Common variable immunodeficiency diagnosed by capsule endoscopy

Common variable immunodeficiency (CVID) is the most frequent primary immunodeficiency, characterized by reduced levels of immunoglobulins [1, 2]. Due to a heterogeneous presentation including a heightened rate of infections, the diagnosis is often delayed for 4–9 years. This delay may contribute to the increased morbidity and mortality in CVID patients [1, 3]. Gastrointestinal manifestations – most often chronic diarrhea [2] – are seen in up to 50% of CVID patients. If performed, capsule endoscopy can reveal characteristic extensive nodular lymphoid hyperplasia of the small intestine in up to two-thirds of patients [4]. If only upper and lower endoscopy are used in the investigation of chronic diarrhea, nodular lymphoid hyperplasia of the duodenum and the terminal ileum can be misinterpreted as celiac or Crohn’s disease [5].

We present the first cases of patients diagnosed with CVID based on capsule endoscopy.

Case 1 was a 40-year-old man with severe symptoms including watery diarrhea, weight loss, recurrent gastrointestinal infections, and grave malnutrition. The symptoms did not fulfill the diagnostic criteria for celiac disease or other sprue-like diseases. The patient’s IgG level was low and IgA not measurable. Capsule endoscopy showed extensive focal nodular lymphoid hyperplasia throughout the entire small intestine (Fig. 1, Video 1). Biopsies from the small bowel revealed intraepithelial lymphocyte infiltration (Fig. 2). Infusion of human IgG improved all gastrointestinal symptoms.

Case 2 was a 60-year-old man with mild symptoms including watery diarrhea, abdominal distension, and recurring upper respiratory tract infections for the previous 25 years. He had low IgG and IgA. Colonic biopsies showed eosinophilia. Upper endoscopy was normal. Capsule endoscopy revealed extensive focal nodular lymphoid hyperplasia throughout the entire small intestine.

These cases emphasize the need for gastroenterologists to be familiar with the link between CVID, chronic diarrhea, and extensive nodular lymphoid hyperplasia in the small intestine, especially because early treatment with human IgG can reduce morbidity and mortality. In evaluation of patients with known CVID or chronic diarrhea of unknown origin, we recommend capsule endoscopy as a diagnostic tool.

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References

Bibliography
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