Table 1a
Characteristics of the patients with classic 21-hydroxylase deficiency

Pat. no.	Gender	Clinical form	Age (yrs)	age at dgs (mo)	Height (cm)	Height SDS	Weight (kg)	BMI (kg/m²)	BMI-Z score	mean HC dose (mg/m²/d)	cHC dose (mg)	Durat. treat. (yrs)	17OHP (ng/dl)	T (xUNL)	DHEAS (xUNL)	Genotype	Genotype group
1	F	SW	11.9	0.75	142.5	-1.1	35.5	17.4	-0.11	21	88530.75	11.8	265	1.3	0.5	del/del	0
2	M	SW	12.1	0.75	137.6	-1.64	35.5	18.7	0.29	30	124282.5	12	757	1.87	1.71	I2G+del8bp/ I2G+Q318X	0
3	M	SW	13.5	0.75	157.5	-0.11	50	20.1	0.53	30	139065	13.4	842	1.72	1.63	I2G+del8bp/ I2G+Q318X	0
4	F	SW	17	0.75	149.2	-2.64	43	19.3	-0.56	20	118625	16.9	241	1.05	1	I2G/del	A
5	M	SW	8.2	2	144	2.62	49	23.6	2.19	17.5	51100	8	1248	2.18	1.52	I2G/I2G	A
6	M	SW	11	1	147.5	0.48	37	17.1	-0.04	28	111398	10.9	1354	2.55	1.37	del/del	0
7	M	SW	8.4	1	126	-0.54	25.5	16.1	-0.11	20	60590	8.2	835	1.3	1.25	I2G/I2G	A
8	F	SW	11.1	0.75	142	-0.28	48	23.8	1.57	18	72270	11	692	1	0.92	I2G/P30L+ I2G+del8bp	A
9	F	SW	7.3	1	114.5	-1.63	30	23.2	2.18	20	52560	7.2	495	1	0.85	I2G/I2G	A
10	F	SW	6	0.75	114.5	-0.28	20	15.2	0.12	20.5	44520.87	5.9	655	1	1	I2G/I2G	A
11	F	SW	4.1	0.5	104.5	0.4	15	13.8	-1.45	15.9	21942	4	223	0.25	0.1	I2G/P30L+ I2G+delbbp	A
12	F	SW	4.2	0.5	90	-3.5	16	19.75	2.24	16.8	22680	4.1	375	0.9	0.2	del/I2G	A
X+/-SD (SW)	**5 M/7F**	**12 SW**	**9.5+/_ 3.3***	**0.8 +/- 0.4 ****	**132.9 +/- 21.2**	**-0.6 +/-1.6**	**33.7+/- 12.5**	**19.0 +/- 3.0**	**0.6 +/- 1.2**	**21.5 +/- 5.0 *****	**75630.3 +/- 40125.6**	**9.4 +/- 3.9**	**665.2 +/- 373.2**	**1.3 +/- 0.6**	**1.0 +/- 0.5**		
13	M	SV	21.6	72	158	-2.81	56	22.49	0.17	12	65700	15.6	792	1	1.1	I2G/I172N	B
14	F	SV	24.6	4	159	-1.03	56	22.2	0.24	15	131400	24.2	3225	4.64	3.1	I2G/I172N	B
15	F	SV	24.6	6	156	-1.53	48	19.7	-0.73	27	206955	24.1	3900	7.57	3.8	I2G/I172N	B
16	F	SV	7.1	24	131.7	2.11	39.5	22.7	2.18	15	27922.5	5.1	756	1	0.97	I172N/P30L+ I2G+del8bp	B
17	F	SV	11.7	60	136	-1.64	37.5	20.5	0.68	15.9	38883.45	6.7	1280	1	0.97	P30L/P30L+ I2G+del8bp	B

18	F	SV	31	180	144	-3.56	46	22.2	-0.26	14.5	127020	16	1120	1	1	P30L/P30L+ I2G+del8bp	B
19	F	SV	28	48	158	-1.19	56	22.4	0.34	27	216810	24	1470	1	1	P30L/P30L+ I2G+del8bp	B
20	M	SV	11	2	147.5	0.48	50	23	1.63	15	60225	10.8	1970	2.8	1.3	I2G/I2G	B
21	F	SV	5.1	1	117.8	2.02	23	16.6	1.05	13.5	24637.5	5	2450	5.1	1.5	I2G/I172N	B
22	F	SV	4.1	7	94	-2.42	13.5	15.34	-0.51	13.7	13563	4	432	0.25	0.1	P30L/P30L+ I2G+del8bp	B
23	F	SV	21.6	3	158	-1.19	45	18	-1.62	15	114975	21.3	3230	7.4	3.9	I2G/I172N	B
24	F	SV	12.7	33	142	-1.66	33.5	16.7	-0.93	17	106215	9.9	2250	2.3	1.2	del/I2G	B
25	F	SV	6.2	2	132	3.82	27.5	15.8	0.17	10	21900	6	1340	1.3	1.25	I2G/I2G	B
26	F	SV	9.4	52	129	-1.22	30	18	0.6	17	31025	5.1	948	1	0.9	I172N/I172N	B
27	F	SV	15.8	128	145	-3.28	57.5	27.3	1.45	15	27375	5.2	1917	1.44	1.43	I172N/I172N	B
X+/-SD (SV)	**2 M/ 13 F**	**15 SV**	**15.6+/- 8.9 ***	**41.1 +/- 52.8 ****	**140.5 +/- 18**	**-0.9 +/- 2.1**	**41.3 +/- 13.5**	**20.1 +/- 3.4**	**0.3 +/- 1.0**	**16.2 +/- 4.7 *****	**80973.8 +/- 66871.0**	**12.2 +/- 7.9**	**1805.3 +/- 1031.9**	**2.5 +/- 2.4**	**1.6 +/- 1.1**		
X +/- SD (SW,SV)	**7 M / 20 F**	**12 SW/ 15 SV**	**12.9 +/- 7.6**	**23.4 +/- 43.6**	**136.2 +/- 19.5**	**-0.8 +/- 1.8**	**37.9 +/- 13.4**	**19.6 +/- 3.3**	**0.4 +/_ 1.1**	**18.5 +/- 5.5**	**78598.9 +/- 55644.9**	**10.9 +/- 6.5**	**1298.6 +/- 982.7**	**2.0 +/- 1.9**	**1.3 +/- 0.9**		

Pat. no. – patient number; F – female; M – male; SW – salt wasting; SV – simple virilizing; yrs – years; mo – months; HC – hydrocortisone; cHC – cumulative durat. treat. – duration of treatment; UNL – upper normal limit

* p = 0.028; ** p = 0.01: *** p = 0.009

Table 1b:

Characteristics of the control group

Control No.	Gender	Age (yrs)	Height (cm)	Height SDS	Weight (kg)	BMI	BMI-Z score
1	F	11.5	145	-0.27	37	17.6	-0.07
2	M	12.0	139.5	-1.38	37.5	19.2	0.5
3	M	13.3	159	0.05	49	19.4	0.3
4	F	16.9	156	-1.48	47	19.3	-0.57
5	M	8.0	138.5	1.65	42	21.9	1.99
6	M	10.8	149	1.15	38	17.1	0.45
7	M	8.2	124.5	-0.8	25	16.1	0.17
8	F	11.0	143	-0.1	47	23.0	1.46
9	F	7.5	114.5	-1.63	29	22.3	2.05
10	F	6.1	118	0.53	21	15.1	-0.11
11	F	4.0	106	0.8	16	14.2	-1.02
12	F	4.3	96	-1.88	15	16.3	0.75
13	M	21.5	160	-2.5	58	22.65	0.19
14	F	24.2	159	-1.03	55	24.35	0.09
15	F	24.5	158	-1.2	50	20.0	-0.62
16	F	7.3	130	1.74	35	20.7	1.79
17	F	11.5	138	-1.33	38.5	20.2	0.79
18	F	31.3	148	-2.88	48	21.9	0.15
19	F	27.9	159.5	-0.94	57.5	22.6	0.5
20	M	11.3	149	0.71	50	22.5	1.47
21	F	5.0	116.5	1.7	22	16.3	0.8
22	F	4.1	94	-2.42	14	15.8	0.44
23	F	21.5	160	-0.86	48	18.75	-1.22
24	F	12.5	145	-1.21	34.5	16.4	-0.97
25	F	6.5	125	2.18	25	16	0.41
26	F	9.4	131	-0.85	32	18.6	0.82
27	F	15.6	150	-2.39	60.5	26.8	1.41
X+/-SD	**7 M/20 F**	**12.8 +/- 7.6**	**137.5 +/-19.8**	**-0.5 +/- 1.4**	**38.2 +/- 13.8**	**19.4 +/- 3.2**	**0.4 +/- 0.9**