

Conclusions

Early treatment with locally applied DHT may have a significant effect on penile growth in 17 β -HSD deficiency.

Introduction

The child was born with ambiguous genitalia at full term.

It is the parent's first child. The mother had been treated for hyperprolaktinemia (Bromocriptin). She had also been investigated for infertility.

In early pregnancy, she was treated with Duphason, (luteal phase dydrogesterone).

The first evaluation of the child was done at 3 days of age due to suspicion of DSD.

Clinical findings:

Prominent labiae/bifid scrotum

Palpable gonads

Phallos/genital tubercle 1.2 cm

No rugae

No increased pigmentation

Vaginal opening?

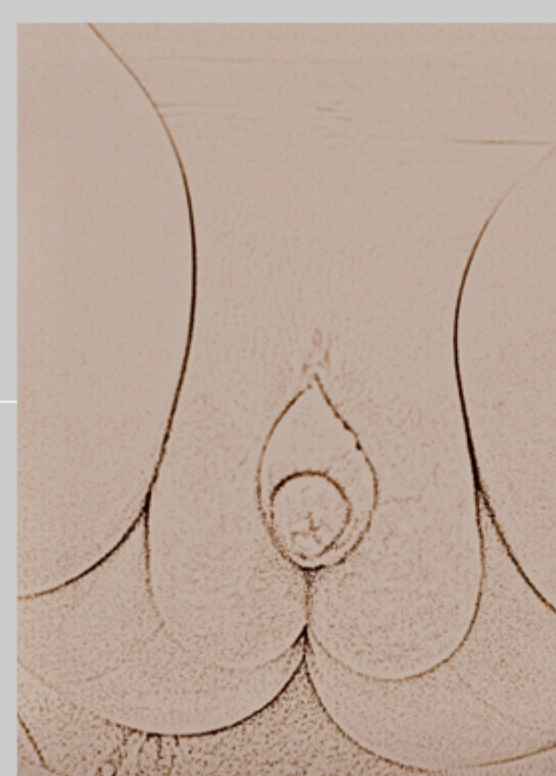
Methods

The penis measured 12 mm at 5 days of age. Local treatment with 2,5% DHT-gel, 0,2 ml daily was started and continued for 8 weeks all together, until the age of 4 months.

Results

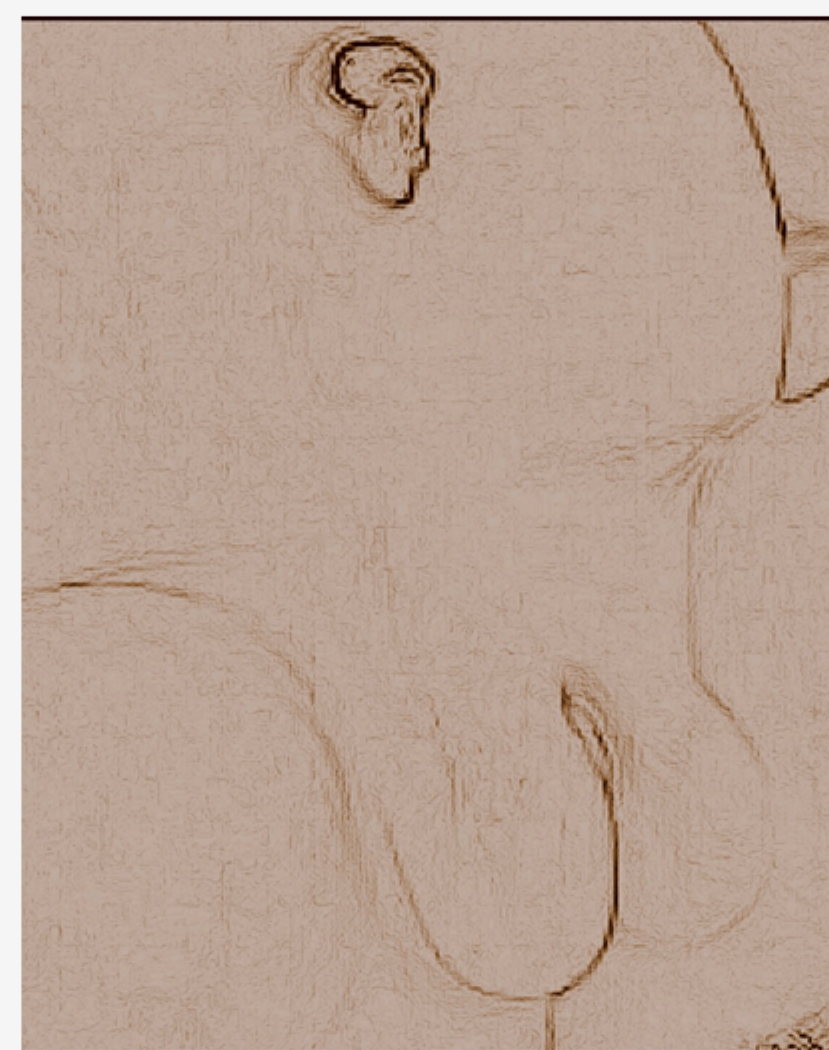
The phallus grew in size to 2,1 cm after hCG stimulation test due to increase in to DHT levels via the backdoor pathway.

After 2 months of therapy with locally applied DHT, the penis had grown to a length of 35 mm and a width of 12 mm. DHT increased in serum to 735 ng/dl.



The path to diagnosis

At 3 days of age



First investigation results

BW/BL	3850 g/52 cm
Karyotype	46, XY
Testosterone	2.5 nmol/L
DHT	<0.1 nmol/L
17-OH-progesterone	1.5 nmol/L
Cortisol	300 nmol/L
Androstenedione	0.7 nmol/L (< 1,8)
DHEAS	6.9 umol/L (2,9 – 16)
AMH	94 ug/L
LH/FSH	0.7/0.5 U/L
Ultrasound	No uterus
Laparscopy	Normal testes

Stimulation test hCG 500 U, 4 d

Testosterone	2.1-3.0 nmol/L
Androstenedione	0.7 – 1.8 nmol/L
DHEAS	2.7 – 3.3 umol/L
T/Androstenedione	1.3
Growth of phallus	2.1 cm

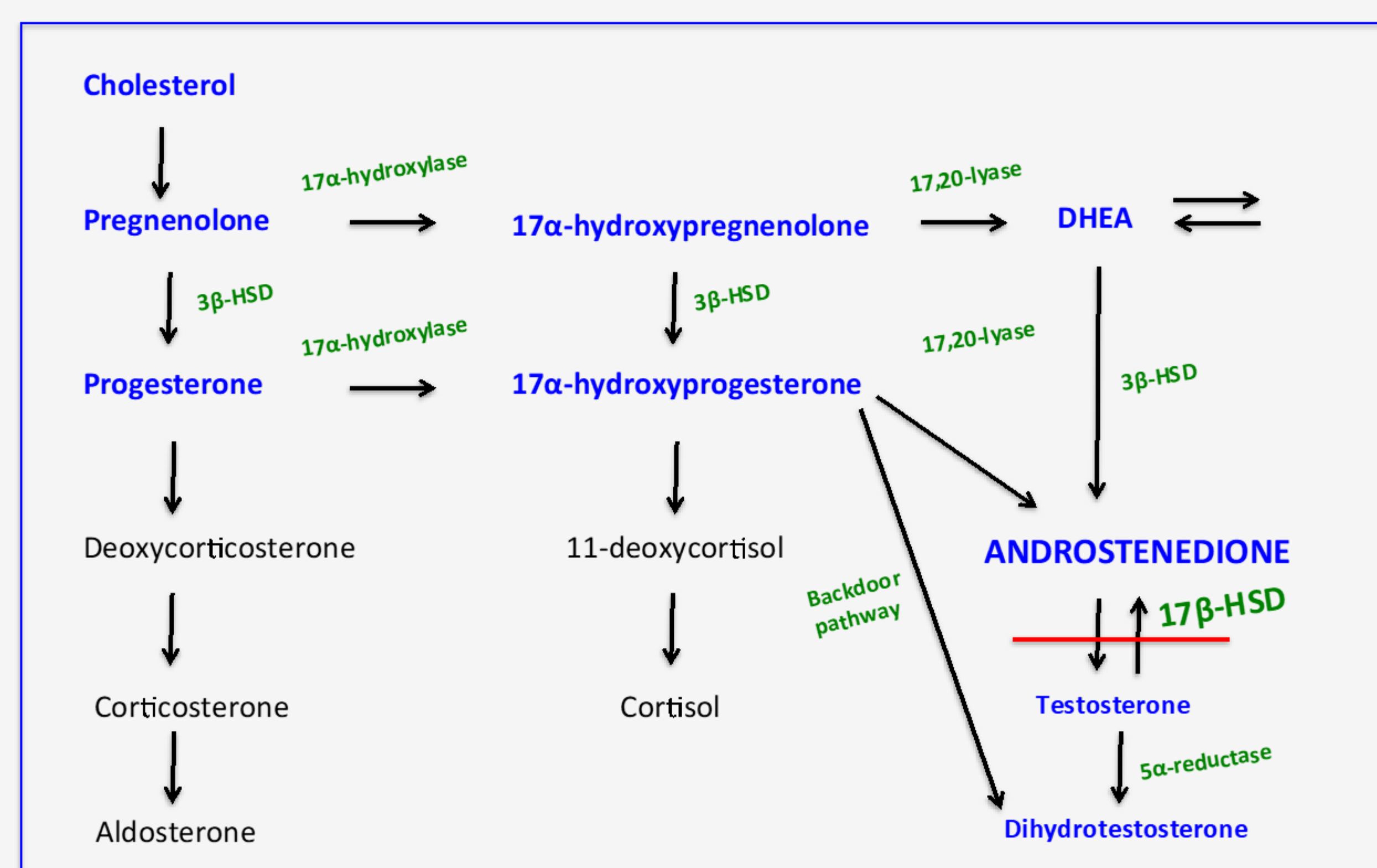
Additional analysis with MS/MS

Progesterone	0.19 ng/ml	0.09
11 deoxycorticosterone	0.03 ng/ml	0.07
Corticosterone	0.4 ng/ml	0.7
Aldosterone	0.35 ng/ml	0.3
17 – OH progesterone	1.15 ng/ml	0.68
11-DOC	0.55 ng/ml	0.35
Cortisol	40 ng/ml	24
Cortisone	38 ng/ml	47
Androstenedione	35 ng/dl (1.22 nmol/L)	41 (1.4 nmol/L)
Testosterone	21 ng/dl (0.7 nmol/L)	63 (2.2 nmol/L)
DHT	14 ng/dl (0,48 nmol/L)	38 (1.3 nmol/L)

Second hCG stimulation

Increased Androstenedione
Low basal Testosterone
Insufficient increase

17 β -HSD deficiency



17 beta-HSD mutations
c.201 G>C, c.506 T>G
p. Glu7Asp, p.Leu169Arg
These were unknown, but resulted in changes in evolutionary conserved aminoacids. The mutations were considered pathogenic.