A mysterious anus

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A 74-year-old man presented at the consultation of gastro-enterology with a painfully ear-shaped swelling (Fig. 1A and 1B) around the anus. He denied fever, weight loss, or diarrhea. His medical history included coronary artery disease, arterial hypertension, atrial fibrillation, melanoma on the lower back with curative resection, a total hip replacement on the left side and knee prosthesis on both sides. Current medication use included acetylsalicylic acid 80 mg, bisoprolol 2,5 mg and acenocoumarol 2 mg. Several biopsies were taken and showed aspecific polypoid granulation tissue without any sign of malignancy. Echo-endoscopy revealed no invasion in the surrounding muscular layers. A left-sided colonoscopy showed no abnormalities. Finally, the lesion was surgically removed.

What is your diagnosis ?

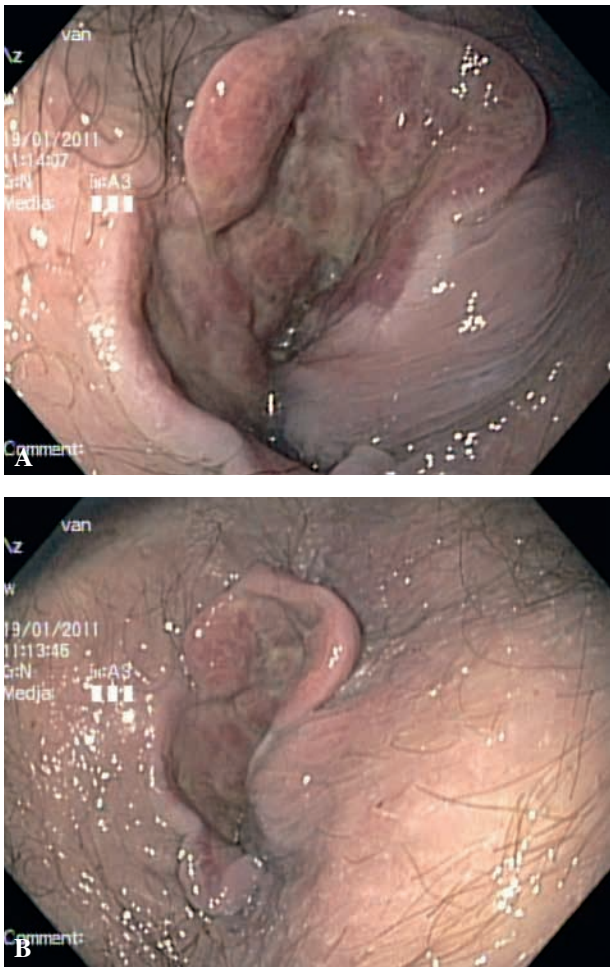

Fig. 1A, B.

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Answer

The diagnosis of amyloidosis was retained. The patient was subsequently referred to a hematologist who performed a bone marrow biopsy, which showed no signs of AL (primary) amyloidosis. Two years later a total colonoscopy was performed in routine follow up which showed small amyloid-containing ulcerations in the sigmoid colon. However given the patient's age, the favourable clinical course and the limited therapeutical repercussion no specific therapy was started until present.

Amyloidosis is a generic term for diseases that have in common the extracellular deposition of insoluble fibrillar proteins in tissues and organs. The two major forms are AL (primary) amyloidosis and AA (secondary) amyloidosis, but more types are described and specification of the subtype can be difficult. Clinical manifestations vary depending upon the type of amyloid and the distribution of deposition.

Tissue biopsy with Congo red staining (in stead of routine staining) is obligatory in all cases to confirm the diagnosis of amyloidosis. Treatment of the different types of amyloidosis varies with the cause of fibril production.

Suspicion of gastrointestinal amyloidosis in patients without known history of amyloidosis is difficult but should be considered in those older than 30 years with gastrointestinal bleeding, dysmotility, malabsorption or protein-losing gastro-enteropathy. Hepatomegaly with or without splenomegaly is a frequent finding. While most gastro-intestinal complications are managed symptomatically, causal therapy is reserved for a selected few from various subtypes of this disorder.