Enterokinase Deficiency: A Case of Pancreatic Insufficiency

Manoj Madhusudan¹ · Srinivas Sankaranarayanan¹ · T. Ravikumar²

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To the Editor: Exocrine pancreatic insufficiency refers to a deficiency of exocrine pancreatic enzymes resulting in malabsorption [1]. The most common cause of exocrine pancreatic insufficiency in infants is cystic fibrosis [1]. A 2-mo-old female infant was born 2nd to non-consanguineous marriage with a birth weight of 3.3 kg. From the 2nd wk of life, she was noticed to pass frequent oily stools with no blood or mucus. There was no history of fever or vomiting. She had just gained 300 g in 2 mo. On inspection, she was found to be pale and having bilateral pitting pedal edema and mild hepatomegaly. Investigations revealed anemia (Hb - 7.7 g%), hypoalbuminemia (1.9 g/dL), normal counts, platelets, and liver and renal functions. Stool microscopy showed fat globules, and fecal chymotrypsin assay showed undetectable activity, suggesting exocrine pancreatic insufficiency. A possibility of cystic fibrosis was considered; however, sweat chloride analysis could not be performed because of inadequate sweat. She was started on pancreatic enzyme and fat-soluble vitamin supplementation, following which, her stools improved and she started gaining weight.

Clinical exome sequencing revealed a novel compound heterozygous mutation in Exome 12 [c.1350dupA(p.Val451SerfsTer21)] and 25 [c.3004G > A(p.Gly1002Arg)] of transmembrane serine protease 15 (*TMPRSS15*) gene at chromosome 21q21.1, which encodes for the enzyme enterokinase.

Enterokinase is an enzyme secreted by the duodenal mucosa. Its deficiency results in reduced activation of pancreatic enzymes trypsin, chymotrypsin, and carboxypeptidase. It is

extremely rare with fewer than 20 cases reported [2]. These children show dramatic response to pancreatic enzyme supplements and prior observations suggests that they gradually outgrow the malabsorptive state without the need for enzyme supplementation [3]. Our child is currently symptom-free with adequate weight and height on minimal pancreatic enzyme supplementation.

This case is being presented, as it is a close mimicker to cystic fibrosis and there has been a drastic improvement in clinical features upon initiating treatment.

Declarations

Conflict of Interest None.

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✉ Srinivas Sankaranarayanan
dr.srinis@gmail.com

¹ Department of Pediatric Gastroenterology, Kanchi Kamakoti CHILDS Trust Hospital, 12A, Nageshwara Road, Josier Street, Nungambakkam, Chennai, Tamil Nadu 600034, India

² Department of Pediatrics, Kanchi Kamakoti CHILDS Trust Hospital, Chennai, Tamil Nadu, India