CASE DESCRIPTION

A 7-month-old girl of average weight and unremarkable medical and family history presented with chronic osmotic diarrhea and failure to thrive. The onset of symptoms appeared shortly after cereals and fruits were introduced into her diet. As part of the investigation of the gastrointestinal symptoms, a biopsy of the small intestine mucosa was sent to the laboratory for biochemical analysis. Upon receipt of the results, the doctor advised the parents to remove sources of sucrose from her diet.

QUESTIONS

1. Which laboratory test performed on the intestinal mucosa is useful in the evaluation of diet-associated osmotic diarrhea?

2. What is a possible etiology for the condition identified by the test?

The answers are below.

<table>
<thead>
<tr>
<th>Table 1. Results of the biochemical analysis.</th>
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<tbody>
<tr>
<td><strong>Disaccharide substrate (glycosidic bond)</strong></td>
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<tr>
<td>Lactase (β-1,4)</td>
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<tr>
<td>Maltase (α-1,4)</td>
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<tr>
<td>Isomaltase*</td>
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<tr>
<td>Sucrase (α-1,2)</td>
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* Also known as palatinase.

* Also known as Palatinose™.

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Carbohydrate malabsorption is a common cause of chronic osmotic diarrhea. Because the small intestine is impermeable to disaccharides, intestinal disaccharidases are required for absorption of monosaccharides. Disaccharidases include lactase-phlorizin hydrolase, sucrase-isomaltase, and maltase-glucoamylase. Measurement of intestinal disaccharidase activities is considered the gold standard test for diagnosis (1). Results for this patient are shown in Table 1. The undetectable sucrase and isomaltase activities combined with markedly reduced maltase activity indicate a sucrose-isomaltase deficiency in this patient. Approximately 80% of maltase activity is accounted for by sucrase-isomaltase (2). Congenital sucrase-isomaltase deficiency is an autosomal recessive disease of low global prevalence, but it may be seen more often in certain populations of the northern hemisphere (up to 10.5% prevalence in Greenland Eskimos) (1, 3). Symptoms may not surface until dietary sucrose is encountered and may easily be attributed to other causes.

Author Contributions: All authors confirmed they have contributed to the intellectual content of this paper and have met the following 3 requirements: (a) significant contributions to the conception and design, acquisition of data, or analysis and interpretation of data; (b) drafting or revising the article for intellectual content; and (c) final approval of the published article.

Authors’ Disclosures or Potential Conflicts of Interest: Upon manuscript submission, all authors completed the author disclosure form. Disclosures and/or potential conflicts of interest:

Employment or Leadership: D.G. Grenache, AACC and Academy of Clinical Laboratory Physicians and Scientists.
Consultant or Advisory Role: None declared.
Stock Ownership: None declared.
Honoraria: None declared.
Research Funding: None declared.
Expert Testimony: None declared.

References