not increased, a specific thiamine-dependent defect in pyruvate decarboxylase was considered. Assay of this enzyme in fresh peripheral leucocytes, or after phytohemagglutination stimulation, at low and high concentrations of pyruvate, with and without thiamine added, revealed no abnormality. Negative results were also obtained with cultured skin fibroblasts. A phenotypic variant of the traits described by BLASS and TADA, but limited to liver in this case, is proposed. (Research supported by Grants MRC 1085 and NHW 604-7-643.)

65 *Low Phosphorylase Activity: A Symptom but not a Diagnosis.* GEORGE HUG, WILLIAM K. SCHUBERT and GAIL CHUCK, Children's Hosp. Res. Fndn. Cincinnati, Ohio.

Since our initial description of deficient activity of hepatic dephosphorylase kinase and increased liver glycogen [Science *153:* 1535, 1966], we have studied a total of 7 patients with this condition. Five of these patients have been reported [J. clin. Invest. *48:* 705, 1969]. They and the 2 additional patients, brothers age 4 and 1 ½ years, always had greatly reduced liver phosphorylase activity (about 10% of control) that could be increased to normal by addition of kinase. This rules out phosphorylase deficiency, or type VI glycogenosis. On the basis of 'one enzymatic defect for one type of glycogenosis', we called deficient kinase activity type IX glycogen storage disease. In type IX glycogenosis there is no hyperlipemia, hypoglycemia, acetonuria, or evidence of X-chromosomal inheritance. There is a typical light and electron microscopic appearance of the liver. Biochemically, the skeletal muscle is normal as is the blood sugar response to glucagon. In contrast, in a girl with hepatomegaly and low hepatic phosphorylase activity there is increased glycogen concentration in the skeletal muscle as well as the liver and the glucagon tolerance curve is flat. In her skeletal muscle the amount of *total* phosphorylase is normal but all of it is in the *inactive* form. Phosphorylase kinase is present in this muscle but we found evidence for deficient activity of 3′5′-AMP dependent kinase that normally activates phosphorylase kinase [J. biol. Chem. *243:* 3763, 1968]. Therefore, we have named this new type of glycogen storage disease with a hitherto undescribed enzymatic deficiency type X glycogenosis. (Supported by NIH grants No. AM 13903 and No. RR 00123.)

66 *Human Liver Galactose-1-phosphate Uridyl Transferase: Activity in the Negro Galactosemic.* STANTON SEGAL and SHIRLEY ROGERS, Children's Hosp. of Philadelphia, Pa.

Since the Negro with congenital galactosemia can metabolize substantial amounts of galactose, assays for the deficient enzyme, gal-1-p uridyl transferase, were performed in homogenates of punch biopsies of liver from two Negro patients. Kinetic characteristics of the normal enzyme were determined in homogenates of liver from four non-galactosemic patients undergoing abdominal surgery and in two 50-fold purified preparations of liver obtained within 24 h of death. Kinetic parameters including concentration dependence and Km were similar in the crude homogenates and partially purified normal enzyme. The specific activity of transferase in mμmoles/min/mg protein was 12 and 1.6 in crude homogenates of the normals and galactosemics, respectively. The latter activity was linear with incubation time but showed no concentration dependence on uridine diphosphoglucose. No uridine diphosphogalactose pyrophosphorylase activity, a possible alternate pathway, was detected in the liver of Negro patients and transferase activity with thymidine diphosphoglucose as substrate was negligible. Either these patients possess a non-nucleotide pathway of galactose metabolism or the residual transferase activity accounts for their galactose metabolic capability. Results of *in vivo* metabolic studies with labeled galactose suggests the latter is most likely.

67 *Idiopathic Hypomagnesemia and Osteochondritis.* WILLIAM G. KLINGBERG, Dept. of Ped., West Virginia Univ. Sch. of Med., Morgantown, W.VA.

A previously asymptomatic 5-year-old boy presented with classic carpopedal spasm of 6 h duration. On examination he also had papilledema, decreased DTR's and a negative Chovstek. Serum Mg was 0.8 and K was 2.8 mEq/L. All other serum electrolytes, proteins, enzymes, Ca, PO_4, Glucose, BUN and Creatinine were normal. The EMG and PEG were normal. The EEG showed a few non-specific low waves and mild disorganization. Unexpectedly x-rays of bones showed a mild osteochondritis of shoulders, knees and hips (Legg-Perthes-like). Creatinine clearance was 70 ml/min/1.73 M^2. Gross renal tubular function was normal by concentration and acidification.

A 6-day balance study showed minimal negative Mg balance. When 20 mEq magnesium acetate (MgA) was added *per os* daily, another 6 day balance study showed a positive balance, but barely so (intake 174 mEq, output 132 mEq = +42 mEq). Little is known regarding Mg balance and metabolism in children, but adults with hypomagnesemia given MgAc would excrete very little in the urine. This child continued to excrete 5–9 mEq/day during the balance studies. Potassium was also in negative balance although not enough to be significant. Other ions were in normal balance.

When MgAc orally was increased to 40 mEq/day serum Mg rose to normal. After 6 months therapy the bony lesions have reverted almost to normal. MgAc therapy is still required as hypomagnesemic tetany again occurred when inadequate therapy was given. Is this a specific, partial renal tubular defect in Mg absorption.

68 *Evaluation of Adenine Therapy for Lesch-Nyhan Syndrome.* JOSEPH D. SCHULMAN, MARTIN L. GREENE, WILFRED Y. FUJIMOTO and J. EDWIN SEEGMILLER, NIH, Univ. of Washington, Seattle, and Univ. of California, San Diego (introduced by W. L. Nyhan).

The use of oral adenine to prevent the devastating neurologic consequences of the Lesch-Nyhan syndrome has a sound theoretical basis, and has been proposed for management of this aspect of the disease. We have attempted therapy of two patients with adenine begun in one patient, during the first month of life. Preliminary experiments in rats explored the toxicity of adenine; pretreatment with allopurinol (10 mg/kg) reduced the nephrotoxic effects of the insoluble adenine metabolite, 2,8-dioxyadenine, at adenine doses of 70 mg/kg. Two patients, a 13-year-old male with established neurologic disease and self-mutilation, and a normal-appearing one-month-old boy with documented hypoxanthine-guanine phosphoribosyltransferase (PRT) deficiency, were given adenine in doses up to 65 mg/kg/day. Toxicity was monitored by daily esti-