BRAZILIAN PATIENT 17 ALFA HYDROXYLASE DEFICIENÇY

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Mutations on CYP17 gene cause 17-hydroxylase deficiency (17OHD), a rare form of congenital adrenal hyperplasia (CAH).Individuals with 17OHD account for roughly 1% of all CAH, generally from consanguineous families. The features of complete 17OHD are hypertension, hypokalemia, and sexual infantilism in phenotypic females in 46, XX and 46, XY subjects. The lack of adrenal 17_-hydroxylase activity drives massive overproduction of the 17-deoxysteroids: 11-deoxycorticosterone (DOC) and corticosterone, which are the mineralocorticoids that cause hypertension and hypokalemia in 17OHD. Concomitant lack of gonadal 17, 20-lyase activity precludes sex steroid production and hence the development of the male phenotype in utero or of secondary sexual characteristics at puberty. We describe a 16 year's old, female raised patient with lack of pubertal signs and primary amenorrhea. Her parents were consanguineous. The family had Portuguese and Italian ancestry. Her skin was darker than her parents. BP: 150/115mmHg, height: 1,51 cm weight= 36,5 Kg, arm span = 163 cm Tanner stage 1: M1P1, infantile external genitalia. Exams: Karyotype : 46XY: Normal Boy . Abdominal-pelvic US: uterus was not seen and so were the gonadas. Cortisol 8h:1,84 ug/dl(6,2 - 19,4),17alfaOHP:0,18 pg/ml,(31 - 2,17),estradiol:<5 pg/ml(7,5 - 42,5),LH:60,1 mUI/ml, FSH:46,6mUI/ml(1,5 -12,4),T Testosterone:<0,1ng/ml(3,5-25),ACTH:476pg/ml(10–52) Androstenedione <0,11ng/dl (0,5 - 3,5), K:3,2meq/l(3,5-5,1),Co2:30meq/l(22-30), Aldosterone: 29,01ng/dl (supine=2,94 to 16,16) , PRA:<0,20ng/ml/h(0,98 to 4,18). XR-Bone Age: CA: 15 years and 9 monthsBA: 11 years old.SD: 9, 23 months. Basal and ACTH –stimulated adrenal steroid values: (VR):Cortisol (ug/dl) 0, 0,(6-25/18-42),Cortisona (ng/dl) 0, 0, (800 -3500), Deoxycorticosterone (DOC) ng/d l18.377, 22030, (0,1-0,5/1,7-4,8).