

Figure S1. The *SRD5A2* mutational spectrum and frequencies in 19 Korean children with 5 α RD2

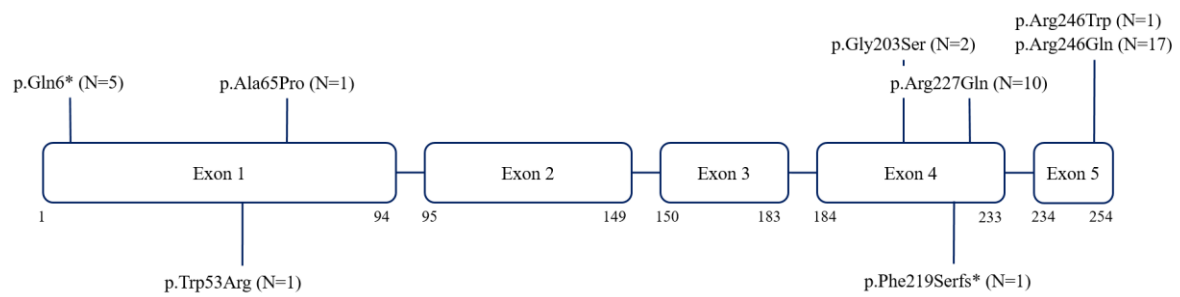


Table S1. The testosterone (T), dihydrotestosterone (DHT), and T/DHT ratio levels at baseline and after human chorionic gonadotropin (hCG) stimulation

	Patient	Age	Baseline			After hCG stimulation			
	No.	(yr)	T	DHT	T/DHT	T	DHT	T/DHT	
			(ng/dL)	(ng/dL)		(ng/dL)	(ng/dL)		
p.R227Q	1	9.7	<2.5	2	<1.3	106.3	8	13.3	
		13.1	224.6	12	18.7				
		13.3	121.3	7	17.3				
		16.0	483.0	10	48.3				
		16.5	468.0	9	52.0				
		17.0	425.0	14	30.4				
	2	1.3	<2.5	5	<0.5	507.0	9	56.3	
	3	0.5	10	2	5.0	217.6	22	9.9	
	4	0.9	<2.5	-	-	309.0	9	34.3	
	5	0.8	<2.5	2	<1.3	610.0	35	17.4	
	6	5.5	<2.5	2	<1.3	300.9	7	43.0	
	7	12.7	559.3	9	62.1	1149.0	18	63.8	
	8	11.8	17.3	5	3.5	203.7	8	25.5	
		13.5	158	11	14.4				
		13.9	182.3	14	13.0				
		17.9	266.0	10	26.6				
	9	0.6	<2.5	2	<1.3	427.1	25	17.1	
	Non-p.R227Q	10	0.0	192.0	44	4.4	587.0	69	8.5
		11	0.7	<2.5	2	<1.3	846.8	24	35.3
		12	0.0	154.0	22	7.0	Not done		
		13	4.6	<2.5	2	<1.3	166.4	7	23.8
		14	16.5	623.3	48	13.0	1267.0	83	15.3
		15	0.1	118.7	23	5.2	458.2	65	7.0
			12.4	170.5	23	7.4	660.7	44	15.0
		16	0.1	240.8	24	10.0	567.3	46	12.3
		17	2.6	4.0	1	4.0	279.7	3	93.2
		18	10.5	7.4	4	1.9	37.0	4	9.3
		19	9.9	<2.5	5	<0.5	330.0	5	66.0
Median		9.8	120.0	9.0	5.2	427.1	18.0	17.4	
Minimum		0.0	<2.5	1.0	<0.5	37.0	3.0	7.0	
Maximum		17.9	623.3	48.0	62.1	1267.0	83.0	93.2	

Table S2. The testosterone (T), dihydrotestosterone (DHT), and T/DHT ratio levels at baseline and after human chorionic gonadotropin (hCG) stimulation according to age

Age	N	Baseline			N	After hCG stimulation		
		T	DHT	T/DHT		T	DHT	T/DHT
		(ng/dL)	(ng/dL)			(ng/dL)	(ng/dL)	
< 6 mon	4*	173.0 (118.7–240.8)	23.5 (22.0–44.0)	6.1 (4.4–10.0)	3*	567.3 (458.2–587.0)	65.0 (46.0–69.0)	8.5 (7.0–12.3)
6 mon–12 yr	12	2.5 (2.5–17.3)	2.0 (1.0–5.0)	1.3 (0.5–5.0)	13	300.9 (37.0–846.8.0)	8.0 (3.0–35.0)	25.5 (9.3–93.2)
p.R227Q	7	2.5 (2.5–17.3)	2.0 (2.0–5.0)	1.3 (0.5–5.0)	8	305.0 (106.3–610.0)	9.0 (7.0–35.0)	21.4 (9.9–56.3)
Non- p.R227Q	5	2.5 (2.5–7.4)	2.0 (1.0–5.0)	1.3 (0.5–4.0)	5	279.7 (37.0–846.8)	5.0 (3.0–24.0)	35.3 (9.3–93.2)
	<i>P</i>				<i>P</i>			
> 12 yr	11 [†]	266.0 (121.3–623.3)	11.0 (7.0–48.0)	18.7 (7.4–62.1)	3	1149.0 (660.7–1267.0)	44.0 (18.0–83.0)	15.3 (15.0–63.8)

All data shows as median (min–max).

*all participants are non-p.R227Q variant allele group.

[†]Only two participants are non-p.R227Q variant group.

Note S1. List of 404 genes included in the custom panel for DSD-related genes

ABCA1, ABCC8, ABCG1, ABCG5, ABCG8, ADRB2, ADRB3, AGL, AGPAT2, AGRP, AIP, AIRE, AKAP2, AKR1C2, AKR1C4, AKT2, ALMS1, AMH, AMHR2, ANGPTL3, ANOS1, AP2S1, APC, APOA1, APOA4, APOA5, APOB, APOC2, APOC3, APOE, APPL1, AQP2, AR, ARAP1, ARL4D, ARL6, ARMC5, ARX, ATP1A1, ATP2B3, ATP7A, ATRX, AVP, AVPR2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, BLK, BMP15, BRAF, BSCL2, BTK, CA2, CACNA1D, CARTPT, CASR, CAV1, CAV2, CBX2, CCND1, CDC73, CDKL5, CDKN1A, CDKN1B, CDKN1C, CDKN2B, CDKN2C, CEL, CEP290, CEP41, CETP, CHD7, CIDEC, CISD2, CLCN7, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COLEC11, COMP, CP, CREB3L3, CREBBP, CRTAP, CRTCL, CRY2, CUL3, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP21A2, DDH2, DHCR24, DHCR7, DHH, DMRT1, DMRT2, DNAJC3, DUSP6, DYRK1B, EGLN1, EIF2AK3, ELN, EMX2, ENPP1, EPAS1, FBP1, FERMT3, FEZF1, FGD1, FGF17, FGF8, FGF9, FGFR1, FGFR2, FH, FIGLA, FKBP10, FLRT3, FOXC2, FOXE1, FOXL2, FOXP3, FRAS1, FREM2, FSHB, FSHR, G6PC, GALNT2, GATA3, GATA4, GATA6, GBE1, GCK, GCKR, GCM2, GDF9, GHR, GHRL, GLI2, GLI3, GLIS3, GLUD1, GNA11, GNAS, GNRH1, GNRHR, GPD1, GPIHBP1, GPR161, GRIP1, GYS2, HADH, HADHA, HADHB, HESX1, HFE, HHIP, HIRA, HNF1A, HNF1B, HNF4A, HOXA13, HRAS, HS6ST1, HSD17B3, HSD3B1, HSD3B2, ICK, IER3IP1, IFIH1, IFITM5, IGFALS, IKBKG, IL17RD, INS, INSL3, INSR, IRF6, IYD, KAT6B, KCNJ11, KCNJ5, KIF1B, KISS1, KISS1R, KLF11, KLHL3, KRAS, LCAT, LDLR, LDLRAP1, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LIMK1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LMNB2, LPIN1, LPL, LRP5, LRP6, MADD, MAGEL2, MAMLD1, MAP2K1, MAP2K2, MAP3K1, MATN3, MAX, MC2R, MC3R, MC4R, MCM4, MEF2A, MEN1, MKKS, MKS1, MLXIPL, MTPP, MYLIP, NEUROD1, NEUROG3, NF1, NIPBL, NKX2-1, NKX2-5, NLGN4X, NNT, NOBOX, NOTCH2, NPC1L1, NR0B1, NR0B2, NR5A1, NRAS, NSMF, NTRK1, NTRK2, OCRL, OPHN1, OSTM1, OTC, OTX1, OTX2, P3H1, PAX4, PAX8, PAX9, PCK1, PCK2, PCSK1, PCSK9, PDX1, PHF6, PHIP, PHKA2, PHKB, PHKG2, PITX2, PLEKHM1, PLIN1, PLOD1, PLOD2, PLTP, PNPLA2, POLD1, POMC, POR, POU1F1, PPARA, PPARG, PPARGC1B, PPIB, PRKAR1A, PROK2, PROKR2, PROP1, PSMC3IP, PTEN, PTF1A, PTH, PTH1R, PTPN11, PTRF, PYGL, PYY, RASGRP2, RB1, RET, RFX6, ROR2, RSPO1, RXFP2, SALL1, SAR1B, SCARB1, SDC3, SDCCAG8, SDHA, SDHAF2, SDHB, SDHC, SDHD, SECISBP2, SEMA3A, SEMA3E, SERPINF1, SERPINH1, SIM1, SLC16A1, SLC16A2, SLC26A2, SLC26A4, SLC2A2, SLC37A4, SLC39A13, SLC5A5, SLC9A3R1, SORT1, SOX10, SOX2, SOX3, SOX9, SP7, SPECC1L, SPRY4, SRA1, SRD5A2, SREBF1, SRXX2, SRXX3, SRXY10, SRY, STAP1, STAR, STK11, STS, STX16, SULT2A1, TAC3, TACR3, TBC1D4, TBCE, TCIRG1, TCTN3, TG, THRA, THRB, TMEM127, TMEM38B, TNFRSF11A, TNFSF11, TP53, TPO, TRHR, TRIB1, TRIM32, TSHB, TSHR, TSPYL1, TTC8, UBR1, UCP1, UCP2, UCP3, VHL, VPS13B, WDPCP, WDR11, WFS1, WNK1, WNK4, WNT1, WNT4, WNT7A, WRN, WT1, WWOX, ZFP57, ZFPM2