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CONGENITAL ADRENAL HYPERPLASIA DUE TO 3 beta-HYDROXYSTEROID DEHYDROGENASE (3bHSD) DEFICIENCY.

An eight weeks old boy, first child to nonconsanguineous parents of Swedish extraction, presented with severe dehydration and salt loss. Plasma Na 109 and plasma K 8.7mval/1. He had a phallus 1,5 cm in length, meatus in the peniscrotal junction and a bifid scrotum with gonads on both sides. Normal blood pressure. After rehydration and hydrocortison i.v. the boy was substituted with 10 mg hydrocortison and 0.05 mg fludrocortison daily. Steroids in 24 hours collection of urine were analysed by capillary gas chromatography mass spectrometry. More than 90% of the steroids were 3beta-5-en steroids mainly androst-5-en-3,17,20-triol, 16 alphahydroxy-DHA and pregn-5-en3,17,20-triol. Plasma levels of 17-OH-P, DHA, androstendion and aldosteron were analysed by RIA. During the salt crisis aldosteron was 1650 pmol/L and decreased to 775 during substitution. During ACTH stimulation 17 OH-P and androstendion were 7 and 2,4 nmol/L respectively which is lower than usually seen in 21 hydroxylasedeficient children in poor control.

We find the case consistent with 3bHSD deficiency of partial but still salt loosing type.

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Melatonin day-night rhythms in subjects with Turner Syndrome

The day-night rhythms of serum melatonin(MEL) were investigated in 10 subjects with Turner syndrome,aged 2-17 years.Five had karyotype 45 XO,4 had 45 XO/46 XX and one 45 XO/46XX/47XXX mosaicism.Serum MEL concentrations were determined according to the RIA procedure of Rollag and Niswander,using the antibody kindly donated by Dr. Niswander.Serum LH,FSH and PRL were measured by routine RIAs.MEL patterns were abnormal in all subjects. 1.Daytime values were higher than normal (135+88 vs. 55+26,$p < 0.05$), as already observed in a previous study(Gupta et al,1983).2.Night peak MEL values were in the prepubertal range irrespective of the age and developmental stage of the subject. 3.Desynchronized ultradian rhythms were present in all subjects except one,a 17-yrs. old XO/XX girl who had spontaneous menstruations. 4.No relationship between MEL serum levels and gonadotrophin concentrations were found. 5.In some of the patients the night elevation in serum PRL levels was very pronounced. These data illustrate the principle that chromosomal abnormality may be antecedent to the development of aberrations in relation to the pineal biorhythm.However,this is simply a hypothesis which should be examined further.

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The effect of L-DOPA on basal LH, FSH and Prolactin (Pr) secretion in subjects with Turner syndrome.

The dopaminergic tuberoinfundibular system has been shown to play a role in the regulation of the pituitary secretion of LH an Pr in experimental animals. In humans,DA and DA agonists have an inhibitory effect on LH and Pr release which differs in the two sexes and in women in the different phases of the menstrual cycle. We examined the effect of L-DOPA in the dose used for GH stimulation on the LH,FSH and Pr levels in 9 subjects with Turner syndrome, who had never received estrogens. We found the following (mean ± SD).

	0	30	60	90	120
FSH mIU/ml	121± 27	114±33	117±30	110±32	112±33
LH mIU/ml	30± 13	28±14	26±11	23± 8	25±13
Pr µU/ml	281±110	154±60	80±16	59±20	44±25

Although there is an intense suppressive effect of L-DOPA on plasma Pr concentration there is no synchronous significant decrease in LH. Thus synchronism in Pr and LH suppression caused by adrenergic stimuli in other experimental settings was not detected in the present one. This finding may indicate that there exists a differential, dose related, sensitivity of LH and Pr to the adrenergic stimulus. It may however indicate that the synchronous suppression of LH and Pr by adrenergic stimulus represents age or estrogen-related maturational event.

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Hypodipsia, osmoreceptor dysfunction, early puberty and abnormal behaviour in two boys

Two unrelated boys (A 13yrs B 18yrs) presenting with early puberty and episodes of aggressive behaviour were found to have hypernatraemia(Na 147-160 mmol/l, osmolality 298-323mosmol/kg) and hypodipsia. Plasma vasopressin(AVP) levels were in the range seen in diabetes insipidus(<2pmol/l), yet 24hr urinary volumes were < 1 litre and the maximal urinary osmolality 1232 in A and 950 in B. Plasma renin activity was elevated (>2000mg/AI/l/hr)and aldosterone normal. Excretion of a water load(20ml/kg)was delayed(minimal urinary osmolality A-166 at 300mins,B-392 at 360mins)but plasma renin and aldosterone fell with increased naturesis. An infusion of 4.5% saline produced a rise in AVP(2.8pmol/l)in A but not in B. Insulin and hypotension resulted in release of AVP. (A 6.1pmol/l, B 4.8 pmol/l) suggesting a selective defect of osmoreceptor function. Hyperprolactinaemia and an exaggerated PRL response to TRH were also noted but no intracranial lesion was demonstrable on CT scan. These boys appear to have a hypothalamic syndrome with early puberty, hyperprolactinaemia, hypodipsia and osmoreceptor dysfunction which may be associated with aggressive behaviour.

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Single vs. repeated doses of hCG in the differential diagnosis of hypogonadotropic hypogonadism.

Serum testosterone (T), 17α -hydroxyprogesterone (17OHP) and 17β-estradiol (E2) were determined in 10 prepubertal (Group 0) and 10 pubertal (Group P) hypogonadotropic boys given 5000IU/1.7m^2 of hCG i.m. on d 0, 4, 7 and 10, before each injection and 4d after the last one. The results were compared with those of appropriate controls, 16 prepubertal (incomplete testicular descent) and 6 pubertal (constitutional delay of puberty). After the first injection T levels increased to 2.0 and 4.6 nmol/l, and then progressively to 5.8 and 11.2 nmol/l, for groups 0 and P, respectively. E2 levels increased slightly in group P only, by the end of the stimulation ($p<0.05$). 17OHP increased gradually ($p<0.05$ and <0.01 for groups 0 and P at the end). Basal T levels did not differ from controls in group 0, but were subnormal in group P ($p<0.001$).In both groups, all stimulated T levels were subnormal ($p<0.01$ or <0.001). With repeated doses of hCG the separation from the controls improved only for group P. Difference in E2 response from controls appeared in puberty. In the controls only group P showed increase in E2 ($p<0.001$). For diagnostic hCG test in prepuberty, a single dose of hCG with determination of T levels on d 4 is the optimal protocol. In pubertal boys, if the basal T levels are inconclusive, repeated doses of hCG are required with determination of both T and E2.

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Bromocriptine treatment in tall adolescents: two years of clinical experience.

29 adolescents referred for excessive height prediction (HP) (9 boys with HP > 195 cm, 20 girls with HP > 180 cm) were treated for 9 to 15 months with bromocriptine (5 to 7.5 mg/day). Minor and transient side-effects were observed in 20% of the subjects at the beginning of the treatment. Treatment had to be stopped in one boy complaining of asthenia and headache. Puberty developed normally, 16 girls experienced menarche during treatment and one continued regular menses. Bromocriptine treatment induced: 1) a significant decrease ($p < 0.001$) in growth velocity from mean ± SEM 8.6 ± 0.6 to 5.2 ± 1.0 cm/year in boys and from 7.1 ± 0.3 to 4.6 ± 0.4 cm/year in girls; 2) a twofold mean increase in skeletal maturation rate. Adult height prediction was reduced significantly from 202 ± 1.6 cm to 195.4 ± 1.2 cm in boys, and from 184 ± 0.7 cm to 179.8 ± 0.8 cm in girls. These results confirm our previous report suggesting that bromocriptine is a valuable alternative to sex steroid treatment in order to limit the final height in excessively tall adolescents.