Pseudohermaphroditism Due to Congenital Adrenal Hyperplasia – A Case Report.

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Background: Congenital adrenal hyperplasia [CAH] due to 21-hydroxylase deficiency is the most common cause of ambiguous genitalia. The hypertensive form is due to 11B- hydroxylase deficiency. The purpose of this report is to apprise clinicians of the presentation of this form of CAH.

CASE REPORT: A 17 – year-old female Nigerian presented with history of a 'male' phallus that was first noticed 3 years prior to presentation. Menarche was at age 14. There was no menstrual irregularity. She was apparently female at birth and was circumcised in early childhood. Her developmental milestones were normal. Her height was 1.60 m and weight was 46. 2kg. BMI was 18 kg/m2. Virilising features accompanied the presence of the phallus. Her breasts were small. Genital examination revealed clitoromegaly. The labia minora were hypo plastic. The vestibule with vaginal orifice and well – formed hymen were present. She was hypertensive with clinical features of hypertensive heart disease. Hormone profile showed elevated testosterone to 3.9ng / ml [0.2 – 0.8 = females]. Elevated Dhea-sulphate [17 ketosteroids] – 6, 400 ng/ml [700-3900, F]. The 17 OH progesterone was 3.71nmol/L [F-0.45 – 3.3]. Cortisol was 192.0 nmol/L [a.m – 171 – 536, p.m – 64 – 340]. Serum electrolytes were normal. Pelvic USS was normal. She was placed on glucocorticoids and lisinopril.

Conclusion: CAH is an uncommon but recognized cause of hypertension and should be considered in the differential diagnosis of young patients with hypertension and virilisation.