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IgG4-Related Disease Presenting as a Neck Lump: A Rare Differential Diagnosis in Primary Care

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Abstract **Case Report**

This case report presents a 67-year-old male with a neck lump, initially suspected of being a malignancy, later diagnosed with IgG4-related disease (IgG4-RD). The patient presented with a non-mobile, firm mass on the right side of the neck, with no associated symptoms such as dysphagia or pharyngeal discomfort. His diagnostic journey included imaging, fine needle aspiration, and histopathological analysis, ultimately revealing characteristics consistent with IgG4-related disease. This condition, though rare, should be considered in the differential diagnosis of neck lumps, particularly in the primary care setting, where early recognition can significantly influence treatment outcomes. The patient was treated with glucocorticoids, and multidisciplinary management was needed in such cases. The case highlights the importance of distinguishing IgG4-RD from other common causes of neck lumps, such as infections or malignancies, and underscores the role of histology in confirming the diagnosis.

Keywords: Neck Lump, Primary Care, IgG4-Related Disease, Autoimmune Disease, Differential Diagnosis, Fine Needle Aspiration.

INTRODUCTION

Neck lumps are a frequent and often concerning presentation in both primary and secondary care, typically raising fears of malignancy. The differential diagnosis is broad, encompassing infections, benign cysts, and malignancies such as squamous cell carcinoma, lymphoma, and thyroid cancers. However, autoimmune diseases can also cause neck masses, though these conditions are less commonly considered in initial assessments. Among these, IgG4-related disease (IgG4-RD) is a systemic fibroinflammatory disorder that can present with neck masses and affect multiple organ systems.

IgG4-RD is a relatively rare but increasingly recognised condition that poses a significant diagnostic challenge. The global prevalence of IgG4-RD is estimated to range between 0.28 to 1.08 per 100,000 population, with an incidence of 0.66 per 100,000 population in Japan, one of the countries with the highest reported rates (Stone *et al*., 2012; Kamisawa & Nagata, 2015). Data on its prevalence and incidence in the United Kingdom are limited, but the condition is generally considered rare. IgG4-RD is an uncommon cause of neck lumps, representing only a small fraction of cases compared to more common etiologies like malignancies or infectious causes (Stone *et al*., 2012; Kamisawa & Nagata, 2015). This rarity, coupled with its ability to mimic other conditions, can delay diagnosis and treatment, especially in the primary care setting, where awareness of the condition may be low.

This case report emphasises the importance of considering IgG4-RD as part of the differential diagnosis in patients presenting with neck lumps. By exploring the diagnostic process and clinical course of this condition, this report aims to enhance awareness and guide primary care practitioners in recognising IgG4-RD.

CASE REPORT

The patient is a 67-year-old male of Vietnamese origin with a medical history significant for psoriatic arthritis well controlled on non-steroidal anti-inflammatory drugs alone, hyperthyroidism, hypertension, hypercholesterolemia, and bilateral deafness secondary to tympanic membrane perforations. He had no family history of autoimmune or connective tissue diseases, and he had been a non-smoker for over a decade. On presentation, the patient expressed concern

about a lump in his neck, which he had noticed several months earlier.

The lump was described as firm, tender, and non-mobile, located on the right side of the neck, but there were no associated symptoms such as dysphagia, odynophagia, or pharyngeal discomfort. Physical examination revealed a 3 x 2 cm mass at right level 1, likely arising from the right submandibular gland. The mass was palpated bimanually from the floor of the mouth, but there was no associated lymphadenopathy, and the oral cavity and oropharynx appeared normal. No signs of systemic illness such as fever or weight loss were present. Given the clinical suspicion of malignancy, the patient was urgently referred to ENT for further evaluation.

Upon evaluation in the ENT clinic, the submandibular swelling was confirmed. The physician ordered imaging studies, including a CT scan of the neck, thorax, and abdomen, along with an ultrasound-guided fine needle aspiration (FNA) of the neck mass. The CT scan revealed a low-density area within the right submandibular gland, suggestive of a submandibular mass or chronic sialadenitis. Additionally, the scan noted several bilateral lung nodules, raising the possibility of a systemic process. The FNA provided tissue for histological analysis, which indicated a differential diagnosis of either a primary mass lesion or complications of chronic sialadenitis.

The patient underwent a surgical excision of the right submandibular gland. Histopathological examination of the excised tissue revealed significant findings. The IgG4 staining showed a density of approximately 55 IgG4+ cells per high-power field (HPF), with an IgG4+/IgG+ ratio of 45%, consistent with IgG4-related disease. The tissue also displayed storiform fibrosis and dense lymphoplasmacytic infiltrates. Importantly, there was no evidence of dysplasia or malignancy in the tissue. These findings confirmed the diagnosis of IgG4-related disease, and the patient was started on a course of glucocorticoids for treatment.

DISCUSSION

IgG4-related disease (IgG4-RD) is a systemic autoimmune condition that can involve multiple organ systems, often presenting with mass-forming lesions (Stone *et al*., 2012; Kamisawa & Nagata, 2015). This disease was first recognised as a distinct entity in 2003 and has since been increasingly identified as a cause of inflammation and fibrosis in various organs, including the pancreas, kidneys, retroperitoneum, and salivary glands (Stone *et al*., 2012; Khosroshahi & Stone, 2016). The disease is characterised by elevated serum IgG4 levels, tissue infiltration by IgG4-positive plasma cells, and a characteristic histological appearance of storiform fibrosis and lymphoplasmacytic infiltrates (Kamisawa & Nagata, 2015; Carruthers & Khosroshahi, 2015).

IgG4-RD is often referred to as a "great mimicker" because it can closely resemble other conditions, particularly malignancies and infections (Stone *et al*., 2012). The presentation of IgG4-RD with a neck mass, as seen in this patient, is relatively uncommon (Khosroshahi & Stone, 2016; Carruthers & Khosroshahi, 2015). The condition most often involves the parotid and submandibular glands but can also affect other head and neck structures (Stone *et al*., 2012; Kamisawa & Nagata, 2015). Its ability to mimic malignancies such as lymphoma or salivary gland cancers can lead to delays in diagnosis, particularly in primary care, where rarer causes of neck lumps are less frequently considered (Kamisawa & Nagata, 2015; Carruthers & Khosroshahi, 2015).

This rarity, coupled with nonspecific symptoms such as fatigue, mildly elevated inflammatory markers (CRP: 44.4 mg/L, ESR: 77), and elevated IgG4 subclass levels (5.220 g/L), makes early recognition of IgG4-RD challenging (Stone *et al*., 2012; Khosroshahi & Stone, 2016). Histopathology remains the gold standard for diagnosis, as it allows for the identification of the characteristic tissue findings of storiform fibrosis and lymphoplasmacytic infiltrates (Carruthers & Khosroshahi, 2015). In this patient, the surgical excision of the submandibular gland provided a definitive diagnosis, as the earlier FNA failed to yield conclusive results.

Given its potential to mimic malignancy and its rarity, IgG4-RD requires a high index of suspicion among primary care physicians. Prompt referral for imaging and histopathological analysis is essential in patients presenting with unexplained neck masses (Stone *et al*., 2012; Kamisawa & Nagata, 2015; Carruthers & Khosroshahi, 2015). Additionally, collaboration with specialists, including ENT physicians and pathologists, is often critical in confirming the diagnosis and initiating treatment.

CONCLUSION

This case underscores the importance of including IgG4-related disease in the differential diagnosis of neck lumps, particularly in primary care. The disease is rare but increasingly recognised, and its ability to mimic malignancy presents a diagnostic challenge. Early recognition and prompt referral to specialists are crucial to achieving a correct diagnosis and initiating appropriate treatment (Stone *et al*., 2012; Carruthers & Khosroshahi, 2015). In this case, the combination of clinical evaluation, imaging studies, FNA, and histopathological analysis allowed for the timely diagnosis and effective management of IgG4-RD, preventing potential complications. Primary care physicians should be aware of the diverse presentations of IgG4-RD and consider it in their differential

diagnosis, especially in cases of unexplained neck masses (Stone *et al*., 2012; Khosroshahi & Stone, 2016).

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