

Case Report

Trichoblastoma arising in nevus sebaceous of Jadassohn: a case report and review of the literature

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Received: 19 September 2026

Revised: 17 October 2026

Accepted: 21 October 2026

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ABSTRACT

Nevus sebaceous of Jadassohn (NSJ) is a congenital hamartoma that may develop secondary neoplasms, most of which are benign. Trichoblastoma (TB) is the most common benign tumor arising in NSJ but is frequently misdiagnosed as basal cell carcinoma (BCC). We report a 34-year-old female with a slow-growing nodule on a pre-existing yellowish, hairless plaque on the right chin, present since childhood. Excision and histopathology showed epidermal hyperkeratosis, acanthosis, and abnormally positioned sebaceous glands opening directly onto the epidermis, consistent with NSJ. Within the dermis, a well-circumscribed tumor composed of basaloid cells with peripheral palisading and papillary mesenchymal bodies was identified, confirming TB. This case highlights the importance of distinguishing TB from BCC to avoid unnecessary aggressive treatment. Pathologists and clinicians should be aware of this entity.

Keywords: Nevus sebaceous of Jadassohn, Trichoblastoma, Basal cell carcinoma, Adnexal tumor, Case report

INTRODUCTION

Nevus sebaceous of Jadassohn (NSJ), first described in 1895, is a congenital, non-hereditary hamartoma composed of malformed epidermal, sebaceous, follicular, and apocrine elements. It typically presents as a well-demarcated, yellowish, hairless plaque on the scalp or face, with an estimated incidence of 0.1 to 0.3 percent in newborns.¹ Historically, basal cell carcinoma (BCC) was thought to be the most common secondary tumor in NSJ, with reported incidences as high as 22 percent.² However, contemporary series with rigorous histopathological re-review have revised this understanding: the majority of secondary neoplasms are benign, with trichoblastoma (TB) emerging as the most frequent.^{3,4} TB is a benign adnexal neoplasm with follicular germinative differentiation that can closely mimic BCC both clinically and microscopically. Misdiagnosis has led to historical overestimation of malignant transformation risk and

unnecessary aggressive surgery. We present a case of TB arising in NSJ on an unusual location (the chin) and discuss distinguishing features.

CASE REPORT

A 34-year-old female presented with a slow-growing mass on the right chin. A hairless, yellowish plaque had been present since childhood. Over the past year, she noticed a nodular growth arising from the plaque, gradually increasing in size. There was no pain, itching, ulceration, or bleeding. The patient sought medical attention due to cosmetic concern. Complete surgical excision with a 3-mm clinical margin was performed under local anesthesia.

Gross pathology: A grey-brown soft tissue piece measuring 1.0 by 0.7 by 0.5 cm. Cut section showed a homogenous white to tan area.

Histopathology: The epidermis showed hyperkeratosis and acanthosis (Figure 1A). The upper dermis demonstrated abnormal localization of pilosebaceous units with some directly opening onto the epidermal surface, a diagnostic hallmark of NSJ (Figure 1B). The dermis also showed a well-circumscribed, non-encapsulated tumor in the mid-to-deep dermis with no connection to the overlying epidermis (Figure 1C). The tumor was arranged in nodules and nests of proliferating basaloid epithelial cells with peripheral palisading (Figure 1D). Individual tumor cells

were round to oval, uniform in size, with open chromatin, small nucleoli, and abundant eosinophilic cytoplasm. Occasional mitosis was seen (fewer than 1 per 10 high-power fields) (Figure 1E). Keratin cyst formation and papillary mesenchymal bodies within the tumor stroma confirmed trichogenic differentiation (Figure 1F). A diagnosis of trichoblastoma arising from NSJ was rendered. At 3 months post-excision, there is no recurrence.

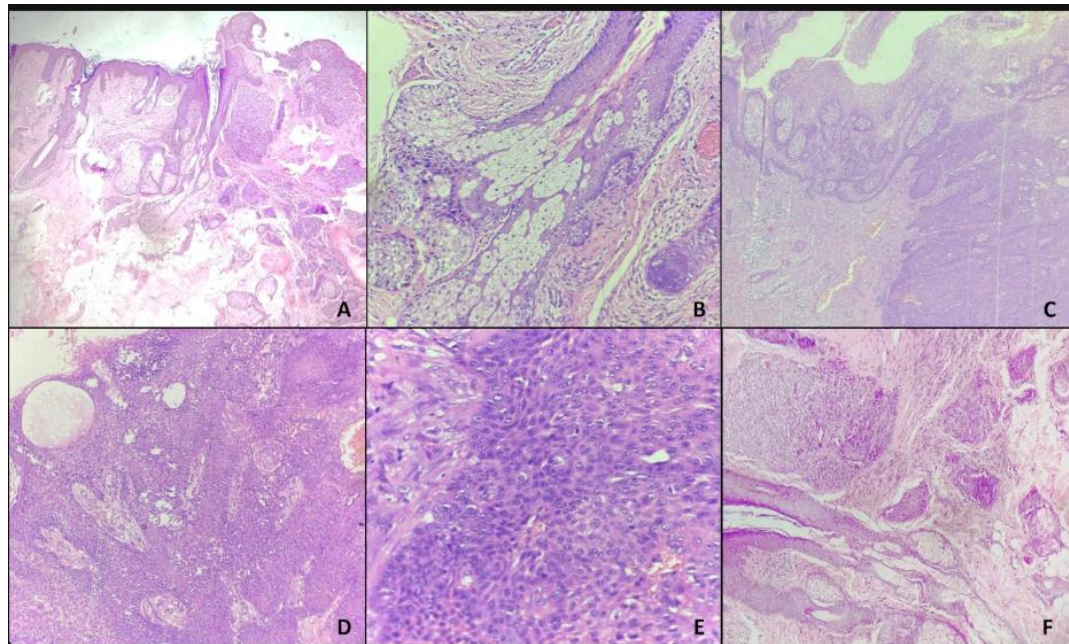


Figure 1: Trichoblastoma arising in nevus sebaceus of Jadassohn. (A) Scanning magnification (4x) shows verrucous epidermal hyperplasia and mature sebaceous glands with an adjacent dermal basaloid tumor. (B) Nevus sebaceus (40x) demonstrates papillomatosis and high dermal sebaceous lobules. (C) The trichoblastoma originates from the lower epidermis (10x). (D) At 20x, the tumor is visible without retraction artifact and cyst. (E) High power (40x) shows peripheral palisading, monomorphic nuclei, and fibrocellular stroma. (F) Papillary mesenchymal bodies (40x) confirm follicular differentiation. (Hematoxylin and eosin; original magnifications as indicated.)

Table 1: Histological and immunohistochemical differences between trichoblastoma and basal cell carcinoma

Features	Trichoblastoma	Basal Cell Carcinoma
Epidermal connection	Usually, absent	Often present
Stroma	Dense, cellular (fibroblastic)	Loose, myxoid
Retraction artifact	Absent	Common (mucin-filled clefts)
Papillary mesenchymal bodies	Present	Absent
Merkel cells (CK20)	Present (scattered)	Absent
Bcl-2	Peripheral staining only	Diffuse, strong
Ber-EP4	Negative or patchy weak	Diffusely positive (over 90 percent)
PHLDA1 (TDAG51)	Positive	Negative
Ki-67 proliferation index	Low (under 5 to 10 percent)	Variable, often higher (10 to 40 percent)

DISCUSSION

Our case illustrates the classic histopathological features of TB arising in NSJ: a well-circumscribed dermal basaloid tumor with peripheral palisading, dense fibrous stroma, papillary mesenchymal bodies, and absence of retraction artifact. The location on the chin is unusual, as

most NSJ occur on the scalp (about 60 percent) or face, but this does not alter the diagnostic approach.^{1,4}

Despite being the most common secondary neoplasm in NSJ, TB remains underrecognized. A large series of 707 NSJ cases found that 18.9 percent developed benign secondary neoplasms, with TB accounting for 7.4 percent,

while malignant transformation to BCC occurred in only 1.1 percent.³ A meta-analysis of 1,892 patients reported a pooled secondary neoplasm rate of 12.8 percent, with benign tumors constituting about 90 percent and BCC in only 1.1 percent.⁵ Papillary mesenchymal bodies, which are stromal papillae projecting into basaloid islands, are considered the single most specific histologic feature of TB and are never seen in BCC.^{4,6} Immunohistochemistry is often essential to distinguish TB from BCC, especially in small biopsies. CK20-positive Merkel cells are present in TB but absent in BCC (rare exceptions). PHLDA1 is positive in TB and negative in BCC.⁷ Bcl-2 shows peripheral staining in TB versus diffuse strong staining in BCC. Ber-EP4 is diffusely positive in more than 90 percent of BCCs but negative or patchy weak in TB.⁸

Genetically, NSJ is caused by post-zygotic activating mutations in HRAS or KRAS. The same mutations are often found in secondary TBs arising within NSJ, indicating common clonal origin. In contrast, BCC is driven by hedgehog pathway mutations (PTCH1, SMO).^{9,10} This genetic divergence further supports the biological distinction between TB and BCC.

Misdiagnosis of TB as BCC leads to overtreatment (wide excisions with 5 to 10 mm margins versus conservative 2 to 3 mm for TB), unnecessary sentinel lymph node biopsy, patient anxiety, and poorer cosmetic outcomes. Conversely, misdiagnosis of BCC as TB could lead to incomplete excision and risk of recurrence. Accurate histopathologic and immunohistochemical diagnosis is therefore critical.

Our case was the conventional nodular type, which is well-circumscribed and easily excised. However, clinicians should be aware that rare plaque variants can be deeply infiltrative and may require Mohs surgery. Complete local excision with conservative margins is curative for typical TB, and recognition of this entity prevents unnecessary aggressive treatment.

CONCLUSION

Trichoblastoma is the most common secondary neoplasm arising in nevus sebaceous of Jadassohn but remains underrecognized and often confused with basal cell carcinoma. Pathologists and dermatologists must be familiar with its characteristic histopathological features particularly papillary mesenchymal bodies, follicular stroma, and CK20+ Merkel cells to avoid misdiagnosis. Complete local excision with conservative margins is curative, and recognition of this entity prevents unnecessary aggressive treatment. Future prospective

studies should focus on documenting the true incidence of TB in NSJ and educating clinicians to reduce the historical overdiagnosis of malignancy.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

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Cite this article as: Ansari SA, Kaur B, Kannaujia S, Swale M. Trichoblastoma arising in nevus sebaceous of Jadassohn: a case report and review of the literature. *Int J Res Med Sci* 2026;14:5448-50.