gutflix
GI learning, personalised

Summary

When it looks like coeliac disease but isn’t: Unmasking the mimickers

Gerd Bouma

Postgraduate Teaching Programme Barcelona 2026 | Presentation

Paediatrics | Small Intestine & Nutrition | Primary Care | Small Intestine & Nutrition

October 17, 2026

This presentation addresses the diagnostic approach to villous atrophy when coeliac disease does not fit, illustrated by a case of indolent T cell lymphoproliferative disease presenting with seronegative villous atrophy.

KEY POINTS

- Coeliac disease is by far the most common cause of villous atrophy, but negative serology does not completely exclude it, though seronegative coeliac disease is very rare.
- Negative coeliac serology combined with absence of increased intraepithelial lymphocytes argues against coeliac disease and should prompt consideration of alternative diagnoses.
- The speaker presented a case of a 40-year-old man with villous atrophy, negative coeliac serology, normal IEL count of 20, and HLA DQ2.5 positivity who did not respond to a gluten-free diet.
- Revised biopsies revealed an abnormal monomorphic T cell population with clonality and a somatic JAK2 mutation, leading to diagnosis of indolent T cell lymphoproliferative disease treated with corticosteroids and a JAK inhibitor.
- The differential diagnosis for unexplained villous atrophy includes infectious causes, drugs, immune-mediated diseases, and neoplastic lesions; refractory coeliac disease type 2 is extremely rare and is actually a low-grade intraepithelial lymphoma.
- The speaker emphasized reviewing biopsy adequacy and orientation with a pathologist, and reconsidering medications and infections before diagnosing rare conditions, and recommended referring challenging cases to specialized centres.

VIEW THIS CONTENT ON GUTFLIX

https://gutflix.eu/search/d/bff5c836-d14f-11f1-91c5-daaa6dbee0a2

AI-generated summary

This summary was generated by an AI large language model based on the content transcript. It is for informational purposes only and should not be considered a substitute for clinical judgment. Always rely on your professional expertise and the full clinical context when making clinical decisions.