Online Supplemental Material

Baseline Characteristics of the Korean Genetic Cohort of Inherited Cystic Kidney Disease

Authors

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Supplementary table 1. Comparison of factors in iCKD, HALT+CRISP, TGESP, and Genkyst cohorts.

	iCKD cohort	HALT + CRISP(1)	TGESP(2)	Genkyst(3,4)
Included disease	CKD	ADPKD	ADPKD	ADPKD
Patient number, N	725	1119	220	741
Age (years), mean ± SD	46.2 ± 14.0	40.7 ± 10.8	38.9 ± 12.7	53.4 ± 14.8
Male, %	48.1	48.3	55.0	43.8
SCr (mg/dL), mean ± SD	1.3 ± 1.4	-	0.90 ± 0.19	-
eGFR (mL/min/1.73m^2), mean ± SD	76.5 ± 32.9	73.4 (52.1–95.8)	-	-
CKD stage, N (%)				
1 (90 ≤ eGFR)	304 (42.5)	357 (31.9)		90 (13.4)
2 (60 ≤ eGFR < 90)	191 (26.7)	388 (34.7)		108 (14.6)
3 (30 ≤ eGFR < 60)	137 (19.1)	353 (31.5)	SCr ≤ 1.4[a]	137 (18.5)
4 (15 ≤ eGFR < 30)	52 (7.3)	21 (1.9)		77 (10.4)
5 (eGFR < 15)	32 (4.5)	-		320 (43.2)
HtTKV (mL/m), median	630.5 (336, 1038)	567.8 (387.6–827.4)	-	-
Age at diagnosis (years), mean ± SD	37.1 ± 13.1	-	-	-
Genetic analysis, N (%)	725 (100)	1119 (100)	188 (85.4)	700 (94)(5)
Mutation detection rate (%)	64.3[d]	92	84.5	89.9
PKD1	343 (47.3)	869 (77.7)[b]	131 (69.7)[c]	527 (75.2)
PKD2	79 (10.9)	165 (14.7)	57 (30.3)	102 (14.5)
NMD	99 (13.7)	85 (7.6)	-	
Other	44 (6.1)	-	-	-
Hypertension, N (%)	543 (74.9)	1051 (93.9)	-	612 (82.5)
Kidney complication, N (%)				
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)

Abbreviations are: CKD, cystic kidney disease; ADPKD, autosomal dominant polycystic kidney disease; eGFR, estimated glomerular filtration rate; SCr, serum creatinine (mg/dL); HtTKV, height-adjusted total kidney volume; NMD, no mutation detected.

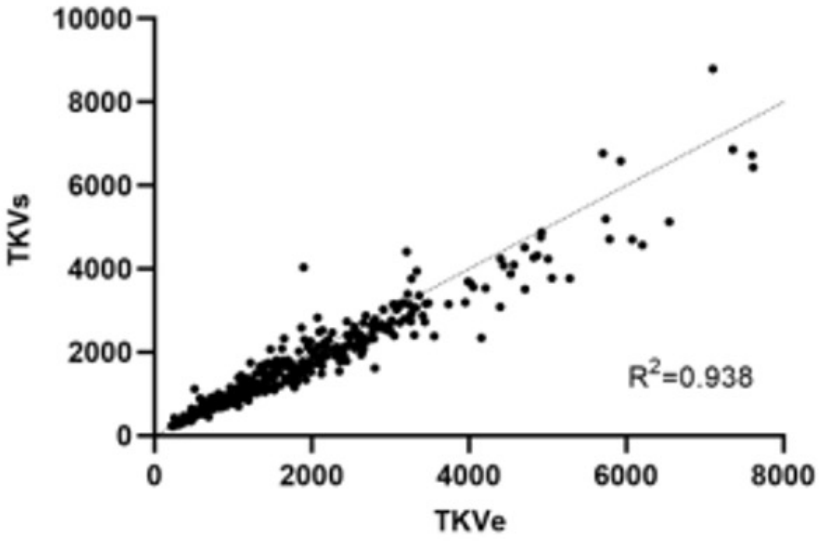
[a] TGESP implemented genetic analysis by configuring a cohort of 220 patients with serum creatinine 1.4mg/dL or less.

[b] PKD1 mutation frequency of HALT + CRISP included PKD1-PT 592 (57.2%), PKD1-NT 277 (26.8%).

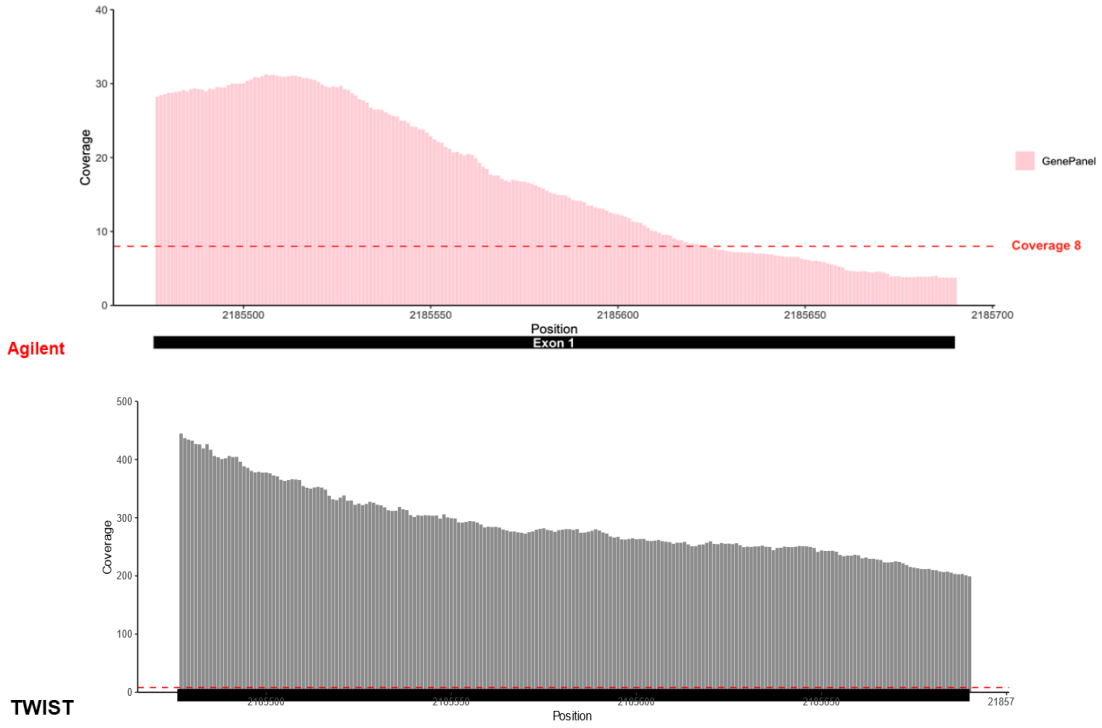
[c] TGESP identified 188 pathogenic mutations in 186 of 220 ADPKD families. PKD1 mutation was classified into PKD1-PT 72 (38.3), PKD1-NT 51 (27.1), IF indel 8 (4.3).

[d] Mutation detection rate of iCKD cohort only includes result of primary screening test by gene panel only.

Supplementary figure S1. Correlation between total kidney volume (TKV) based on either stereology or ellipsoid) by linear regression analysis (R2 = 0.938)



Supplemental figure S2. Coverage of polycystic kidney disease (PKD1) using TWIST versus Agilent probes. The gene panel was performed using a TWIST or Agilent probe. As the result, the average coverage was 902.8X and the median coverage was 999.0X for the TWIST probe, which was comparable to that using Agilent probe (985.9X and 999.0X, respectively). However, TWIST gene panel demonstrated higher coverage for PKD1 exon 1.



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