Abstracts

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Intestinal phosphate transport in familial hypophosphatemia.

Renal inorganic phosphate (Pi) transport is impaired in familial hypophosphatemic rickets (FHR). Short et al. (Science, 179,700, 1973) have reported that the mutation was also expressed in the gut. We have examined Pi uptake in vitro by jejunal mucosa from 7 (4 female, 3 male) FHR mutants (from 5 pedigrees) and 6 controls. Peroral samples were incubated for 5 to 40 minutes in TRIS buffer, pH 7.4 with substrate concentrations from 0.003 to 3 mM. Pi uptake was concentrative and energy dependent reaching mean distribution ratios (intracellular/extracellular ^{32}P) of 4.2 ± 0.9 in controls and 5.0 ± 0.5 in patients after incubation with 0.003 mM ^{32}Pi for 40 minutes. Incorporation of Pi in the organic pool was rapid and equilibrated after 10 minutes at a ^{32}Pi/total ^{32}P ratio of 0.5, at all substrate concentrations, in both groups. Only one mediated transport system for Pi was present in control subjects, with a Michaelis constant $\simeq 0.2$ mM and a maximum velocity $\simeq 0.7$ mmoles per liter per 40 minutes. Similar kinetic values were obtained in the group of FHR patients. These observations do not support the thesis that a significant defect for Pi uptake is present in the jejunal mucosa of FHR mutants.

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Renal handling of phosphate in vitamin D resistant rickets.

In 9 children with vitamin-D-resistant rickets (VDRR) urinary phosphate excretion (U_PV), phosphate clearance (C_P) and tubular reabsorption (T_P) were measured before and during intravenous phosphate loading. The children with VDRR exhibited significantly lower values for T_P, T_P/C_{in} and Tmp, while the rate of endogenous phosphate excretion was not higher than in age matched controls. In 5 of these children the effect of 200 I.U. PTH (Parathormone Lilly) on U_PV, C_P, and Tmp was investigated. PTH has no significant effect on the tubular phosphate reabsorption under these conditions. Cyclic AMP, a presumable transmitter of PTH-mediated phosphate-transport, was significantly increased after PTH-injection in all children with VDRR. These results indicate, that in Vitamin D resistant rickets the PTH-sensible component of tubular phosphate transport system is disturbed.