

Table 2. Clinical findings of study population at diagnosis of congenital adrenal hyperplasia

Patient No	Karyotype	CYP21A2 Gene Analyses	Age (year)	Bone age, (year)	Height, (cm)	Predicted height, cm	Mean parental height, cm	BMI kg/m ²	Pubic hair	Breast stage	Penil length, cm	Testicular size, mL	Uterus length, cm	Right ovary, mL	Left ovary, mL	Adrenal hypertrophy
1	XX	V282L, Homozygous mutation	7.0	8.8	130.5	167	160	19.6	3	1			18	1.8	.90	no
2	XX	No mutation	8.8	11	141.5	160.8	158.5	16.2	3	2			36	6.5	2.7	no
3	XX	P31L, Homozygous mutation	6.8	12	134.5	149.2	161.5	17.5	4	1			15	1.0	1.3	no
4	XX	V282L, Homozygous mutation	7.8	10.5	127	148.3	153.5	15.5	3	1			35	1.2	0.8	no
5*	XX	12G, Homozygous mutation	4.6	11.6	125	ND	155.5	17.5	3	1	7.5*		40	0.5	0.4	yes
6	XY	No mutation	4.0	12	115.5	ND	167	18.3	3		8.5	4				no
7	XY	No mutation	2.4	6	82	ND	176.5	16.3	1		11.5	4				yes
8	XY	p. I173N, Homozygous mutation	8.4	13	136.2	160.3	161	16.7	4		10.0	10				yes
9**	XY	No mutation	4.1	9	124	ND	171	15.6	2		11.0	4				no
10**	XY	No mutation	5.5	11	142	ND	171	16.8	3		8.5	5				yes
11	XY	I172N, Homozygous mutation/del	5.8	13.4	138.7	158.5	157.95	16.1	3		8	3				no
12	XX	No mutation	2,5	3,48	96	ND		16,2	1	1			20	1,58	2,35	no

*: Reared as a male, **: siblings.