# A CASE OF CONGENITAL SUCROSE AND ISOMALTOSE INTOLERANCE

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## Review of the literature.

Much has been learned about digestion of disaccharides since the first clinical description by Durand in 1958[1] in an infant with fatal lactose intolerance. Disaccharides are confined to the outer cell layer of the intestinal epithelium and their separation from the intestinal mucosa was pioneered by Dahlqvist and many others[2-6]. Auricchio, Rubino and Murset by studying intestinal glycosidase activity in the human embryo, foetus and new born, found these activities present by the third month of intrauterine life[7]. All alpha – disaccharidase activities reach a maximum during the sixth or seventh month of intrauterine life, the beta – disaccharidases develop less rapidly during antenatal existence and reach their peak in the early perinatal period. Dahlqvist and Linberg showed that in man there appears generally to be a diminution of all disaccharidase activities in the duodenum and terminal ileum with relatively uniform levels of activity for each disaccharidase through out the remainder of the small intestine[8].

Congenital malabsorption of sucrose has been recorded in the Pediatric literature since 1960. It is thought to be present congenitally under a genetic control. While initially claimed by Prader to be an autosomal dominant inherited disorder, the more recent study suggests to be of an autosomal recessive[10]. Characterized by absence or low level of the intestinal enzyme sucrase, and almost always associated with deficiency in enzyme isomaltase, there is a characteristic malabsorption of sucrose. With the exception of a few cases in whom the symptoms appeared in later life[11], diarrhea began as soon as sucrose or dextrins were added to the milk formulae. Moreover, it disappeard when glucose, lactose or other sucrose isomaltose– free diets were substituted. Rosenthal and Cornblath (1962) reported a case of sucrose intolerance in a 21–month–old caucasian female with the symptoms of chronic low pH diarrheal stools since early infancy, who had a flat response to sucrose tolerance test but a normal blood sugar response after glucose, fructose and maltose ingestion[12,13]. A mixed form of lactose and sucrose intolerance reported by Sunshine (1964) in an infant also showed a good response to the special diets described[15].

## Report of A Case.

A $8\frac{1}{2}$ month–old–caucasian female was admitted to the Department of Pediatrics, University of Kansas Medical Center because of chronic diarrhea. She was the only child of the healthy, unrelated parent. Her birth weight was 7 lbs 5 ounces. She was placed on Enfamil during the first four days of life and after that was changed to whole milk. At 3 weeks of age, baby cereals were added. Vege-

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tables and fruits were added at 6 weeks. She then began to have increased in number of stools which were described as being loose, explosive, watery and foul-smelling and usually occured within 20-30 minutes after feedings. At 3 months of age the formular was changed to Mullsoy. The diarrhea had persisted despite numerous changes of formulae namely in sequences; skimmed milk, boiled whole milk, Nutramigen, and Neo-mullsoy. Despite the fact that the onset of diarrhea was at 6 weeks of age the weight gain had been within the average of 50 percentile until at 5 months of age when it had become plateau ever since. The weight at 5 months old and at $8\frac{1}{2}$ months old were 15 pounds even. The developmental milestones were within normal limit. The child's mother and father were in good health. Neither gave a history of diarrhea in infancy.

On admission, she was somewhat thin, undernourished but well developed, The weight was 15 lbs (3-10%) and the height was 28 inches (50-75%). The remainders of the physical examination were unrevealing.

The laboratory results were as follows: hemoglobin 13.4 gm/100 ml, white blood cell count 15,790/cu.mm. with 50 segmented neutrophils, 45 lymphocytes, 4 monocytes and 1 eosinophil, total serum protein 6.5 gm/100 ml with gammaglobulin 13%, beta-globulin 9%, alpha 2 globulin 11%, alpha 1 globulin 5% and albumin 60%. Fasting blood sugar, serum electrolytes and urinalysis were normal. The upper and lower GI series were noncontributory. Stool examination for ova and parasites were negative. The D-Xylose absorption test (0.3 gm/kg) was normal (25% of the ingested dose excreted in 5-hours urine).

During the first 5 days of her hospitalization she was fed with Neomullsoy and she had number of stools varied from 4-13 times a day. The stools pH varies from 4.5-7 and showed 2+ to 4+ positive reaction with Clinitest. Since the sixth hospital day she had been placed on glucose water and carbohydrate-free formula (Borden). The number of stools were 0-3 times/Day. The oral glucose tolerance test done on the 8th hospital day showed that the peak of absorption at $1\frac{1}{2}$ hrs specimen was abnormally high which was probably reflecting the condition following chronic diarrhea and malnutrition and/ or slow absorption of glucose through the bowel. The lactose tolerance test done on the 10th hospital day demonstrated a 43% increase in blood sugar level from the fasting. The sucrose tolerance test done on the 12th hospital day showed definitely a flat curve Fig I. Approximately 9 hrs. following the test she became fussy, irritable and refused to eat. She had one loose, foul-smelling stool within 15 minutes following the ingestion and had four more loose, watery, pasty coloured and foul-smelling stools within 24 hours. The small-bowel biopsy was performed under fluoroscopy and a piece of tissue of the terminal portion of the duodenum was obtained for the histopathologic study and also for the assessment of the intestinal enzymes content. The light microscopic picture of the duodenum was normal. The values of the intestinal enzymes are shown in figure II.

Fig. I Changes in levels of blood sugars after ingestion of glucose, lactose, & sucrose

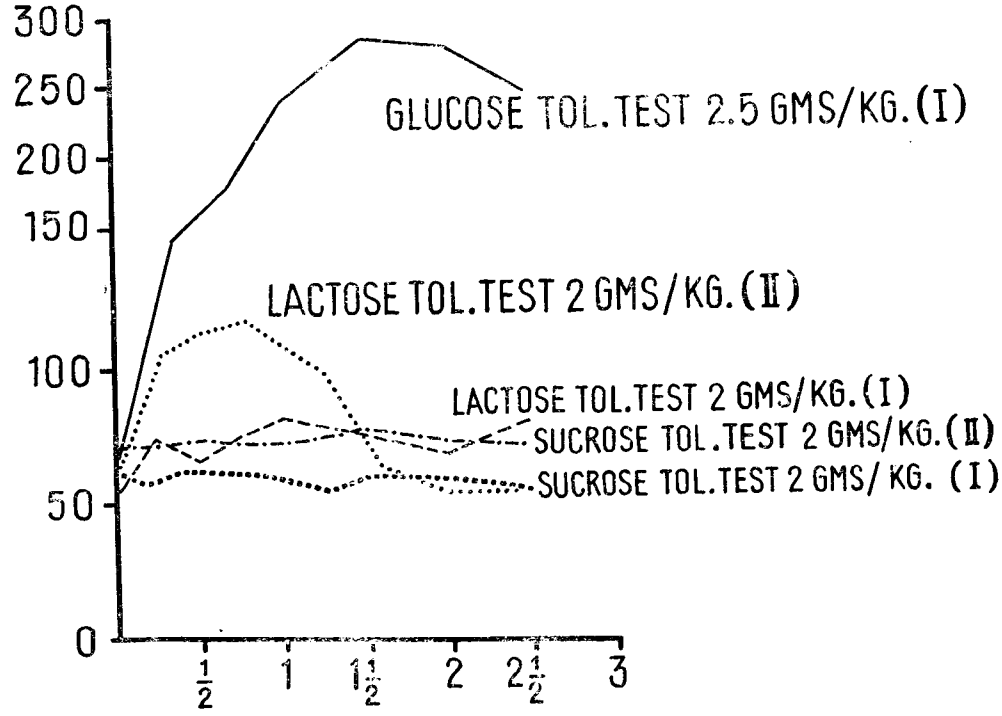


Fig. II. Disaccharidase levels in intestinal mucosa of the patient

| | *Enzyme Levels (Units)* | | | |
|---|---|---|---|---|
| | *Lactase* | *Maltase* | *Sucrase* | *Isomaltase* |
| Normal Values | 0.2–19 | 13–45 | 6–17 | 1.5–4 |
| Ist. Admission | 2 | 11 | 0.68 | – |
| 2nd. Admission | 3.2 | – | 0.06 | 0.3 |

She was dismissed and readmitted again 2 months later at which time she gained 1850 grams in weight while she had been on special diet containing less than 1.5 % of sucrose and isomaltose. The stools ran 2–3 times/day, varied from mushy brown to formed but never watery. She became more alert, active and much more enjoyable. Repeated absorption and intestinal enzymes studies were unchanged.

## Discussion

In general, patients with sucrose intolerance or isomaltose intolerance usually presented with profused low pH, watery diarrhea as soon as cane sugar was introduced into the diets. This symptom will coincide in the breast-fed infant with the start of weaning, and in the bottle-fed infant with the addition of sucrose and starch to the formulae used or with the regular administration of sugar-cane water between feedings.

In this case, she had done fine during the first 6 weeks of life while she had been on Enfamil, whole milk and cereals. At 6 weeks of age vegetables, bananas and oranges were added to the diets. The stools were then loose, explosive, watery, and foul-smelling with the frequency of 10–12 times a day and usually occurred within 20–30 minutes after the feedings and oftenly came during the feedings. It was quite plausible that these additional diets aggravated the symptoms since several of fruits such as apricots, bananas, dates, melons, oranges, pineapples have relatively high content of sucrose and among vegetable is green peas[14]. Since then she was placed on several kinds of formulae without any beneficial result. One may wonder why she didn't do any better with skimmed milk or boiled whole milk in which contains only the lactose. The additional history of her previous diets simply explained that the formulae had been switched at any given times, from one to anothers but she had consistently been exposed to vegetables and fruits.

The presence of low intestinal enzyme sucrase found in the initial study may either represent the primary defect in production of such enzyme of the intestinal mucosa in this child or be secondary to some underlying conditions such as cow-milk induced malabsorption, gluten enteropathy, infectious agents namely giardiasis or amebiasis. The latter possibility so called the secondary disaccharidase deficiencies is less likely because of the following remarks. In such cases, (1) the intestinal enzyme lactase is generally more affected than the intestinal enzyme sucrase and it quite often shows flat curves on absorption of both monosaccharides and disaccharides as well (2) the intestinal mucosa usually shows some change in the architecture such as blunting of the microvilli (3) symptoms and signs of the GI upset can be aggravated by being exposed to the particular agents such as to gluten in patients with gluten induced enteropathy, to giardia lamblia in the case of giadiasis; or be subsided after removal of such associated causes. And (4) the intestinal enzymes levels usually return back to normal after removal of the noxious agents.

Concurring with the above statements, the clinical manifestations and results of the absorption and intestinal enzymes studies indicated that this case represented

a Sucrose and Isomaltose Intolerance. Since the repeated absorption study of sucrose and intestinal enzyme sucrase during the state of well-being of the patient still demonstrated a flat absorption curve of sucrose and persistently low level of sucrase and isomaltase respectively, it renders a strong support that the Sucrose and Isomaltose Intolerance in this child representing a congenital deficiencies or defects in maltase and isomaltase.

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