Table S1. *PKD1* and *PKD2* mutations identified in study patients without apparent family history

Family	Gene	Sequence Variant	Codon Change	Predicted Effect	Domain	PKDB	Polyphen	SIFT	Segregation	Present in Parent
De Novo Disease										
TOR254	PKD1	c.11343C>G	p.Y3781X	Nonsense		2x				No
TOR279	PKD1	c.3628G>T	p.E1210X	Nonsense		1x				No
TOR303	PKD1	c.11393C>A	p.Ser3798X	Nonsense		Novel				No
TOR189	PKD1	c.5887_5888insG	p.V1963fs26X	Frameshift		Novel				No
TOR186	PKD1	c.7666C>T	p.Q2556X	Nonsense		7x				No
TOR253	PKD1	c.2618_2621del4	p.V873fs23X	Frameshift		1x				No
TOR293	PKD1	c.11378delG	p.G3792fs33X	Frameshift		Novel				No
TOR135	PKD1	c.2605delC	p.R869fs29X	Frameshift		Novel				No
TOR361	PKD1	c.9957_9958del2	p.Ser3319fs69X	Frameshift		1x				No
TOR403	PKD1	c.5014_5015del2	p.R1672fs97X	Frameshift		13x				No
TOR271	PKD1	c.1202-9G>A	Ala401fs	Atypical Splicing*		1x				No
TOR241	PKD1	c.1306T>C	p.C436R	Missense	C-lectin	1x	0.313	0	++	No
TOR351	PKD1	c.1367T>C	p.L456P	Missense	C-lectin	Novel	0.99	0		No
TOR358	PKD1	c.11257C>T	p.R3753W	Missense	Extracellular	4x	0.994	0	++	No
TOR130	PKD2	c.2159_2160insA	p.Asn720fs4X	Frameshift		Novel				No
TOR257	PKD2	c.1_2907del	p.Met1fs	Large Deletion		Novel				No
Indeterminate Family History										
TOR244	PKD1	c.4746G>A	p.W1582X	Nonsense		3x				Unknown
TOR309	PKD1	c.11766G>A	p.W3922X	Nonsense		3x				Unknown
TOR328	PKD1	c.10525_10526del2	E3509fs225X	Frameshift		Novel				Unknown
TOR329	PKD1	c.2016_2017insG	p.G672fs40X	Frameshift		1x				Unknown
TOR331	PKD1	c.4042_4043del2	p.H1347fs82X	Frameshift		Novel	0.995			Unknown
TOR379	PKD1	c.1674delG	p.P559FS	Frameshift		Novel				Unknown
TOR235	PKD1	c.10838T>C	p.L3613P	Missense	Cytoplasmic	Novel	0.987	0.01		Unknown
TOR256	PKD1	c.5995G>A	p.G1999S	Missense	PKD16	2x	0.999	0.3	++	Unknown

TOR307	PKD1	c.7088T>A	p.V2363E	Missense	REJ	1x	0.993	0.03		Unknown
TOR319	PKD1	c.5809A>G	p.N1870S	Missense	PKD14	Novel	0.999	0		Unknown
TOR330	PKD1	c.10972A>G;	p.K3658E	Missense	Cytoplasmic	Novel	0.214	0.03	++	Unknown
TOR368	PKD1	c.8464G>A	p.V2822M	Missense	REJ	2x	0.906	0		Unknown
TOR219	PKD2	c.2407C>T	p.R803X	Nonsense		6x				Unknown
TOR232	PKD2	c.973C>T	p.R325X	Nonsense		6x				Unknown
Positive Family History in Retrospect										
TOR323	PKD1	c.4049C>T	p.T1350M	Missense	PKD8	1x	0.971	0.09	++	Yes
TOR343	PKD1	c.7409C>T	p.P2470L	Missense	REJ	Novel	0.998	0.01	+	Yes
TOR395	PKD1	c.2695C>G	p.L899V	Missense	PKD3	Novel	0.784	0.02	+	Yes

*Presence of atypical splice site was confirmed by rtPCR

Table S2. Characteristics of study patients with asymmetric polycystic kidney disease*‡

Proband	Age[a]	Sex	Family History (Y/N)	De novo (Y/N)	Ccr (mL/min)[b]	TKV (mL)	LKV (mL)	RKV (mL)	RKV/ LKV	*PKD1/2* mutation findings
TOR186.1	26	F	Y	N	105	784.9	238.1	546.8	2.3	*PKD1*: c.7666C>T; p.Q2556X
TOR225.1	18	F	Y	N	104	722.0	502.4	219.6	0.4	*PKD1*: c.856_862delTCTGGCC; p.S286fs1X
TOR320.1	26	F	Y	N	101	875.2	634.6	240.6	0.4	NMD
TOR233.1	44	F	Y	N	94	801.7	634.6	167.1	0.3	*PKD2*: c.567G>A; p.W189X
TOR204.1	34	F	Y	N	104	638.1	453.5	184.6	0.4	*PKD2*: c.1249C>T; p.R417X
TOR176.1	19	M	Y	N	98	506.7	148.3	358.4	2.4	*PKD2*: c.595+1G>A; p.Gly199fs
TOR212.1	40	F	Y	N	134	1265.6	418.1	847.5	2.0	*PKD2*: c.973C>T; p.R325X
TOR216.1	53	F	Y	N	84	811.8	545.9	265.9	0.5	*PKD2*: c.C1024T; p.R320X
TOR8.1	44	F	Y	N	99	552.9	379.0	173.9	0.5	*PKD2*: c.2160InsA; p.D724fs4X
TOR350.1	68	M	Y	N	79	1019.5	715.0	304.5	0.4	*PKD2*: c.2614C>T; p.R872X
TOR184.1	55	M	Y	N	116	3632.0	3184.5	447.5	0.1	NMD
TOR135.1	53	F	N	Y	73	885.5	698.8	186.7	0.3	*PKD1*: c.2605delC; p.R869fs29X
TOR293.1	21	M	N	Y	53	1314.6	424.9	889.7	2.1	*PKD1*: c. 11378delG; p.G3792fs33X
TOR271.1	30	M	N	Y	92	1391.0	1005.2	385.8	0.4	*PKD1*: c.1202-9G>A; p.A410fs
TOR180.1	58	M	N	Y	92	1215.3	963.9	251.4	0.3	NMD
TOR297.1	38	F	N	Y	138	635.3	164.1	471.2	2.9	NMD
TOR222.1	47	F	N	Y	89	463.0	324.2	138.8	0.4	NMD
TOR368.1	72	M	IND	IND	102	245.2	182.0	63.2	0.4	*PKD1*: c.8464G>A; p.V2822M
TOR407.1	74	M	IND	IND	65	2235.0	318.9	1916.1	6.0	NMD
TOR282.1	51	M	IND	IND	154	854.9	173.1	681.8	3.9	NMD
TOR333.1	55	M	IND	IND	77	922.9	266.2	656.7	2.5	NMD
TOR349.1	57	M	IND	IND	76	2083.9	630.9	1453	2.3	NMD

*Asymmetric PKD is defined as diffuse cystic disease involving one or both kidneys with >50% difference in volume between the two kidneys by CT/MRI, or >50% difference in length between the two kidneys by ultrasound.

[a]Age at MRI or ultrasound; [b]measured creatinine clearance at time of renal imaging.
NMD: no mutation detected; IND: indeterminate.
‡The diagnosis of asymmetric PKD in the 23rd case (TOR189.1) was based on ultrasound findings: 56 year-old female patient with negative family history, Ccr 46 mL/min, a ratio of left/right kidney length of 2.29, and a *PKD1* mutation (c.5887_5888InsG; p.V1963FS26X).

Table S3. Results of *GANAB* mutation screen in 32 mutation-negative cases

DNA ID#	Rare GANAB variants	Alt/Alt+Ref	MAF in 1000G	MAF in ExAC	Damaging calls on SNV by 7 programs
5995	exon10:c.991C>T:p.R331C (rs1063445)	48.439%	1.0000%	0.8000%	6/7
5583	no mutation found				
5506	no mutation found				
5873	no mutation found				
1320	exon10:c.991C>T:p.R331C (rs1063445)	50.932%	1.0000%	0.8000%	6/7
5916	exon5:c.460C>T:p.R154W	51.003%	1.0000%	0.4900%	1/7
5994	no mutation found				
7002	exon23:c.2614C>T:p.H872Y (rs114915323)	48.939%	0.8000%	0.3000%	4/7
5744	no mutation found				
5146	no mutation found				
5560	no mutation found				
5968	no mutation found				
6132	exon23:c.2614C>T:p.H872Y (rs114915323)	46.803%	0.8000%	0.3000%	4/7
6045	no mutation found				
5748	no mutation found				
2021	no mutation found				
5577	no mutation found				
5850	no mutation found				
5708	no mutation found				
5524	no mutation found				
5833	no mutation found				
5958	no mutation found				
5517	no mutation found				
6048	no mutation found				
5989	no mutation found				
5510	no mutation found				
5176	exon10:c.991C>T:p.R331C (rs1063445)	48.723%	1.0000%	0.8000%	6/7
5176	exon3:c.160C>T:p.R54W	52.519%	NA	0.0050%	4/7
5586	no mutation found				
6216	no mutation found				
6917	no mutation found				
5767	no mutation found				
5170	no mutation found				

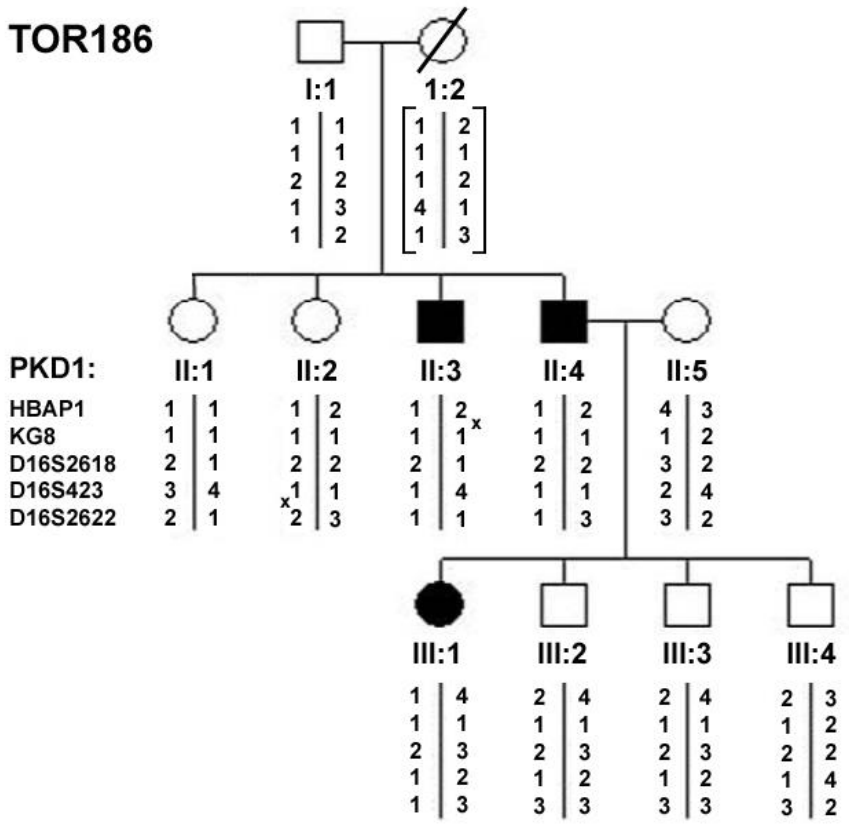

Figure S1. *PKD1* haplotype analysis of TOR186. Only one *PKD1* haplotype (1-1-2-1-1) co-segregates with the two affected siblings (II:3, II:4) and the affected daughter (III:1) of II:4, all carrying the *PKD1* (c.7666C>T; p.Q2556X) mutation. This haplotype originated from the unaffected grandfather (I:1) and is also present in one unaffected (II:2) sibling. These data strongly support germline mosaicism in I:1. The location of simple sequence repeat markers relative to *PKD1* are as follows (the number between markers denotes inter-marker distance in cM): HBAP1-2.0-KG8/*PKD1*-0.8-D16S2618-1.2-D16S423-1.3-D16S2622. KG8 is an intragenic marker located within the 3' end of *PKD1*. x denotes inter-marker recombination.