

Pseudotumoral Anal Beta2-Microglobulin Amyloidosis: About an Observation

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Abstract

Beta2-microglobulin amyloidosis is a serious complication of chronic hemodialysis. The usual manifestations are osteoarticular and ligamentous, cutaneous and mucosal involvement being relatively infrequent. We report an exceptional aspect of perianal pseudocondylomatous lesions secondary to amyloidosis in chronic hemodialysis associated with amyloid arthropathy. The diagnosis of amyloidosis is histological, and in the absence of effective treatment, management remains symptomatic in order to improve quality of life.

Keywords: Amyloidosis; Amyloid Arthropathy; Chronic Hemodialysis; Perianal

Introduction

Amyloidosis corresponds to the pathological aggregation of proteins due to a folding defect. Rudolf Virchow was the first to describe what he called the amyloid substance. It can be deposited in all major organs (kidney, heart, skin, central and peripheral nervous system) including the digestive organs (esophagus, stomach, small bowel, colon, rectum, liver, spleen, pancreas) [1]. We then owe Samuel Wilks the first description of primary amyloidosis [2]. In 1867, Weber established the link between amyloidosis and multiple myeloma [3].

More than fifteen types of amyloidosis have been described. Primary (AL) or secondary (AA) systemic forms are by far the most common. Beta2-microglobulin amyloidosis is a serious complication of chronic hemodialysis. The usual manifestations are osteoarticular and ligamentous, cutaneous and mucosal involvement being relatively infrequent. We report an exceptional aspect of perianal pseudocondylomatous lesions secondary to amyloidosis in a chronically hemodialyzed patient treated for chronic hepatitis C.

Case Report

This is the case of a 70-year-old man who has been chronically hemodialyzed on vascular nephropathy since 1995. In his history, a cholecystectomy in 1994, a parathyroid adenoma operated in 2007, a chronic hepatitis C genotype 2a with fibrosis assessed by Fibroscan as minimal to moderate F1-F2, treated with pegylated dual therapy in 2010 with a sustained viral response 2 years after the end of the treatment and an amyloid arthropathy in 2011 for which he was put on steroids with analgesics. The patient presented an isolated anal pain without rectal bleeding nor transit disorders since 2015. Clinical examination found a hard nodular lingual infiltration with a difficulty of protraction of the tongue while proctological examination found perianal lesions with a whitish keratotic surface, hard and ulcerated in places extending to the intergluteal fold (Figure 1). 2 sets of biopsies of the anal lesions performed with a forceps concluded to a fibroepithelial polyp. Pelvic MRI did not show any suspicious anal lesions. Further investigation with an esophagogastroduodenoscopy,

colonoscopy and an abdominal ultrasound along with a biological assessment in search of a gammopathy returned negative. Anatomopathological examination of anal surgical biopsies performed under sedation found pseudotumoral amyloidosis with no histological signs of malignancy (Figure 2A and 2B). The patient was put on a symptomatic treatment with a steady evolution to date.



Figure 1: Keratotic and whitish anal lesions.

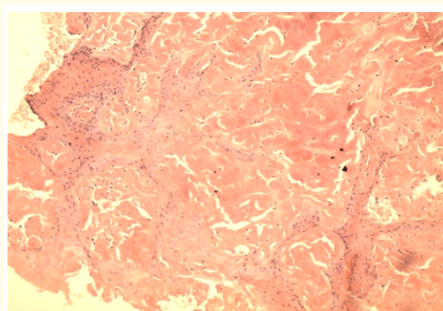


Figure 2A: Pseudo-tumoral amyloidosis after Congo Red staining x 10.

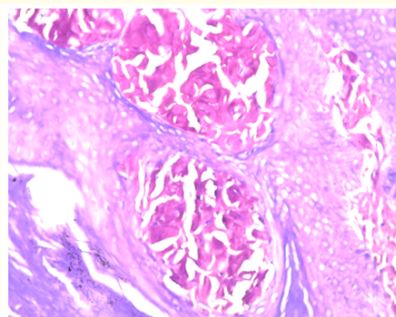


Figure 2B: Amyloid deposits with Crystal Violet staining.

Discussion

Amyloidosis is a rare disease. In the United States, there are between 1275 and 3200 new cases per year [4]. B2-microglobulin amyloidosis is a long-term complication of dialysis regardless of the type of treatment (hemodialysis or peritoneal dialysis). It is secondary to the deposition of an amyloid substance composed mainly of b2-microglobulin which, normally eliminated by the kidneys, accumulates in dialyzed patients and is mostly responsible for an osteoarticular symptomatology [5]. Its incidence is higher when the duration of hemodialysis exceeds 10 years, one study showed that the overall prevalence reaches 48% of the patients on HD for a median of 47 months [6], another study conducted in Italy demonstrated that the age of onset of HD treatment is considered as an one of the independent risk factor promoting Dialysis-related amyloidosis(DRA) meaning that younger people are less liable to develop this complication than older people, and given that the survival in hemodialyzed patients is more long-lasting, is seen more frequently in older patients [7]. The characteristic lingual involvement is a nodular infiltration of yellowish color as the case of our patient. Cutaneous involvement is characterized by β -2-microglobulin deposits in the dermis, observed in hemodialyzed patients after an average of 8 to 10 years of dialysis. Clinical manifestations are mainly osteoarticular and ligamentous: carpal tunnel syndrome and destructive arthropathies that are present in our patient. Cutaneous signs are not very specific and rare: subcutaneous nodules on the buttocks, frequently bilateral [8] as in our patient, hyperpigmentation, lichenoid papules, infiltrated digital lesions and prurigo [8,9].

Involvement of the anogenital region can mimic condylomas and often leads to a late diagnosis. The confirmation of the diagnosis of amyloidosis is histological. Under an optical microscope, after Congo Red staining, which is indispensable for the diagnosis, a "green-apple" appearance corresponding to a birefringence under polarized light, very specific to the amyloid substance, is observed [10]. Electron microscopy shows helical fibrils. X-ray diffraction analysis shows the pathognomonic arrangement in flat sheets perpendicular to the axis of the fibril. It is then necessary to carry out typing using immunohistochemistry (anti-amyloid protein antibody), immunofluorescence on frozen sample and more recently "immuno-gold" in electron microscopy, or mass spectrometry, in order to specify the type of amyloidosis.

According to literature, 1 case of condyloma-like perianal vegetation was reported during the amyloidosis of a hemodialyzed patient in 2013 at the dermatology department in Rabat [11]. Such an aspect has already been described in an AL amyloidosis related to multiple myeloma [12]. A similar appearance at the vulvar level was reported in 4 patients during systemic or localized immunoglobulin amyloidosis. The treatment is based on low-dose corticosteroids and more importantly, the use of high-flux dialysis membranes with columns allowing absorption of the amyloid precursor. The use of these membranes would be useful in preventing the progression of amyloidosis [13]. Renal transplantation allows a rapid regression of joint symptoms, but its action on amyloid deposits remains poorly understood.

Conclusion

We report an original case of pseudotumoral anal amyloidosis in a patient treated and cured for chronic hepatitis C, on chronic hemodialysis complicated by amyloid arthropathy caused by β -2-microglobulin deposits. The diagnosis of amyloidosis remains histological and, in the absence of a current effective treatment, management is symptomatic in order to improve quality of life.

Bibliography

1. Kyle RA and Bayrd ED. "Amyloidosis: review of 236 cases". *Medicine (Baltimore)* 54.4 (1975): 271-299.
2. Wilks S. "Cases of lardaceous disease and some allied affections. With remarks". *Guy's Hospital Reports* 2 (1856): 103.
3. Weber H. "Mollities ossium, doubtful whether carcinomatous or syphilitic". *Transactions of the Pathological Society of London* 18 (1867): 206.
4. Feng D, et al. "Intracardiac thrombosis and anticoagulation therapy in cardiac amyloidosis". *Circulation* 119.18 (2009): 2490-2497.

5. Bardin T., *et al.* "Synovial amyloidosis in patients undergoing long-term hemodialysis". *Arthritis and Rheumatology* 28.9 (1985): 1052-1058.
6. Jadoul M., *et al.* "Histological prevalence of beta 2-microglobulin amyloidosis in hemodialysis: a prospective post-mortem study". *Kidney International* 51.6 (1997): 1928-1932.
7. R Scarpioni., *et al.* "Dialysis-related amyloidosis: challenges and solutions". *International Journal of Nephrology and Renovascular Disease* 9 (2016): 319-328.
8. Shimizu S., *et al.* "Subcutaneous nodules on the buttocks as a manifestation of dialysis-related amyloidosis: a clinico- pathological entity? *British Journal of Dermatology* 149.2 (2003): 400-404.
9. Uenotsuchi T., *et al.* "Cutaneous and lingual papules as a sign of beta 2 microglobulin-derived amyloidosis in a long-term hemodialysis patient". *European Journal of Dermatology* 13.4 (2003): 393-395.
10. Modiano P. "Amyloses cutanées". *Annales de Dermatologie et de Vénérologie* 132.1 (2005): 62-70.
11. W Raffas., *et al.* "Végétations péri-anales pseudocondylomateuses au cours d'une amylose chez un hémodialysé chronique". *Annales de Dermatologie et de Vénérologie* 140.12 (2013): 607-608.
12. Roterman I., *et al.* "Why Congo red binding is specific for amyloid proteins - model studies and a computer analysis approach". *Medical Science Monitor* 7.4 (2001): 771-784.
13. Gejyo F., *et al.* "Arresting dialysis-related amyloidosis: a prospective multicenter controlled trial of direct hemoperfusion with a beta2-microglobulin adsorption column". *Artificial Organs* 28.4 (2004): 371-380.

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