Accepted Manuscript

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PII: S0016-5107(18)30041-5

DOI: [10.1016/j.gie.2018.01.011](https://doi.org/10.1016/j.gie.2018.01.011)

Reference: YMGE 10903

To appear in: *Gastrointestinal Endoscopy*

Received Date: 3 November 2017

Accepted Date: 11 January 2018

Please cite this article as: Struyve M, Ferrante M, Van Overbeke L, Meersseman W, Cassiman D, Unusual yellow scaly colonic mucosal appearance: Tangier disease, *Gastrointestinal Endoscopy* (2018), doi: 10.1016/j.gie.2018.01.011.

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Unusual yellow scaly colonic mucosal appearance: Tangier disease.

Short title – running head: Colon mucosa in Tangier disease.

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Conflict of interest: None declared.

Electronic word count: 180

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Mathieu Struyve	-	Disclosures:
	-	Speakers fee: AstraZeneca, Janssen-Cilag NV
	-	First author of this article.
Marc Ferrante	-	Disclosures:
	-	Speakers fee: Abbvie, Boehringer-Ingelheim, Chiesi, Ferring, Janssen, MSD, Pfizer, Tillotts, Zeria
	-	Consultancy: Abbvie, Boehringer-Ingelheim, Ferring, Janssen, MSD, Pfizer
	-	Research grant: Takeda
	-	Endoscopist in this case.
Lode Van Overbeke	-	No disclosures
	-	Endoscopist in this case
Wouter Meersseman	-	No disclosures
	-	Physician specialized in metabolic disorders that settled the diagnosis of Tangier disease.
David Cassiman	-	Disclosures
	-	On behalf of David Cassiman, the University of Leuven and University Hospitals Leuven have received research grants, travel and conference bursaries, speaker fees and advisory board compensations from Sanofi-Genzyme, Shire, Actelion, Bayer, Roche, BMS, Schering-Plough, Synageva, Chiesi, Alexion. Unrestricted research grants were received from Sanofi-Genzyme, Shire and Actelion.
	-	Physician specialized in metabolic disorders that settled the diagnosis of Tangier disease.

Key words: Colon – Cholesterol – Hypo-alphalipoproteinemia – Tangier disease

A 51-year-old man was referred for a colonoscopy because of abdominal discomfort and loose stools as seen with irritable bowel syndrome. His medical history consisted of atherosclerosis and a tonsillectomy. Blood analysis revealed an abnormal lipid level with a total cholesterol level of 80 mg/dL (normal < 190 mg/dl), an undetectable low (<3 mg/dl) HDL cholesterol level (normal > 40 mg/dL), a high triglycerides level of 807 mg/dl (normal < 150 mg/dl) and an undetectable low apolipoprotein A1 (< 0.20 g/L) (normal 1.04 – 2.02 g/L) and apolipoprotein A2. Colonoscopy revealed an irregular discoloration of the colonic mucosa with a yellow-orange appearance (A, B), innumerable small yellow-brown dots with a scaly to cobblestone pattern, and multiple polyps throughout the colon (C). Histological analysis revealed colonic mucosa with focal aggregates of foam cells and foamy histiocytes in the lamina propria and submucosa. Most polyps were hyperplastic deformed colonic mucosa with an excess of foamy histiocytes or so-called xanthomateous lesions, other polyps were adenomatous lesions (D, E, [orig. mag. x50] F, G [orig. mag. x400], CD68 staining. Genetic testing showed a homozygous variant in the ABCA1-gene (c.5785G>T,p.(Val1929Phe)). The diagnosis of hypo-alpha lipoproteinemia due to Tangier disease was made.

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