

Chronic Diarrhea in a 5-Year-Old Girl: Pitfall in Routine Laboratory Testing with Potentially Severe Consequences

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CASE

A 5-year-old girl was referred because of recurrent watery diarrhea, abdominal pain, and flatulence. She was the second of 3 children. Her 10-year-old sister was normally developed and healthy. Her younger brother has meningomyelocele and hydrocephalus.

At 1 year of age, the patient was admitted to the hospital for recurrent bronchitis, otitis media, food refusal, mild diarrhea, and abdominal distension since weaning. A sweat chloride analysis excluded cystic fibrosis. At that time, the patient's laboratory results showed increased C-reactive protein (139 mg/L; reference interval, <5 mg/L) and moderate leukocytosis ($14.6 \times 10^9/L$; reference interval, $4.5\text{--}13.5 \times 10^9/L$). Values for all other variables were within the reference intervals. Since then, the patient experienced recurrent abdominal pain, mild diarrhea, and occasional nausea. Stool samples were repeatedly analyzed for pathogenic bacteria without consistent relevant results. Beginning at the age of 3 years, the patient developed increasingly anomalous behavior, which included frequent episodes of fatigue and apathy, persistent enuresis, and encopresis (fecal soiling), delayed language development, inability to integrate in her kindergarten group, and continual food intake in parallel with the avoidance of sweets and fruit. Fasting glucose concentrations were measured several times, and all measurements were in the lower part of the reference interval [3.05–6.38 mmol/L (55–115 mg/dL)]. Interestingly, the parents were not aware of the patient's eating habits, because the mother was largely occupied with the care of her youngest child. Consequently, the 2 older children helped themselves most of the time.

A physical examination revealed abdominal distension, but the findings for the patient were otherwise unremarkable: blood pressure, 112/62 mmHg; temperature, 36.2 °C; height, 110cm (43.3 in, 38th percentile); weight, 19 kg (41.9 lb, 48th percentile).

Questions to Consider
• What are common causes of diarrhea in a child?
• What inborn metabolic errors may cause aversion to sweets and fruit?
• What tolerance testing may be hazardous in these metabolic disorders?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the May 2009 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol55/issue5> and follow the link to the Clinical Case Study and Commentaries.

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