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Diarrhea Caused Possibly By Deficiency of Sugar Splitting Enzymes

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Carbohydrates taken through foods are classified into two main groups: The disaccharides and polysaccharides. In the small intestine the disaccharides have to be broken into monosaccharides to be absorbed. Any deficiency in the amount of disaccharidase results in the appearance of certain clinical syndromes.[1 2 3]

Disaccharidases are usually found localized in the microvilli of the superficial areas of the mucosal cells.[2] In recent research work which were done employing the electron microscope, intestinal disaccharidase were observed to be localized on the surface of the microvilli in small knot-like forms.[4]

In this report, we present a case suffering from diarrhea, caused possibly by a deficiency in sugar splitting enzymes.

Case Report

A three and half year old boy was brought to the Out-Patient Clinic of Hacettepe Children's Hospital on February 28, 1968. with complaints of frequent diarrhea Starting at the age of one and a half, the boy had suffered from severe intractable diarrhea, causing failure to thrive.

The boy's mother had a normal pregnancy and labor. His weight at birth was also within normal limits. He was breast fed and given yoghurt until he was one and a half years old. While being breast fed his stools seemed to be normal. However, following introduction to supplementary foods, they became loose and continued to be as such. He received about 50 gm of sucrose and 40 gm starch each

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day in a cow's milk feeding. The child had five or six bowel movements a day, and sometimes experienced watery stool. The stool often seemed to contain an unusual amount of fluids.

On physical examination, the child was found to be underdeveloped for his age. He was pale and his muscles were weak. He weighed 11.5 kg. and his height measured 98 cm. He gave the impression of a malnourished child; his abdomen was found to be normal and systematic findings within normal ranges.

Laboratory Findings: Routine blood counts and several urinalyses revealed normal results. The findings gathered from various chemical blood analyses also were found within normal limits

Chest X-ray also was normal. Gastrointestinal series showed no abnormalities. Rectosigmoidoscopy carried out under general anesthesia revealed normal results. The probability of Giardia Lamblia was eliminated by the examination of stools. Results were negative on three consecutive occasions. Stool and throat cultures yielded no pathogenic organisms. There was no growth in the blood culture.

The Lactose Tolerance Test carried out with 2 gr. per kg. of 12 percent lactose in solution was given by mouth after eight hours of fasting. Following this, in 15, 30, 60 and 120 minute intervals, blood was taken from the finger of the patient.

After two days of rest following Lactose Tolerance Test, the child was again subjected to a period of fasting for 8 hours and then 2 gr. per kg. of weight of the 12 percent solution of glucose was given. Following a further two days rest, a Sucrose Tolerance Test was performed in exactly the same manner.

Within the 5 hour period after the tests, the urine and stool of the child were checked for pH contents, using pH hydrion paper.

Absorption seemed to be normal in Lactose and Glucose Tolerance Test (Figure 1). The Sucrose Tolerance Test showed a very "flat" curve, (Figure 2). Some diarrhea followed the oral load of sucrose. Stool pH content was 5 during the 5 hour period. The patient's family did not give permission for mucosal biopsy.

A sucrose and starch free diet was given to the patient and he progressed moderately well on this. His stools became firm and numbered, at the most, to two for each day.

On March 7, 1970, a physical examination showed the child's weight to be 17 kg. and height 109 cm. Follow-up clinic visits throug-

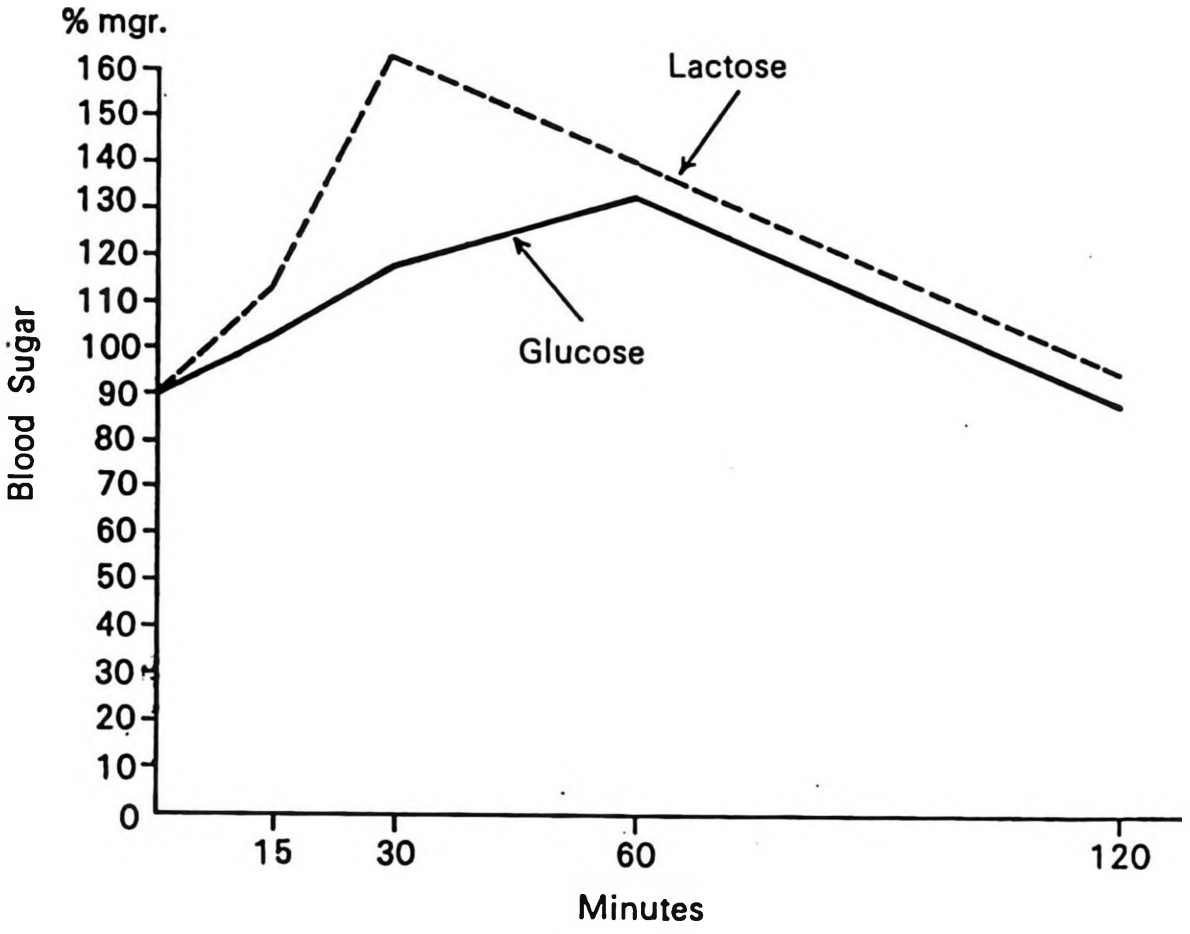

Figure 1

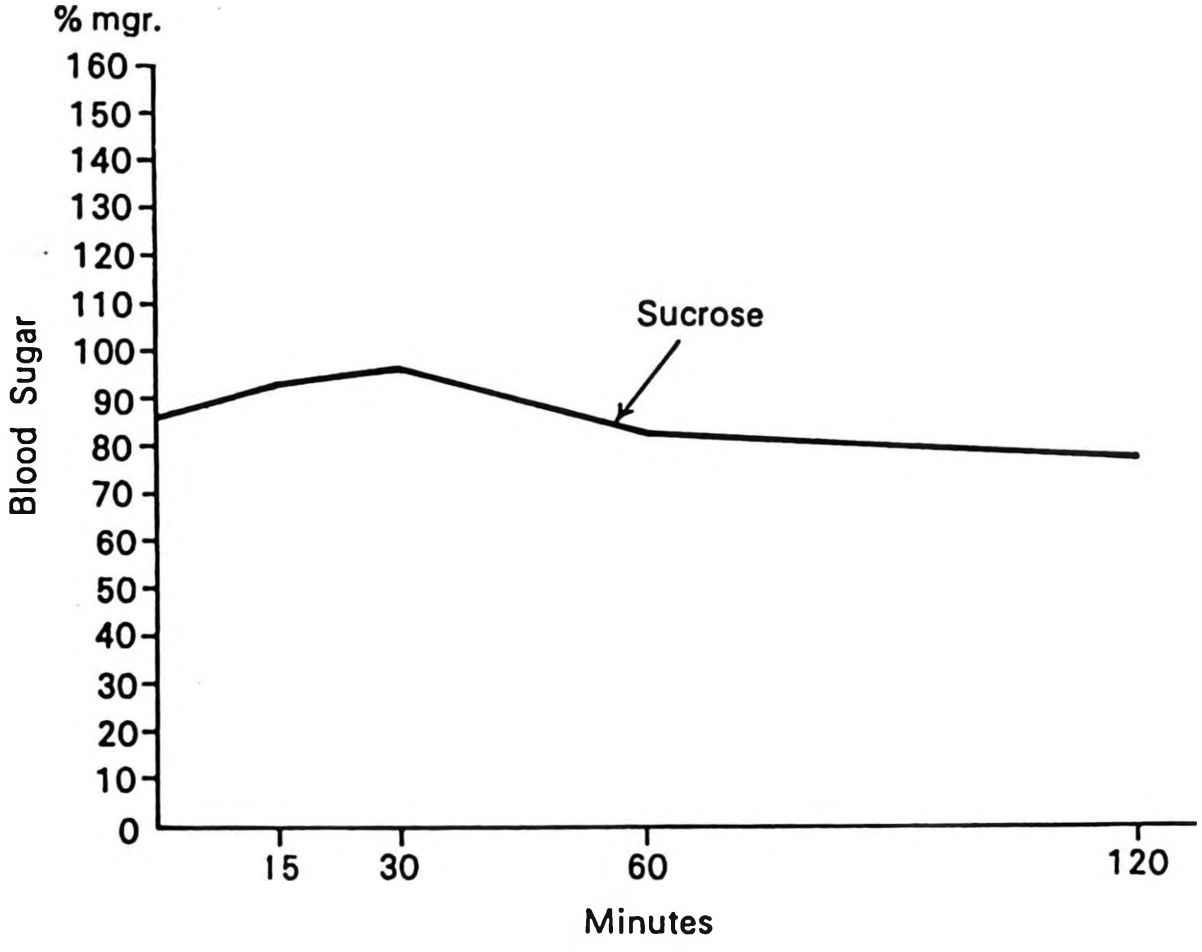

Figure 2

out that year showed a continued normal growth and development. On February 2, 1971, a physical examination indicated his weight to be 21 kg. and height 115 cm. (Figure 3). At that time, the age of the boy was 5 years 10 months. The sucrose and starch content of the child's diet have now been increased slightly and the bowel movements decreased in number to one or two each day and the stools are firmer.

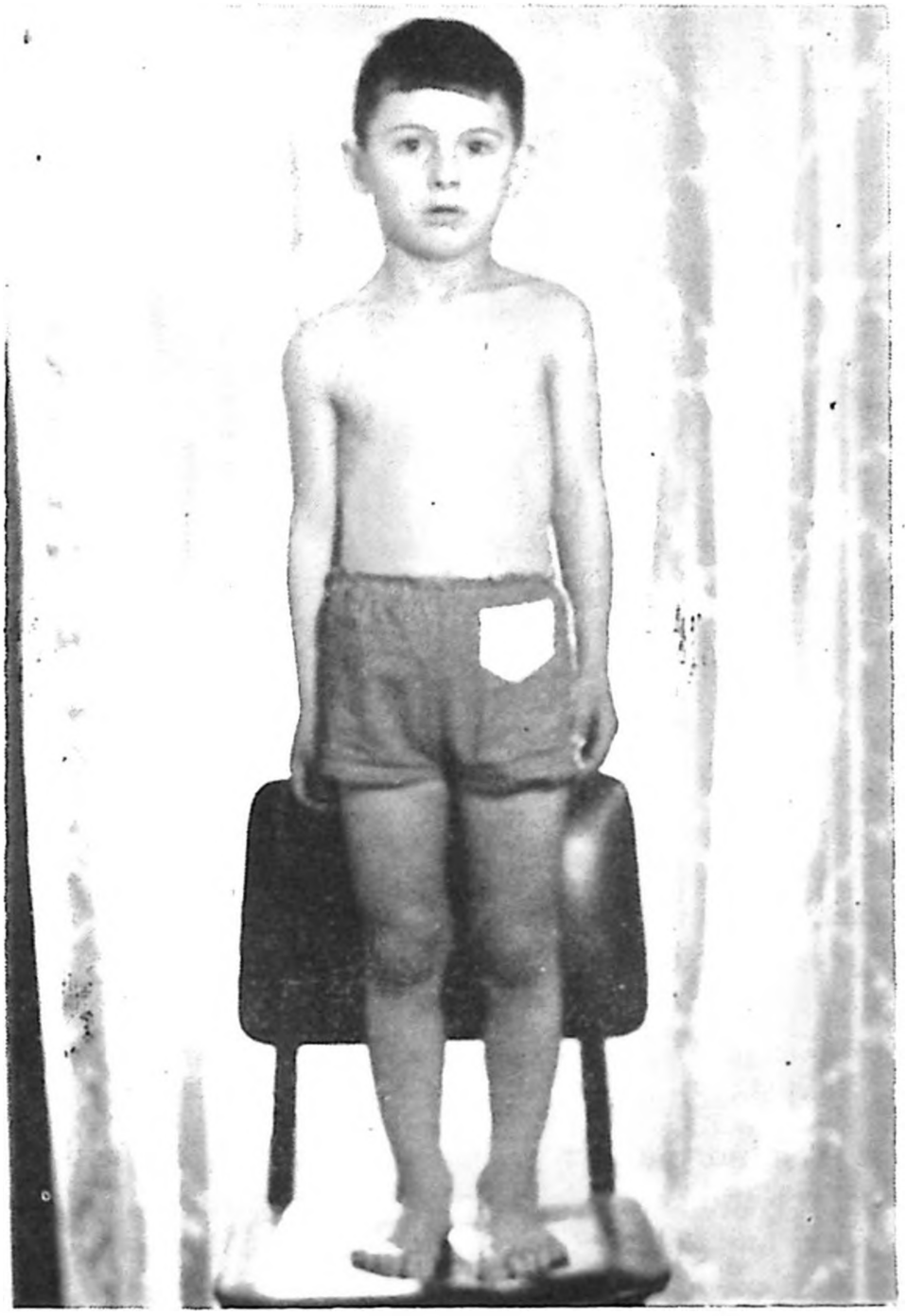

Figure 3

Discussion

Research on disaccharidases has been continuing for quite a number of years. In 1921, John Howland published a study on a case of con-

genital absence of disaccharidases.[9] Thirty-seven years later, in 1958, Durant reported a case under the title of "Lactosuria".[9] In 1959, Holzel, Schwarz and Sutcliffe published a paper describing two cases of defective lactose absorption causing malnutrition.[9]

In 1961, Weijers and his colleagues reported a case of sucrose and isomaltase deficiency.[13] In 1963, Anderson and his colleagues studied two cases showing a deficiency of intestinal sucrase and isomaltase.[1]

The results of the tolerance tests in our case are in agreement with those gathered in other reported cases of primary sucrase deficiency. There was no rise in blood sugar with sucrose and a normal rise with oral glucose and lactose load tests. The stool pH did not reach the expected low levels, probably because of the speedy movement of food through intestines or reduction of the enteric flora as a result of excessive use of antibiotics.

The intestinal disaccharidases are found localized in the microvilli of the superficial areas of the mucosal cells. A definitive diagnosis of disaccharidase deficiency therefore depends on the direct demonstration of the absence of these enzymes from the intestinal mucosa of the patient.[1]

In this case, the diagnosis was reached with the help of dietary exclusion of the offending disaccharides and oral loading of disaccharides tests. The patient's parents were uncooperative and they did not give permission for the examination of the intestinal mucosa of the patient.

Auricchio et al[7] have suggested that their patients with sucrose intolerance also had isomaltase deficiency. The absence of this enzyme from the small intestine may resul in diarrhea when large amounts of starch were consumend. When a sucrose and starch free diet was given to our patient he progressed well on it and his stools became firm and less in number each day.

Auricchio and Dahlaqvist have suggested that sucrose intolerance is an inherited condition. Recessive types of the disease also exist.[7]

When the history of our patient was checked, no familial diarrhea and sucrose intolerance was encountered It is possible that parents and other children are all heterozygous. The child may have berome homozygous through a mutation of the gene carried by parents in heterozygous forms. Father and mother were first cousins.

Summary

In this study, a chronic case of diarrhea was presented in a three and half year old boy, caused possibly by deficiency of sugar-splitting

enzymes. Diagnosis of the intestinal disaccharidase deficiency was reached with the help of indirect methods. Definite improvements were observed in the patient following the reduction of the amount of foods rich in sucrose and starch.

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