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Congenital nephrosis cause of neonatal hypothyroidism

Nephrotic syndrome is known to induce significant changes in thyroid economy due to the increased thyroid hormones and thyronine-binding proteins urinary loss,without influences on euthyroid status(Gavin et al.JCEM,1978).In congenital nephrosis(CN),proteinuria starts in utero and is resistant to any treatment.CN is not generally reported as a cause of neonatal hypothyroidism;neverthless we observed biochemical and clinical evidence of hypothyroidism in 3 infants suffering from CN.In serum,taken from A.A.on the 5th day of life(proteinuria > 200 mg/hr/m^2),TSH was 44 µU/ml,on the 15th day,TSH was 27 µU/ml,T4,4.6 µg/dl,T3,75 ng/dl, FT4,6.5 pg/ml,FT3,4.0pg/ml and TBG,11 µg/ml;Béclard nucleus was absent and a severe somatic and psycomotor retardation became evident during the first months of life.At the time of the first observation(5 months) showed clinical and biochemical features of hypothyroidism(T4,0.6 and 1.4 µg/dl, T3,50 and 85 ng/dl,respectively).In the three infants thyroid replacement therapy,started from 3 to 6 months,produced a meaningful improvement of clinical and biochemical features(A.A.12 months,TSH 12, T4 9,T3 200,TBG 18;P.D.10 months,TSH 25,T4 3.2,T3 190,TBG 10; V.D.9 months,TSH 13,T4 4.4,T3 150,TBG 12)in spite of persistent and marked proteinuria and thyroxinuria.

60 SLS DROP*,GJ BRUINING*,HKA VISSER,WG SIPPEL* Dept Paed Erasmus Univ Rotterdam;Div.Paed.Endocr.Univ. Munich. Prolonged galactorrhoea in a girl with isosexual precocious puberty due to an adrenal tumour. From a 6 yrs old girl with isosexual precocious puberty (Tanner M_4P_4) treated with cyproterone-acetate (CA) for 6 mo a large cystic adrenocarcinoma was removed in toto. Capsular invasion was present but no metastasis found. Radiation (3000R) was given and CA therapy continued. An x-sella was normal. Following surgery, severe galactorrhoea developed, subsiding under bromocriptine (BC) given from 3-6 mo post-op. Cystic tumour fluid (CTF), pre-and serial post-op plasma samples were analysed by multisteroid radioimmunoassay. In comparison to adult female plasma levels, in CTF estrogens were elevated 1000x; 17 OH progesterone, compound S and desoxycorticosterone 100x; DHEA(S), androstenedione and testosterone 10x. In pre-op plasma samples these parameters (both basal and after ACTH stimulation) were highly elevated and post-operatively returned to normal. Prolactin levels, however, remained high but could be fully suppressed by BC. The prolonged galactorrhoea is most likely the result of excessive estrogen "priming", whereas the hyperprolactinaemic effect of CA therapy remains speculative.

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ACTH stimulation studies in two children with isolated hypoaldosteronism and their relatives.

In two children with isolated congenital hyperreninaemic hypoaldosteronism, plasma levels of aldosterone (Aldo), corticosterone (B), desoxycorticosterone (DOC), 18-OH-B and 18-OH-DOC were measured before and after an i.v. bolus of 0.25 mg Synacthen®.

Results: (ng/ml)			nml range:
pat.1:	basal/max.	p.2 basal/max.	basal/max.
DOC	0,23/0,82	1,27 /3,32	0,03-0,2 /0,08-0,5
18-OH-DOC	1,0/4,8	0,8 /3,8	0,04-0,25/ -1,0
B	11,9 /15,3	7,4 /23,7	0,05-5,0 /1,0 -12,2
18-OH-B	8,3 /10,2	0,6 /1,3	0,2 -0,5 /2 -1,2
Aldo	0,012/0,056	0.004/0.017	0,14-0,9 /0,2 -1,2

An 18-dehydrogenase deficiency is clearly demonstrated in pat.1. The normoreninaemic parents (no consanguinity) had plasma values consistent with heterozygosity. The consanguine parents of pat.2. had normal Aldo levels but one asymptomatic sib had steroid levels and plasma renin activity similar to patient values. An impairment of the 18-oxidation system possibly limited to the zona glomerulosa could be postulated.

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Urinary excretion of iodine and thyroid hormones in neonates.

Total iodine and thyroid hormones were studied longitudinally at first and fifth day in the serum and urine of 9 brest feeding normal newborns.Thyroxine(T_4),triiodothyronine(T_3),T_4 binding globulin and thyrotropin(TSH)were determined by radioimmunoassay,total iodine by an automatic micromethod.Serum TSH was less than 5 µu/ml at the fifth day;T_4 and T_3 concentrations were in the range of the normal newborns of the same age,TBG mean level was 21,6 mg/l the first day and increased to 31,6 mg/l the fifth day.The difference assayed by paired t test was significant($p<0.001$).The mean of total iodine content of the first urination was 0.94 µg; the mean of the first 24 hours iodine excretion by urine was 1.7 µg and 5.7 the fifth day,corresponding to the level of iodine milk ingested.Mean T_4 excretion was 44 ng the first and 22 ng the fifth day,decreasing according to the serum concentrations during the same period.Mean T_3 excretion was 47.1 ng the first day and 93.4 ng the fifth.On weight body basis the fifth day T_3 values were similar to the quantity of T_3 excreted by 2-13 years children. The most interesting data arising from this study are:the rapid onset of the iodine excretion and the T_3 excretion as a characteristic of growth since the fifth day of life.

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An endocrine study of six patients with cerebral gigantism (Sotos' syndrome).

The auxological and clinical basis for the diagnosis will be shown. Basal prolactin levels were measured and a prolonged glucose tolerance test, an L-dopa test, a TRH test and an LH-RH test performed. Abnormalities were found in five of the six patients. These included high basal prolactin levels, a pre-diabetic blood sugar curve, inadequate suppression of growth hormone or a paradoxical rise in growth hormone in response to glucose, an excessive or continuing rise of TSH following TRH, an excessive response of prolactin to TRH, and, in one child, a pubertal response of LH and FSH to LH-RH. The endocrine abnormalities demonstrated, although not consistent, suggest possible hypothalamic dysfunction in cerebral gigantism, in which a variable aetiology is likely.

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Familial Athyreotic Cretinism.

Multiple athyreotic cretinism is described in a family of Turkish origin with frequent intermarriage. In 2 siblings aged 9 and 11 years, respectively, history revealed that the disease must have existed since birth. Both had marked ptosis of the eyelids but no other signs of hypothyreotic myopathy, the electromyogramm was normal. Two other sisters (20 and 23 years) were euthyroid and had no goiter. One of these sisters gave birth to a normal child, whereas the other sister - also married consanguineously - was delivered of 2 girls with congenital cretinism. In 3 of these 4 affected children the diagnosis of athyreosis was established by complete lack of 131-iodine retention (less than 3% after 3 days) measured with a whole body counter. The normal handling of 99-m Technetium-Pertechnetate by the salivary glands excluded a trapping defect. No microsomal and no thyroglobulin (Tg) antibodies (AB) could be found in these 3 subjects who came to registration at a later age. In contrast, the youngest girl who is in our control since birth had positive microsomal AB-titers which disappeared after 3 months. Her mother had highly positive microsomal AB titers but also no Tg AB. Whether the appearance of these antibodies plays a pathogenetic role remains unclear. No association to the HLA-A,B,C system could be demonstrated. The evaluation of the pedigree suggests an autosomal recessive inheritance of this rare disease.