

Table S3: Clinically Reportable Variants Identified in the Diagnostic Cohort

Pt. ID	Age at test (yrs)	Typical or Atypical PKD	Enlarged Kidneys	CKD Stage	FHx of PKD	Gene	Exon^	Coding Change ^s	Protein Change ^{&}	Variant Classification [#]	Novel
D038	30	T	Y	1	Y	<i>PKD1</i>	23	NM_001009944.2:c.8458_8459del	p.(Ser2820*)	P	Y
D041	42	T	Y	NR	N	<i>PKD1</i>	IVS26	NM_001009944.2:c.9398-1G>C	p.?	P	Y
D128	34	A	N	1	Y	<i>PKD2</i>	IVS2	NM_000297.3:c.709+1G>T	p.?	P	Y
D158	24	T	N	1	Y	<i>PKD1</i>	IVS18	NM_001009944.2:c.7489+5G>A	p.?	VUS	N
D163	31	T	N	1	N	<i>PKD1</i>	23	NM_001009944.2:c.8447T>C	p.(Leu2816Pro)	VUS	N
D164	71	A	N	4	Y	<i>GANAB</i>	11	NM_198335.3:c.1208G>C	p.(Ser403Thr)	VUS	Y
D165	44	A	N	3	N	<i>HNF1B</i>		Whole gene deletion		P	N
D166	39	A	N	1	N	<i>PKD1</i>	15	NM_001009944.2:c.4592A>G	p.(Asn1531Ser)	VUS	Y
D329	58	T	Y	1	Y	<i>PKD2</i>	9	NM_000297.3:c.1973del	p.(Pro658fs)	LP	Y
D335	0	A	Y	5	N	<i>PKD1</i>	23/ 42	NM_001009944.2:c.[8302G>A];[11579C>T]	p.[(Val2768Met)]; [(Ala3860Val)]	VUS /VUS	N/ Y
D3936	28	A	N	3	Y	<i>PKHD1</i>	3/ 30	NM_138694.3:c.107C>T(;);3467C>T	p. (Thr36Met) (;)(Ser1156Leu)	P/ LP	N/ N
D429	34	A	Y	1	N	<i>PKD1</i>	15	NM_001009944.2:c.4828_4830del	p.(Ile1610del)	VUS	N
D443	0	A	Y	5	N	<i>PKHD1</i>	IVS17 /61	NM_138694.3:c.[1602+1G>A];[10174C>T]	p.[?];[(Gln3392*)]	P/ P	N/ N

D501	42	T	Y	1	N	<i>PKD2</i>	5	NM_000297.3:c.1258A>G	p.(Arg420Gly)	LP	N
D5183	0	A	Y	5	Y	<i>PKD1</i>	16/40	NM_001009944.2:c.6994_7000dup(;):11399C>G	p.(Val2334fs)(;)(Pro3800Arg)	P/VUS	N/Y
D598	64	T	NR	NR	Y	<i>PKD1</i>	45	NM_001009944.2:c.12326_12335del	p.(Arg4109fs)	LP	Y
D599	18	T	N	1	Y	<i>PKD1</i>	5	NM_001009944.2:c.856_862del	p.(Gly287*)	P	N
D6038	34	T	NR	1	Y	<i>PKD1</i>	40	NM_001009944.2:c.11338_11344dup	p.(Asp3782fs)	P	N
D646	50	T	N	5	Y	<i>PKD1</i>	IVS42	NM_001009944.2:c.11712+1G>C	p.?	LP	Y
D648	25	A	NR	NR	N	<i>PKD1</i>	IVS33	NM_001009944.2:c.10405+5G>T	p.?	VUS	N
D688	3	A	Y	4	N	<i>PKD1</i>	23	NM_001009944.2:c.8251C>T	p.(Gln2751*)	P	N
D702	2	A	Y	1	N	<i>PKD1</i>	15/37	NM_001009944.2:c.[3746del];[10939C>T]	p.[(Asp1249fs)];[(Arg3647Trp)]	P/VUS	Y/Y
D709	58	T	Y	3	Y	<i>PKD1</i>	14	NM_001009944.2:c.3202C>T	p.(Gln1068*)	P	N
D710	57	T	Y	3	Y	<i>PKD1</i>	15	NM_001009944.2:c.5855G>A	p.(Gly1952Asp)	VUS	N
D711	63	T	N	5	Y	<i>PKD1</i>	15	NM_001009944.2:c.6806C>G	p.(Ser2269*)	P	N
D719	40	A	N	1	N	<i>PKD1</i>	6	NM_001009944.2:c.1260C>G	p.(Tyr420*)	LP	Y
D720	35	A	Y	1	Y	<i>PKD1</i>	38	NM_001009944.2:c.11105A>C	p.(Gln3702Pro)	VUS	Y
D721	50	T	NR	NR	Y	<i>PKD1</i>	25	NM_001009944.2:c.8955_8962del	p.(Arg2985fs)	LP	Y

D722	37	T	NR	2	Y	<i>PKD1</i>	IVS43	NM_001009944.2:c.12004-2A>G	p.?	LP	N
D730	37	T	NR	NR	Y	<i>PKD1</i>	11	NM_001009944.2:c.2180T>C	p.(Leu727Pro)	LP	N
D738	15	T	Y	1	Y	<i>PKD1</i>	37	NM_001009944.2:c.10958C>T	p.(Ala3653Val)	LP	N
D830	48	A	N	2	N	<i>PKD1</i>	18	NM_001009944.2:c.7409C>T	p.(Pro2470Leu)	VUS	N
D159	34	A	N	1	N	<i>PKD1</i>	23/ 43	NM_001009944.2:c.8531T>C(;) 11972C>T	p.(Val2844Ala)(;) (Ala3991Val)	VUS/ VUS	Y/ N
D837	49	A	N	1	N	<i>PKD1</i>	15	NM_001009944.2:c.3994G>A	p.(Asp1332Asn)	VUS	N
D869	48	A	Y	3	N	<i>PKD1</i>	15	NM_001009944.2:c.3883G>T	p.(Glu1295*)	LP	Y
D870	33	T	N	NR	Y	<i>PKD1</i>	11	NM_001009944.2:c.2180T>C	p.(Leu727Pro)	LP	N
D871	79	A	N	3	N	<i>TSC2</i>	35	NM_000548.3:c.4534G>A	p.(Asp1512Asn)	VUS	N
D873	46	A	N	1	Y	<i>HNF1B</i>	2	NM_000458.3:c.516C>G	p.(Tyr172*)	P	Y
D897	54	T	Y	1	Y	<i>PKD2</i>	4	NM_000297.3:c.964C>T	p.(Arg322Trp)	LP	N
D913	33	A	N	1	N	<i>GANAB</i>	10	NM_198335.3:c.916G>A	p.(Val306Met)	VUS	Y
D917	28	A	N	1	N	<i>PKD1</i>	15	NM_001009944.2:c.3483G>T	p.(Trp1161Cys)	VUS	Y
D925	26	T	Y	2	Y	<i>PKD1</i>	16	NM_001009944.2:c.7008C>G	p.(Tyr2336*)	P	Y
D927	40	T	N	2	Y	<i>PKD2</i>	4	NM_000297.3:c.916C>T	p.(Arg306*)	P	N
D932	28	T	N	1	Y	<i>PKD1</i>	15	NM_001009944.2:c.4070del	p.(Leu1357fs)	P	N
D933	45	A	Y	1	N	<i>TSC2</i>	32	NM_000548.3:c.3816G>A	p.(Val272Val)	VUS	N

D936	25	T	Y	3	Y	<i>PKD1</i>	15	NM_001009944.2:c.374 5del	p.(Asp1249fs)	P	Y
D939	30	T	Y	1	Y	<i>PKD1</i>	IVS21	NM_001009944.2:c.801 7-2_8017-1del	p.?	P	N
D069	31	T	Y	2	N	<i>PKD1</i>	15	NM_001009944.2:c.344 1C>G	p.(Tyr1147*)	P	N
D069						<i>GANAB</i>	18	NM_198335.3:c.2159G> A	p.(Arg720Gln)	VUS	Y
D095	35	A	N	3	N	<i>PKHD1</i>	3/ 41	NM_138694.3:c. 107C>T(:)6800T>C	p.(Thr36Met) (:)(Leu2267Pro)	P/ VUS	N/ Y
D947	6	A	N	1	N	<i>PKD1</i>	25	NM_001009944.2:c.904 7del	p.(Gln3016fs)	P	Y
D962	39	A	N	1	Y	<i>PKHD1</i>	56	NM_138694.3:c.8776C> T	p.(Arg2926Trp)	VUS	N
D968	49	A	N	1	N	<i>PKD1</i>	6	NM_001009944.2:c.136 0C>T	p.(Arg454Cys)	VUS	N
D978	36	T	NR	NR	Y	<i>PKD1</i>	34	NM_001009944.2:c.104 41del	p.(Val3482fs)	P	Y
D979	33	T	Y	1	N	<i>PKD1</i>	18	NM_001009944.2:c.748 3T>A	p.(Cys2495Ser)	VUS	Y
D980	29	T	Y	1	N	<i>PKD1</i>	10	NM_001009944.2:c.209 6C>A	p.(Ser699*)	LP	Y
D981	38	T	N	1	Y	<i>PKD1</i>	10	NM_001009944.2:c.198 7del	p.(Gln663fs)	P	N
R811	35	T	NR	NR	Y	<i>PKD2</i>	13	NM_000297.3:c.2407C> T	p.(Arg803*)	P	N
D002	35	T	N	3	Y	<i>PKD1</i>	5	NM_001009944.2:c.827 _830del	p.(Thr276fs)	LP	Y
D004	51	T	Y	5	NR	<i>PKD1</i>	15	NM_001009944.2:c.363 1G>T	p.(Glu1211*)	LP	Y
D024	1	A	N	1	N	<i>HNF1B</i>		Whole gene deletion		P	N

D851	69	A	NR	2	N	<i>PKHD1</i>	61	NM_138694.3:c.10174C>T	p.(Gln3392*)	P	N
D809	57	T	Y	NR	Y	<i>PKD1</i>	15	NM_001009944.2:c.6025T>C	p.(Ser2009Pro)	VUS	Y
D828	33	T	Y	NR	Y	<i>PKD1</i>	23	NM_001009944.2:c.8311G>A	p.(Glu2771Lys)	LP	N
D808	25	T	NR	NR	Y	<i>PKD1</i>	46	NM_001009944.2:c.12506del	p.(Lys4170ArgfsTer28)	LP	Y
D807	55	A	NR	NR	Y	<i>DNAJB11</i>	4	NM_016306.5:c.400del	p.(Ile134fs)	LP	Y
D763	35	T	Y	1	N	<i>PKD1</i>	16	NM_001009944.2:c.6970dup	p.(Thr2324fs)	LP	Y
D579	35	T	NR	NR	Y	<i>PKD2</i>	IVS9	NM_000297.3:c.2019+1G>A	p.?	P	Y
D683	34	T	N	1	Y	<i>PKD1</i>	27	NM_001009944.2:c.9568G>A	p.(Gly3190Arg)	LP	Y
D644	75	T	N	NR	N	<i>PKHD1</i>	32/32	NM_138694.3:c.3761_3785delinsGCCCCAGATACCCGATGCGGGCGT(;) 5134G>A	p.(Gln1254fs)(;)(Gly1712Arg)	LP/VUS	Y/N
D608	32	T	NR	NR	Y	<i>PKD2</i>	5	NM_000297.4:c.1235dup	p.(Asn412fs)	LP	N
D607	40	T	NR	NR	N	<i>PKD1</i>	20	NM_001009944.2:c.7743_7767dup	p.(Gly2590fs)	LP	
D684	39	T	NR	NR	Y	<i>PKD1</i>	28	NM_001009944.2:c.9592C>T	p.(Gln3198*)	P	N
D67	11	A	N	1	N	<i>PRKCSH</i>	16	NM_001001329.2:c.1260C>G	p.(Tyr420*)	VUS	
D573	27	T	N	NR	Y	<i>PKD1</i>	11	NM_001009944.2:c.2289_2299del	p.(Cys767fs)	LP	Y

D544	76	A	NR	3	N	<i>PRKCSH</i>	16	NM_002743.3:c.1390A>G	p.(Lys464Glu)	VUS	Y
D462	39	T	NR	NR	Y	<i>PKD1</i>	19	NM_001009944.2:c.7642G>T	p.(Glu2548*)	P	Y
D296	59	A	N	3	N	<i>DNAJB11</i>	6	NM_016306.5:c.616C>T	p.(Arg206*)	P	N
D297	24	T	N	2	Y	<i>PKD2</i>	2	NM_000297.3:c.637C>T	p.(Arg213*)	P	N
D263	31	A	N	NR	Y	<i>PKD1</i>	35	NM_001009944.2:c.10524del	p.(Glu3509fs)	LP	Y
D254	36	T	N	1	y	<i>PKD1</i>	15	NM_001009944.2:c.4070del	p.(Leu1357fs)	P	N
D428	30	T	NR	NR	Y	<i>PKD1</i>	14	NM_001009944.2:c.3294_3295delinsTT	p.?	VUS	Y
D436	31	T	N	1	Y	<i>PKD1</i>	23	NM_001009944.2:c.8543T>C	p.(Val2848Ala)	VUS	Y
D009	33	A	N	1	N	<i>PKD1</i>	15	NM_001009944.2:c.6229G>C	p.(Ala2077Pro)	VUS	Y
D377						<i>PKD2</i>	15	NM_000297.3:c.2807del	p.(Asn936fs)	VUS	Y
D377	58	T	Y	3	N	<i>PKD1</i>	34	NM_001009944.2:c.10407T>A	p.(Asp3469Glu)	VUS	Y
D421	59	T	N	4	N	<i>PKD1</i>	15	NM_001009944.2:c.3437_3439del	p.(Phe1146del)	VUS	Y
D048	47	A	NR	1	N	<i>HNF1B</i>		Whole gene deletion		P	N
D916	20	A	N	2	N	<i>PKD1</i>	3	NM_001009944.2:c.348_352del	p.(Asn116fs)	P	N
D520	63	T	N	3	Y	<i>PKD2</i>	IVS1	NM_000297.4:c.709+6T>G	p.?	VUS	Y

D605	32	T	N	1	Y	<i>PKD1</i>	17	NM_001009944.2:c.710 8T>C	p.(Cys2370Arg)	LP	N
D961	24	T	NR	NR	NR	<i>PKD1</i>	5	NM_001009944.2:c.856 _862del	p.(Gly287fs)	P	N
D914	34	A	N	NR	Y	<i>PKD2</i>	4	NM_000297.3:c.1039del	p.(Val347fs)	P	Y
D106	52	A	NR	NR	Y	<i>PKD1</i>	6	NM_001009944.2:c.136 0C>T	p.(Arg454Cys)	VUS	Y
D781	27	T	NR	1	Y	<i>PKD1</i>	16	NM_001009944.2:c.699 4_7000del	p.(Ala2332fs)	Path	N
D573	41	T	NR	1	Y	<i>PKD1</i>	15	NM_001009944.2:c.450 7G>A	p.(Gly1503Arg)	VUS	N
D207	30	A	NR	NR	N	<i>TSC2</i>	3	NM_000548.4:c.175C>T	p.(Arg59Trp)	VUS	Y
D404	20	A	N	1	N	<i>PKD2</i>	IVS6	NM_000297.3:c.1549- 1G>A	p.?	P	Y
D732	42	A	NR	1	Y	<i>DNAJB11</i>	4	NM_016306.5:c.430G>T	p.(Glu144*)	LP	Y
D461	48	A	NR	1	Y	<i>PKD2</i>	4	NM_000297.4:c.875A>G	p.(Tyr292Cys)	LP	N
D057	34	A	N	2	N	<i>HNF1B</i>		Whole gene deletion		P	N
D717	38	T	N	1	Y	<i>PKD2</i>	11-13	NM_000297.3:c.2119_2 522del	p.?	P	Y
D561	60	A	NR	NR	N	<i>PKD1</i>	45	NM_001009944.2:c.123 07del	p.(Ala4103fs)	P	Y

T = Typical disease; A = Atypical disease; Enlarged Kidney = Bilateral Kidney length ≥ 14.5 cm, or reported 'enlarged' by clinician, Y = Yes; N = No; NR = Not Reported; FHx = family History; PKD = polycystic kidney disease ^{\$}*PKD1* NM_001009944.2, *PKD2* NM_000297.3, *PKHD1* NM_138694.3, *GANAB* NM_198335.3, *HNF1B* NM_000458.3, *TSC2* NM_000548.3, *DNAJB11* NM_016306.5; ^Exons are numbered like in *PKD1* NG_008617, *PKD2* NG_008604.1, *PKHD1* NG_008753.1, *GANAB* NG_053018.1, *HNF1B* NG_013019.2, *TSC2* NG_005895.1, *DNAJB11* NG_034024.1; & *PKD1* NP_001009944.2, *PKD2* NP_000288.1, *PKHD1* NP_619639.3, *GANAB* NP_938149.2, *HNF1B* NP_000449.1, *TSC2* NP_000539.2, *DNAJB11* NP_057390.1; #as per ACMG criteria; P = Pathogenic; LP = Likely Pathogenic; VUS = Variant of Uncertain Significance