

Table 1. Diagnostic and clinical features of the four affected siblings

	Sibling 1	Sibling 2	Sibling 3	Sibling 4
Karyotype	46XX	46XX	46XY	46XX
Guthrie TSH (mU/L)	213	-	188	-
Venous TSH on recall (mU/L)	310.2	-	248.3	-
Venous FT4 on recall (pmol/L)	<8	-	17.6	-
Basal 17-OHP at diagnosis (nmol/L)	276	10.1	302	49
Peak of 17-OHP 30 min post-Synacthen (nmol/L)	-	71.9	-	144
Age at diagnosis of CAH	Day 3	15.5 yrs	Day 3	9.8 yrs
Presenting feature of CAH	Virilised genitalia (Prader 3)	Asymptomatic	Absent (positive elective screening)	Pubarche, increased growth rate, advanced bone age
Complications during follow-up	Precocious puberty Short stature Polycystic ovarian disease Right adrenal adenoma	-	Precocious puberty Short stature	-
Additional features	Learning difficulties	-	Developmental delay	-
Clinical form of CAH	Salt-wasting	Non-classical (cryptic)	Salt-wasting	Non-classical

