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A FORM OF GLYCOGEN STORAGE DISEASE (GSD) WITH HYPERTENSION (HP). Marcos Kotoyan, Edwin T. Holloway, and Duncan R. MacMillan, (Spon by Billy Andrews) University of Louisville School of Medicine, Department of Pediatrics.

T.S. was a 5 wk. prem. W/F whose neonatal period was complicated by hypoglycemia and prolonged jaundice. She presented at 3 1/2 mos. with hepatomegaly, hypoglycemia, lactic acidosis, hyperuricemia, ↑cholesterol and triglycerides. She also had systolic HP, B.P. varying between 140-200 mm Hg. Glucagon stimulation caused paradoximal hypoglycemia. Light and electron microscopy of liver biopsy were consistent with GSD. Enzyme assays (courtesy of Dr. G. Hug) revealed the following: G-6-P'-tase 5.6 u moles P/min/g tissue (N = 4.7 ± 1.9); liver and muscle debrancher enzyme was present; liver phosphorylase was 12.0 u moles P/min/g tissue (N = 25.1 ± 6.5); active muscle phosphorylase was 9.71 (N = 47.7 ± 13.2) and total muscle phosphorylase was 53.5 (N = 78.0 ± 21.1); liver phosphorylase kinase was 0.067 n moles b⟶a/min/mg prot. (N = 0.175 ± 0.068). Catecholamines, VMA, HVA, 17-OH, 17-KS, Aldosterone, plasma renin, C_{cr}, IVP, and aortogram were all normal. At 11 mos. she developed generalized xanthomas; serum cholesterol was 906 mg% and triglycerides 3620 mg%. Clofibrate was started and a month later there was resolution of her xanthomas with fall in cholesterol to 259 mg% and triglycerides to 1624 mg%. Presently she is being maintained on propranolol, diazoxide, allopurinol, Lasix, clofibrate. In summary, our pt. has the clinical picture of Type I A or B GSD but the enzyme findings are suggestive of Type IX. The HP does not fit either type; it could be a coincidental finding, however a true association can not be ruled out at this time.

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DWARFISM DUE TO IMMUNOREACTIVE BUT BIOLOGICALLY INACTIVE GROWTH HORMONE. A. Avinoam Kowarski, Jill Schneider, Virginia V. Weldon, Ehud Ben-Galim and William H. Daughaday. The Johns Hopkins University School of Medicine, Baltimore, Maryland and Washington University School of Medicine, St. Louis, Missouri.

In the syndrome of familial dwarfism with high plasma immunoreactive growth hormone (GH)(Laron Syndrome), elevated levels of GH are associated with low levels of plasma somatomedin that do not increase following administration of GH. These patients do not respond to treatment with GH.

The two subjects of this report were three-year-old boys with dwarfism (height ages 1 3/12 and 1 6/12 years) and delayed bone ages (1 3/12 and 1 9/12 years). Both had normal GH response after stimulation associated with undetectable levels of somatomedin. However, unlike patients with Laron Syndrome, the two patients generated normal levels of somatomedin after intramuscular administration of GH. Treatment with GH (2 IU every other day) brought a significant increase in the growth rate of both patients. The growth rate of the first patient increased from 2 cm/year before treatment, to 16 cm/year on therapy. The growth rate of the second patient was 4.5 cm/year before treatment, and 11.0 cm/year while on treatment.

The two cases represent a new syndrome of dwarfism due to biologically inactive, immunoreactive GH. If erroneously diagnosed as having Laron Syndrome, these patients may be denied the benefit of treatment.

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SUSTAINED EFFECT OF HUMAN GROWTH HORMONE (hGH) THERAPY ON CHILDREN WITH INTRAUTERINE GROWTH RETARDATION (IUGR). Roberto Lanes, Leslie Plotnick, Peter A. Lee. Johns Hopkins University School of Medicine, Department of Pediatrics, Baltimore, Maryland.

Previous studies have not clarified the issue of whether hGH therapy can significantly increase the height of patients with IUGR. In order to determine whether the initial increase in growth rate shown by Foley (J. Pediatr. 84: 635, 1974) is sustained through subsequent treatment, 19 prepubertal IUGR patients (term gestation, height <43.7 cm, weight <2kg) were treated with hGH. Ten of them received a second treatment course.

Growth rates in cm/year were 4.8 ± 1.4 (mean ± SD) for the pre-treatment period (mean duration 14 months), 7.6 ± 2.3 for the first treatment period (mean 13.7 months), 4.2 ± 2.5 for the interval between treatments (mean 11.1 months), 5.9 ± 1.4 for the second treatment period (mean 13.7 months) and 4.3 ± 2.6 for the post-treatment period (mean 13.6 months). Growth rates for the two treatment periods were significantly greater than the pre-, interval between, and post-treatment rates. The SD below the mean in height increased significantly between the onset of treatment and the most recent measurement. Sixteen untreated IUGR patients followed for ≥ 5 years did not show this difference.

These data indicate that hGH has a sustained positive effect on increasing growth rates in children with IUGR, although the magnitude of the effect may decrease with further treatment. Furthermore, with the presently increased availability of hGH, therapy appears to be indicated in children with IUGR.

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BROMOCRYPTINE (CB-154) THERAPY IN CHILDHOOD ACROMEGALY Elmer S. Lightner, Jeremy S. Winter. University of Arizona, Health Sciences Center, Department of Pediatrics, Tucson and University of Manitoba, Winnipeg, Canada.

A 9 2/12 male with McCune-Albright Syndrome and acromegaly (length-178cm) had continued elevation of growth hormone(GH) and prolactin(Pr) despite surgery and radiation for a Pituitary Adenoma at 7 11/12 yrs. He perspired freely and constantly needed larger shoes. His continued rapid growth(>10cm/yr) and severe fibrous dysplasia led to recurrent pathological fractures of his femurs. A 5 mo. trial of CB-154 was initiated. The results of sequential resting GH and Pr concentrations and an oral glucose tolerance test (OGTT) were:

GH(ng/ml)		Pr(ng/ml)		OGTT mg%	
Pre CB-154	Post CB-154	Pre CB-154	Post CB-154	Pre	Post
14.6	7.5	210	<5	69	66
18.3	8.4	205	<5	122	130
15.1	8.9	226	<5	130	130
20.9	5.5	171	<5	160	84
17.7	9.1	178	<5	92	94
17.4	9.1	225	<5	71	72

CB-154 significantly lowered GH and Pr concentration, and improved the OGTT. While on CB-154 a 24 Urine excretion of hydroxyproline fell 10 fold; he perspired less, and his shoe size and length did not increase. However, left heel pad thickness did not change. No side effects from CB-154 were noted. CB-154 appears to be an effective supplementary form of therapy for acromegaly in childhood.

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Thyroid Antibodies and HL-A Antigens in Children with Thyroid Disease: McLAUGHLIN J, MACLAREN N*, KRUPP B, TAYLOR G, KIM C, HUANG S-W, Baltimore, MD

Antibodies to thyroid cytoplasm (T.C.A.), to thyroid nuclei (ANA) and to thyroglobulin (T.G.A.) were determined in 30 children with thyrotoxicosis (Tx), in 13 with euthyroid goiter (EG), in 9 with hypothyroidism and goiter (H.G.) and in 6 with hypothyroidism with no goiter (H). Histocompatibility (HL-A) antigens were also studied. The results (as percentages) were:

Patients	T.C.A.			T.G.A.		ANA
	1:1	1:10	>1:100	1:10	>1:20	
Controls	8	2	0	8	1	0
Tx	93	86	48	21	14	23
Tx (B8)	100	100	80	60	40	32
Tx (other)	100	100	50	0	0	11
EG	71	45	28	40	20	38
H.G.	86	29	29	60	40	30
H	17	17	0	0	0	16

Patients with Tx had increased HLA-B8 (45% of controls 25% $p < 0.05$) whereas the other thyroid patients did not. Siblings, one with Tx and one with juvenile diabetes were HLA identical (B8+) whereas an unaffected sibling shared none of the types.

Families of Tx had increased Tx, rheumatoid arthritis and S.L.E. In summary, Tx is strongly inherited and related to HLA-B8. Patients with B8 have more T.G.A. and higher titers of T.C.A., but show less tendency to remit. We speculate that Tx is an antibody mediated disease and thyroiditis a predominantly cell mediated disorder. Supported by NIH grants AM19286 and AM05745.

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NEUROENDOCRINE EVALUATION OF A PATIENT WITH KEARNS-SAYRE SYNDROME (KSS). Sharon L. Maby, Marilyn L. Cowger and H. Lawrence Vallet, Albany Medical College Department of Pediatrics, and Birth Defects Institute, New York State Department of Health, Albany, New York.

In Kearns-Sayre Syndrome (KSS) (progressive external ophthalmoplegia, pigmentary retinal degeneration, and heart block) the growth failure and delayed sexual maturation have not previously been studied in detail. We investigated a 15½ year old boy with KSS. A CAT scan revealed loss of the basal ganglia and of cerebellar white matter. His bone age was 10 years. An OGTT was abnormal at 120 and 180 minutes (161 and 164 mg/dl respectively). The IV GTT was normal: K value 2.31. Thyroid function was normal in the basal state; following TRH, the TSH and prolactin responses were normal, but the T_4 had not increased by 90 minutes. The growth hormone response was normal to both insulin and glucagon. The plasma cortisol and ACTH showed normal diurnal variation; after release from metapyrone block, neither the plasma cortisol nor the 17-OH excretion increased appropriately, although the ACTH response was normal. The LH, FSH and testosterone were normal for his BA; following three days of high dose HCG, the testosterone did not increase. After water deprivation, the urinary osmolality was 1011 mosm/kg.

Thus in our patient with KSS there is evidence to suggest lack of functional reserve or end organ unresponsiveness of the thyroid, adrenal and testis; hypothalamic-pituitary function appears to be intact to provocative stimuli.