Inborn metabolic disorders in differential diagnosis of cyclic vomiting syndrome in children

Ana Smajo, can. med.[a], Stipe Dumančić, can. med.[b], Mario Ćuk, MD, PhD[a,b]

[a]*School of Medicine, University of Zagreb, Zagreb, Croatia*

[b]*Department of Pediatrics, Division for Genetics and Metabolic diseases, University Hospital Centre Zagreb, Zagreb, Croatia; Assistant Professor of Pediatrics, Consultant in Pediatric Endocrinology and Metabolic Medicine, Research Associate, Senior authorship and mentor*

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INTRODUCTION Cyclic vomiting syndrome is a rare chronic disorder of unknown cause characterized by recurrent periods of frequent vomiting. The episodes are often triggered by infection and stress.Some metabolic disorders appear to hide under this clinical presentation. CASE PRESENTATION The patient is a 10-year-old girl with episodic vomiting since early childhood, born prematurely at 33 weeks and soon diagnosed with esophageal atresia with tracheoesophageal fistula which was operated the second day. Since the age of 3 she was frequently hospitalised due to episodic vomiting and poor weight gain. She underwent the Nissen fundoplication procedure at the age of 9. At the age of 10 she suffered from pneumonia accompanied by repeated vomiting episodes. Endoscopy, laboratory and radiology findings were unremarkable. Numerous infective episodes with vomiting and poor weight gain raise suspicion for metabolic etiology. The metabolic workup included several tests, all of which were negative. However, metabolic tests may not have been performed during the vomiting attack itself when they are most informative. We suggest performing metabolic tests during attacks when they are most valuable, including orotic acid in urine and allopurinol loading test to exclude urea cycle disorders. If these tests would also be negative, with persistent symptoms, we suggest OTC gene analysis. CONCLUSION The inborn errors of metabolism should be considered in the broad spectrum of differential diagnosis of recurrent vomiting in children, especially if accompanied with developmental delay, poor growth, and triggers such as infection.