

Table 1. Summary of clinical information of women followed with a diagnosis of NCCAH

Patient characteristics:	N or mean	% or IQR
Index case	23	79.3
Age at first symptom (years, mean \pm SD)	15.8	8
Number of symptoms, median (IQR)	2	1 to 3
Most common manifestations:	N	%
Hirsutism	20	69
Oligomenorrhea	14	48.3
Infertility	4	13.8
Accelerated linear growth	4	13.8
Acne	3	10.3
Alopecia	3	10.3
Precocious puberty	3	10.3
Hormonal work-up at diagnosis:	Availability, n (%)	Concentration (nmol/l), median (IQR)
Basal 17 OH-P	22 (75.9)	14 (5.75 to 41)
ACTH stimulated 17 OH-P	8 (27.6)	142 (38.5 to 177.75)
Pharmacological treatment #	N	%
None	38	12.1
Corticosteroids:	233	74.2
Dexamethasone	188	59.8
Hydrocortisone	44	14
Prednisone	1	0.3
Other treatments, alone or a combination:	43	13.7
Oral contraceptives	27	8.5
Spirolactone	15	4.8
Flutamide	1	0.31
Genetic analysis:	N	%
No mutations	3	10.3
Heterozygous, mild mutation	8	27.6
Homozygous, mild mutation	9	31
Compound heterozygous, mild/severe	9	31
Most frequent mutations:	N ‡	%
Mild exon 7, Val281Leu	28	62
Mild exon 10, Pro453Ser	6	11.1
Deletion	5	8.8
Severe exon 10, R483P	3	6.6
Severe IVS2, 290-13 C>G	2	4.4
Severe exon 7, His282Tyr	1	2.2

IQR: Interquartile range; # in 314 follow-up visits; ‡One patient had 3 mutations.