Supplementary Table 2. Phenotype and genotype of subjects with VUS variants

Patient ID	Age	Phenotype	Transcript ID	Genetic variants	Zygosity	Family ineheritanc	Inheritance mode	Classification	gnomAD frequency	Popmax	Reference
1909	6y3m	predominantly female external genitalia, cryptorchidism	NM_000233.3	*LHCGR* c.1541G>C, p.Cys514Ser	homo	F+M	AR	VUS	/	/	/
2036	6y	micropenis, hypogonadism	NM_000044.3	*AR* c.142G>T, p.Ala48Ser	hemi	M	XR	VUS	9.492E-06	0	/
2473*	12y	micropenis, small testes	NM_032551.4	*KISS1R* c.631G>A, p.Ala211Thr	het	M	AR	VUS	0.0001812	0.001506 (East Asian)	/
2692	1y1m	hypogonadism, micropenis, cryptorchidism	NM_144773.2	*PROKR2* c.533G>C, p.Trp178Ser	het	F	AD	VUS	0.0002227	0.00231 (East Asian)	17054399
2786*	1y7m	micropenis, cryptorchidism	NM_000348.3	*SRD5A2* c.702C>G, p.Phe234Leu	het	F	AR	VUS	0.0002037	0.004319 (East Asian)	15266301
3828*	10y	micropenis	NM_000348.3	*SRD5A2* c.457T>C, p.Phe153Leu	het	F	AR	VUS	/	/	/
3874*	7y7m	prepenile scrotum, hypospadias	NM_021044.2	*DHH* c.968G>A, p.Gly323Asp	het	M	AR	VUS	/	/	/
4274*	13y	gynecomastia (Tanner B4)	NM_000145.3	*FSHR* c.227A>C, p.Glu76Ala	het	F	AR	VUS	/	/	/
4431*	14y	gynecomastia (Tanner B4)	NM_000941.2	*POR* c.1196_1204delCCTCGGAGC, p.Pro399_Glu401del	het	M	AR	VUS	/	/	21843508
4500	3y	micropenis, small testes	NM_144773.2	*PROKR2* c.892C>T, p.Arg298Cys	het	M	AD	VUS	/	/	/
4940	7y	clitorimegaly, absent Müllerian structures, cryptochidism	NM_000102.3	*CYP17A1* c.785T>G, p.Met262Arg	het	F	AR	VUS	/	/	/
				CYP17A1 c.1193C>T, p.Ala398Val	het	M	AR	VUS	/	/	14552333
5337	4m	micropenis, perineal hypospadias	NM_000044.3	*AR* c.626G>A, p.Gly209Glu	hemi	M	XR	VUS	/	/	/
5421	12y	gonadal dysgenesis, micropenis, perineal hypospadias	NM_005921.1	*MAP3K1* c.2062C>G, p.Leu688Val	het	M	AD	VUS	/	/	/
6500	5y	idiopathic short stature, gonadal dysplasia, female external genitalia, utero-like tissue	NM_000163.4	*GHR* c.136+1Gly>Ala	homo	F+M	AD/AR	VUS	/	/	/
6928*	6m	perineal hypospadias, penile curvature	NM_000197.1	*HSD17B3* c.466T>A, p.Cys156Ser	het	M	AR	VUS	/	/	/
8585	13y	micropenis, hypogonadotropic hypogonadism	NM_144773.2	*PROKR2* c.1004C>T, p.Thr335Met	het	M	AD	VUS	0.00000701	0.00000701 (European non-finnish)	/
9171	10y	micropenis, unilateral cryptorchidism, hypogonadotropic hypogonadism	NM_023110.2	*FGFR1* c.1961A>G, p.Tyr654Cys	het	F	AD	VUS	/	/	/
10724*	2y5m	micropenis, unilateral cryptorchidism, hypospadias	NM_000348.3	*SRD5A2* c.408C>A, p.Tyr136*	het	de novo	AR	VUS	/	/	/
13317	6y4m	micropenis, bilateral cryptorchidism, gonadal dysplasia	NM_012082.3	*ZFPM2* c.1124_1127del, p.Gln375Argfs*40	het	M	AD	VUS	/	/	/
17641	2m	short stature, penile curvation, hypospadias, bilateral cryptorchidism, micropenis, penile scrotal translocation, scrotal division, special features (high hairline, forehead protrusion), premature delivery, small for gestational age	NM_000435.2	*NOTCH3* c.2129A>G, p.Tyr710Cys	het	M	AD	VUS	/	/	24000151
18385	7m	micropenis, bilateral cryptorchidism	NM_144773.2	*PROKR2* c.533G>C, p.Trp178Ser	het	M	AD	VUS	0.0002227	0.00231 (East Asian)	17054399
20685	10y3m	gynecomastia, micropenis	NM_000044.6	*AR* c.1159A>T, p.Ile387Phe	hemi	M	XR	VUS	/	/	/
21257	8y7m	micropenis	NM_144773.2	*PROKR2* c.533G>C, p.Trp178Ser	het	M	AD	VUS	0.0002227	0.00231 (East Asian)	17054399

Abbreviations: hemi, hemizygote; het, heterozygote; homo, homozygote; F, father; M, mother; XL, X chromosome-linked; AR, autosomal recessive; AD, autosomal dominant; P, pathogenic; LP, likely pathogenic.

*: Variant zygosity does not match the inheritance mode of detected genes.