

Abstract

Ann Endocrinol (Paris). 2007 Dec;68(6):456-9.

Doctor, my son is so tired... about a case of hereditary fructose intolerance.

Guery MJ, Douillard C, Marcelli-Tourvieille S, Dobbelaere D, Wemeau JL, Vantyghe MC.

Service d'endocrinologie et métabolisme, clinique Marc-Linquette, CHU de Lille, 6, rue du Professeur-Laguesse, 59037 Lille cedex, France.

BACKGROUND: We present the case of a 17-year-old male who was diagnosed at birth with hereditary fructose intolerance (HFI). The patient complained of morning-time asthenia and post-prandial drowsiness despite a correct sleep pattern. The physical examination and biological check-up only showed severe vitamin C deficiency (<10 mol/l; normal range: 26-84). The patient's tiredness was attributed to this vitamin C deficiency, which is a frequent side-effect of the fructose-free diet. A change in diet associated with a supplementation in vitamin C was advised, with an increase in vegetable intake, principally avoiding carrots, onions, leaks and tinned sweet-corn.

DISCUSSION: This case offers the opportunity for a review of this rare disease. Two kinds of fructose metabolism disorders (both autosomal recessive) are recognized: 1) essential fructosuria caused by a deficiency of fructokinase, which has no clinical consequence and requires no dietary treatment; 2) HFI, linked to three main mutations identified in aldolase B gene that may be confirmed by fructose breath test, intravenous fructose tolerance test, and genetic testing. In HFI, fructose ingestion generally induces gastro-intestinal (nausea and vomiting, abdominal pain, meteorism) and hypoglycemic symptoms. Fasting is well tolerated. If the condition remains undiagnosed, it leads to liver disease with hepatomegaly, proximal tubular dysfunction, and slow growth and weight gain.

CONCLUSION: In conclusion, endocrinologists should be aware of this rare metabolic disease in order to provide careful follow-up, particularly important when the patient reaches adulthood. Moreover, hypoglycemia induced by fructose absorption, unexplained liver disease, irritable bowel syndrome or familial gout in an adult is suggestive of the diagnosis.

PMID: 18035330