg1951Gln	Missense	VUS	Likely Benig	Uncertain significance	(Last reviewed: Jan 5, 201	rs137905643	0.000503 (115/228510, GnomAD)				
	DY1893, DY108, DY2238	c.5968_5969delAG	p.Arg1990fs	Frameshift Indel	P	Pathogenic		Definitely Pathogenic						
	B2	c.5970delG	p.Asn1991fs	Frameshift Indel	P	Likely Pathogenic								
	DY1344, DY1754	c.5976_5978delCAC	p.Phe1992_Thr199	In-frame Indel	P	Uncertain Significance	Highly Likely Pathogenic	Likely Pathogenic						
	DY294, DY295, DY1176, DY1436, DY1675, DY2140, DY2192	c.5995G>A	p.Gly1999Ser	Missense	P	Likely Patho	Likely path	Highly Likely Pa	+		Likely Pathogenic			
	DY1882	c.6018G>C	p.Trp2006Cys	Missense	P	Uncertain Significance		+						
	DY1149	c.6024C>G	p.Phe2008Leu	Missense	VUS	Uncertain Significance					rs770185977	0.000012 (3/241618, GnomAD)		
	DY1926	c.6037G>A	p.Val2013Ile	Missense	VUS	Uncertain Significance					rs778658448	0.000004 (1/242002, GnomAD)		
	DY339, DY747, DY1533,	c.6040C>T	p.Gln2014*	Truncating	P	Pathogenic		Definitely Patho	+					
	DY1694, DY1704, DY1751	c.6102delG	p.Leu2035fs	Frameshift Indel	P	Pathogenic								
	DY2173	c.6108G>T	p.Leu2036Phe	Missense	VUS	Uncertain Significance_P								
	DY19, DY837, DY1657	c.6109dup	p.Glu2037fs	Frameshift Indel	P	Pathogenic								
	DY1669	c.6224G>C	p.Arg2075Pro	Missense	P	Likely Pathogenic								
	DY1450, DY1483, DY1497,	c.6293A>C	p.Asp2098Ala	Missense	P	Likely Pathogenic								
	DY1346	c.6296_6319delGGTC	p.Gly2099_Asp210	In-frame Indel	P	Likely Pathogenic								
	DY1648	c.6341A>G	p.Tyr2114Cys	Missense	P	Likely Patho	Likely pathogenic	(Last revi	+					
	DY1554	c.6341_6344delACCT	p.Tyr2114fs	Frameshift Indel	P	Pathogenic								
	DY1163, DY1845	c.6371T>A	p.Val2124Glu	Missense	P	Likely Pathogenic								
	DY1485	c.6395T>G	p.Phe2132Cys	Missense	P	Likely Benig	Uncertain	Likely Pathoger	+		rs150154235	0.000325 (61/187642, GnomAD)		
	DY1684, DY1944	c.6497G>T	p.Arg2166Leu	Missense	P	Likely Pathogenic					rs754882717	0		
	DY1379	c.6499A>G	p.Asn2167Asp	Missense	P	Likely Pathogenic		+						

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DY1938	c.7694C>T	p.Ala2565Val	Missense	P	Likely Pathogenic				rs774163518	0			
DY1654	c.7829_7836delAGTAC	p.Glu2610fs	Frameshift Indel	P	Pathogenic								
DY713, DY1259, DY1337, DY1542, DY1608	c.7833C>G	p.Tyr2611*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogeni	rs138871063	0.000215 (27/125568, TOPMED)		
DY728 (in-cis)	c.5705G>A	p.Gly1902Glu	Missense	VUS	Uncertain Significance								
	c.7831T>C	p.Tyr2611His	Missense	P	Likely Pathogenic					rs1281912678	0.000014 (2/140226, GnomAD)		
DY2151 (in-cis)	c.7844T>G	p.Leu2615Arg	Missense	P	Likely Pathogenic				p.Leu2615Pro				
	c.6706T>C	p.Phe2236Leu	Missense	VUS	Likely Pathogenic/Uncertain Significance					rs764698239	0.000008 (2/264690, TOPMED)		
F4	c.7886_7887insC	p.Ala2630fs	Frameshift Indel	P	Pathogenic								
2_H1	c.7969A>C	p.Thr2657Pro	Missense	P	Likely Pathogenic								
DY2174	c.8017-2A>G	p.?	Splicing site	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogeni	rs1567180640	0		
DY1166	c.8203C>T	p.Gln2735*	Truncating	P	Pathogenic								
DY1831	c.8213_8214delIAG	p.Glu2738fs	Frameshift Indel	P	Pathogenic				Definitely Pathogenic				
DY1194	c.8291_8308delITCGC	p.Met2764_Leu276in-frame Indel		P	Likely Pathogenic								
DY1443	c.8299C>G	p.Arg2767Gly	Missense	P	Likely Pathogenic								
DY1530	c.8302G>A	p.Val2768Met	Missense	P	Likely Patho	Uncertain	Indeterminate	+		rs1456510041	0.000008 (2/246906, GnomAD)		
DY2065	c.8306_8310delTCAAC	p.Leu2769fs	Frameshift Indel	P	Pathogenic								
DY108, DY327, DY1909	c.8311G>A	p.Glu2771Lys	Missense	P	Pathogenic	Pathogenic	Highly Likely Pa	+	Likely Path	rs1057518897	0 (0/10680, ALFA Project)		
DY1652	c.8327T>C	p.Leu2776Pro	Missense	P	Likely Pathogenic								
DY1637	c.8359C>G	p.Arg2787Gly	Missense	P	Likely Pathogenic								
DY1494	c.8362_8371delTCGG	p.Ser2788fs	Frameshift Indel	P	Pathogenic							PMD: 23624871	c.8362_8363 ins34
DY42	c.8377C>G	p.Leu2793Val	Missense	P	Likely Pathogenic					rs752736484	0.000024 (3/125568, TOPMED)		
DY1668	c.8419T>C	p.Ser2807Pro	Missense	VUS	Uncertain Significance								
DY1294	c.8536A>C	p.Thr2846Pro	Missense	VUS	Uncertain Significance			+					
DY1207, DY2276	c.8537C>T	p.Thr2846Ile	Missense	P	Uncertain Significance	Likely Pathogen	+			rs1261349563	0.000008 (1/125568, TOPMED)		
DY751	c.8590G>T	p.Glu2864*	Truncating	P	Pathogenic	Pathogenic	+			rs374629549	0.000076 (20/264690, TOPMED)		
DY1096	c.8657delG	p.Gly2886fs	Frameshift Indel	P	Likely Pathogenic		Definitely Pathogenic						
DY1736, DY1782 (in-cis)	c.8606A>C	p.Glu2869Ala	Missense	VUS	Uncertain Significance					rs1370676628	0.000004 (1/233202, GnomAD)		
DY1544	c.8663G>A	p.Arg2888His	Missense	VUS	Likely Benig	Uncertain significance	(Last reviewed: Apr 3, 201	rs200168879	0.000192 (51/265390, GnomAD)				
DY1881, DY1883	c.8751_8761delIGGCC	p.Ala2918fs	Frameshift Indel	P	Pathogenic								
DY1496	c.8792-1G>C	p.?	Splicing site	P	Pathogenic		Definitely Pathogenic						
DY57	c.8932_8933delTTinsA	p.Phe2978Thr	Missense	P	Likely Pathogenic		Likely Pathogenic						
DY132	c.8984T>C	p.Leu2995Pro	Missense	VUS	Uncertain Significance					rs762182312	0.008568 (989/115428, ExAC)		
DY1542, DY1608	c.9033C>G	p.Tyr3011*	Truncating	P	Pathogenic								
DY1113	c.9074G>A	p.Trp3025*	Truncating	P	Pathogenic								
DY1380	c.9103_9105delIGAG	p.Glu3035del	In-frame Indel	P	Likely Pathogenic			+					
DY1521	c.9124_9130delIGTCTC	p.Val3042fs	Frameshift Indel	P	Pathogenic								
DY1720	c.9129C>A	p.Cys3043*	Truncating	P	Pathogenic								
DY1501, DY1908	c.9158C>A	p.Ala3053Asp	Missense	P	Likely Pathogenic								
DY1745	c.9173C>T	p.Pro3058Leu	Missense	P	Likely Pathogenic					rs765531129	0.0000261 (5/191614, GnomAD)		
DY1785	c.9201+4A>G	p.?	Splicing site	VUS	Uncertain Significance								
1_E1	c.9240delA	p.Cys3081fs	Frameshift Indel	P	Likely Pathogenic								
DY2263	c.9338G>A	p.Gly3113Glu	Missense	VUS	Uncertain Significance	LP							
DY1191	c.9388C>T	p.Arg3130Trp	Missense	P	Benign		Likely Pathogen	+		rs111244530	0.0000611 (11/180080, GnomAD)		
DY1684	c.9400A>G	p.Thr3134Ala	Missense	P	Likely Pathogenic					rs779771874	0.000004 (1/246260, GnomAD)		
DY804	c.9404C>T	p.Thr3135Met	Missense	P	Likely Patho	Conflicting	Likely Pathogen	+		rs1555449635	0		
DY1134	c.9507delG	p.Ile3170fs	Frameshift Indel	P	Likely Pathogenic								
DY1105	c.9541A>G	p.Lys3181Glu	Missense	P	Likely Pathogenic								
DY2144	c.9548G>A	p.Arg3183Gln	Missense	P	Likely Patho	Uncertain significance				rs79648977	0.000295 (78/264690, TOPMED)		
DY1151, DY1160, DY1231, DY1231, DY1986	c.9547C>T	p.Arg3183*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogeni	rs1485297878	0.000016 (2/125568, TOPMED)		

DY1274	c.9585G>T	p.Trp3195Cys	Missense	P	Likely Pathogenic												
DY1745	c.9644T>C	p.Val3215Ala	Missense	P	Likely Pathogenic												
DY1792	c.9746T>C	p.Leu3249Pro	Missense	VUS	Uncertain Significance												
DY840	c.9758T>C	p.Leu3253Pro	Missense	VUS	Uncertain Significance												
DY1851	c.9789G>A	p.Trp3263*	Truncating	P	Pathogenic			+									
DY1749, DY1913, DY1947	c.9827C>T	p.Thr3276Ile	Missense	VUS	Uncertain Significance												
DY1729	c.9827delC	p.Thr3276fs	Frameshift Indel	P	Pathogenic												
DY1182	c.9835C>T	p.Gln3279*	Truncating	P	Pathogenic												
DY1541, DY1733	c.9859_9861delCTC	p.Leu3287del	In-frame Indel	P	Pathogenic			Highly Likely Pathogenic	Likely Pathogenic								
DY2128, DY2186, DY2195	c.10086G>T	p.Gln3362His	Missense	VUS	Uncertain Significance						rs767064899	0.000053 (14/264690, TOPMED)					
DY728, DY1067, DY1091, DY1112, DY1167, DY1181, DY1270, DY1945	c.10102G>A	p.Asp3368Asn	Missense	P	Likely Benign	Likely Pathogen	+				rs762866943	0.000250(63/25188, GnomAD)					
DY2058	c.10139T>C	p.Phe3380Ser	Missense	VUS	Uncertain Significance LP												
DY1977	c.10151C>G	p.Ser3384*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+			rs1383556063	0.000008 (1/125568, TOPMED)					
DY1766 (in-cis)	c.10151C>G	p.Ser3384*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+			rs1383556063	0.000008 (1/125568, TOPMED)					
	c.9884A>G	p.Asn3295Ser	Missense	VUS	Likely Benign		Indeterminate	+			rs751447044	0.000048 (8/166260, GnomAD)					
DY2075	c.10168C>T	p.Gln3390*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+									
DY298, DY299, DY300	c.10188dup	p.Lys3397fs	Frameshift Indel	P	Likely Pathogenic												
KMUH_HD213, DY1927	c.10220+1G>A	p.?	Splicing site	P	Pathogenic								PMD: 30042192		c.10220+2T>G		
DY1952	c.10280dup	p.Ser3428fs	Frameshift Indel	P	Pathogenic			+									
DY1725	c.10306C>T	p.Gln3436*	Truncating	P	Pathogenic												
DY52, DY856, DY1094, DY1411, DY1441, DY1540	c.10315C>T	p.Arg3439Trp	Missense	VUS	Likely Benig	Uncertain significance(Las	+				rs374486955	0.000185 (51/275532, GnomAD)					
DY356 (in-cis)	c.10321C>T	p.Gln3441*	Truncating	P	Likely Pathogenic			+									
	c.5132C>T	p.Thr1711Ile	Missense	P	Uncertain Si	other(Last	Likely Pathogenic				rs750076336	0.000064 (8/125568, TOPMED)					
KMHK120	c.10406-2A>G	p.?	Splicing site	P	Pathogenic								PMD: 30230107		c.10406-1G>C		
DY1530	c.10407T>A	p.Asp3469Glu	Missense	VUS	Uncertain Si	Uncertain significance(Last reviewed: Dec 20, 2013	rs373139880	0.000005 (1/207844, GnomAD)									
DY2220	c.10441delG	p.Val3481fs	Frameshift Indel	P	Pathogenic	Pathogenic	Likely pathogenic										
DY780	c.10585G>A	p.Glu3529Lys	Missense	VUS	Uncertain Significance												
DY2061	c.10616C>T	p.Thr3539Ile	Missense	P	Likely Patho	p.Thr3539Ala, Conflicting interpretations of pathogenicity											
DY156, DY1173, DY1768	c.10710_10715delGGC	p.Ala3571_Val3571del	In-frame Indel	P	Likely Patho	Uncertain si	Highly Likely Pathogenic				rs777460677	0.000005 (1/202676, GnomAD)					
DY1991, DY2267	c.10710_10712delGGC	p.Ala3571del	In-frame Indel	P	Likely Pathogenic								PMD: 20642692		10710_10715del6		
DY1173	c.10711delG	p.Ala3571fs	Frameshift Indel	P	Likely Pathogenic								PMD: 22936402		non-PKD related		
DY1197	c.10764G>A	p.Trp3588*	Truncating	P	Pathogenic												
DY2031, DY2115	c.10838T>C	p.Leu3613Pro	Missense	VUS	Uncertain Significance LP			+									
DY1515	c.10951G>A	p.Gly3651Ser	Missense	P	Pathogenic	Pathogenic	Highly Likely Pe	+									
DY1204	c.11017-10C>A	p.?	Splicing site	P	Uncertain Si	Pathogenic	Highly Likely Pathogenic				rs555703777	0.000016 (2/125568, TOPMED)					
DY1290	c.11156+2T>C	p.?	Splicing site	P	Pathogenic												
KMUH1799	c.11172G>A	p.Trp3724*	Truncating	P	Pathogenic			+									
DY1147, DY1553, DY1653, DY1765	c.11249G>A	p.Arg3750Gln	Missense	P	Likely Patho	Conflicting interpretations of pathogenicity					rs1327414405	0.000004 (1/247254, GnomAD)					
DY2062	c.11256dup	p.Arg3753fs	Frameshift Indel	P	Pathogenic								PMD: 32457805		c.11257_11269+3del		
DY1470	c.11257C>T	p.Arg3753Trp	Missense	P	Likely Patho	Pathogenic	Highly Likely Pathogenic				rs1167476946	0.0000 (0/8988, ALFA Project)					
DY1703	c.11258G>A	p.Arg3753Gln	Missense	P	Likely Patho	Conflicting	Highly Likely Pe	+			rs1555446330	0					
DY1504, DY1728	c.11274_11275delCT	p.Tyr3759fs	Frameshift Indel	P	Pathogenic						rs1555446105	0					
DY1604	c.11399C>T	p.Pro3800Leu	Missense	VUS	Uncertain Significance												
DY1384	c.11411+1dup	p.?	Splicing site	P	Pathogenic								PMD: 29633482		c.11411+1_11411+5delGTG		
DY2148	c.11411+5G>A	p.?	Splicing site	VUS	Uncertain Significance												
DY1942, DY2052	c.11453G>A	p.Gly3818Asp	Missense	P	Likely Patho	Likely pathogenic		+			rs1555445740	0					
DY1584	c.11461C>T	p.Gln3821*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+			rs1325300747	0.000008 (1/125568, TOPMED)					
DY2143	c.11482G>T	p.Glu3828*	Truncating	P	Pathogenic			Definitely Pathc	+								
DY2123, DY2156	c.11510T>C	p.Leu3837Pro	Missense	P	Likely Pathogenic						p.Leu3837Arg, likely pathogenic						

DY56, DY124, DY710	c.11524_11535delGGG	p.Trp3841_Asn3844del	In-frame Indel	P	VUS	Uncertain Significance_P													
DY1140	c.11526G>T	p.Trp3842Cys	Missense	P	VUS	Likely Pathogenic													
DY1534	c.11566C>T	p.Arg3856Cys	Missense	P	VUS	Uncertain Significance						rs746316553	0.000008 (1/125568, TOPMED)						
DY1294	c.11603C>T	p.Thr3868Met	Missense	P	VUS	Uncertain Significance						rs1320643987	0						
DY2132	c.11632C>T	p.Arg3878Cys	Missense	P	VUS	Uncertain Significance LP						rs1207741763	C=0.000004 (1/264690, TOPMED)						
DY1257	c.11687_11694dup	p.Leu3899fs	Frameshift Indel	P	P	Pathogenic									PMID: 24611717			c.11694_11695insA	
DY1272	c.11713-1G>A	p.?	Splicing site	P	P	Pathogenic	Pathogenic	(Last reviewed: Mar 16, 2017)				rs867092741	0 (0/466, ALFA Project)						
DY2180	c.11725_11727delCTG	p.Leu3909del	In-frame Indel	P	P	Uncertain Significance_P													
DY2230	c.11728T>C	p.Phe3910Leu	Missense	P	VUS	Uncertain Significance_P						rs759162173	0.000060 (16/264690, TOPMED)						
DY1537	c.11758dup	p.Arg3920fs	Frameshift Indel	P	P	Likely Pathogenic													
DY2269	c.11774A>G	p.Glu3925Gly	Missense	P	VUS	Uncertain Significance_LP													
DY105, DY293	c.11782delIT	p.Trp3928fs	Frameshift Indel	P	P	Likely Pathogenic									PMID: 29529603			c.11772_11775dupGGG	
DY12	c.11808G>A	p.Trp3936*	Truncating	P	P	Likely Pathogenic		+											
DY1710	c.11876C>A	p.Ala3959Asp	Missense	P	VUS	Uncertain Significance													
DY2182	c.11884C>T	p.Gln3962*	Truncating	P	P	Pathogenic	Definitely Pathc+		Pathogenic										
DY1629	c.11927G>T	p.Ser3976Ile	Missense	P	P	Uncertain Significance	Likely Pathogenic												
DY1251, DY1838, DY2070, DY2112	c.11944C>T	p.Gln3982*	Truncating	P	P	Pathogenic	Pathogenic	Definitely Pathc+		Pathogenic		rs1161012209	0						
DY1373	c.11955dup	p.Ala3986fs	Frameshift Indel	P	P	Likely Pathogenic													
DY1889	c.12003G>C	p.Lys4001Asn	Missense	P	P	Likely Pathogenic						rs772496324	0.000009 (2/226208, GnomAD)						
DY1588	c.12003+1dup	p.?	Splicing site	P	P	Likely Pathogenic													
DY1903	c.12010C>T	p.Gln4004*	Truncating	P	P	Pathogenic	Pathogenic	Definitely Pathc+				rs766551411	0 (0/10680, ALFA Project)						
DY1808	c.12013C>T	p.Gln4005*	Truncating	P	P	Pathogenic		Definitely Pathc+				rs1567148587	0.00003 (1/31372, GnomAD)						
DY1171, DY1578	c.12013_12016delCAG	p.Gln4005fs	Frameshift Indel	P	P	Likely Pathogenic									PMID: 27499327			c.12011dupA	
DY1589	c.12123_12124dup	p.Gln4042fs	Frameshift Indel	P	P	Likely Pathogenic													
DY1278, DY1855	c.12124C>T	p.Gln4042*	Truncating	P	P	Pathogenic	Pathogenic	Definitely Pathc+		Pathogenic		rs199476094	0 (0/470, ALFA Project)						
DY1179	c.12138G>A	p.Leu4046=	Splicing site	P	P	Likely Pathogenic (splicing effect)						rs762641368	0.000016 (2/125568, TOPMED)						
DY1615	c.12139_12146delGCC	p.Leu4047fs	Frameshift Indel	P	P	Pathogenic													
DY2153, DY2198	c.12220_12221delCT	p.Leu4074fs	Frameshift Indel	P	P	Likely Pathogenic		Definitely P	Pathogenic	Pathogenic									
DY1535	c.12245delIT	p.Leu4082fs	Frameshift Indel	P	P	Pathogenic													
DY746	c.12286T>C	p.Trp4096Arg	Missense	P	VUS	Uncertain Significance		Indeterminate				rs1274235394	0.000008 (1/125568, TOPMED)						
DY39	c.12299_12300insTAG	p.Leu4100fs	Frameshift Indel	P	P	Pathogenic									PMID: 31844813			c.12301delC	
DY1108, DY1136, DY1425, DY1990, DY2023, DY2127, DY2252	c.12391_12393delGAG	p.Glu4131del	In-frame Indel	P	P	Uncertain Significance	Conflicting	Likely Pathogenic		Likely Pathoge	rs1555444468	0							
DY47	c.12395T>A	p.Leu4132*	Truncating	P	P	Pathogenic													
DY1878	c.12424G>T	p.Gly4142Cys	Missense	P	P	Likely Pathogenic						rs1406023463	0.000004 (1/245366, GnomAD)						
DY1388	c.12431G>A	p.Ser4144Asn	Missense	P	VUS	Uncertain Significance						rs765611740	0.00000 (0/14050, ALFA)						
DY2067	c.12442delG	p.Glu4148fs	Frameshift Indel	P	P	Pathogenic									PMID: 26139440; PMID: 2			c.12444G>	
DY2061, DY2146	c.12448C>T	p.Arg4150Cys	Missense	P	P	Likely Pathogenic	Pathogenic	Highly Likely P	+	Likely Pathoge	rs747127796	0.000004 (1/264690, TOPMED)							
DY120	c.12453_12454delCA	p.His4151fs	Frameshift Indel	P	P	Pathogenic													
DY733	c.12456delA	p.Val4153fs	Frameshift Indel	P	P	Pathogenic									PMID: 27499327			c.12458_12459delITC	
DY1477, DY1488, DY1888, DY2119	c.12460C>T	p.Arg4154Cys	Missense	P	VUS	Likely Pathogenic	Uncertain	Likely Pathoge	+	Likely Pathoge	rs115538130	0.00101 (270/268194, GnomAD)							
DY1177	c.12471delG	p.Met4158fs	Frameshift Indel	P	P	Pathogenic									PMID: 11773467			12470InsA	
DY1142, DY1415	c.12604_12631delGGC	p.Gly4202fs	Frameshift Indel	P	P	Pathogenic	Pathogenic	Definitely Pathogenic		Pathogenic									
DY1612	c.12608_12635delGGC	p.Arg4203fs	Frameshift Indel	P	P	Pathogenic		Definitely Pathogenic											
DY1835	c.12697A>C	p.Thr4233Pro	Missense	P	VUS	Uncertain Significance	Uncertain significance	(Last reviewed: May 15, 2019)		rs1230641065	0.000004 (1/264690, TOPMED)								
DY26	c.12721C>T	p.Gln4241*	Truncating	P	P	Pathogenic		Definitely Pathc+											
DY1349	c.12728T>A	p.Leu4243Gln	Missense	P	VUS	Uncertain Significance													
DY853, DY1188	c.12751_12823delAGC	p.Ser4251fs	Frameshift Indel	P	P	Pathogenic													
2_H1	c.12818G>C	p.Arg4273Pro	Missense	P	VUS	Uncertain Significance													

[illegible]

	DY1089	c.2159dupA	p.Asn720fs	Frameshift indel	P	Pathogenic		Definitely Pathogenic										
	DY1424	c.2276T>C	p.Phe759Ser	Missense	VUS	Uncertain Significance												
	DY1546, DY1793	c.2320G>T	p.Glu774*	Truncating	P	Pathogenic												
	DY1966	c.2380_2392delAGTT	p.Ser794fs	Frameshift indel	P	Pathogenic												
	R803* Table	c.2407C>T	p.Arg803*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogeni	rs778235410	0.0000119	(3/251064, GnomAD)					
	DY1208, DY1242, <u>DY1472</u> , DY1942, DY2080	c.2522+1G>C	p.?	Splicing site	P	Pathogenic	Pathogenic											
	DY1198	c.2544delG	p.Met849fs	Frameshift indel	P	Pathogenic					rs761705522	0.000004	(1/251186, GnomAD_exome)					
	DY1608	c.2596A>G	p.Lys866Glu	Missense	VUS	Uncertain Significance					rs1480227336	0.000004	(1/251138, GnomAD_exome)					
	DY336	c.2614C>T	p.Arg872*	Truncating	P	Pathogenic	Pathogenic	Definitely Pathc	+	Pathogeni	rs755226061	0.000016, 0.00000398	(2/125568, TOPMED; 1/251096, GnomAD)					
	<u>2_E1</u> , DY106, DY130, DY335, DY359, LSL	c.2629A>G	p.Arg877Gly	Missense	VUS	Uncertain Significance												
	DY1110, DY1158, DY1237, DY1662	c.2671-2A>G	p.?	Splicing site	P	Pathogenic		Definitely Pathogenic										
	KMUH2086	c.2726T>C	p.Val909Ala	Missense	VUS	Uncertain Significance												
PKHD1																		
	DY1068	c.218G>A	p.Arg73Gln	Missense	VUS	Uncertain Significance					rs577541526	0.000040	(10/251358, GnomAD_exome)					
		c.3953A>T	p.His1318Leu	Missense	VUS	Uncertain Si	Uncertain significance				rs200733734	0.000108	(27/250190, GnomAD_exome)					
	DY1461	c.2507T>C	p.Val836Ala	Missense	P	Likely Patho	Pathogenic/Likely pathoge		+		rs199568593	0.000080	(20/251222, GnomAD_exome)					
		c.3313delT	p.Ser1105fs	Frameshift indel	P	Pathogenic	Pathogenic/Likely pathogenic				rs1057516922	None						
	DY1346	c.2795G>A	p.Cys932Tyr	Missense	VUS	Uncertain Si	Uncertain significance				rs1329209110	0.000008	(1/125568, TOPMED)					
		c.3463C>T	p.Gln1155*	Truncating	P	Pathogenic	Pathogenic		+		rs1554200780	0.0	(0/10680, ALFA Project)					
	DY1398	c.4197_4199delTTCins	p.Ser1400Arg	Missense	P	Likely Pathogenic												
		c.4202delC	p.Ala1401fs	Frameshift indel	P	Likely Pathogenic												
	DY1528	c.8432T>C	p.Leu2811Pro	Missense	P	Likely Pathogenic					rs779827172	0.000024	(6/250940, GnomAD_exome)					
		c.9368C>T	p.Ala3123Val	Missense	VUS	Uncertain Significance					rs779827172	0.000024	(6/250940, GnomAD_exome)					
	DY1550	c.227C>A	p.Pro76His	Missense	P	Likely Pathogenic					rs753364440	0.000056	(7/125568, TOPMED)					
	DY1209	c.439G>A	p.Gly147Ser	Missense	P	Likely Pathogenic					rs1442794854	0.000004	(1/251188, GnomAD_exome)					
	DY1220	c.448+1G>A	p.?	Splicing	P	Pathogenic												
	DY1360	c.704delG	p.Gly235fs	Frameshift indel	P	Pathogenic												
	DY1665, DY2047	c.778+1G>C	p.?	Splicing	P	Pathogenic	Likely pathogenic				rs753471298	0.000004	(1/251432, GnomAD_exome)					
	DY1366	c.1046G>A	p.Gly349Glu	Missense	P	Likely Pathogenic					rs746153656	0.000008	(2/251292, G found in cancer)					
	DY2184	c.1124G>A	p.Arg375Gln	Missense	P	Likely Pathogenic				p.Arg375Tp	Arg375Trs767232748	0.000004	(1/264690, TOPMED)		PMID: 27415407		p.Arg375Trp	
	DY1678	c.1487G>A	p.Arg496Gln	Missense	VUS	Uncertain Significance					rs181391485	0.000076	(19/251216, GnomAD_exome)					
	DY1368	c.1766_1786delGTCAC	p.Arg589_Val595del	In frame indel	VUS	Uncertain Significance					rs745731220	0.000020	(5/251098, GnomAD_exome)					
	DY1432, DY1660, DY2126	c.1849T>C	p.Tyr617His	Missense	VUS	Uncertain Si	Conflicting interpretations of pathogenicity			Likel	rs141177165	0.000299	(75/251252, GnomAD_exome)					
	DY1306	c.2201T>C	p.Val734Ala	Missense	VUS	Uncertain Significance												
	DY1774	c.2291C>T	p.Thr764Ile	Missense	VUS	Uncertain Si	Uncertain significance				rs190315828	0.000095	(24/251372, GnomAD_exome)					
	DY1788	c.2341C>G	p.Arg781Gly	Missense	P	Uncertain Si	Pathogenic/Likely pathogenic				rs398124478	0.000040	(10/251408, GnomAD_exome)					
	DY2237	c.2530T>C	p.Tyr844His	Missense	VUS	VUS_LP												
	DY1299, DY1384	c.2507T>C	p.Val836Ala	Missense	P	Likely Patho	Pathogenic/Likely pathoge		+		rs199568593	0.000080	(20/251222, GnomAD_exome)					
	DY2260	c.3299T>C	p.Phe1100Ser	Missense	P	Likely Pathogenic												
	DY1337, DY1795, DY1813	c.3953A>T	p.His1318Leu	Missense	VUS	Uncertain Si	Uncertain significance				rs200733734	0.000108	(27/250190, GnomAD_exome)					
	DY1831	c.4039_4091delGTGAC	p.Val1347fs	Frameshift indel	P	Pathogenic												
	DY2038	c.6091delG	p.Ala2031fs	Frameshift indel	P	Pathogenic	Pathogenic/Likely pathogenic				rs1057516804	0						
	DY1599	c.7802A>G	p.Asn2601Ser	Missense	VUS	Uncertain Significance					rs1423175594	0.000004	(1/251400, GnomAD_exome)					
	DY1214	c.8870T>C	p.Ile2957Thr	Missense	P	Pathogenic	Pathogenic			Likely Pat	rs760222236	0.000052	(13/251136, GnomAD_exome)					
	DY1177	c.9282C>A	p.Asn3094Lys	Missense	VUS	Uncertain Significance												
	<u>DY1116</u> , <u>DY1114</u>	c.9472A>G	p.Met3158Val	Missense	VUS	Uncertain Significance					rs764138084	0.000004	(1/250882, GnomAD_exome)					
	DY1385	c.9629C>G	p.Ser3210Cys	Missense	VUS	Uncertain Si	Conflicting interpretations of pathogenicity		+	VUS	rs141081295	0.000104	(13/125568, TOPMED)					
	DY1397	c.10058T>G	p.Leu3353Arg	Missense	VUS	Uncertain Si	Conflicting interpretations of pathogenicity			Likel	rs777377414	0.000012	(3/251236, GnomAD_exome)					
	DY1390	c.10284T>G	p.Tyr3428*	Truncating	P	Pathogenic												
	DY1785	c.11741G>A	p.Arg3914Gln	Missense	VUS	Uncertain Significance					rs776601687	0.000008	(2/251220, GnomAD_exome)					
	DY1396	c.11882G>A	p.Arg3961Gln	Missense	VUS	Uncertain Significance					rs749579192	0.000056	(7/125568, TOPMED)					

GANAB														
	DY1591	c.11_16delTAGCGG	p.Val4_Ala5del	In-frame Indel	P	Likely Patho	Pathogenic				rs750723025	0.000036 (10/280422)		
	DY716	c.193G>T	p.Asp65Tyr	Missense	VUS	Uncertain Significance								
	DY1299	c.252+2T>C	p.?	Splicing site	P	Pathogenic								
	DY2272	c.252+8_252+9delGAirp	p.?	Splicing site	VUS	Uncertain Significance								
	DY1732	c.1389G>A	p.Trp463*	Truncating	P	Pathogenic								
	DY2160	c.1803+1G>T	p.?	Splicing site	P	Pathogenic								
	DY1919	c.1900+1G>C	p.?	Splicing site	P	Pathogenic								
	DY778	c.2116C>T	p.Arg706*	Truncating	P	Pathogenic								
	DY2043	c.2504G>A	p.Trp835*	Truncating	P	Pathogenic								
ALG8														
	DY858	c.164_165delGGinsTC	p.Trp55Phe	In-frame Indel	VUS	Uncertain Significance								
	DY2242	c.175-2A>G	p.?	Splicing site	P	Pathogenic				Pathogenic				
	DY158	c.396delA	p.Val133fs	Frameshift Indel	P	Pathogenic								
	DY1591, DY1826	c.460G>A	p.Gly154Arg	Missense	VUS	Uncertain Significance					rs201359142	0.000016 (4/251222)		
	DY1159, DY1499, DY1513, DY1538	c.482T>C	p.Ile161Thr	Missense	VUS	Uncertain Significance					rs373493408	0.000100 (27/282280)		
	DY2275	c.599A>G	p.His200Arg	Missense	VUS	Uncertain Significance_LP					rs776497184	C=0.000008 (2/264690, TOPMED)		
	DY1787, DY1854, DY1920, DY2090	c.824delG	p.Gly275fs	Frameshift Indel	P	Pathogenic					rs745894763	0.000004 (3/282444)		
	DY704, DY1761	c.1169T>G	p.Leu390Arg	Missense	VUS	Uncertain Significance								
	DY855	c.1241A>G	p.His414Arg	Missense	VUS	Uncertain Significance					rs1013838215	0.000008 (1/125568)		

ACMG: American College of Medical Genetics and Genomics, ALFA: Allele Frequency Aggregator, gnomAD: The Genome Aggregation Database, HGMD: Human Gene Mutation Database, LOVD: Leiden Open Variation Database, LP: likely pathogenic, P: pathogenic, TOPMed: The Trans-Omics for Precision Medicine, VUS: variant of unknown significance. Underscore indicated more than one variant was identified in the index case.

Supplementary Table 3. Summary of *PKD2* p.R803* Identified in Database and Previous Publication

	Database	p.R803* Number	Cohort Number	Allele Frequency	Note
	TOPMed	1	125568	0.0001635	East Asian
	GnomAD	3	251064	0.00001195	
	Mayo PKDB	7	2494	0.0028	
Publication Year	Country	p.R803*N umber	Cohort Number	Percentage in cohort (%)	Reference
Asian					
2005	Taiwan	1	20	5	1
2005	China	1	24	4.2	2
2006	Korea	4	91	4.3	3
2013	Taiwan	3	46	6.5	4
2014	Korea	1	20	5	5
2019	Korea (HOPE-P KD)	4	524	0.76	6
Non-Asian					
2001	Cyprus	1	-	-	7
2012	France	2	700	0.29	8
2016	Canada (TGESP)	1	220	0.46	9
2017	France (Genkyst)	6	293 (PKD2 only)	2.05	10
2018	Spain	1	101	0.99	11
2019	Canada (eTGESP)	7	612	1.1	12

TGESP: Toronto Genetic Epidemiology Study of PKD, HOPE-PKD: The coHort for genotype-PhenotypE correlation in ADPKD.

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Supplementary Table 4. Annual renal function decline in patients with *PKD2* p.Arg803* and non-p.Arg803* truncation

	Non-p.Arg803*			p.Arg803*		
	Coefficient	Standard error	<i>p</i> -value	Coefficient	Standard error	<i>P</i> -value
Intercept	72.91	6.62	<.0001	89.95	3.94	<.0001
Follow-up year	-2.67	0.77	0.0019	-2.09	0.39	<.0001
Follow-up year ²	0.10	0.09	0.26	-0.06	0.01	<.0001
Follow-up year ³	-0.01	0.003	0.002	-	-	
Follow-up year ⁴	-	-		-	-	

The estimated glomerular filtration rate (eGFR) was calculated using the CKD-EPI equation. The model was developed by mixed model incorporating random intercept and slope for longitudinal eGFR change. A *p*-value less than 0.05 was considered statistical significance.

Supplementary Table 5. Primers Used in This Study. Including primer sequences used for long-range PCR for PKD1 pseudogene regions, gene panel, and PKD2 microsatellite markers.

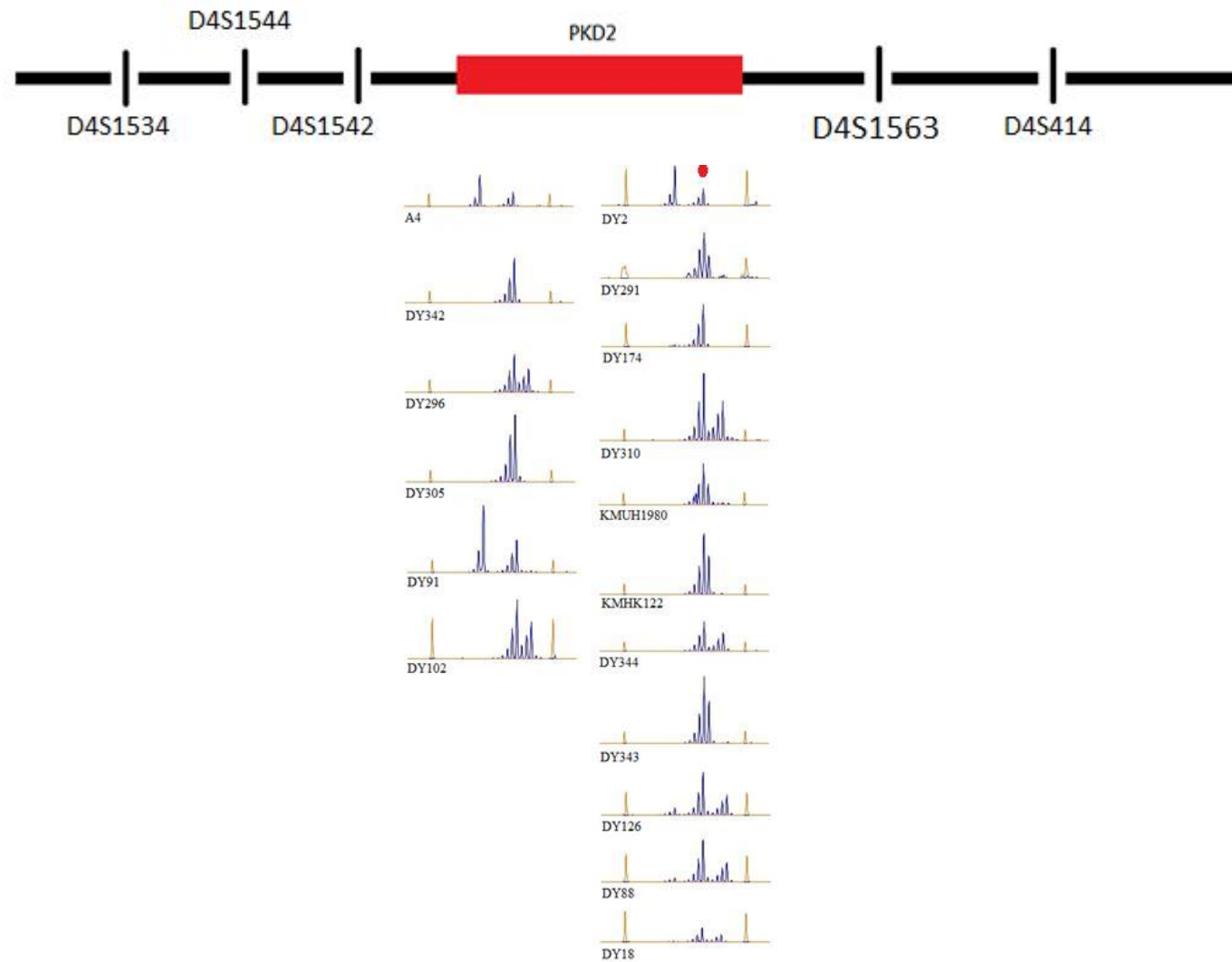
	Primer Name	Sequence	Length (bp)
<i>PKD1</i> Long_Range Primer	PKD1_LA_Ex	CGGGGCCCCGCACTGCAGCGCCAG	587
	PKD1_LA_Ex	AGGCGTTCTTATTTAGCAGGGCCGCC	
	PKD1_LA_Ex	CCCCGAGTAGCTGGAACACAGTTAC/	4041
	PKD1_LA_Ex	CGTCCTGCTGTGCCAGAGGCG	
	PKD1_LA_Ex	ACGCTCTGCGAGCTGCAGCCC	3893
	PKD1_LA_Ex	CTGCAGGGACAGGCGTCAGTGA	
	PKD1_LA_Ex	TGGAGGGAGGGACGCCAATC	4391
	PKD1_LA_Ex	GTC AACGTGGGCCTCCAAGT	
	PKD1_LA_Ex	ATCCCTGGGGGTCTACCATCTCTTA	5253
	PKD1_LA_Ex	ACACAGGACAGAACGGCTGAGGCTA	
	PKD1_LA_Ex	ATGCTTAGTGAGGAGGCTGTGGGGGTC	3276
	PKD1_LA_Ex	GCTTAAAGGGGAATGGCTTAAACCCG	
	PKD1_LA_Ex	CGGGTCACCGGTTGTGGCA	3916
	PKD1_LA_Ex	ATGAGGCTCTTTCCACAGACAACAGAGTT	
	Primer Name	Sequence	Sequence
<i>PKD1</i>	Exon1.1	CTGAGCTGCGGCCTCC	GGCCCCGCTACTCACAGC
	Exon1.2	TGCCGCGTCAACTGCTC	AGGCGTTCTTATTTAGCAGGG
	Exon2	GGATTCGGCAAAGCTGATG	CTGGCAATGTGTGGGATG
	Exon3	GAAGGATATTGGGGGCCTG	CTGGCAGAGCTGTGAGTGTC
	Exon4	CTGGGAAGGACAGAGCTGG	CTGGCATAGACCCCTCCAC
	Exon5.1	GTACCAGGCTCTGCCCCATC	CTGCCTGGGCGCTATCAC
	Exon5.2	CTCCACCTGCACGCTGTGTC	CCTCCTCCAGCACGCTTTC
	Exon5.3	CCACCAGGGTGGCCC	CCTCCCTGACAACAGCTCAG
	Exon5.4	CCGGTGGAGAAGCAGAAGG	GAGCCAGGAGGAGCAGAAC
	Exon6	TTCAGAGATCTCCCAACCTATGG	CACACTGACCGTTGACACCC
	Exon7	GTCTCAGGCCCTGCC	TGCCCTGAGCCCCCTG
	Exon8	TTTTTGGCGAGACCCACAG	CCAGTCTGTTCTCCTGGTG
	Exon9	ACGTGAAAGCTCAGAGAGGC	CTCTTCTGGGAAGTTCGGG
	Exon10.1	GAGATGCAGGGAACAGACCC	ACAGGGACCCCGGAGAAC
	Exon10.2	CGGGAACGGAGAAGAGGAAC	GTTGGGCATCTCTGACGGTG
	Exon11.1	TGGTGGTGGAGCCTCGG	GCCCGCTTTGAGAATGTCTG
	Exon11.2	GGTGGCCACCAGGGCAG	AGGCCCAACCCTGGACTG
	Exon11.3	GGACCTCATAGCGCCCAG	GGATGTCTCATGCTCCCTG
	Exon11.4	AGCCATGACGAGGCGTTG	CTCCTTGAGGTGTGGCTGAC
	Exon12	AAGCAGAGCAGAAGGCAGAG	CAGGAGGCGACAGGCTAAG
	Exon13	CTGGTGCCCAACCCAAAC	GAGGGAGGGACGCCAATC
	Exon14	CTCAGAGCCTGAAAGGCAGTG	CCTGTCCCGGTTCACTCAC
	Exon15.1	CGTCAGGTTCTCGAAGGC	CTCTGCTGCCCCGAGTG
	Exon15.2	CGTGTGTTGACCTCCAGGC	GTACCTCCTGACCGTGCTG
	Exon15.3	GCTGGCCGCACCCAC	CCTCGAGGGGCACCTACC
	Exon15.4	GGACAGCACCAGCGCC	GGCACAGAAGTGCACAGTG
	Exon15.5	TGACTGTCTACAAGATAGGAGCC	CGGCACGTTCCCCCTG
	Exon15.6	GGCCACCCTAACGGTGAAG	CGTTCATCTACCGAGACCCAG
	Exon15.7	CGATGATATTGAAGGTGCC	CACGCTTACAACAGCACAGG
	Exon15.8	GTAGGTGCCGGCCTCG	CTCTTACACCTTCCGCTCCG
	Exon15.9	CAAGTGCAGGCCGGGTG	CTTCTCGCTACCGTGCTC
	Exon15.1	AGAAGGTGCCAGCATCC	CACCCATAGCTTCCCCAC
	Exon15.11	TGGTCTCCGACGCGGG	TCATGTCACCATGGTCTTCCC
	Exon15.12	TGTAGGTGACGTGCGGG	CCCGTTTCTCCACAGCTTC
	Exon15.13	CATCTGTGTCTGCCCTGG	CGACTCGCTGGTCATCCTG

	Exon15.14	CAGTCTGGTAGGTGACGCAG	GGCCTACCACTGGGACTTTG
	Exon15.15	ACCCTCAATGATGGGCACC	GCAACTACTTGGAGGCCAC
	Exon15.16	TCTCTGGGCTCATGGGTG	CCACTGACACAGAGCATCCAG
	Exon16	TAGATGACCAGGGAGGCTGG	AAACTGGATGGGGCTCTCAG
	Exon17	CCTGTCAGCCCCACTTCTG	TCCAGCAGGCCAAATAGACC
	Exon18.1	TCACAGAGTCGGGGGATC	ATGAGACCACCACATCCAG
	Exon18.2	CGTGAGCGTGAAGGTGTATC	GAGGGGTGAGTGTGAGCG
	Exon19	GAGCAGGTGGCAGTCTCG	CTCCCGTGA TGCCGTGG
	Exon20	GTGAGCAGGTGGCAGTCTC	GTGATGCCGTGGGGACC
	Exon21	CAGGGGTACAGGTCTTGGTC	CGAGACTGCCACCTGCTC
	Exon22	GGGTGGCATGGGGCAC	GTGGGCTCTCCAGTGCAG
	Exon23.1	CCTACGAGAAACGCCTTCCC	GCGCCCAGATCCCCATC
	Exon23.2	CGAGTTGTTGGGCACCTTC	CGGAGCCTGCTGTGCTATG
	Exon23.3	GGGGATGGAGAAGTGCAG	CACTGACCTCACGCATGTCTG
	Exon24	GCAATGCTGACCCATGATGC	GGGTGGTTGAGCTTCCCG
	Exon25.1	CTCCCAGGAGCACAGGGTC	GAGACCCAGCGGGGAGTTAC
	Exon25.2	CACAAAGCGGACATGGCTTG	CACGTCCGCTACCCCTTC
	Exon26	CACAGCCAGTGAGAGCAGG	AGCTTGCACGTGACCTCC
	Exon27	CAGAGGGGCAGAGCTTGG	AGATGACTTGCCCTGGGATGC
	Exon28	CCACGGAGTGGGAACATGG	GTGTGGCAGCACAACAAAGG
	Exon29	GAAGGGCTGGGCAGGAAG	CCTCCTGCAGTGCGGC
	Exon30	GCTCCATTCCCAGTACTCCC	GTTGGCGACTCTGCCTACAG
	Exon31	TCCAACAAAGGCCTGCTGAG	GGTGGCTTCTCATCCCTGTC
	Exon32	TGACCACATGGAGCCACAG	AGGTGAGGACTCTACTGGGG
	Exon33	TACAGGAGGCATAGGGTGGG	CTGGTACCTTGTGACCCG
	Exon34	TTCAGAGAAGTGAAGTGGTGC	GGCTCTGAAGCTACCCCTTG
	Exon35	CTGGGGAAGCAGAGACAGAC	GGTTAACATGGGCTTGGCTG
	Exon36	GTGATGGAGGCCTGTAGCC	GAGCTGCCTCTCACAGGTC
	Exon37	CAAGAGACGGAGGTGGCAG	GCTACAGGCCTCCATCACG
	Exon38	CAAAGCCCTGCTGTCACTG	ACATGTCCCCTAGGGTCTGG
	Exon39	GTTCCCTGGGTCTCTGGTG	GCTGCTCTCTCAACAAGAGG
	Exon40	CACTCCTGGAGAACTACTCCCTTG	AAACACTCCTGTTGGGTTTTG
	Exon41	CTGGCTGGTGA CTGCGG	CGCCAAGGACAAGGGAGTAG
	Exon42	GGTCTGGCCGGGGAC	CTTCCACAGCCCCTCAGC
	Exon43.1	GTCTTGCCAAAGACGGACC	CCACGGCACTGGTACGC
	Exon43.2	CACGGGCTGCGGAGC	CCCTCCCGCCCTCCTG
	Exon44	TGAGCTGAGCTAAGACGCC	CTTCTGCTTTTGGTCAAGG
	Exon45.1	GGTACAGCTCTCCACGCAAG	GGGAGGGCGTCTTAGCTC
	Exon45.2	CACAGGGGCTCAGTCAGTC	TGTCACCCCTGCTGTGTG
	Exon46.1	TGTCCACTCCGACTCCACG	CCTCCAAGCCGTGTTCGA
	Exon46.2	GTCCGTCAAAC TGGGTGAG	GAGGGACACGCCCTGG
PKD2	Exon1.1	TCCTGAGGCGCACAGC	AGCGGAGGAGAAGGGGAG
	Exon1.2	GGCCTGGAGATCGAGATGC	CGCCGTGGTAGCCCC
	Exon1.3	GCAGCCGGAGGTCGG	CCGGCCGTCTCTGGTTC
	Exon2	TGATTTGGATAGGTCAAATCTTTTC	TTTATTTTCCCTTTTGCCATTG
	Exon3	AGGTCCTGTCGATACTCATGC	CTGCTGGTATGTGAATGTGTGC
	Exon4.1	TCATCTCTCAAGTCCTGGGG	GTTATGCAAACGATGCAGGC
	Exon4.2	TGAATGGTGGGAGTTCAGAG	CAACCGAAGTTTCATCTTCTATG
	Exon5.1	CCTCAAGTGTCCACTGATTG	AAAAGTTGCCCTGGTTCCTC
	Exon5.2	GGTAGTAGCCACTGGGGAATC	CTGGCTAACCCCAAGATAGG
	Exon6.1	GCTGTATTTCATGTGTTGTTTG	GCGAATTTCCAATATCTCTTCC
	Exon6.2	CCTTTAAAGCTGATCCGATATG	TGCTGAGGAGATCAAAGACTC
	Exon7.1	GAGCCCTTATAATTAATACATTGGTG	ACTGTGACAGCAGCTATATTG
	Exon7.2	GCTTTTCAGCTGTCA GTGG	AGCTTTGGCTGGTCAC TTG
	Exon8.1	AATTTCTTTTCAGGATGAAATAATG	GTGCCAAAGACAAGGTATGC
	Exon8.2	TAACAGGACCATGAGCCAGC	CTTGAGAAGCAGTGACA ACTCTG
	Exon9	GTTGCATCAACTAGTGGACATTG	GTGTGAGAGAAAAGAGAAGACAA
	Exon10	TCTTCCTTTAATTTTGCCCTCC	AATCTGGGTGAAACAATGCTC
	Exon11	GAGCCCTTGGGCTAGAAATAC	GAAAGTAAACAGATGCAAAAAGG
	Exon12	TGTTTGATACATCTGTGGTGTG	CTGTGTTGAGG GTGAACTGG
	Exon13	CTGCCTGGTCTCATGTGGAC	TCCTTGGTGAGGCTTCTGTG

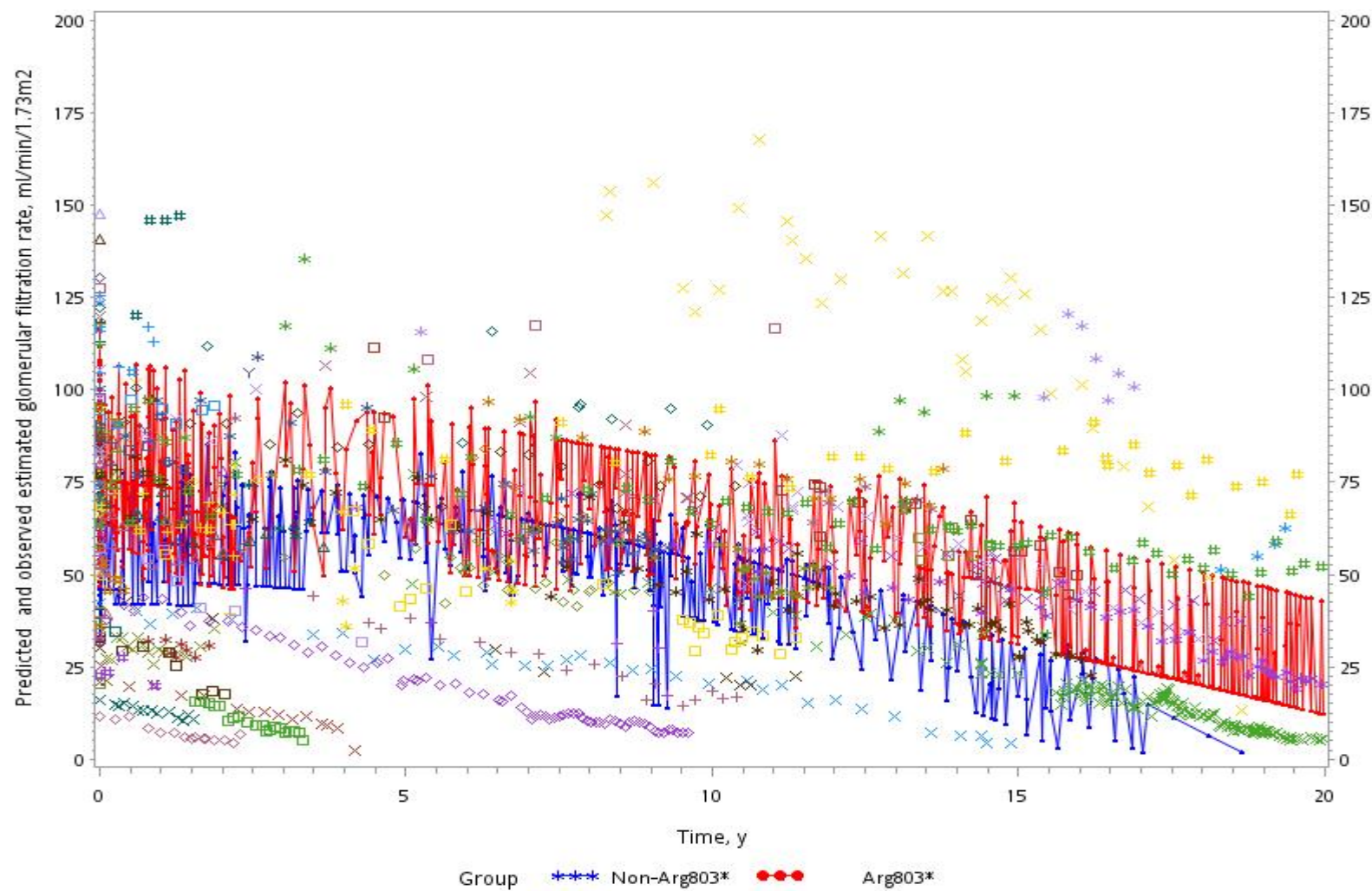
	Exon14	ACTTCAAATACAACGTGTCAGCAAC	TGGGGCTGAAAAGACAATG
	Exon15.1	GACTTCCTAAGGCATTTCTTCTAC	AGCTTCTGGGGCGAGG
	Exon15.2	GTGAAGAGTTGGAACGCTGG	CAGGACAGCCACTTCTCTCAC
PKHD1			
	Exon2	AACACCTGCTCTGAAAACAGT	AGCTATGCTGCTCCAATCAA
	Exon3	TCAGGCCCACTTTTACACCT	TCTGGTGATTCTGAGGCAGG
	Exon4	CAGGATCTTAGCACAGTTTCACAC	ATCCCTCATCCTGTCTGGTCT
	Exon5	TAACCCGGTCAAGCAGACAC	GGGCTGCTAGCTTTGGGAAT
	Exon6	AATTTTCCCACTCATAAAAACCACT	TTACCTTCAAGTAATGCTGTCTGA
	Exon7	GACCTGCTTTGCCATATTGAG	TTTATTGCCATCAGGGAAGC
	Exon8	TGGTGAGTGGGGAGAGAAAGA	CCAGAGGAGAGTGTCATAGCC
	Exon9	TCCTCATGAGAACCAATCTTGC	TCTCTGTCTACCATTTTCTCTAGC
	Exon10	AGGACACAACCTTCATTCACCCA	CCCAGAAAGACTCGTGCAAT
	Exon11	AGCTTCGGGTGTTAATGGTC	TTCCCAATCTTCCTTTCAGGCA
	Exon12	TTGCTGGACTTAGAAAAGTTAAAGTAAG	TGCCATACAGACATATAATCTCCT
	Exon13	ATCTCCCGACTGCctctctc	ATTGAACAGCCCTGGTGG
	Exon14	CCAATTTGGGAAGGTGCG	CCTTCCTTATCTGTCTCCTAGCC
	Exon15	CATGGGTAATGGGACTGGCAA	AGACAAAGTACAAGGGCAGTCA
	Exon16_1	GAGATGCCTGGAAGCTGCAT	AGGATTCAAGCCAGGTGTTG
	Exon16_2	AAGTTGGAGCTGTTGGGTGG	CAGTGCCTCCTGCTACATGGG
	Exon17	TGAGGAGGAATGTCCTGTGTT	AACTCCTCAATGTTGTTTGAATC
	Exon18	CTAGTCAGCCAAATCCCTGCAA	ACAATCAGAATGAAGCCACGG
	Exon19	GCACACATGTAGGGAAGCTCT	TACCTACCCACCTGACCCAG
	Exon20	TGTTTGAATGACTGCTTCTTGC	CCTGAGGTGGGTAAGTGTCC
	Exon21	CTCCCTGAGAGCCCAAGTCT	ATGTGACCGCTGTGGAG
	Exon22	TCCACACAGCAAGTCTACCA	AGTCACACCTGTGTCCCTCA
	Exon23	CCTAGGTGCACACCCCAAC	ACCTCCCAGGATGTTGTTCC
	Exon24	GAGGATGAAACTCTGTAAGGTGGA	GCAGCAAATCCATGCCACTA
	Exon25	TTAGACGAGATTAGATTTTCGGTTC	tcocatgttacaatcagtgagg
	Exon26	CATCACCAAGCTACATGGCCT	GTACGCTTGGGAGCACTTCA
	Exon27_1	GGCAGAGAAGGAACATTTGATG	ACCAGAGGGTCTCACCAACA
	Exon27_2	CCGGAACGTGTTTCTCTGGT	TGTGAGACCCTCCCCAGATT
	Exon28_1	CCTGCAGTTTGTAGCATGCCC	CCACTTACCCTGGGTGGAAC
	Exon28_1	ACCATCCGAGGCTCTAGTTTG	TGGGGTCAGTTTCCCACTTC
	Exon29	TTCTCTTCCCTTAAGTCAGTCCTAC	TTTCAGAGGACATTGATTGCC
	Exon30	CCCCACATGTCAGAGGCTAT	AATGCAAGTGGTCACCTCAC
	Exon31	CAGACAGTTTGCTCTTCTCCTC	CCCTCTCTCTGACCTCACTGG
	Exon32_1	GGACTTCCACAGGTGCTATGAA	GGCGAAGAACCTGTTGCCAG
	Exon32_2	CACTGTTCCAGCTGCCGT	GTTGCCCTGGAAGGACTGTG
	Exon32_3	TCCTTCTGGGGAACCTGAAC	AAGAAGCAACCCCTCACAG
	Exon32_4	CATCTTCCCATCGCAGGGTT	TGCAATTCCCCTGACACTCG
	Exon32_5	CTCCCTGAACGTACAGTCC	GCCAAGTCTCTGTCTGGCA
	Exon32_6	AATGCAAGCCACGTTGTGT	TTACCCGTCAGGCAGGTCT
	Exon32_7	GGTGGAAGCCTCTTGACCAT	GAGATCTGGGCCACTGCAAA
	Exon32_8	TAACCTTTGCAAGTGGCCAG	CCAGAAAGTGAAGGAGCTACCA
	Exon33	GAGTTGAGTAAAGAAGGGAGATTG	AGAATTAACCAAAGAATATCATTTC
	Exon34_1	GGCCTTAGGTTCTCTGTGG	AGAGGCCCGAGAATGATGGA
	Exon34_2	CTGAAGCGTGAGGAGGACAG	GGTCCATTGGCCAAAGCATTT
	Exon35	ACACCCCATTTAACTCCCC	ATCGCTGCCATTTGGACTAA
	Exon36	gaagcaacagaccagccaac	AAAGTTTCCCTCCTCCATCC
	Exon37	CCCAGGGACTGACAAATTTTC	TGCTAGACACAGCTCTACTTCATT
	Exon38	TGGACAACTTTCTCTCGGGC	AACTTAGCCAAGCTGACACTCA
	Exon39	TTGGAGTGATGTCCTCAGTTCT	CAACCACAGCAATGCCATCT
	Exon40_1	TGGGAGATTTGTCTGGAAGCA	ACCTCTCATAGTCCCACCA
	Exon40_2	TCAGGTAATCTGCAGCACTGT	TCCACCATGCATGTAGTGCC
	Exon41	GGTCAACAGAATCTCAGGAGC	TTTGGGGAGAATTCAATTGTG
	Exon42	ACATTTTGCCATCAGGCTTGT	AGATCTGTATCTCTTTCCTGATT
	Exon43	TGCCCTCTCAGTTCTGGTCT	TGCATTATGTGAACTCTGATGTG
	Exon44	AAACAAGAGTCTTCTTTCCAGC	AACAAATGCCAAAGTGCTC
	Exon45	TGGCTTAATTTTCAATCTGTTTCACT	CCCCTCAAGGGCAAGTCAAT
	Exon46	AAAAGCTACAGGAAGTCCGC	GCCCAGCACATGTAATTTTG

	Exon47	TGAATTGTATTTTCATGTGTCCAG	TGATTCACAAAGTATTTGCCACTA
	Exon48_1	AGTTGCTATTTGTGCCATTGTG	AAGAACACCTCCTCCACAGG
	Exon48_2	TGTGAAGACCAGCCAGTTGA	ATGCCCATCGGCAAGCTAAA
	Exon49	TGAGCAAAATATCTCTCAACCCA	TGAGAATGCAGCATAACCACT
	Exon50_1	GGTGGAAATGATGGGGTTCCCT	ACCCAGATAGGTGAGTTGCC
	Exon50_2	CCTTTGACAACTTTGCTCCTGG	TGGAATTGAAGGGTGATTGGTG
	Exon51	TTAACTAACAAGGATTTCAATTTTCTG	AACAGTATGACAAGGTGGAATTC
	Exon52	GCATCCTAAAAAAGGATAGAAATTTG	AATCAGATCTGGCTGGGTTT
	Exon53	TCCCAAACCTGCTGGGAGAAC	CTGTTTCCCAGAGCCCCCTT
	Exon54	ACTGCAATTTCTCCCTCTCTTTC	AAAGGGTGTTTGGAATTTAGCAT
	Exon55	CAAAATCTCAAGCAGAAGCAACC	TTTTGCACAACAGCCTCTGG
	Exon56	AGATGATTTGTGCTGCACTG	CCAAACATGGTCTTCTCTAGCTC
	Exon57	TTCTCAGGTTTGTGGGTCC	AGGCTCCAACCTGGTAATGGC
	Exon58_1	GCCTTTTGTGGGAAGAGGA	GTGACAGTATAGGCCTGACCC
	Exon58_2	AGCTGGCCATGGCATAGATTT	GGCCATGAAGACTTGAATGCG
	Exon58_3	CTTTCACATCCGAGGCCACA	GACCGAATTTTGTGGAGCAGA
	Exon58_4	TTGGTCTTTTGGCAGTAGTGT	CTTTGTGCCATGGCTCCTGA
	Exon58_5	GAGCTCCCTCCAATCCAAGAG	ACAGCAAGCACTAGACCACA
	Exon59	TCGATAATTTGTGGCTGGTGGT	AGGGTTTGAAGAATTGCCAAGTA
	Exon60	CCAATGTATATTTTCTTCTTGCTGC	TGCCTCTACCACAGGCATTG
	Exon61A	GGTCCATGCTCCAACCCATT	GAGGGCGGGCATGAGTAAAG
	Exon61_1	TCACTTGTTTTGCTTCTTTCTTTT	AGAAAGTAGACACTGACCCAGA
	Exon61_2	TGTGACTAGTGGTTTTGTTGATGTC	AGCTGACTGAACCAGAGTGG
	Exon61_3	GAGCCCCACGTCTTCTTAG	GATTTGGTTTTGGCCAACTGT
	Exon61_4	CCTCACTGTGATGGTTTCAGTC	CCACAGTCATTGGGGGTGAA
	Exon61_5	GTTGGTCAACGTAGGCCTCT	ACCCATAGCCAATGACTCCC
	Exon61_6	TCGCCAACAGTAAGGAGCAC	TCAGTGTAAGTAGATTGACATTTGC
	Exon62	GGATTGTGGAAAATTGCTACC	AGGCTGAATGCTACATGCTAC
	Exon62A	TGATGCTCAGGTATTAGAAAAAGTG	ACAGCATCAGGGTAGACTAGGT
	Exon63	TCTGATCCATCTTTCTAACTTCAC	AATTCAGTATTACCAATTTTGCTAT
	Exon64	GTCCAAATGTCTGCCTTCGC	TCCCACTATAAGGGAGAAAGGA
	Exon65	ACATCTTCTTTTCCCCGCCA	GTCTTTGGGGAAAGAAACAGAAATC
	Exon66	ACCTAAATGCTGATGGTCCC	AGGAGAGGGAGGCTCAGACC
	Exon67_1	CCATTCTTCCOCTATTTCTTTCC	CCTCCTCCAGTTCCCAGTC
	Exon67_2	CATCTGTGCTCCAGGTGCT	CGGGTGTACTGAATGAAGGCA
	Exon67_3	CTGCTGCTATGCCAGACTT	AGTCCTCACTTCCCCAGCTT
GANAB	Exon1	GCCTCCAGAGGAACAAAGGT	CGCTCAGGTGGAACCAATC
	Exon2	CCTTAGGCACTCCATCTGC	AGTGCTATGACAGGAAGAAGCT
	Exon3	GGCCACAATTTCCCCCTT	ATCTGTGGCAGATGGAGTGC
	Exon4	ATCAGGAAGGAGAAAGGGGC	TTAGGCAAAGGGCTCAAAGG
	Exon5	ACCTCACAAACCCCCATGC	TGTCATTACTCCTCTTGGCCC
	Exon6	GGGTGAGTGAAAGGCTGGAA	CCCCCATCTCTGGAAGTTTGT
	Exon7	CCCGCTCCCATCAACTAAA	TCGTTCTTTGGACCCCTG
	Exon8	TGGGGCCTGGAAGAAAAAC	GGAGAGGCCCATGAGAAACT
	Exon9	TGACCCATGCTTGCTTAGAG	GACAGCAAGCCGTATGGTGA
	Exon10	CCAGTTAGCAGCCCCAGTCT	TGCCCCTAGTATACCTATTTGCC
	Exon12-1	GGTCCCAGGTGAAATACCG	TTATACCCCTCAGCCTGCCT
	Exon11	GTCTTGGGGTTGAAAATGG	GGCACAGGATCTCTGAAAAAC
	Exon12-2	CCTTGATGTGGGGTCTACG	GAGGCTGATGTGCTGGAAGT
	Exon13	CCTAAGGGGCAAAAGAGCC	TCACGCTTGGTCTTCTCAGG
	Exon14	AAGGCCCCCTGATATGTTG	GGGAGAGTTGATTGGCCAG
	Exon15	AGTGAGGAGCAAAAGCCAGC	TCTGTCCCCATTTTCTGCC
	Exon16	CATGGCTCCTGAGATCTGG	GGGCCTGAGAGAGGAGATT
	Exon17	TTGGTCTGTGCCCTCTACT	ATCTTCAGCAGGGTTCTGGG
	Exon18-1	CCAGTCCCTAATACCCCA	CAAAAACCCAGAGCCAGAGC
	Exon18-2	GTGTACCAGAAGGGCAGCAA	CAAGTTAGGGCCTTGGGAA
	Exon19	GGGGGATTAGTTCCAGAACAA	CCAGTTCTGGAGGAGCATT
	Exon20	ACCACACCTGTGAGTGACAA	TAGTGAAGCTGGTGGTGAG
	Exon21	CCCACAGCATGTTTGTCTT	CATGGTGGGGAAAGATGGT
	Exon22	GCACAACCCAGAGAACTGA	TGCCTTAGGGTCCAGTACCT

	Exon23	AGAGCACAGTGAAC TGGGAG	GCGAGTCTCCTCAGCCATTT
	Exon24	AACAACAACGTTCC TACAGGC	GACCTTGCTTTGGGTCTCC
	Exon25	TGCCTGTAGGAACG TTGTTG	TAAGTGAAGTCTGGGGAGGG
<i>DNAJB11</i>	Exon1	CTGTCTCTGCGGAC CAAGG	AGCAAAGGCTGACCCACAG
	Exon2-1	TCTCTCCCTCTACT TCCCAGAC	GAGCTGAAAGGAGTGTTTCTCT
	Exon2-2	CCTCTTTGCTCAGAT ACAAACC	CCTCATAAGCAGCACCCAGA
	Exon2	GAAGTATTCTCTCC CTCTACTTC	GTGAGCTGAAAGGAGTGTTTCTC
	Exon3	ACCCACTGTCAAAGT AAAAACAAAC	AATCGCTAAGTGACCCCTCTCT
	Exon4-1	CCCTCGTCAGCAAG ACAGAA	TCAGTGTATCATGAACCAAGCA
	Exon4-2	AAATACCTTGTAGTT TGCACAATG	CCTGCATATACTTCTTCCAAAGTG
	Exon5	AGTCCAGGCATATGT GCAGAG	CTTGGTAGAACAAAGAAGGTGCT
	Exon6	GGCACACTACAGTCT TCCAAGAAA	TTCACAAACAAGCAGAGAGCA
	Exon7	GAGACACTGCCGTCA CAGAT	AGCTAACTGCCTGTGGTGGC
	Exon8	GTTTGAAAGAGTGGT GTTCACCTT	GCTTCAAAGGATGCAGAACGA
	Exon9	CTGCTGCAGTACATT CCCTT	GTTCTTCTATTGCCATCCTTTTCT
	Exon10	TGCAATTACGTTGG AGATTGGT	ACCTTGACAGATAATAAATATCGCT
<i>ALG8</i>	Exon1	ACCCAGGGATATCCA CACCT	GGCTTCAGAGGGCTTTCTCC
	Exon2	GCACCCAGCCAGAAA CATT	TGTGACAGCTTAGGGAATGTCA
	Exon3	AATCGTTGCACCATC TTTGTTT	CCTTCCATACAAAATGACATGCTC
	Exon4	GCAGAAGTGATGTTCC GATG	CACCCCACTCATTTTTCAG
	Exon5	GCACTTGGCAGCTCA AAAACA	ACTCCATCAGCTGGTATAGTTCA
	Exon6	cattattgcatttCAT TTACCCTG	AAGCAAGCCACCAAGTCAAC
	Exon7	TGTTCCTTTCCATCT TCCTGGT	GGGAGTCACGTTGGGTAAGG
	Exon8	CCCAGACCCCATAA CTTTCTAC	TCTCCATGTGCCAAGTCCT
	Exon9	TCTTAGCCTTTAAGC AGCAATAAGA	GAAATGAGCACCATCTGTTGAGT
	Exon10	GGCTGTC TTTTCAGAGATGATGC	ATGCCAAAATGCTCACTGGC
	Exon11	GTGGGCCTCGATGAAA AGGT	TCTATGCCTCAGCCAGTTCT
	Exon12	AGCTTATTTGACCAC TTAGGTGTA	CTCAGCATCTCTAGATGTCACAA
	Exon13_1	TGTTTGCTAAACCAG GATGACT	TTGCTTGCCAATAGCAGAGT
	Exon13_2	GCCTCTGGAAGTCTG CTGTG	GGCAACTTTCTTCAAGTTCTGG
<i>PKD2</i> Microsatellite Primer	D4S231	GAGAGAAAGGGAAGG GATGCTAGAGTTCCTAGTG AGATGAGTATGTTATTATACC	
	D4S1534	GAGAGAAAGGGAAGG GACCATGTGTGATTTCAGTTTCAGCC TAGACCAGCCCAAGGTAGAGGAG	
	D4S1542	GAGAGAAAGGGAAGG GAAAGATCGACTCCAGTGCATTTCC GATTTTCAGTTTCTTCAAATTCTCCC	
	D4S1563	GAGAGAAAGGGAAGG GAGCTGCCTGACACACTGG ACTATTGCTGTTGCTGACCC	
	D4S1544	GAGAGAAAGGGAAGG GATGCCATAAATATCAAAATAGTCACAG GCTAGTAGTATTAAGGCTGTTGG	
	D4S414	GAGAGAAAGGGAAGG GATCTTGCACAAAGCATCAGCCCTC TCAGGAACCTCAGCCCATTTAAG	



Supplementary Figure 1. Microsatellite Analysis of 24 PKD2 p.R803* families. a Position of 5 microsatellite markers, including D4S1534, D4S1542, D4S1563, D4S1544, and D4S414. The region between D4S1534 and D4S414 is around 0.6 cM. b Microsatellite D4S1563 analysis showed a peak in the 230 bp position (red dot) in 11 samples. Figure not drawn to scale. cM: centimorgan.



Supplementary Figure 2. The observed and predicted trajectory of estimated glomerular filtration rate in individuals with PKD2 p.Arg803* and non-p.Arg803* truncation. The estimated glomerular filtration rate (eGFR) was calculated using the simplified Modification of Diet in Renal Disease equation. The predicted eGFR was generated by the mixed model for repeated measures incorporating random intercept and slope after putting the age at baseline, sex, follow-up year, quadratic follow-up year, and the interaction term between time and group of with and without p.Arg803 mutation using the forced entry approach.

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