Chronic Watery Diarrhea in a Toddler: Think Beyond Toddler's Diarrhea

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To the Editor: An 18-mo-old toddler, born to 2⁰-consanguineous parents was referred for passing large volume, watery, yellow stools, 10 times/d associated with abdominal distension and poor weight gain since 7 mo of age. He was exclusively breastfed upto 6 mo. Diarrhea was aggravated by syrups and rice-based feeds. On examination, he had perianal excoriation and was hydrated. He weighed 8.6 kg [< 3rd centile]. Systems were normal. Complete blood counts, liver and renal function tests, serum IgE and ultrasonography of the abdomen were normal. Stool reducing substance was positive. Based on the above features, a diagnosis of osmotic diarrhea probably due to carbohydrate-malabsorption was suspected. Endoscopy and duodenal histology was normal. Genetic testing revealed a homozygous nonsense mutation in exon 41[c.4825C>T] of *SI* gene confirming congenital sucrase-isomaltase deficiency (CSID). He was started on specific diet (avoiding foods rich in sucrose, maltose, isomaltose, and starches, allowing protein and fat-rich foods with dairy products). At 2 mo follow-up, he is asymptomatic and has gained 600 g of weight.

CSID results from mutations in sucrase-isomaltase (*SI*) gene [1]. Symptoms occur when sugar/cereals is introduced in the diet and include diarrhea, flatulence, and abdomen distension [2]. CSID is misdiagnosed as irritable bowel syndrome (IBS) type-D or toddlers diarrhea and is hence diagnosed late. Disaccharidase assay from the intestinal biopsy remains the gold standard but is not widely available. Gene testing is confirmatory. Restriction of sucrose and starch is necessary to alleviate symptoms [3]. Baker's yeast

(lyophilized *Saccharomyces cerevisiae*) has appreciable sucrase and isomaltase activity and a mustard seed size given after a sucrose containing meal may ameliorate the symptoms [4]. Sacrosidase, an oral enzyme replacement therapy, is very expensive. The severity of CSID depends on the nature and type of *SI* gene mutations. While some patients may show signs of sucrose tolerance during the second decade of life, majority exhibit life-long sucrose intolerance [1].

Declarations

Conflict of Interest None.

References

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