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DYNAMIC BIOCHEMICAL TESTING IN THE PRELIMINARY DIAGNOSTIC EVALUATION OF PATIENTS WITH GLYCOGEN STORAGE DISEASE.
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Rapid confirmation of the probable type of enzyme defect in patients with GSD is important to limit the number of tests, select the most useful tissue for biopsy and institute appropriate dietary measures. Eleven children with GSD were initially evaluated using the screening procedures originally proposed by Fernandes. This algorithmic approach involves the study of changes in blood glucose, lactate and ketones during fasting, the use of lactate curves following oral hexose loads and the glycaemic response to glucagon in the fed and fasting states. An early tentative diagnosis of specific enzyme defects was possible in all children (5 with type IA, 2 with type IB, 2 with type III, 1 with type VI & 1 with type IX) and later confirmed in most by direct enzyme assay. In a few the diagnosis could only be inferred from the dynamic studies in view of the parents' refusal of an invasive procedure. Preliminary screening with step-wise biochemical testing was useful in all our cases. It confirmed the presence of a metabolic defect, limited further unnecessary testing, yielded an early tentative diagnosis with rapid institution of appropriate dietary therapy and allowed us time to plan a confirmatory diagnostic procedure for direct enzyme assay.

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VISCERAL FAT DETECTED BY MRI AND METABOLIC ABNORMALITIES IN OBESE CHILDREN.

Visceral fat amount (VF) detected by computed tomography (CT) has been related to obesity complicances in adults. Ethical reasons exclude CT use in children studies. Magnetic Resonance Imaging (MRI) is a safe, not invasive technique suitable for body composition and fat distribution analysis even in children.

MRI (S.E. T1 300/200) was performed at lumbar level (L4) in 22 obese children (11 males and 11 females, RBW 121-205%) and 19 normalweight children, aged 10 - 15 years.

In contrast to adults, in obese children subcutaneous fat (SF) was predominant, about 55 to 75% of total L4 area. VF ranged 10 to 15% of SF, without differences regarding sex and pubertal stage. As morbidity risk index we evaluated visceral fat/intrabdominal area (VF/IA). Total cholesterol level and insulin response to OGTT were significantly related to VF/IA ($p<0.02$ and $p<0.05$, respectively), while glucose response to OGTT, triglyceride, HbA1c levels and blood pressure were not. Our data suggest that childhood obesity is predominantly subcutaneous, without sex differences, even during puberty. However VF seem to be already related to morbidity risk. Follow up studies will clarify changes in fat distribution leading to adult visceral pattern.

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WEIGHT GAIN FOLLOWING PROLONGED REMISSION IN PATIENTS WITH ACUTE LYMPHOBLASTIC LEUKAEMIA.
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There is a clinical impression that patients who have been successfully treated for acute lymphoblastic leukaemia(ALL) become obese young adults. A retrospective analysis of the medical records of 130 patients in first prolonged continuous remission from ALL who had reached final height was performed. The body mass index (BMI) was assessed at commencement of treatment (V 1), immediately after treatment was completed (V2), and at final height (V3). All patients were treated with standard UK regimen consisting of combination cytotoxic chemotherapy and employing 1800 cGy or 2100-2500 cGy cranial irradiation for CNS prophylaxis . The mean BMI standard deviation score at V1,V2 and V3 for the groups were as follows.

1800 cGy	V1	V2	V3	
GIRLS	-0.2(0.5)	0.4(1.1)	0.8(1.0)	n=27
BOYS	-0.3(1.5)	-0.5(0.6)	-0.2(0.9)	n=27
2100-2500 cGy				
GIRLS	-0.2(1.2)	0.03(0.8)	-0.1(0.8)	n=40
BOYS	0.3(1.7)	0.2 (0.8)	-0.03(0.9)	n=36

Statistical analysis was carried out employing a Friedmann's non parametric two way analysis of variance to assess the significance of the differences. There was a tendency to an increase in BMI SDS in all four groups. However, this reached statistical significance only in the girls who received 1800 cGy cranial irradiation.($p<0.005$) The BMI in this group did not reach levels defined as obesity for adults more frequently than in the general population as only 10% reached a value of 27. In conclusion,patients successfully treated for ALL do not become obese more frequently than the general population.

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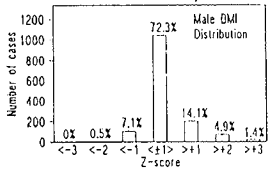
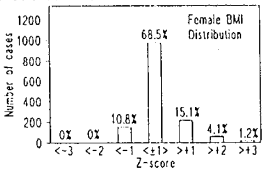
DISTRIBUTION OF BMI IN 2858 CHILDREN AGED 6-14.

Preview: well-balanced body proportions characterize good health; weight to height ratio, used with a power of 2 (BMI = kg/m^2), is considered a good anthropometric index. Aim: to know data about the pattern of BMI distribution. Sample size: 2858 children (1440/M-1418/F), aged 6-14 years; data collected in 1991. Analysis method: 1) calculation of values for Body-Mass Index in kg/m^2 by eight class of age (as BMI varies with age) and each sex; 2) calculation of Z-score, using the following formula: Z = (Q - M)/SD on which Q is the observed weight/height^2 index, M is the median, SD is S x M when S is the coefficient of variation; 3) division of the sample for each of the eight class of age in seven Z-score subgroups, according to the following cut off points: *a)* <-3; *b)* ≥ -3 <-2; *c)* ≥ -2 and <-1; *d)* ≥ -1 and $\leq +1$; *e)* $> +1$ and $\leq +2$; *f)* $> +2$ and $\geq +3$; *g)* $> +3$. Results: histograms of BMI distribution data, according to the above method, have been showed in the figures. Discussion: the data analyzed in this way show no evident problems of severe "under" stoutness conditions, beside to the presence of severe "over" stoutness ones. Comment about the analysis method: anthropometric data are not often normally distributed, tending to be skew, usually with the right tail of the distribution longer than the left. As the above method does not take in account the degree of skewness, the overweight conditions could have been over-estimated. Additional analysis in this way should be undertaken.





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METABOLIC AND ENDOCRINE DISORDERS IN CARBOHYDRATE DEFICIENT GLYCOPROTEIN SYNDROME. W. Bryant, MD , D. Zimmerman, MD, and M. Patterson, MD, Department of Pediatrics and Section of Pediatric Neurology, Mayo Clinic, Rochester, MN 55905, USA

Carbohydrate-deficient glycoprotein syndrome [CDGS] was recently described and includes acquired microcephaly, developmental delay, olivopontocerebellar atrophy, congenital hypotonia, progressive peripheral neuropathy and retinal degeneration, failure to thrive, hepatic steatosis, skeletal abnormalities and lipodystrophy. Secretory glycoproteins are deficient in carbohydrate moities; elevated levels of disialotransferrin and asialotransferrin are diagnostic. Three patients with this condition manifest the following metabolic and endocrine disorders:

	Case 1	Case 2	Case 3
Hypothyroidism	+	+	?
TBG Deficiency	+	+	+
Hypocortisolemia	?	?	+
Cryptorchidism	+	n/a	n/a
Hypotriglyceridemia	+	+	+
Hypocholesterolemia	+	+	+

Conclusions:(1) Mild primary hypothyroidism occures in CDGS; (2) TBG throxine binding is low in CDGS; (3) Cortisol levels may be low in CDGS, possibly from lowered affinity of cortisol binding protein; (4) Cholesterol levels are low in CDGS, possibly because of increased hepatic removal.

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FLUIDITY AND COMPOSITION OF PLASMA LIPOPROTEINS (PL) IN HYPERCHOLESTEROLEMIC CHILDREN

Aims: to evaluate possible changes in PL fluidity and composition in hypercholesterolemic children. *Patients & Methods*: 18 hypercholesterolemic children (mean total plasma cholesterol [TPC] 286 mg/dL, SD 86.0), mean age 10.3 years (SD 2.0), normal weight (mean BMI 17, SD 2.0); 14 controls (mean TPC 158.7 mg/dL, SD 22.0), mean age 10.3 (SD 1.0), normal weight (mean BMI 16, SD 1.0). TPC in hypercholesterolemic children significantly higher than in controls ($p<0.001$). Analysis of PL composition; study of LP fluidity by fluorescence polarization (FP) of 1,6-diphenyl-1,3,5-hexatriene (DPH). *Results*: significant increase of LDL-associated cholesterol ($p<0.003$) and VLDL-associated cholesterol ($p<0.003$) in hypercholesterolemic children compared to controls; FP values significantly higher in VLDL ($p<0.01$) and VLDL ($p<0.001$) of hypercholesterolemic children compared to controls, indicating lower LP fluidity in patients. *Conclusions*: our data suggest early changes in LP composition and fluidity in hypercholesterolemic children.