

¹³C-SUCROSE BREATH TEST: ARE WE READY FOR POINT OF CARE TESTING YET?

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BACKGROUND

- Congenital sucrase-isomaltase deficiency (CSID) is absence or deficiency of sucrase and isomaltase (SI)
- Absence of SI → an osmotic effect → bloating and diarrhea
- Endoscopic duodenal biopsy assays are the gold standard
 - Biopsies are difficult to interpret
- ¹³C-sucrose breath testing can confirm CSID
 - Small intestinal bacterial overgrowth (SIBO) can lead to false positives
- Trio-Smart® breath testing can rule out SIBO
- Goal: Confirm CSID diagnosis on biopsy with breath tests and monitor clinical response to a trial of Sucraid®

METHODS

- Case-series of 3 teenagers with sucrase <25 μM/min/g protein on assays
- Breath tests completed at home
- Medical history:
 - Patient 1: Irritable bowel syndrome, type 1 diabetes, polycystic ovarian syndrome
 - Patient 2: Crohn’s disease (in remission)
 - Patient 3: Previously healthy
- A trial of Sucraid® replacement therapy with assessment of symptoms 4 weeks later

RESULTS

| Parameter | Ref. Ranges | Patient 1 | Patient 2 | Patient 3 |
|-------------------------------------|----------------|----------------|-------------------|--------------|
| Age (years) | | 14 | 15 | 17 |
| Gender | | Female | Female | Male |
| Gastrointestinal Symptoms | | Pain, diarrhea | Pain, weight loss | Loose stools |
| ¹³ C-Sucrose Breath Test | | | | |
| Result | | Abnormal | Abnormal | Abnormal |
| Sucrose Digestion (%) | M >3.9; F >5.1 | 1.88 | 1.96 | 3.23 |
| Trio-Smart Breath Test | | | | |
| Result | | Normal | Normal | Normal |
| Hydrogen (ppm) | <28.16 | 17.85 | 12.35 | 15.62 |
| Methane (ppm) | <10 | 1.32 | 0 | 0.5 |
| Hydrogen Sulfide (ppm) | <3 | 1.6 | 1.85 | 1.53 |
| Biopsy Assay (μM/min/g protein) | | | | |
| Lactase | 15 to 45.5 | 9 | 3.7 | 2 |
| Sucrase | 25 to 69.9 | 14.4 | 12.7 | 8.7 |
| Maltase | 100 to 224.4 | 54 | 39.6 | 37.4 |
| Palatinase | 5 to 26.3 | 5.6 | 4 | 3.7 |
| Duodenal Biopsies | | Normal | Normal | Normal |
| Improvement with Sucraid | | Partial | Partial | Partial |

Figure 1: Results with References Ranges



1. Box: Serves as a self-mailer for returning samples to lab

2. Prepaid USPS return mailing label: This label must be applied to box for shipment back to lab

3. Step-by-step instructions for conducting a successful test

4. Lab test return form
Note: This form must be filled out by patient and returned with samples
5. Inside sleeve with QR code links to How-To instructional video

6. Tape: To seal box closed when mailing back to the lab

7. Sucrose (sugar) packet: Pour into 8-ounce glass of water and mix thoroughly until dissolved

8. Test tubes (4 total)

9. Straw

Figure 2: Metabolic Solutions ¹³C-sucrose breath test kit (completed at-home)

CONCLUSION

- Combination ¹³C-sucrose and Trio-Smart® breath testing was useful to confirm CSID and screen for confounders in this small case series
- Initiation of Sucraid® enzyme replacement improved but did not resolve symptoms
- Point of care ¹³C-sucrose breath testing may be close, but larger scale studies are necessary
 - Pros: Likely lower-cost, less invasive, may replace disaccharidase assays or be complementary, may be used for follow-up testing
 - Cons: Potential compliance issues, test validity when performed at home