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Case Report

A JAPANESE CASE OF PENTOSURIA

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Essential pentosuria (McKusick 26080; Hiatt, 1978) is a rare, autosomal recessive genetic metabolic disorder characterized by the extretion of gram quantities of L-xylulose in the urine. It is a benign disturbance which occurs principally in Jews.

The pentosuric subject is a healthy Japanese male, 23 years old. We accidentally found that the fasting urine gave a positive Benedict's test and a negative enzymatic method specific for glucose. The urinary sugar was identified as L-xylulose by paper chromatogram (solvent system: isopropanol–water, 160/40, R_g 1.55) and was determined by the enzymatic method (Hickman and Ashwell, 1957). In the urine nothing else of note was found. A tolerance test with 50 g of glucose showed a normal disappearance rate of glucose. Urine L-xylulose levels before and after oral administration of 10 g D-glucuronolactone were 5.40 mmol l^{-1} (fasting) and 15.19 mmol l^{-1} (90 min after administration). Serum AST, ALT, LDH, CK, urea, creatinine, uric acid, protein, albumin and globulin were all within the reference ranges.

The activities of L-xylulose reductase (EC 1.1.1.10) in erythrocytes were measured fluorometrically as follows: Reaction mixtures contained 500 µl of 0.2 mol l^{-1} Tris–HCl buffer, pH 8.0, 250 µl of 20 mmol l^{-1} $MgCl_2$, 250 µl of 20 mmol l^{-1} nicotinamide, 100 µl of 42 mmol l^{-1} NADP, 100 µl (Hb 0.2 mg) of haemolysate, 250 µl of 1.2 mol l^{-1} xylitol and 550 µl of water for the subject cuvette and 800 µl of water for the reference cuvette (without xylitol). Fluorescence was determined in a Shimadzu Difference Spectrofluorometer, RF-503A, circulating thermobath (37 °C), with an excitation wavelength of 365 nm and an emission wavelength of 450 nm. The activity obtained from the pentosuric subject was 38.0 nmol min^{-1} $(g\,Hb)^{-1}$ and activities obtained from controls ($N = 5$) were 56.7 ± 8.2 nmol min^{-1} $(g\,Hb)^{-1}$, (mean ± SD). When the enzyme activities were fluorometrically measured under the conditions used in Wang's glutathione method (1970), the activity of the pentosuric subject was 9.9 nmol min^{-1} $(g\,Hb)^{-1}$ and the activities obtained from controls were 27.1 ± 4.0 nmol min^{-1} $(g\,Hb)^{-1}$. These values are similar to the values of Wang's glutathione method. Michaelis constants for NADP of L-xylulose reductase in the pentosuric subject and control were 1.1×10^{-3} mol l^{-1} and 1.0×10^{-4} mol l^{-1}, respectively. Thus the K_m value for NADP was about ten times higher in the pentosuric subject. This result is in agreement with that of Wang and Eys (1970). Therefore, in our case also, pentosuria results from a molecular abnormality that decreases NADP affinity of the enzyme.

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