CASE REPORT

Benign anlage tumour: a very unusual neck mass

Shivani Parihar,¹ Rohit Gohil,¹ Richie Oparka,² Ceilidh Kennedy¹¹Department of Otolaryngology, Ninewells Hospital and Medical School, Dundee, UK²Department of Histopathology, Ninewells Hospital and Medical School, Dundee, UK

Correspondence to

Rohit Gohil,
rohit.gohil@nhs.net

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SUMMARY

A 44-year-old woman presented with a slow-growing asymptomatic neck swelling at the left medial clavicle. Haematological and biochemical work up was normal and an ultrasound confirmed the swelling, but needle aspiration was non-diagnostic. As lymphoma was the main differential diagnosis, the swelling was completely excised. Immunohistochemistry yielded a rare lesion, suspected to represent a myoepithelial/mixed cellularity tumour of soft tissue. The extreme rarity of these tumours required a confirmatory secondary opinion, which ultimately led to it being identified as a benign anlage tumour (previously known as an ectopic hamartomatous thymoma). This case highlights the fact that thorough assessment of patients with neck swellings should be undertaken to rule out sinister causes—keeping in mind more rare differentials—helping to guide final management.

BACKGROUND

Neck lumps are the most common surgical disease of the neck and a frequent presentation to ear, nose and throat (ENT) departments. The underlying diagnosis is often benign in origin and is usually due to persistent reactive lymphadenopathy following infection.¹ Among all other benign causes, lumps can sometimes be congenital in nature. It is critical to consider that a neck lump can be the first sign of neoplasia. Thorough history-taking, including enquiry into 'red flag' symptoms, along with a systematic examination of a neck swelling, is essential in differentiating underlying pathology, and can therefore help guide further level of investigation, multidisciplinary involvement and definitive management. In this report, we present the case of a neck lump presenting in an unusual location and its associated rare identity.

CASE PRESENTATION

A 44-year-old woman, a non-smoker, presented to the ENT department, reporting of an 18-month history of a swelling at the base of her neck, to the left, which she described as a ganglion, and wished to have removed.

There was no history of neck trauma, weight loss, fevers or intercurrent illnesses, nor any changes in appetite, voice or breathing. There were no recognised superficial skin changes. The swelling was, however, slow-growing and was now causing her mild discomfort, but was otherwise asymptomatic.

On examination, the swelling was smooth, rubbery and non-tender; it seemed to be fixed to the medial clavicle/sternocleidomastoid insertion.

There were no other swellings in the neck or elsewhere.

INVESTIGATIONS

Haematological and biochemical work up was normal and an ultrasound confirmed the swelling, but needle aspiration was non-diagnostic (figure 1).

The main differential diagnosis that needed exclusion was of lymphoma; the other differentials included benign neck lumps associated with the skin and subcutaneous tissues.

TREATMENT

An excisional biopsy of the swelling was performed. The swelling was well circumscribed and associated with the clavicular head and left sternocleidomastoid tendinous insertion.

Initial microscopy described a non-encapsulated lesion with a predominant spindle cell pattern in a haphazard arrangement, but with focal 'herringbone'-type structures. There was limited infiltration into surrounding fat and minimal evidence of cellular atypia, and few mitoses (figure 2). The tumour had been completely excised.

Immunohistochemistry yielded positive assays for AE1–3, actin, BCL2, vimentin and p63, and confirmed a myoepithelial nature of the underlying cellular tissue. It was suspected that the lesion might represent a myoepithelial/mixed cellularity tumour of soft tissue. Owing to the extreme rarity of these tumours, a secondary opinion was sought. On review, the swelling was identified as a benign anlage tumour.

OUTCOME AND FOLLOW-UP

Owing to the non-sinister nature of the lesion, no further surgery/management was required. The patient was re-assured to this end and discharged

Figure 1 Ultrasound shows a non-specific well-defined ovoid heterogeneous mass (length (+ to +)=2.12 cm; width (x to x)=1.11 cm).

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Figure 2 H&E stained sections of the lesion. The images highlight the spindle cell morphology of the tumour as well as areas of adipose tissue and epithelial areas. It can be noted that the field is highly cellular, but there is a lack of generalised cellular atypia.

after satisfactory follow-up, revealing a well-healed wound with no clinical evidence of recurrence.

DISCUSSION

A benign anlage tumour, previously known as an ectopic hamartomatous thymoma (EHT), was first described by Smith and McClure in 1982, as an 'unusual subcutaneous mixed tumour exhibiting adipose, fibroblastic, and epithelial components'.² It was then given the name EHT by Rosai *et al*³ 2 years later, due to its histopathological findings, which combine properties of a hamartoma with those of a neoplasm, also thought to be derived from abnormal thymic tissue.

It is now known as a rare benign tumour that presents predominantly in the left lower neck region, located either superficial or deep to the sternocleidomastoid muscle, near the sternoclavicular joint. In the few dozen cases reported over the years, it has shown a marked male predominance with a mean reported age of 47.8 years. Sizes of the tumours have ranged from 0.8 cm to 19 cm. In addition, none have shown any association with the thyroid gland, given their inferior cervical location.⁴

No cases have yet reported any recurrences following complete tumour excision. Two reports of recurrence were noted after incomplete excision. In these cases, definitive re-excision resulted in the patients being disease-free thereafter.⁵ It is therefore important to recognise EHTs and to distinguish them from other malignant differentials, as they follow a benign clinical course and can be optimally treated with limited surgical management.

Pathologically, this tumour comprises a combination of spindle cells, epithelial cells, mature adipose tissue and lymphocytes, in varied proportions. Usually, in these cases, the spindle cell population predominates. Any features of malignancy such as mitotic activity, necrosis or atypia are not generally seen.⁶

Immunohistochemistry shows the spindle cells staining positive for keratin.

As an ectopic hamartomatous tumour most often presents with only a non-tender, slow-growing neck lump, knowledge of this tumour is essential to differentiate it from various differentials such as a lymphoma, mixed tumours of skin adnexal, spindle cell carcinoma, synovial sarcoma, peripheral nerve sheath tumour and teratoma.⁵ The key to this differentiation lies in the specific morphology and immunohistochemistry of EHT and thus can lead its prompt diagnosis.

The histogenesis of EHTs still remains a controversial topic. During initial investigations, it was thought an EHT originated from a developmental anomaly of the thymus that arose from the third branchial pouch.²⁻³ This idea was supported by the fact that normal components of the thymus include adipose cells, lymphocytes and epithelial cells that frequently present in a spindle arrangement. This theory was later challenged by the observation that normal thymic tissue has never been linked with tumours, and lesions similar to it have never been found in the thymus or mediastinum. However, as these tumours are specifically localised to the lower neck region, the general consensus remains that it originates from a branchiogenic origin.

Other models of the histogenesis of this tumour have associated it with a heterotopic salivary gland, based on the fact that they arise in the same anatomical location and also possess a marked male predominance.⁷ Others have suggested a growth in response to androgenic stimuli, due to the observed androgen receptor expression in the spindle cells of the tumour. The latest theory has proposed an origin from the remnant of the cervical sinus of His.⁸

The cervical sinus of His grows during the fifth and sixth week of development when the second branchial arch expands caudally, and then, in most cases, regresses completely by the end of the seventh week. The development of epithelial tissues in the cervical sinus, if persistent, could explain the presence

and growth of any structure potentially derived from the primitive ectoderm.⁹ In order to better explain the origin of this tumour, it is now known as a 'branchial anlage mixed tumour', as suggested by Fetsch *et al.*⁵

Learning points

- ▶ We present a case of a suspicious neck swelling, initially thought to be a lymphoma but after thorough investigation found to be a rare benign anlage tumour.
- ▶ The knowledge of this tumour and its characteristics can be useful when confronted with a lower neck swelling.
- ▶ It is important, however, to always keep in mind what is common and rule out the more severe underlying diagnosis at initial investigation.
- ▶ Ultimately, it is vital to possess understanding of these rare cases while remembering to consider the more common differentials and investigate these expeditiously.

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surgical care and management of this patient. RO provided the histopathological report and further advice on this case.

Competing interests None declared.

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REFERENCES

- 1 British Association of Otorhinolaryngology. *Head and neck cancer: multidisciplinary management guidelines*, 4th edn. London: Royal College of Surgeons of England, 2011.
- 2 Smith PS, McClure J. Unusual subcutaneous mixed tumour exhibiting adipose, fibroblastic, and epithelial components. *J Clin Pathol* 1982;35:1074–7.
- 3 Rosai J, Limas C, Husband EM. Ectopic hamartomatous thymoma. A distinctive benign lesion of lower neck. *Am J Surg Pathol* 1984;8:501–13.
- 4 Sakurai H, Kaji M, Mukai K, *et al.* Ectopic hamartomatous thymoma—a truly rare neoplasm: report of a case. *Surg Today* 2010;40:146–9.
- 5 Fetsch JF, Laskin WB, Michal M, *et al.* Ectopic hamartomatous thymoma. A clinicopathologic and immunohistochemical analysis of cases with data supporting reclassification as a branchial anlage mixed tumor. *Am J Surg Pathol* 2004;28:1360–70.
- 6 Fletcher CDM, Unni KK, Mertens F, eds. *World Health Organization classification of tumours. Pathology and genetics of tumours of soft tissue and bone*. Lyon: International Agency for Research on Cancer (IARC) Press, 2002.
- 7 Michal M, Zámecník M, Gogora M, *et al.* Pitfalls in the diagnosis of ectopic hamartomatous thymoma. *Histopathology* 1996;29:549–55.
- 8 Fetsch JF, Weiss SW. Ectopic hamartomatous thymoma: clinico- pathologic, immunohistochemical, and histogenetic considerations in four new cases. *Hum Pathol* 1990; 21:662–8.
- 9 Weinreb F, O'Malley D. Ghazarian ectopic hamartomatous thymoma: a case demonstrating skin adnexal differentiation with positivity for epithelial membrane antigen, androgen receptors, and BRST-2 by immunohistochemistry. *Hum Pathol* 2007;38:1092–5.

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