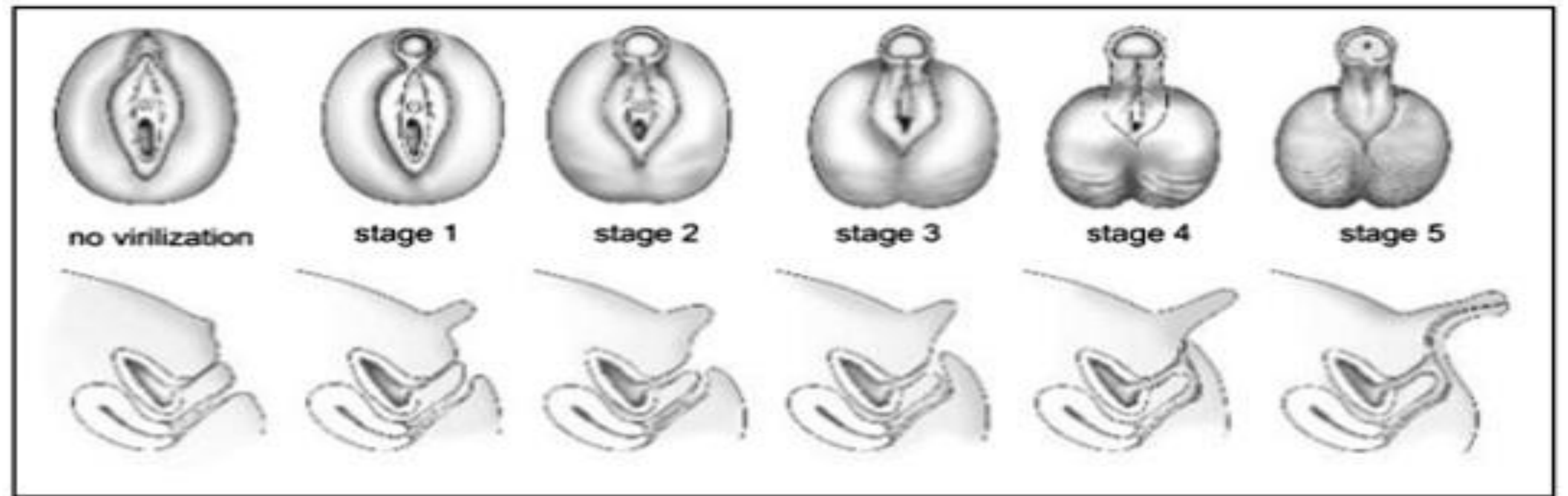


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We report a case of a female newborn with DSD, due to Congenital Adrenal Hyperplasia (CAH).

The mother, affected by hypothyroidism and autoimmune hepatitis, was treated during the whole pregnancy with levothyroxine and prednisone (5 mg/day).

At birth mild ambiguous genitalia (Prader stages I-II) were noticed and the baby was admitted to neonatology department for clinical assessment.



At 3 days of life				At 28 days of life					
17-OH Progesterone	510 ng/ml	Hydrocortisone	30 mg/m <sup>2</sup>	Sodium	131 mmol/l	Fludrocortisone	0.15 mg/die		
ACTH	329 pg/ml			Potassium	5.9 mmol/l				
Cortisol	202 nmol/l			Aldosterone	88.5 ng/dl			NaCl	2 gr/die
Testosterone	9 ng/ml			Renin	>500 uUI/ml				

## ABDOMEN ULTRASOUND

Normal uterus (18.5 x 9.4 mm; cervix 10.2 mm) and ovaries (R 11.5x 4.9 mm; L 9.9 x 5.8 mm)

## GENETIC ANALYSIS

Karyotype: 46, XX

Molecular analysis SRY: negative

Molecular analysis CYP21: **[655 I2 Splice (C>G) intron2; R356 W (exone 8)]**

At 4 months of life									
17-OH Progesterone	1.1 ng/ml	Hydrocortisone	15 mg/m <sup>2</sup>	Sodium	136 mmol/l	Fludrocortisone	0.15 mg/die		
ACTH	6.1 pg/ml			Potassium	4.5 mmol/l				
Cortisol	336 nmol/l			Aldosterone	19.3 ng/dl			NaCl	2 gr/die
Testosterone	0.01 ng/ml			Renin	413 uUI/ml				

In CAH deficient fetal cortisol production leads to overproduction of ACTH, stimulating the disordered fetal adrenal to produce excess androgens, virilizing female fetuses at 7-12 weeks gestation; however development of the ovaries, uterus and fallopian tubes remains normal.

Supraphysiological doses of exogenous steroids are required to suppress androgen excess and it was shown that prenatal dexamethasone treatment of fetuses at risk of congenital adrenal hyperplasia can prevent virilization of a female fetus. This treatment has since been offered at the dose of 20 mcg/kg maternal body weight per day, to avoid the need for feminizing genital reconstructive surgery and the risk of complications, and remains controversial. However, this treatment has shown a success rates of 85%. The long-term effects of steroids in early pregnancy remain unclear, with potential detrimental effects on fetal programming, brain function and congenital anomalies.

In this case, although the baby has a severe form of salt-wasting CAH, at birth she has minimal virilization (Prader stages I-II). We hypothesize that maternal therapy with prednisone at low dose since the beginning of the gestation, partially prevented virilization of the child genitalia.