Gastrointestinal Sarcoidosis with Esophageal Involvement- A Rare Clinical Experience

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Abstract: Sarcoidosis is a multisystemic chronic granulomatous disease of unknown etiology. Primary gastrointestinal disease is uncommon and esophageal involvement is extremely rare. In symptomatic esophageal sarcoidosis only limited case reports have been described in literature with well-documented histological evidence of noncaseating granulomas. We present a case of 20 yrs old Male with dysphagia and swelling over right lower face whose mucosal biopsies revealed noncaseating granulomas consistent with sarcoidosis. Treatment with oral steroids along with azathioprine showed good clinical response in alleviating his symptoms on serial follow up. These rare manifestations reflect the known pathophysiology, clinical course of more common and easily recognizable systemic sarcoidosis. Hence alternative diagnoses must always be excluded when there are deviations from established manifestations for this disease.

Keywords: Sarcoidosis, granulomatous, granulomas, dysphagia.

INTRODUCTION

Sarcoidosis is an inflammatory, granulomatous, multisystem disease with overall incidence and prevalence of 8 and 60 cases per 100,000 respectively [1]. Primary involvement of cardiac, musculoskeletal, lymphatic, neurological and gastrointestinal system is rare.

Old clinicopathological investigations of sarcoid patients did not find any evidence of gastrointestinal involvement [2]. The trend in recent decades has shown presence of subclinical as well as clinically significant and identifiable gastrointestinal disease in 0.1 to 0.9% of patients with the disease [3]. Esophageal involvement is an extremely rare occurrence. We report this case of esophageal involvement in sarcoidosis and the relevant literature for this rare manifestation of disease.

CASE REPORT

20 year old Saudi male university student was referred to gastroenterology clinic with 8 month history of dysphagia and swelling over right lower face. This swelling was painless without any change in size since onset. There was no history of fever or weight loss. He gave past history of contact dermatitis, intermittent asthma and was having thalassemia trait. On examination this swelling was well-defined firm, nontender, measuring 3X4 cms, with normal overlying skin; There were no lymph nodes palpable elsewhere; no skin lesions; no hepatosplenomegaly. His baseline laboratory data showed anemia with hemoglobin 7.8; normal white cell count, Renal, liver, coagulation profile and inflammatory markers. (Table1). Further radiological workup with MRI Face and Neck (Fig 1, Fig 2 and Fig 3) showed abnormal diffuse gingival surface thickening of right maxillary, mandibular alveolar margin, gingival-buccal fold and buccal mucosa infiltrating right buccinator muscle with extension to retro-molar area and prominent right sub-mandibular lymph node. The initial radiological impression was lymphoma for which biopsy of this accessible lesion from right tonsillar area was done which revealed granulomatous morphology with multinucleated giant cells and calcification. It was negative for neoplasia, acid fast bacilli and fungal
infection. In order to complete further workup of other differential diagnosis of this granulomatous lesion upper and lower gastrointestinal endoscopy was done in which a small linear ulcer at lower esophagus was seen along with nonspecific inflammation in terminal ileum suggestive of ileitis. The histopathology of these endoscopic lesions also showed focal granulomatous inflammation. A multidisciplinary consensus was achieved to further work out this as a case of gastrointestinal sarcoidosis. Chest CT was unremarkable (Fig 4). Angiotensin converting enzyme level was normal. Slit lamp examination of eyes did not show any signs of uveitis. Patient was treated with oral steroids followed by introduction of azathioprine. He was closely followed up during which he showed good regression in symptoms along with swelling size. A follow-up MRI showed significant improvement (Fig 5 and Fig 6). In the forthcoming year on follow-up he was symptoms free maintained on Prednisolone 12.5 mg and Azathioprine 150 mg. When steroid was stopped after tapering in 2\textsuperscript{nd} year he relapsed again with reappearance of dysphagia and facial swelling. Prednisolone was reintroduced with azathioprine and repeat upper gastrointestinal endoscopy was done which showed esophageal candidiasis that was treated with fluconazole, but the previously seen lower esophageal ulcer was absent. MRI findings on follow-up remained same as one year ago. Patient was last seen 1 yr ago symptom free on low dose steroid and Azathioprine.
Fig-3: MRI Contrast Axial T1

Fig-4: Chest CT4

Fig-5: Followup Axial T1 Contrast

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DISCUSSION
Sarcoidosis rarely involves luminal gastrointestinal tract and is usually asymptomatic. It is diagnosed on the basis of clinical presentation, diagnostic workup, imaging and histopathology. The gastrointestinal features depend on the affected organ. Esophageal involvement is classified based upon level of involvement and the layer of involvement. It can involve in four distinct patterns like superficial mucosal, myopathic involvement of the esophageal musculature, direct myenteric involvement and lastly extrinsic compression from mediastinal lymphadenopathy [4]. Clinical features in esophageal sarcoidosis are dysphagia from impaired motility, esophagitis, stricture or extrinsic compression from enlarged lymph nodes. The other gastrointestinal features like abdominal discomfort, malabsorption, gastrointestinal bleed; colitis, polyposis or intestinal obstruction depends on the site of involvement [5]. There is no confirmatory test for sarcoidosis. The diagnosis is based on excluding other causes of granulomatosis such as primary biliary cirrhosis, Crohn’s disease, tuberculosis, brucellosis, viral hepatitis, fungal infections, lymphoma and drugs.

A general rule is to start treatment when organ function is threatened. Oral prednisone 20-40 mg daily for 8 weeks with slow taper based on clinical response has been recommended [6]. Due to the rarity of esophageal involvement in sarcoidosis, the role of other immunosuppressive therapy has not been defined. Recent literature supports use of Methotrexate or Azathioprine as second line therapy in those with side effects due to systemic steroid therapy or in those with worsening of symptoms despite corticosteroids. Other cytotoxic drugs infrequently used in the treatment are cyclophosphamide, mycophenolate and leflunomide.

CONCLUSION
Sarcoidosis can have variable presentation. The patients who are highly likely to develop specific organ dysfunction must be kept on regular follow up for timely detection and intervention as required. Esophageal sarcoidosis is under-diagnosed due to the slow progression of the disease and difficulties in diagnosing the same. The use of steroid sparing drugs in sarcoidosis will need more evidence. A treating physician should be aware of this rare but clinically important disease process and an interdisciplinary team approach is essential for a favorable outcome.

REFERENCES