Complete virilisation without salt wasting in a 7 year-old Haitian child with congenital adrenal hyperplasia

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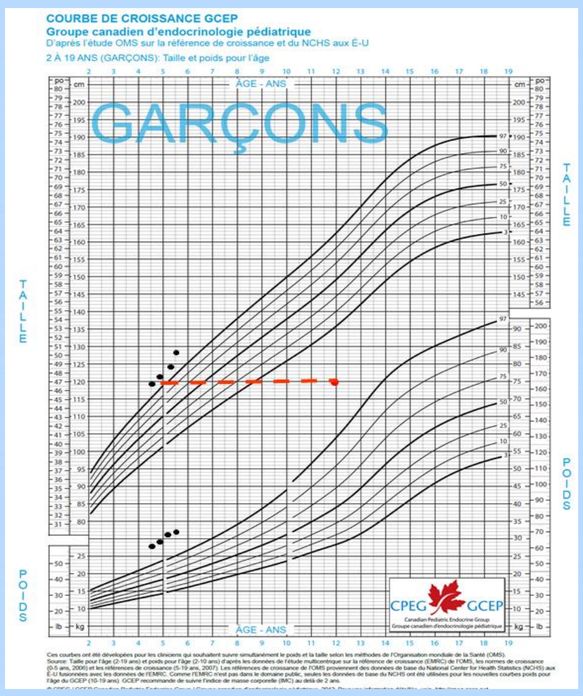
BACKGROUND

Genetic females with congenital adrenal hyperplasia (CAH) from severe 21-hydroxylase deficiency may be declared at birth as cryptorchid boys. Neonatal salt wasting leads to early reassignment but in its absence, the condition may go unrecognized.

HISTORY

A 5 year-old child presented with sexual precocity since age 4 years 8 months and underwent clinical assessment and diagnostic evaluations. The past medical history was notable for a term newborn with non-palpable gonads but normally formed penis and penile urethra. He was declared a male with cryptorchidism.

EXAMINATION



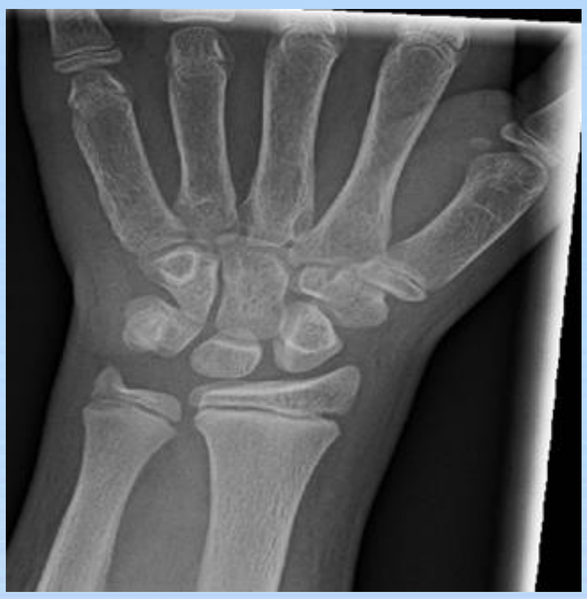
Height and weight >97percentile (ht +2.3 SDS)

The child appeared older than his chronological age. His build was muscular and his voice was deep. There was no facial hair, but increased terminal hair over the extremities. Adam's apple was visible, thyroid was not enlarged. No breast development. Phallus was of adult male appearance, length 9 cm, mid-shaft diameter 4.5 cm. Gonads were not palpable. Pubic hair was Tanner stage 3, and axillary hair was 2+ bilaterally.

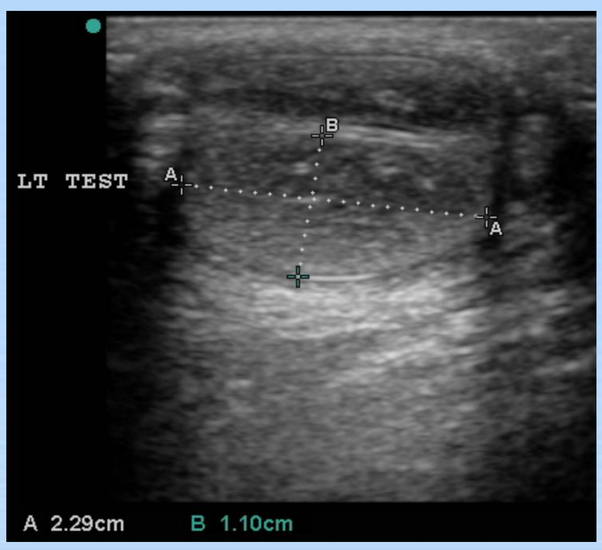
INVESTIGATIONS

Lab Test	Values w/Units	Normal range
17-OH-progesterone	7440 ng/dl	7-69
DHEAS	139.9 mcg/dL	5.0-40.0
Testosterone	224 ng/dL	
LH	< 0.1 mIU/L	
FSH	0.49 mIU/L	

NB: Karyotype non available in Haiti



Bone age (unfortunately obtained as a wrist X-ray) at age 4 years 8 months was 13 years 6 months



On ultrasound (at age 4 years 8 months), no uterus was identified and a gonad was described as a testicle.

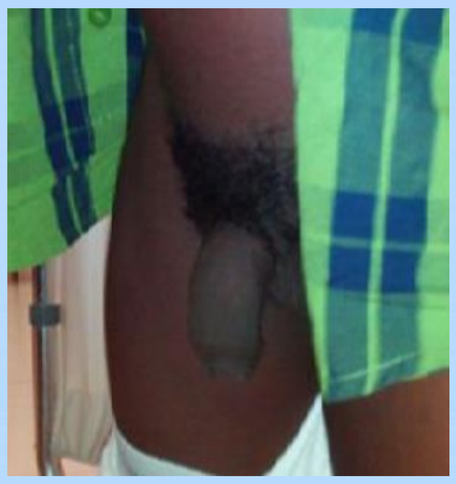
Head CT without contrast was normal, no evidence for brain tumor,the sella was normal in configuration

DIAGNOSIS & TREATMENT

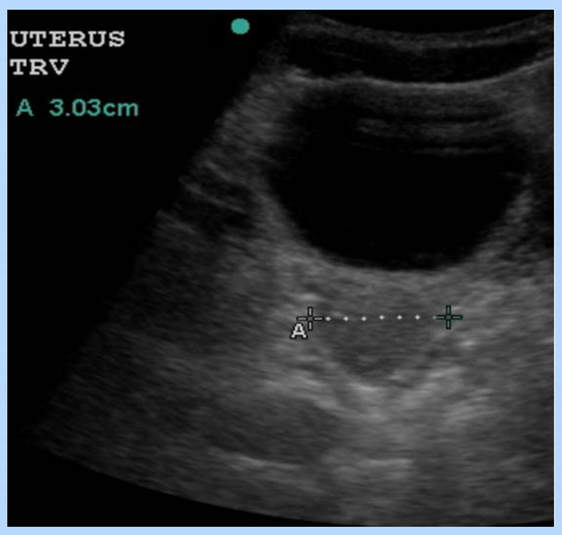
Diagnosis: Simple virilising congenital adrenal hyperplasia (CAH).
Treatment: Prednisone (5 mg/m2/day)

FOLLOW-UP

Central precocious puberty as evidenced by LH and FSH increase developed at age 5 years 3 months. GnRH therapy was unavailable (despite international efforts). Thelarche was noted by age 5 years 9 months, and was interpreted as gynecomastia. Monthly urethral bleeding lasting 3-4 days ensued at age 6 years 9 months, prompting repeat pelvic ultrasound that showed a pubertal uterus and ovaries.



Appearance of genitalia at age 6 years 9 months



Presence of uterus and ovaries on ultrasound age 6 years 9 months (1.9 years after treatment initiation)

Upon informing the mother, she stated that she had known all along that her child was 'female inside and male outside'. She did not consider sex reassignment. Prednisone was discontinued in an attempt to stop menstruation and reduce breast development.

DISCUSSION

Term newborns with a male phallus but non-palpable testes should undergo pelvic ultrasound. While the neonatal uterus, stimulated by maternal estrogens, should be visible, it may become hypoplastic from gonadotropin suppression and thus missed in older genetic females with CAH. When karyotype analysis is unavailable, the search for a Barr body should be considered; indeed, our observation has led to this technique being introduced in Haiti. Late sex reassignment is often unacceptable to the parents and needs to be viewed in the cultural context. Long-term follow-up of this patient to adulthood with a focus on psychosexual development is planned.

CONCLUSION

Diagnostic and treatment options for CAH in resource limited settings remain suboptimal. To make a correct diagnosis and provide appropriate care in such settings, close follow-up and reassessment are essential.

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