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Case Report

Primary Localised Duodenal Amyloidosis: A Case Report

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Abstract: Amyloidosis is a condition resulting from extracellular deposition of insoluble fibrillar protein formed from various precursor proteins causing structural and functional disruption. We present a patient with primary localised duodenal amyloidosis who presented with gastrointestinal haemorrhage. Endoscopy showed friable mucosa with raised telengiectatic lesion in duodenum. Biopsy showed the presence of amyloid deposition. IHC studies showed positive lambda light chain staining. Patient had no evidence of an underlying monoclonal plasma cell disorder or other organ involvement. Therefore, we concluded the case as localised primary amyloidosis probably due to extramedullary monoclonal proliferation of plasma cells.

Keywords: Localised Primary amyloidosis, Duodenum, AL amyloid.

INTRODUCTION

Amyloidosis is a condition resulting from extracellular deposition of insoluble nonbranching fibrillar protein formed from various precursor proteins causing structural and functional disruption in tissues (Inayat, F., & Hurairah, A. 2016; Park, S. W. *et al.*, 2018; & Hokama, A. *et al.*, 2011).

Primary amyloidosis is caused by plasma cell or a lymphoplasmacytic neoplasm in which the monoclonal plasma cells secrete intact or fragments of abnormal immunoglobulin light chains that deposit in various tissues and form a beta pleated sheet structure (amyloid light chain) (Swerdlow, S. H. *et al.*, 2017). Secondary amyloidosis or Reactive systemic amyloidosis is characterised by deposition of Amyloid associated (AA) protein formed by incomplete breakdown of serum amyloid A, an acute phase reactant. Hence, it is associated with chronic inflammatory conditions such as Rheumatoid arthritis, Crohn's disease, familial Mediterranean fever, leprosy and tuberculosis. Both types are usually systemic in distribution (Park, S. W. *et al.*, 2018). Localised amyloidosis is a condition in which amyloid infiltration is limited to a single organ in the absence of any systemic disease and plasma cell dyscrasia (Cowan, A. J. *et al.*, 2013).

Although amyloidosis of gastrointestinal tract is not uncommon, the variable clinical features and endoscopic findings make this condition difficult to diagnose clinically (Hokama, A. *et al.*, 2011). In addition, amyloidosis localised to small intestine, with complete sparing of colon and in the absence of underlying systemic disease is only rarely observed (Choi, J. H. *et al.*, 2014). We present a patient with primary localised duodenal amyloidosis, with no evidence of any underlying systemic disorder, who presented with gastrointestinal haemorrhage.

CASE REPORT

A 74 year old elderly man presented with complaints of early satiety, fatigability and bilateral leg swelling for the past 1 month and black coloured stools for the past 1 week. He had a history of sputum positive pulmonary tuberculosis 2 years back with completed antituberculous therapy. He had no history of chronic alcoholism. On examination, the patient was found to be moderately built and poorly nourished with a BMI of 17.5kg/m2. He had pallor, glossitis, cheilitis, tachycardia and bilateral pitting pedal oedema. He had normoactive bowel sounds with no abdominal tenderness, hepatomegaly or splenomegaly. Laboratory workup revealed anaemia (Hb-8.3g%, MCV-70.3fL,

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MCH- 20.7pg, MCHC- 31.5g/dL) mild leucocytosis (WBC count-11.800/mm^3) and thrombocytosis (Platelet-6.8Lakh/mm^3). He had a raised ESR-80mm in 1st hour and elevated CRP- 20.5mg/dL. His total protein was reduced (5.4 g/dL) with hypoalbuminemia (1.7g/dL) and reversal of Albumin Globulin ratio (0.47). Serum Creatinine and blood Urea levels were normal. Other liver enzymes were also normal. Urine analysis revealed trace albumin, microscopy showed no casts and 24 hour urine protein was normal (148mg/dL). CEA was normal (2.16ng/ml). USG abdomen showed a normal study. CECT abdomen showed diffuse wall thickening with post contrast enhancement in duodenum causing luminal narrowing. Colonoscopy revealed normal study up to terminal ileum except for Grade 2 external haemorrhoids. Oesophagogastroduodenoscopy revealed friable mucosa with telengiectatic raised lesions with no active oozing in first and second part of duodenum only [Fig 1.0]. Biopsy was taken from the same.

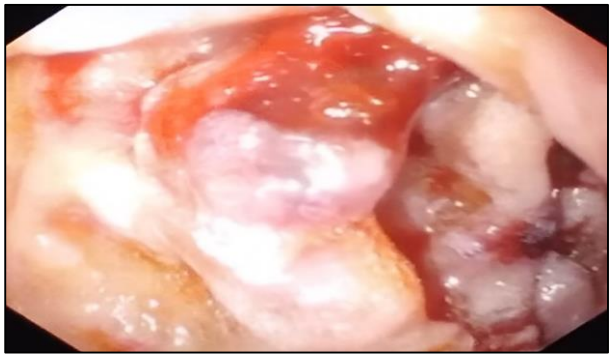

Fig 1.0 Endoscopy showing telengiectatic raised lesion

Histopathological examination of bits of duodenal mucosa showed lymphoplasmacytic infiltration and extracellular deposition of homogenous eosinophilic material in lamina propria. [Fig 2] Congo Red staining revealed red coloured amyloid deposition [Fig 3] and apple green birefringence under polarised light microscopy [Fig 4]. To differentiate between AL and AA amyloidosis, Immunohistochemical study was done which showed lambda positivity, suggesting AL amyloidosis [Fig 5.1, 5.2].

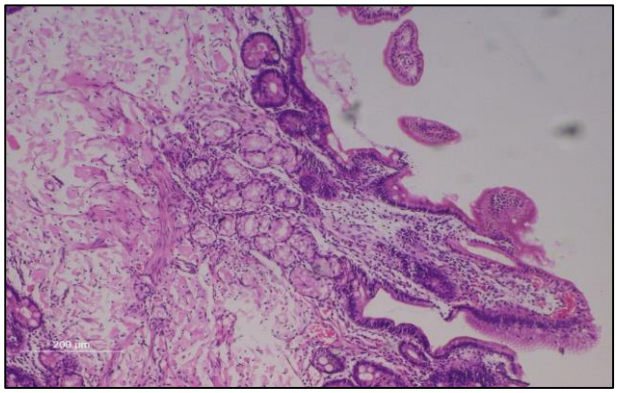

Fig 2.0 Biopsy No: 3526/2019: Duodenal biopsy- lymphoplasmacytic infiltration and extracellular deposition of homogenous eosinophilic material in lamina propria.

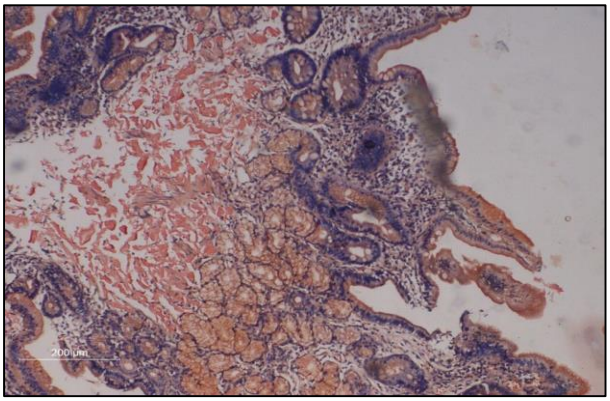

Fig 3.0 Congo red staining – red coloured amyloid deposition

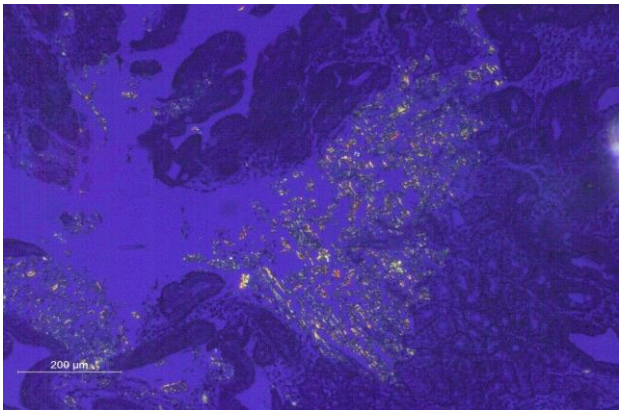

Fig 4.0: Apple green birefringence under polarised light microscopy

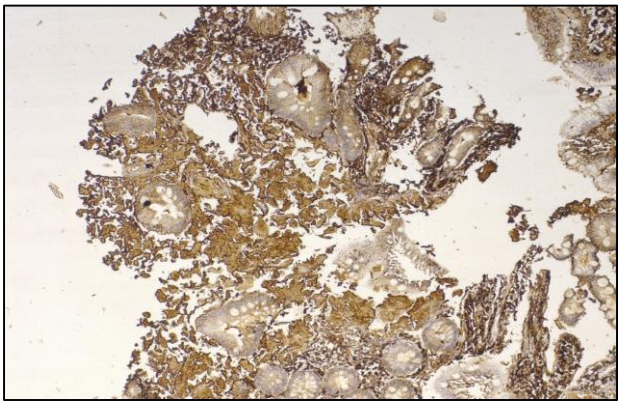

Fig 5.1. IHC 507/2019 Lambda light chain positivity

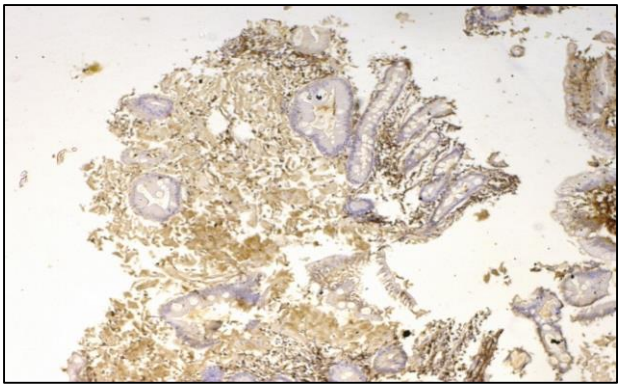

Fig 5.2. IHC 507/2019 Kappa light chain negative

His Serum Calcium was within normal limits (10.4g/dL). Skeletal survey was done which showed no focal punched out lytic lesions. Serum Protein Electrophoresis was done which showed hypoalbuminemia with polyclonal gammopathy. Bone marrow study was done which showed a mild increase in plasma cells (8% plasmacytosis) scattered in the interstitium and IHC study of Bone marrow showed polyclonal kappa and lambda positivity. Biopsies were carried out from other parts of intestine- ileal and rectal mucosa which showed no amyloid deposition or other inflammatory process.

Follow up: He had no active oozing on follow up endoscopy and was discharged on haematinics. At present, his anemia is corrected and is symptomatically better.

DISCUSSION

Gastrointestinal amyloidosis is common in individuals with systemic amyloidosis, small intestine being most common site (Hokama, A. *et al.*, 2011). Gastrointestinal symptoms are diverse and nonspecific and include macroglossia, dysphagia, abdominal pain, haemorrhage, malabsorption and altered bowel habits (Park, S. W. *et al.*, 2018; Hokama, A. *et al.*, 2011). Gastrointestinal haemorrhage occurs as a presenting complaint in 25-45% of patients. The various causes may be ulceration, infarction, infiltration or generalized oozing without a particular source (Inayat, F., & Hurairah, A. 2016). In the present case, the patient presented with gastrointestinal haemorrhage, microcytic hypochromic anemia, reactive thrombocytosis and features of malabsorption- hypoproteinemia and hypoalbuminemia. Malabsorption is seen as the early sign of disease in about 5% of individuals with primary amyloidosis (Swerdlow, S. H. *et al.*, 2017).

Studies show that varying clinical presentation and endoscopic findings are associated with the patterns of amyloid deposition. Primary systemic amyloidosis is associated with AL type amyloid deposition in muscularis mucosa, submucosa and muscularis propria of bowel wall which may lead to polypoidal protrusions and thickenings so that the patient may present with symptoms of mechanical obstruction and constipation. Secondary amyloidosis is associated with deposition of AA type amyloid protein in the mucosa alone which cause mucosal friability and fine granular appearance in endoscopy and diarrhoea or malabsorption as symptoms (Hokama, A. *et al.*, 2011; Tada, S. *et al.*, 1994). In our case, endoscopy showed telengiectatic raised protrusions into the lumen with surface ulceration in duodenum.

Tissue biopsy is essential for the diagnosis of amyloidosis. Presence of amyloid should be confirmed by polarised light microscopy using Congo red stain. Once amyloid is detected, the protein type should be determined by Immunohistochemistry (Park, S. W. *et al.*, 2018). All subtypes of amyloid shows positive Amyloid P staining, hence it is useful in diagnosis of amyloidosis. Positive kappa or lambda light chain staining indicates primary amyloidosis and positive amyloid A staining indicates secondary amyloidosis. In our case, eosinophilic deposition was seen in the mucosa, which showed Congo red positivity and apple green birefringence under polariser. No deep biopsy was available to assess the deposition in deeper layers of duodenum. As the above patient showed positive results for lambda light chain, he was diagnosed with primary localised AL amyloidosis (Choi, J. H. *et al.*, 2014).

Primary systemic amyloidosis should be determined by testing for the presence of monoclonal immunoglobulin in serum and urine (Park, S. W. *et al.*, 2018). In the present case, this was excluded by Serum protein electrophoresis, renal function tests, and skeletal survey and bone marrow studies.

In Localised amyloidosis, amyloid infiltration is limited to a single organ in the absence of any systemic disease and plasma cell dyscrasia (Cowan, A. J. *et al.*, 2013). Primary localised amyloidosis is due to local population of plasma cells producing amyloidogenic immunoglobulin light chain with absence of monoclonal immunoglobulin in serum or urine (Choi, J. H. *et al.*, 2014; Akamatsu, T. *et al.*, 2007). Thus, extramedullary monoclonal proliferation of plasma cells could be the underlying cause of localised amyloidosis in this patient.

Therapeutic goal of systemic amyloidosis include treatment of underlying condition and control of symptoms. Patients with localised amyloidosis should be monitored and treated symptomatically as the rate of progression to systemic disease is low and they do not warrant systemic therapy (Cowan, A. J. *et al.*, 2013). The above patient had no active oozing on follow up endoscopy and was discharged on haematinics.

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