Breath hydrogen test and sucrase isomaltase deficiency

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SUMMARY Sucrose breath hydrogen tests were performed on 7 children with proved sucrase isomaltase deficiency. All children had raised breath hydrogen excretion. The amount of hydrogen produced and symptoms experienced increased with increasing sucrose loads. The sucrose breath hydrogen test appears to be a reliable indicator of sucrose malabsorption in sucrase isomaltase deficiency.

Sucrase isomaltase (SIM) deficiency is an autosomal dominant disorder causing malabsorption of sucrose. Affected children usually develop diarrhoea in the first years of life when sucrose is introduced into the diet and there may be associated abdominal distension and failure to thrive. The diagnosis is usually made when low concentrations of these disaccharidases are found in mucosal samples obtained by small bowel biopsy.

There has been disagreement over the value of the sucrose breath hydrogen test in the diagnosis of this condition. Perman et al. described 4 patients with disaccharidase proved SIM deficiency who all developed positive sucrose breath hydrogen tests. They stated that this test was an accurate method of evaluating specific carbohydrate malabsorption. By contrast, Gardiner et al. reported 5 children with SIM deficiency who all had negative sucrose breath hydrogen tests and concluded that this test was of no value in diagnosing primary sugar malabsorption in children. Our experience of the sucrose breath hydrogen test in 7 children with homozygote SIM deficiency is similar to that of Perman et al. and we believe it to be a reliable index of this disorder.

Patients and methods

Between 1976 and 1981, 15 children with SIM deficiency were diagnosed by disaccharidase assay. Only 7 of these children could be contacted subsequently, but in each case their parents agreed to allow them to be tested. Patient details and disaccharidase values are given in Table 1. All biopsy specimens showed normal histology and all patients except the girl in case 6 had normal lactase concentrations. Her normal histology excluded coeliac disease.

After an overnight fast, oral sucrose ranging from 0.6 to 2.2 g/kg was given in a 20% solution. The dose was varied because of concern that large sucrose loads might cause unacceptable symptoms. End expiratory breath samples were taken by nasal prong at 30 minute intervals over 2 hours and the samples were analysed immediately.

Table 1 Patient details and disaccharidase values

<table>
<thead>
<tr>
<th>Case No</th>
<th>Sex</th>
<th>Age at diagnosis (yr/mo)</th>
<th>Disaccharidase values (units/wet weight)</th>
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<tr>
<td></td>
<td></td>
<td></td>
<td>Maltase</td>
</tr>
<tr>
<td>1</td>
<td>F</td>
<td>1/9</td>
<td>3.1</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>1/2</td>
<td>2.3</td>
</tr>
<tr>
<td>3</td>
<td>M</td>
<td>3/8</td>
<td>6.1</td>
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<tr>
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<tr>
<td>6</td>
<td>F</td>
<td>6/4</td>
<td>3.8</td>
</tr>
<tr>
<td>7</td>
<td>F</td>
<td>7/4</td>
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Normal values:

<table>
<thead>
<tr>
<th>Normal values</th>
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<tbody>
<tr>
<td>&gt;9.0</td>
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</tr>
<tr>
<td>&gt;1.0</td>
</tr>
<tr>
<td>&gt;2.5</td>
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</tbody>
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ND = not done
Hydrogen, oxygen, and nitrogen concentrations were measured by gas chromatography using a Packard 427 chromatograph. A sample volume of 5 ml delivered via a gas sampling valve was injected through a 3 m × 6.35 mm (outside diameter) stainless steel column packed with Molecular Sieve 13X. Argon carrier gas was run at 15 ml/min at ambient temperature. A thermal conductivity detector was used, filament temperature was 350°C, and the signal was read by a Hewlett-Packard 3390A reporting integrator. Run time was 13.5 minutes. By using this technique hydrogen concentrations could be measured to 1 part per million (ppm). Observed hydrogen concentrations were corrected to values expected in an ideal sample containing alveolar air. A test was considered positive if there was a rise in hydrogen concentration of 10 ppm or greater.

Results

Results of the sucrose breath hydrogen tests in the 7 children are given in Table 2. All were hydrogen producers. Three children had a second test 1 year later. Symptoms of diarrhoea with or without abdominal pain were reported during all but 1 test. The symptoms came on between 1 and 8 hours and were more severe with increasing loads of sucrose. No patient had serious symptoms.

Eight tests were done in 5 patients using a sucrose load of 1 g/kg or greater. These tests were unequivocally positive, with increased hydrogen excretion becoming evident by 60 minutes in all but 1 test. The 2 oldest children (cases 6 and 7) were given sucrose loads of less than 1 g/kg, but although they both showed an increase in hydrogen concentration by 60 minutes, the results could be regarded as equivocal. Unfortunately they did not agree to have the test repeated with a larger sucrose load.

Discussion

The breath hydrogen concentrations in 5 of 7 children with sucrase isomaltase deficiency who were given oral sucrose were well above the normal range. The other 2 children showed equivocal rises (12 ppm and 16 ppm) after small challenge doses of sucrose (0-8 g/kg and 0-6 g/kg) that would be accepted as positive by some. Our study confirms the report by Peman, but the findings are in marked contrast to those of Gardiner et al. who reported that all 5 children with sucrase isomaltase deficiency whom they studied failed to produce hydrogen—possibly owing to altered activity of colonic flora. In both these studies a sucrose load of 2 g/kg was used. We used a range of doses of sucrose and when the dose was greater than 1 g/kg, clear results were obtained. Although the results might be regarded as equivocal in the 2 tests where a smaller dose was used, there was still a definite rise in breath hydrogen concentration.

This non-invasive test appears to be a reliable indicator of sucrose malabsorption in children with sucrase isomaltase deficiency but its place in diagnosis is not yet clear. It cannot replace the need for small bowel intubation and biopsy if other conditions causing diarrhoea, including coeliac disease and giardiasis, are to be detected. On the other hand a negative test may simply mean that the child does not harbour hydrogen producing bacteria in the colon. Perhaps its usefulness will lie in following changes in sucrose tolerance with time.

Sucrase isomaltase deficiency is an example of sugar intolerance due to virtual absence of the appropriate disaccharidase enzyme. It is encouraging that the breath hydrogen test in a group of children with this disorder gave results that could be predicted by the theory on which the test is based.

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References


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Fifty years ago

Some observations on 300 fatal cases of tuberculosis in children
A V NEALE (Birmingham)

Observations on the exact necropsy findings in 300 cases of tuberculosis in infants and young children were reported. Practically all had died of miliary tuberculosis and tuberculous meningitis. Detection of the primary route of infection was determined by careful search for the primary focus—intrathoracic (Ghon tubercles and hilar glands) or abdominal (intestinal). The results were: (a) primary thoracic (aerogenous infection) 215 or 71.6%; (b) primary abdominal (infection) 46 or 15.3%; (c) no primary focus detectable 39 or 13.0%. In the last group, although no primary focus could be detected, it was almost certain that there had been a rapid generalisation after aerogenous infection. In cases under 3 years of age, the numbers in the three groups were 141, 30, and 33 respectively.

Archives of Disease in Childhood 1933; 8: 362.

(300 necropsies on children with tuberculosis in less than 5 years! Victor Neale became the first Professor of Child Health in Bristol. PRE.)

The medical treatment of obstetric fractures in the newborn
E PRITCHARD (London)

Dr Pritchard exhibited a number of x-ray photographs illustrating the possibility of treating obstetric and other fractures in the newborn without splints, bandages, extension apparatus or other appliances commonly used by surgeons. Some of the films demonstrated the failure of surgical means to improve the position of the fractured bones. Others showed the astonishing reparative power of unaided nature to reduce deformities and restore the axis and alignment of the bones by throwing out an enormous quantity of provisional callus, and subsequently moulding it and shaping it that the original form and shape of the bone is ultimately attained. In most unpromising cases, with extensive overlapping and deformity the bones are restored to their original shape within a few months with no shortening or other evidence of the original fracture.

Archives of Disease in Childhood 1933; 8: 363.

(I wonder if they are still left alone as he recommended? Eric Pritchard founded the Westminster Children's Hospital in Vincent Square, London. PRE.)