

FRUCTOSURIA IN A SUDANESE FAMILY

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Abstract A Sudanese family who has three boys affected with fructosuria is reported and discussed. The index case, a 10-year-old boy, presented with vague abdominal pain, periodic attacks of polyuria and a reducing sugar in his urine that did not react with glucose oxidase. A sucrose load (50 g) lead to substantial excretion of fructose in the urine and to polyuria. The absence of classical symptoms of hereditary fructose intolerance suggests essential fructosuria as the most likely abnormality in this family.

Key words Essential fructosuria; Child; Sudan.

INTRODUCTION

Though the sugar fructose is found in free form in certain fruits and vegetables, its major source in the diet is the disaccharide sucrose. The latter is hydrolysed in the intestine by sucrase into glucose and fructose before absorption. Fructose is easily utilized in the human body, particularly in the liver where the greater part is metabolized¹. As shown in Fig. 1, the breakdown of fructose in the liver follows the same metabolic pathway as glucose with the exception of the first two reactions. Thus metabolic defects specifically interfering with fructose utilization could result from deficiency of fructokinase or fructose-1-phosphate aldolase. Such defects have been reported in the literature^{2,3}.

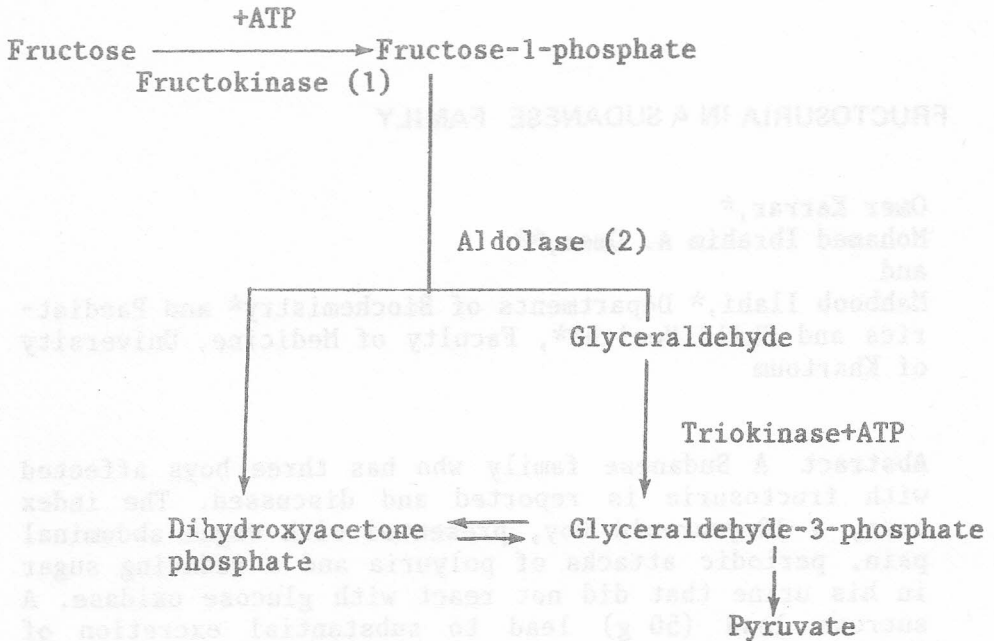


Figure 1. The metabolic pathway for breakdown in the liver. Relation (1) and (2) are catalysed by fructokinase and fructose-1-phosphate aldolase, respectively. Blockage at (1) or (2) leads to fructosuria.

Fructokinase deficiency gives rise to a condition known as essential fructosuria while deficiency of aldolase causes hereditary fructose intolerance. The former condition is asymptomatic and may pass unnoticed while the latter shows distinct symptoms and could prove fatal⁴.

This paper reports three cases of essential fructosuria in a Sudanese family.

Case report

The patient, a 10-year-old boy living in Sahafa of Khartoum presented to hospital with a vague abdominal pain and periodic attacks of polyuria. He did not suffer from any significant illness in the past and his brothers, four in all, together with his parents were apparently healthy. Clinical examination did not reveal any significant findings. The only abnormal result in the laboratory investigations was the presence of a reducing sub-

stance in the patient's urine that did not react with glucose oxidase paper strips. Furthermore, it was noticed that the reducing substance appeared after the intake of tea or sweet lemon juice, but not after the intake of glucose solution. These observations suggested that the substance was most likely fructose and to confirm this finding further investigations were carried out.

The child was given an oral load of 50 g of sucrose and the excretion of fructose in the urine was followed at half-hourly intervals. Urinary fructose was then determined by the procedure described by Roe⁵. Blood glucose was determined after an overnight fast and 2 hours after the intake of 50 g glucose. The estimation was done using glucose oxidase method described by Hugget and Nixon⁶.

RESULTS AND DISCUSSION

Chain et al⁷ and Miller and Crane⁸ observed that fructose was absorbed from the intestine more efficiently when administered as sucrose than when administered as an equimolar mixture of glucose and fructose. It is for this reason that a sucrose load instead of fructose was given to the child. Ingestion of 50 g of sucrose led to a substantial excretion of fructose in the urine. This heavy fructosuria was accompanied by the excretion of more than a litre of urine during the test period of 150 minutes. This diuresis was due to the osmotic effect exerted by the unmetabolized fructose and largely explains the periodic attacks of polyuria experienced by the child. The observation that the blood glucose of the patient 2½ hours after the intake of 50 g of glucose fell to the fasting level of 55 mg/dl shows that the child's tolerance for glucose was normal. In view of the fact that the child did not present any of the classical symptoms of hereditary fructose intolerance, his abnormality would most likely be essential fructosuria. This result prompted us to carry out screening tests on the rest of his family. The tests revealed that two of his four brothers were passing fructose in urine, but the parents were not.

As inborn errors leading to the excretion of reducing sugars in urine could considerably confuse doctors, it is essential to screen all infants and children admitted to the paediatric wards so that early advice and assurance could be given to the parents of affected children or infants.

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