performed first, and will reveal the etiology if abnormal. If the result is normal, and a genetic cause is suspected, genetic testing could involve the analysis of multiple genes implicated in sexual differentiation.[12]

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Restricted diet in fruits causes scurvy in a child of 7 years old☆

Dieta restrictiva en frutas como causa de escorbuto en niño de 7 años

Scurvy is a disease caused by vitamin C deficiency. Humans do not synthesize this vitamin, so it needs to be taken in the diet. Scurvy is very rare in industrialized countries; cases reported in children may be caused by inadequate diet in patients with neuropsychiatric disorders such as autism or infantile cerebral palsy. Some cases have also been reported in children with celiac disease[1] and in infants given inadequate diets such as almond beverages instead of adapted formulas.[2]

We report the case of a boy with multiple allergies, multiple food intolerances, and fructose malabsorption-intolerance who had scurvy despite not being on a vegetable-and fruit-restricted diet.

This was a 7-year-old Caucasian boy diagnosed two years previously with an allergy with anaphylaxis to nuts and stone fruits, in addition to intolerance to other food such as tomatoes, kiwis, oranges, milk and dairy products (which the child refused to eat due to abdominal pain). As he had been diagnosed with fructose malabsorption and intolerance by a hydrogen breath test, his diet also restricted the intake of fruit juices, honey, baked goods and manufactured products containing fructose. During monitoring at the clinic, malabsorption diseases such as celiac disease and inflammatory bowel diseases had been ruled out, and there was no relevant family history of the disease.

At his yearly nutritional check-up, the boy had normal body measurements as follows: weight, 22 kg (p50–85); height, 123 cm (p85–97); BMI, 14.2 kg/m^2 (p85–97), with no weight or height stagnation. The mother reported that the boy was tired and complained of severe bone pain in his lower limbs and lumbar region, for which reason the pediatrician had referred him to the orthopedic surgeon. She also mentioned gum bleeding and small perifollicular ecchymoses on the legs and arms, which led the pediatrician to request coagulation tests and a complete blood count, which were normal. The physical examination was unremarkable except for the small ecchymoses on his legs. A very low vitamin C level of 1 mg/L (4.6–14.9 mg/L) was seen in the tests requested for the check-up at the clinic. Levels of all other vitamins were within normal ranges: folic acid, 11 ng/mL (2.8–20 ng/mL); vitamin B_{12}, 545 pg/mL (239–931 pg/mL); vitamin E, 8 mg/L (3–9 mg/L); vitamin A, 0.25 mg/L (0.2–0.4 mg/L); vitamin D, 32 ng/mL

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(30–100 ng/mL). His mother reported that the child ate vegetables occasionally (but not daily), and that the only fruit he ate was watermelon. A vitamin C dietary intake count of the previous three days revealed a mean vitamin C intake of 31 mg. Treatment with ascorbic acid 500 mg/day for one month was prescribed, and the symptoms subsided and his blood levels of vitamin C became normal (7 mg/L). The mother was advised to provide vegetables and two pieces of tolerated fruit each day, in addition to a vitamin C supplement of 30 mg daily.

Ascorbic acid or vitamin C is a lactone synthesized from glucose.[3] Most mammals and plants synthesize endogenous vitamin C from glucose and galactose. However, humans lack the enzyme gulonolactone oxidase, and are thus unable to synthesize the vitamin, which needs to be obtained from food.[4] The main sources of vitamin C are fruits and vegetables. The recommended intakes are 75 mg/day for women, 90 mg/day for men, 50–60 mg/day for infants under one year of age, and 25–50 mg/day for older children.[5] The current incidence of scurvy is very low. There are isolated cases especially in high-risk groups such as the elderly, alcoholics, malnourished individuals, and infants under the age of one year who are fed low-quality milk.[2,6] The clinical signs of scurvy are varied, consisting of fatigue[7] and skin manifestations such as ecchymoses, petechiae, bruising, and bleeding. Follicular hyperkeratotic papules and changes in hair morphology (corkscrew, kinky hair) can also occur. There may be gum edema and bleeding, and even tooth loss.[6] In contrast to adults, bone manifestations are very common in children,[6] who may limp and complain of bone pain. Radiologic findings may include osteoporosis, cortical thinning, and periosteal detachment. In advanced cases there may be heart muscle involvement, marrow disorders, and brain and adrenal hemorrhages which may lead to death.[4,8] Treatment has not been standardized, but the usual therapeutic doses are 100–300 mg/day for children and 500–1000 mg/day for adults for one month or until the symptoms subside. Vitamin C doses greater than 2 g cause side effects such as diarrhea, vomiting, abdominal pain, headache, and insomnia. With this treatment, bleeding ceases within 24 h, and muscle and bone pain disappear in a few days.[7] After treatment, a varied diet should be followed to meet daily requirements.[9] A daily intake of five servings of fruits and vegetables provides more than 60 mg/day.[10]

Scurvy is currently an uncommon disease, but should be kept in mind not only in the abovementioned risk groups, but also in any patient with a fruit-restricted diet such as the one reported here. In cases with a severely fruit-restricted diet, close dietary follow-up should be implemented, including the monitoring of vitamin levels and/or the provision of preventive vitamin C supplements.

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