Prevalence of Disaccharidase Deficiencies in Adults with Irritable Bowel Syndrome and Functional Diarrhea: Interim Analysis from a Multicenter, Prospective US Trial

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Abstract

Disaccharidases play an important role in the digestion of carbohydrates and starches. **Disaccharidase deficiencies can present similarly to IBS-D and functional diarrhea**. The prevalence of disaccharidase deficiencies have not been well defined amongst adults.

Symptomatic adult patients with IBS-D or functional diarrhea undergoing **upper endoscopy and duodenal biopsy revealed a disaccharidase deficiency rate of 36.4%. Nearly 1 in 7 of these patients had multiple forms of disaccharidase deficiency**. These results suggest that disaccharidase deficiency may be an important and overlooked cause of symptoms in US patients with IBS-D or functional diarrhea.

Introduction

Disaccharidases are brush border enzymes that digest disaccharides into monosaccharides for absorption across the intestinal epithelium (figure 1).[1] Deficiencies or dysfunction of disaccharidases can cause carbohydrate maldigestion resulting in symptoms including post-prandial abdominal pain, bloating, flatulence, and diarrhea.[2] Previous study has shown that disaccharidase deficiencies (DDs) can mimic symptoms seen with IBS-D and functional diarrhea (FD).[1,2]

Specific forms of DDs, in addition to generalized enzymatic deficiencies, have been well characterized in children.[3] However:

- DD prevalence in symptomatic adults is poorly defined
- A recent multicenter study found that 17% of adult patients produced positive breath testing suggestive of specific DDs[4]
- There is growing suspicion that DD could provide an underrecognized explanation for some patients suffering from apparent IBS-D or FD

The aim of this poster is to provide updates on an active US multicenter study assessing the prevalence of DDs in US adult patients with IBS-D and FD.

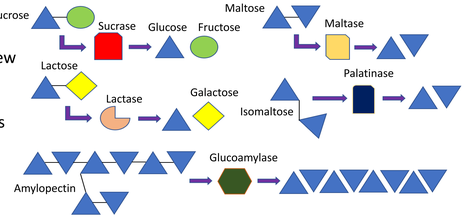


Figure 1. Overview of brush border disaccharidase activity pathways

Methods and Materials

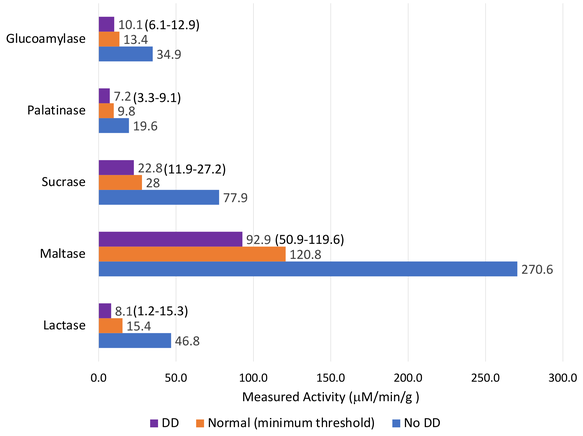
Adult (≥18 years) patients fulfilling the ROME IV diagnostic criteria for IBS-D or FD were recruited at Michigan Medicine (UM) and the University of Texas Health Science Center at Houston (UTH). Exclusion criteria included pregnancy, lactation, severe GI or abdominal co-morbidities, & history of previous GI surgery (excluding cholecystectomy or appendectomy).

Eligible patients completed upper endoscopy (EGD) with 2-4 small intestinal biopsies collected from the duodenum distal to the ampulla of Vater. Biopsy samples underwent disaccharidase assay (DA) to measure disaccharidase activity (minimum normal limits within figure 2) using validated protocols at experienced reference labs (Arnold Palmer Hospital Labs & Joli Diagnostics, Inc.).

Results

- There were no significant demographic, etiological, or diagnostic differences between patients recruited at UM (n: 113) and UTH (n: 41) (figure 3)
- All patients with a non-lactase form of DD had ≥1 additional form of DD
  - Pan-DDs were identified in ≥27% of these patients (table 2)

Figure 2. Mean Enzymatic Activity by Disaccharidase Type & Diagnosis Group

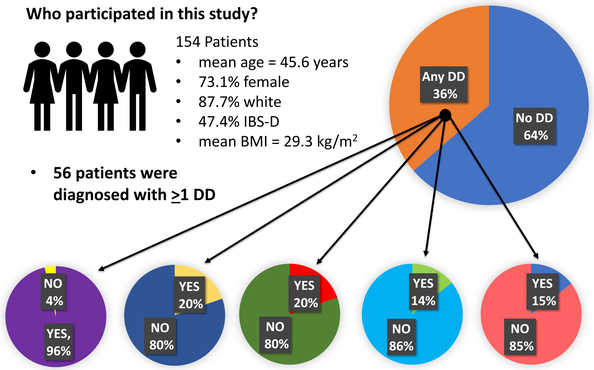


Results (cont.)

Who participated in this study?

154 Patients
- mean age = 45.6 years
- 73.1% female
- 87.7% white
- 47.4% IBS-D
- mean BMI = 29.3 kg/m^2

- **56 patients were diagnosed with ≥1 DD**

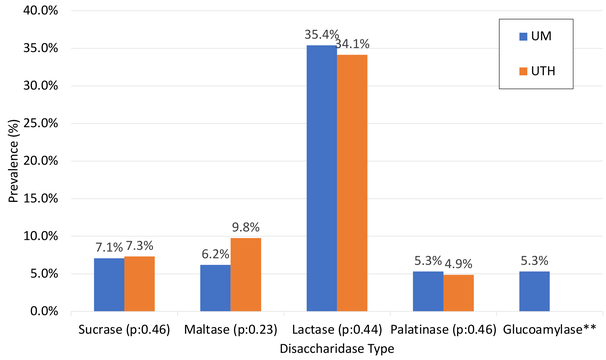


Lactase was the most common DD at 96% (54/56) within group, 35.1% (54/154) overall, and the only form to occur in isolation (table 2)

Sucrase-Isomaltase deficiency was identified in 20% (11/56) of patients within group, and 7.1% overall

Palatinase & Glucoamylase deficiencies were identified in 14% (8/56) & 15% (6/41) of patients within group, and at 5% overall

Figure 3. Disaccharidase deficiency prevalence rates by study site



**Glucoamylase was not measured for patients recruited and screened at UTH

Results (cont.)

Table 2. Characterization of Disaccharidase Deficiencies
A) Prevalence of single or multi-enzymatic deficiencies by disaccharidase
B) Prevalence of enzymatic deficiency overlap by disaccharidase

A	Total	<1 DD	1 DD	2 DD	3 DD	4 DD	5 DD
Total	154	63.64%	27.27%	2.60%	1.95%	2.60%	1.95%
Suc	11	0.00%	0.00%	9.09%	27.27%	36.36%	27.27%
Mal	11	0.00%	0.00%	18.18%	27.27%	27.27%	27.27%
Lac	54	0.00%	77.78%	5.56%	3.70%	7.41%	5.56%
Pala	8	0.00%	0.00%	12.50%	12.50%	37.50%	37.50%
Gluc	6	0.00%	0.00%	16.67%	0.00%	33.33%	50.00%

B	Suc	Mal	Lac	Pala	Gluc
Total	11	11	54	8	6
Suc	X	90.91%	16.67%	87.50%	83.33%
Mal	90.91%	X	16.67%	75.00%	66.67%
Lac	81.82%	81.82%	X	87.50%	100.00%
Pala	63.64%	54.55%	12.96%	X	66.67%
Gluc	62.50%	57.14%	15.00%	66.67%	X

DD: Disaccharidase Deficiency
Suc: Sucrase
Mal: Maltase
Lac: Lactase
Pala: Palatinase
Gluc: Glucoamylase

Discussion/Conclusions

- More than a third of US adult patients diagnosed with IBS-D or FD had at least 1 DD
- Lactase deficiency was most common and most likely to occur in isolation
- Sucrase & maltase deficiencies were identified in approximately 1 of 14 symptomatic patients (7.1%), and were typically associated with other forms of DD
- DD prevalence rates were nearly identical at both sites
- Lactase and non-lactase DDs may be important and overlooked causes for symptoms in US adult patients with IBS-D or FD

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