

Supplementary Table 3. Comparison of characteristics between cystic kidney disease cohorts

Characteristics	Our cohort	HALT/CRISP [1]	TGESP [2]	Genkyst [3,4]
Disease entity	PKD	ADPKD	ADPKD	ADPKD
No. of patients	725	1,119	220	741
Age (yr)	46.2 ± 14.0	40.7 ± 10.8	38.9 ± 12.7	53.4 ± 14.8
Male sex (%)	48.1	48.3	55	43.8
sCr (mg/dL)	1.3 ± 1.4	-	0.90 ± 0.19	-
eGFR (mL/min/1.73 m ²)	76.5 ± 32.9	73.4 (52.1–95.8)	-	-
CKD stage (eGFR)			sCr ≤ 1.4 ^a	
1 (≥90)	304 (42.5)	357 (31.9)		90 (13.4)
2 (≥60, <90)	191 (26.7)	388 (34.7)		108 (14.6)
3 (≥30, <60)	137 (19.1)	353 (31.5)		137 (18.5)
4 (≥15, <30)	52 (7.3)	21 (1.9)		77 (10.4)
5 (eGFR < 15)	32 (4.5)	-		320 (43.2)
HtTKV (mL/m)	1,161.00 (631.2–1,895.7)	567.8 (387.6–827.4)	-	-
Age at diagnosis (yr)	37.1 ± 13.1	-	-	-
Genetic analysis	725 (100)	1,119 (100)	188 (85.4)	700 (94)(5)
Mutation detection rate (%)	64.3 ^d	92	84.5	89.9
<i>PKD1</i>	343 (47.3)	869 (77.7) ^b	131 (69.7) ^c	527 (75.2)
<i>PKD2</i>	79 (10.9)	165 (14.7)	57 (30.3)	102 (14.5)
<i>NMD</i>	99 (13.7)	85 (7.6)	-	-
Others	44 (6.1)	-	-	-
Hypertension	543 (74.9)	1,051 (93.9)	-	612 (82.5)
Kidney complication				
Flank pain	40 (5.5)	-	-	114 (38.9)
Macroscopic hematuria or intracystic hemorrhage	134 (18.5)	-	-	91 (31.1)
Kidney stone	68 (9.4)	-	-	57 (19.5)
Cyst infection	21 (2.9)	-	-	45 (15.3)

Data are expressed as number only, mean ± standard deviation, percentage only, number (%) or median (interquartile range).

ADPKD, autosomal dominant polycystic kidney disease; eGFR, estimated glomerular filtration rate; HtTKV, height-adjusted total kidney volume; NMD, no mutation detected; PKD, polycystic kidney disease; sCr, serum creatinine.

^aTGESP conducted genetic analysis by configuring a cohort of 220 patients with serum creatinine 1.4mg/dL or less. ^b*PKD1* mutation frequency of HALT/CRISP included PKD1-PT 592 (57.2%), PKD1-NT 277 (26.8%). ^cTGESP identified 188 pathogenic mutations in 186 of 220 ADPKD families. *PKD1* mutation was classified into PKD1-PT 72 (38.3), PKD1-NT 51 (27.1), IF indel 8 (4.3). ^dMutation detection rate of our cohort only includes result of primary screening test by gene panel only.