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Biphenotypic Branchioma of the Anterior Neck: A Case Report

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Abstract: ***We report a case of biphenotypic branchioma, formerly termed ectopic hamartomatous thymoma (EHT), occurring in the suprasternal region of a 54-year-old Asian man. The patient presented with a three-year history of a slowly enlarging, painless nodule. Microscopically, the tumour comprised spindle cells, epithelial nests, and mature adipose tissue. The spindle cells were arranged in fascicular and storiform patterns, epithelial areas formed squamous islands, and adipocytes were distributed in irregular clusters and sheets. Immunohistochemistry showed AE1/AE3 and CK7 positivity in both spindle and epithelial cells, and the spindle cells also expressed vimentin. All components were negative for CK20 and S100. The Ki-67 proliferation index was less than 10%.***

Keywords: Biphenotypic branchioma, Ectopic hamartomatous thymoma, Branchial apparatus

1.Introduction

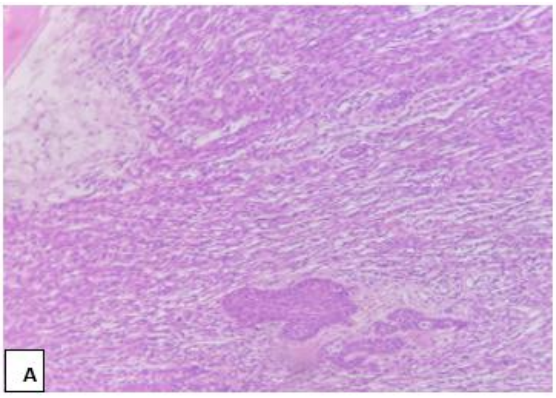
Biphenotypic branchioma is a rare, benign tumour predominantly arising in the deep, lower anterior neck, most frequently the supraclavicular or presternal region, with fewer than 100 cases reported in the literature [5,7]. In 1982, Smith and McClure characterized the lesion as "a mixed tumour exhibiting mesenchymal, lymphangiomatous, and squamous elements" [8]. Rosai et al. reported that it may be a spindle cell thymic anlage tumour and subsequently named it "ectopic hamartomatous thymoma" in 1984 [6]. The 2022 WHO classification introduced the term "biphenotypic branchioma" to reflect a tumour composed of spindle (mesodermal), epithelial (endodermal), and adipose elements, recognising both its dual germ-layer derivation and its genuine neoplastic character [9]. Biphenotypic branchioma most commonly arises in the supraclavicular, suprasternal, and sternoclavicular regions. Less frequently, it has been reported in atypical sites such as the chest wall and posterior axilla [4].

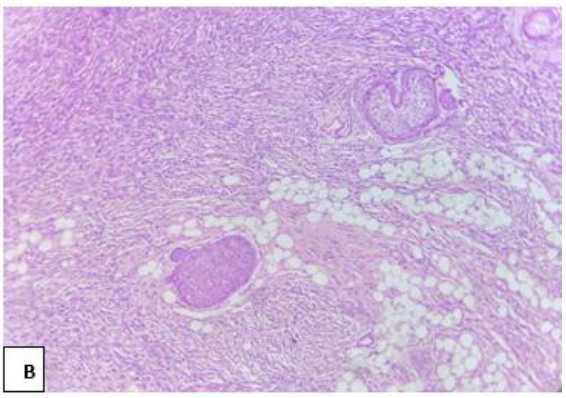
2.Case Presentation

A 54-year-old Asian man was evaluated for a slowly enlarging, painless nodule in the suprasternal region that had been present for approximately three years. He denied any history of trauma, systemic symptoms, or prior similar lesions. On physical examination, the mass was firm, well-circumscribed, and non-tender. No additional abnormalities were detected on further physical examination.

Ultrasonography revealed a well-defined, encapsulated heteroechoic lesion with no evidence of calcification. The patient subsequently underwent wide local excision under general anaesthesia, and the mass was removed in its entirety. At 12 months post-resection, the patient remained free of recurrence or metastasis.

Gross examination of the specimen demonstrated a circumscribed, lobulated tumour measuring 2.0 × 1.5 × 1.0 cm, located 0.3 cm from the closest resection margin. The cut surface appeared grey-white and solid. Microscopically (Figure 1), the lesion comprised three distinct elements: spindle cells, epithelial nests, and mature adipocytes. The spindle cell component displayed fascicular and storiform arrangements, while squamous islands and irregular sheets of mature adipocytes were also observed.





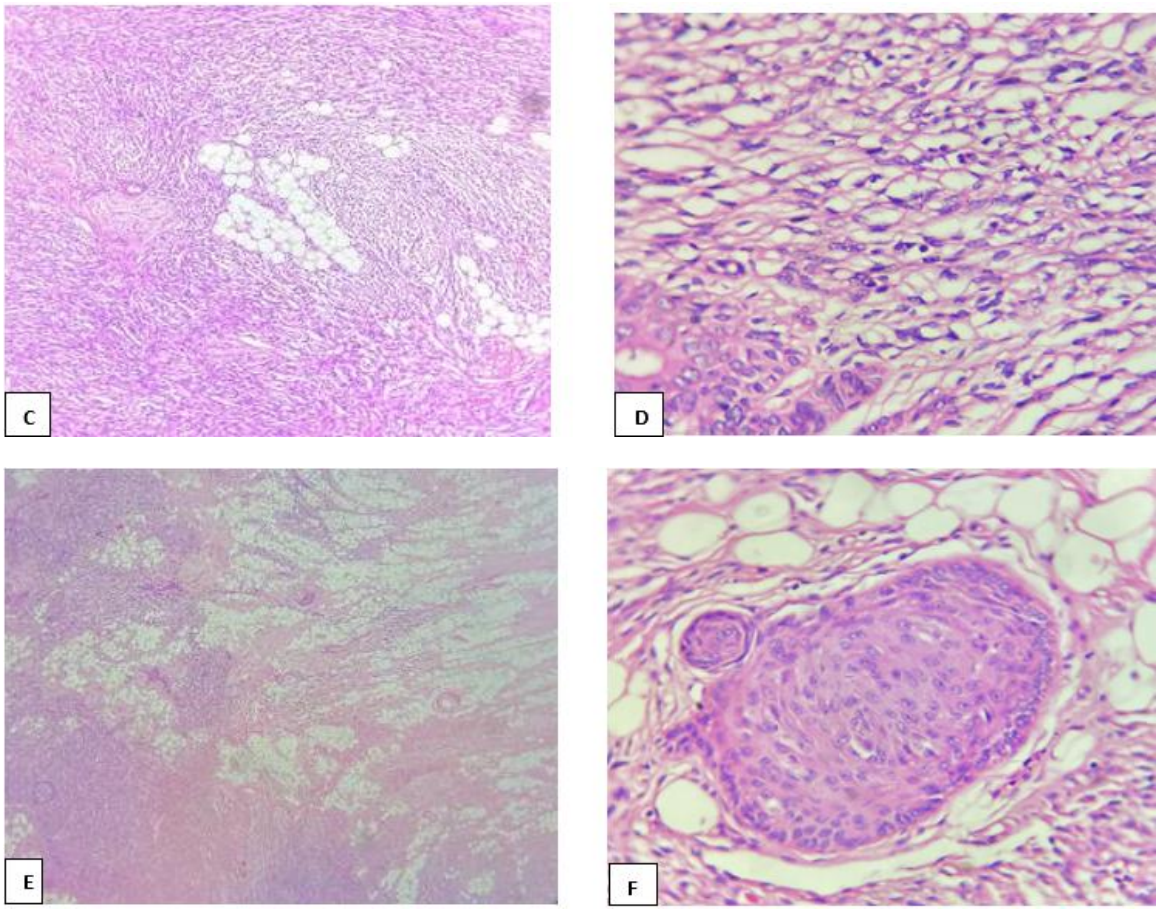
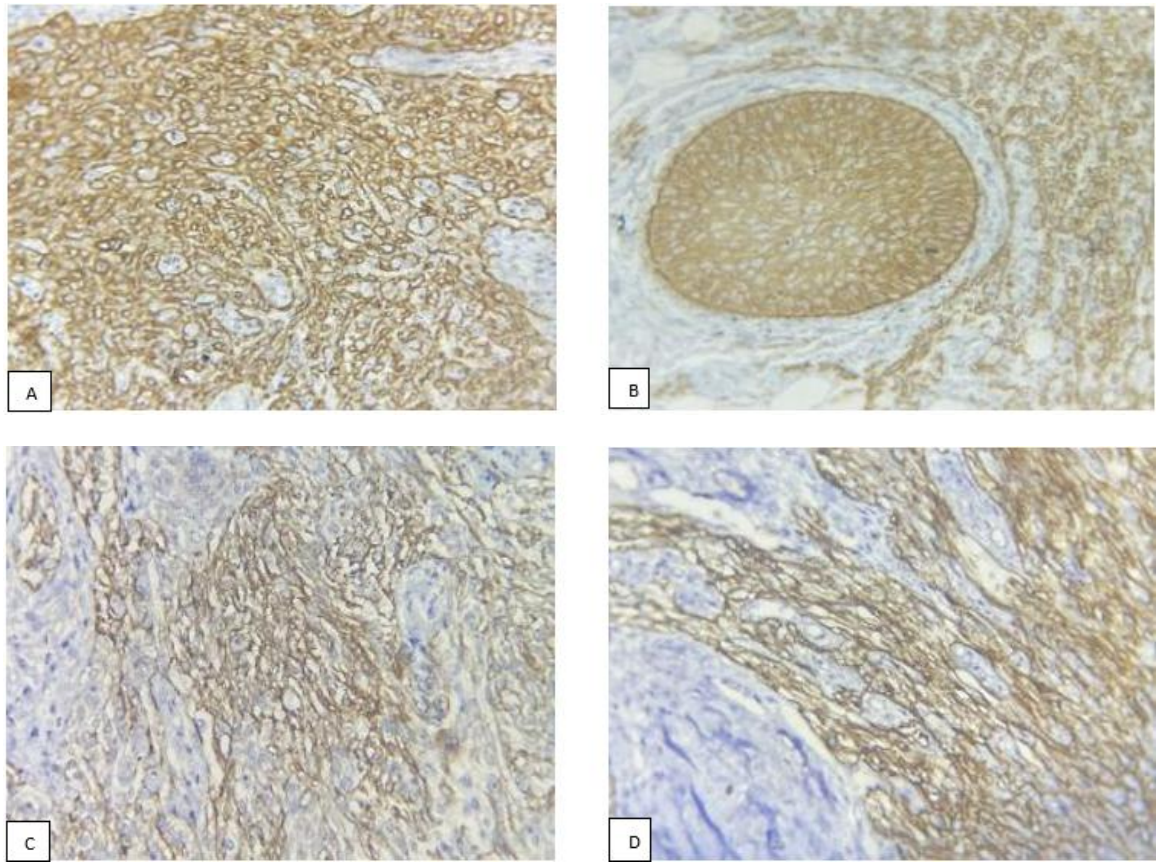


Figure 1: Microscopic features of the tumour (H&E staining). The tumour consists of spindled cells, epithelial islands, and mature adipocytes (A x 100) (B x 100). The spindle cells of the tissue are clearly arranged in a fascicular manner (C x 100) and (D x 400). Mature adipocytes as sheets admixed with spindly cells (E x 100). Keratinizing squamous nests (F x 100).



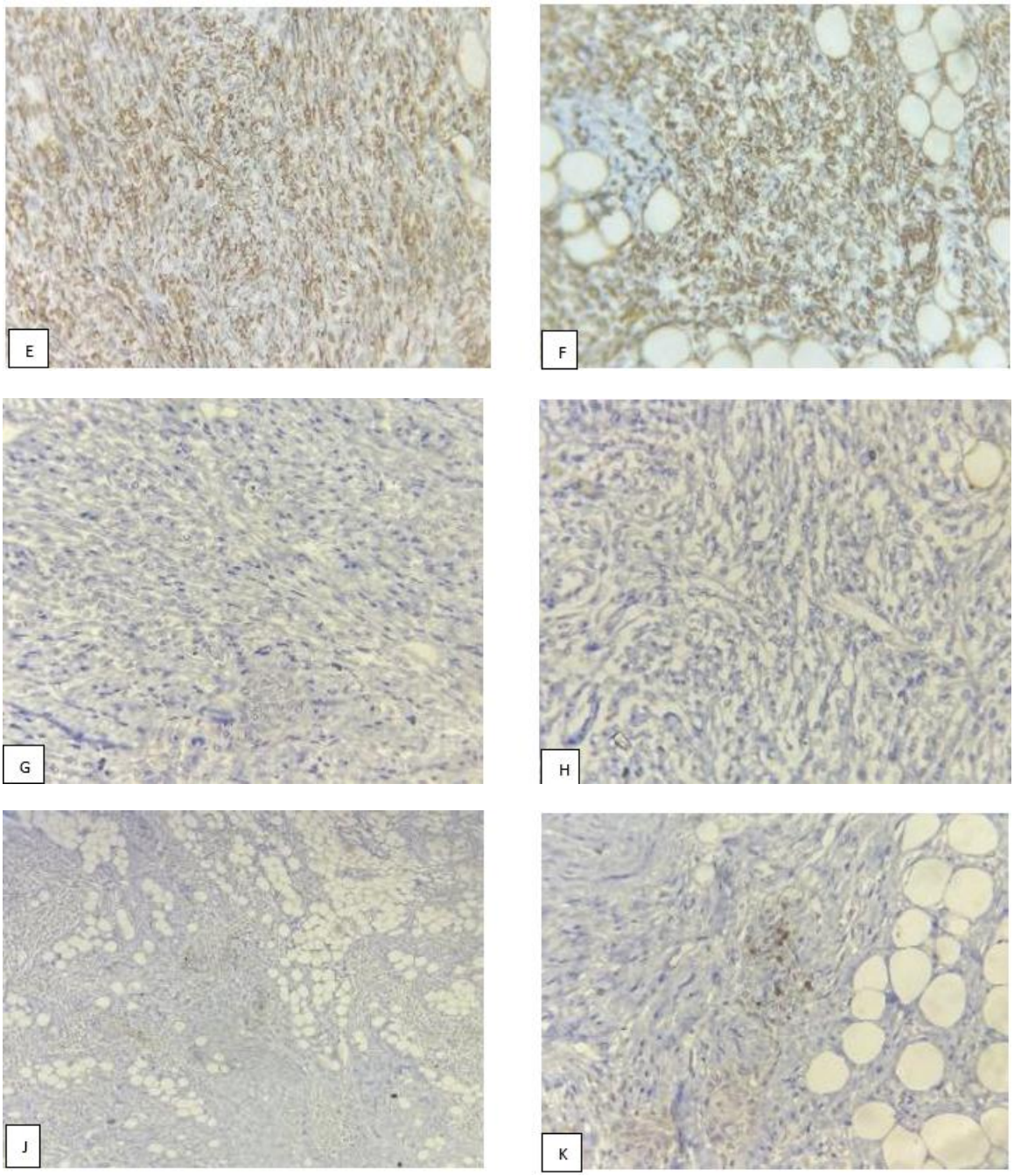


Figure 2: Immunohistochemistry findings of the tumour. Spindle and epithelial islands were strongly positive for AE1/AE3 and CK7 (A, C-100x, B, D-400x). Spindle cell components were positive for Vimentin (E-100x, F-400x). Negative for CK20 (G-100x) and S100 (H-100x). Ki-67 was less than 10% (J-100x, K-400x).

3.Discussion

Rare tumours arising in the soft tissues of the neck and thyroid gland have been described under various terminologies in the literature [9]. They demonstrate partial or complete histological similarity to the fetal, mature, or involuted thymus, as well as to mediastinal thymomas [4,9]. These lesions encompass a broad histopathological and biological spectrum, ranging from entirely benign entities to malignant neoplasms with metastatic potential [9]. At the benign extreme lies biphenotypic branchioma, which characteristically occurs in the soft tissues of the lower neck.

Although biphenotypic branchioma is historically designated as "thymoma," no morphological, immunophenotypic, or molecular evidence substantiates a thymic origin or differentiation [2]. Rather, the embryologic foundation appears to be the branchial apparatus, which contributes to the formation of multiple complex and heterogeneous structures within the neck.

These branchial derivatives comprise spindle cells, epithelial nests, and adipose tissue, reflecting both mesodermal and endodermal differentiation [1]. A review of reported cases indicates that biphenotypic branchioma most often arises in middle-aged men and typically presents as a low anterior neck mass in the supraclavicular or suprasternal region [9], similar to the above case.

Biphenotypic branchioma is frequently misdiagnosed clinically as lipoma, with confirmation relying on histopathological and IHC assessment [2]. The primary challenge is differentiation from other biphasic tumours such as mixed tumour, myoepithelial adenoma, thymic lipoma, spindle epithelial tumour with thymus-like differentiation (SETTLE), malignant peripheral nerve sheath tumour, and biphasic synovial sarcoma [3]. The distinguishing features in biphenotypic branchioma, from mixed tumour are the absence of hyalinized or chondromyxoid stroma, the mediastinal location of thymic lipoma, mucinous epithelium in SETTLE, and marked spindle cell atypia in synovial sarcoma and nerve sheath tumours [2].

In our case, histology showed spindle cells, epithelial nests, and adipose tissue, resembling other biphasic neck tumours, so IHC was essential. The epithelial cells were positive for AE1/AE3 and CK7, and spindle cells for vimentin, indicating biphasic differentiation. Negativity for S100 and CK20 excluded myoepithelial tumours, malignant peripheral nerve sheath tumours, and CK20-positive metastases. This immunoprofile, along with the classic lower neck location, triphasic morphology, and a Ki-67 index below 10%, supports a benign biphenotypic branchioma and distinguishes it from histologic mimics.

4.Conclusion

Biphenotypic branchioma is a rare, benign tumour of the lower anterior neck characterized by dual epithelial and mesenchymal differentiation [6]. Recognition of its distinctive triphasic morphology and immunoprofile is essential to avoid misdiagnosis [3]. Complete surgical excision remains the treatment of choice and is usually curative, with recurrence mainly associated with incomplete removal [2,6]. Awareness of this entity and its preferred nomenclature support accurate diagnosis and optimal management in clinical practice [9].

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