

CASE REPORT

Is it just a scar? A case report of a diagnostic and management challenge of desmoplastic melanoma

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Abstract

A 73-year-old female presented with a progressively enlarging pink nodule on the left anterior upper arm resembling a keloidal scar with accelerated growth and pain in recent months. Clinically, the lesion resembled a keloid scar. Ultrasound imaging revealed a well-demarcated, hypoechoic lesion located within the dermis and extending into the subcutaneous tissue. An excision biopsy confirmed the diagnosis of mixed desmoplastic melanoma, characterised by a spindle cell morphology and significant stromal desmoplasia, with a Breslow thickness of 8.1 mm. Notably, perineural invasion was identified, a recognised adverse prognostic feature associated with this subtype. Immunohistochemical analysis showed strong positivity for SOX-10 (SRY-box transcription factor 10) and minimal staining for HMB-45 (Human Melanoma Black), thereby supporting the diagnosis. Molecular genetic analysis did not reveal pathogenic variants in BRAF, NRAS, or KIT, thereby limiting options for targeted therapy. Staging imaging studies, including positron emission tomography-computed tomography (PET-CT) and Magnetic resonance imaging (MRI) scan of the brain, did not detect any metastatic disease. The wide local excision and sentinel lymph node biopsy were negative for residual disease, local spread, and regional nodal involvement.

This case exemplifies the diagnostic challenges posed by desmoplastic melanoma, an uncommon and often amelanotic subtype that can clinically mimic benign scar-like lesions. Such similarities may contribute to delayed diagnosis and result in an advanced Breslow thickness upon presentation. Histopathological evaluation remains paramount in establishing an accurate diagnosis, particularly given the overlap in clinical features with benign conditions. The absence of actionable molecular mutations underscores the importance of surgical intervention and the necessity of a multidisciplinary clinical approach.

Keywords: desmoplastic melanoma; melanoma; skin cancer; dermatoscopy; dermatopathology

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Introduction

Desmoplastic melanoma is a rare subtype of cutaneous melanoma, often presenting as an amelanotic spindle cell lesion that mimics benign or malignant soft tissue tumours.^{1,2,3} Its subtle dermoscopic features and predilection for sun-exposed skin contribute to diagnostic delay.⁴ Prognosis is strongly influenced by tumour thickness, perineural invasion, and margin status.³ Unlike other melanoma subtypes, desmoplastic melanoma is less likely to harbour actionable mutations such as BRAF V600, limiting targeted therapeutic options.⁵ This case demonstrates the diagnostic challenges and the importance of a multidisciplinary approach in management, particularly when faced with high Breslow thickness and co-existing cutaneous malignancies.

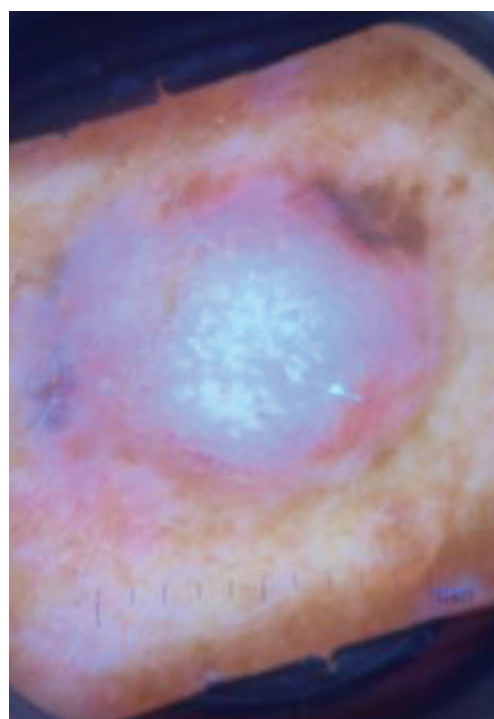
Case Presentation

We report a case of a 73-year-old woman of Fitzpatrick skin type II, with a medical history of peripheral neuropathy, non-diabetic hypoglycaemia,

and hypercholesterolaemia, who was referred to the emergency skin cancer pathway, known as the Two-Week Rule (TWR) clinic, due to a pink raised lesion on her left anterior upper arm (Figure 1A). This lesion had been present for approximately 1 year but had increased in size and become significantly sore over the past 4 months. She described a dragging pain and some degree of soreness that extends deep into her arm. She had been a regular sunbather with significant sun exposure but had no personal or family history of melanoma or any other skin cancers. Upon clinical examination with a dermatoscope, a 15 × 10 mm pink nodule with a focal pigmentation at the base and polymorphic blood vessels was observed (Figure 1B). The lesion had characteristics resembling a keloid scar in both appearance and symptoms, such as pain, although its morphology was unusual. There was no palpable lymphadenopathy detectable in the ipsilateral antecubital fossa, axilla, or cervical regions. The full skin examination revealed an 8mm pink-coloured patch on her right lateral thigh, clinically suspicious for basal cell carcinoma.



A



B

Figure 1. (A) Initial presentation of the desmoplastic melanoma. (B) Dermoscopic appearance of the nodule showing focal pigmentation at the base and polymorphic blood vessels.

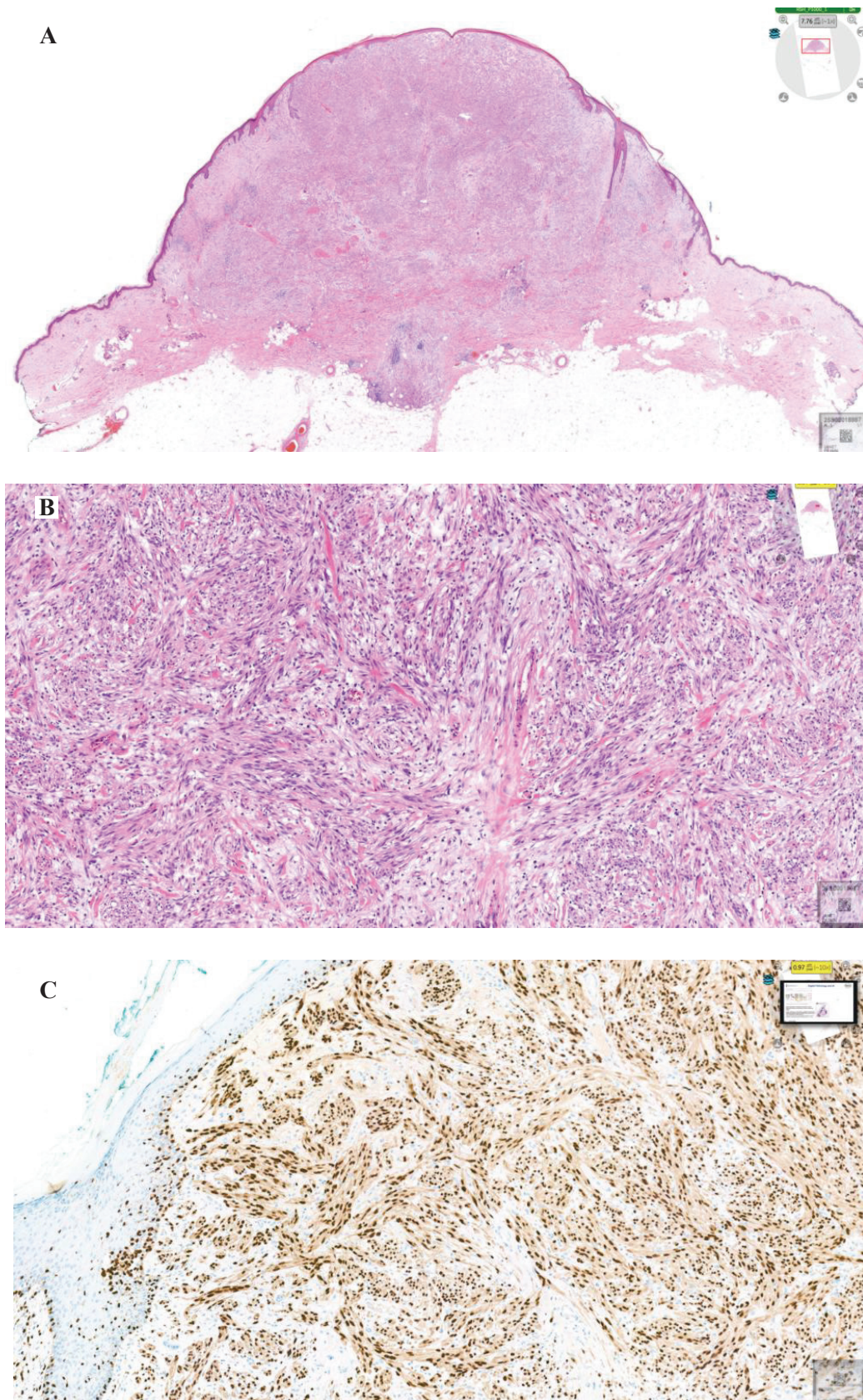


Figure 2. Histopathology of the lesion. (A) Dermal tumour (H&E $\times 20$). (B) Evidence of spindle cell proliferation within the dermis (H&E $\times 40$). (C) SOX-10 positive immunohistochemistry staining ($\times 40$).

Differential Diagnosis: The initial clinical presentation of the amelanotic nodule necessitated a careful consideration of differential diagnoses. A keloid scar was considered based on its characteristic raised, smooth surface and pink colour, along with the patient's report of pain and a dragging sensation. A dermatofibrosarcoma protuberans was considered in the differential diagnosis, particularly in light of recent changes in its appearance and the accompanying deep-seated neurological symptoms. The possibility of amelanotic melanoma was also considered, especially given the recent growth observed and the presence of polymorphic blood vessels identified during dermatoscopic examination. Other spindle cell neoplasms were ultimately less likely. The definitive diagnosis relied heavily on histopathological analysis and immunohistochemical studies, which were instrumental in confirming it.

Investigations: A comprehensive series of investigations was conducted throughout the management of the case, leading to the definitive diagnosis of an atypical nodule and subsequent treatment protocols. Initial imaging with ultrasound revealed a well-demarcated, heterogeneous, hypoechoic lesion located within the dermis, extending into the subcutaneous tissue, with no evidence of intramuscular invasion. An urgent excision biopsy was carried out with standard margins for suspected melanoma for the diagnosis via histology. The histopathology was reported as a lentiginous proliferation of atypical melanocytes in the epidermis, and the dermis was occupied by a poorly circumscribed tumour consisting of elongated spindle cells, requiring further evaluation with immunohistochemical markers. Histopathology also showed an occasional small lymphoid aggregate in the periphery of the dermal tumour (Figure 2A-B). The cells were strongly positive for SOX10 and negative for HMB45 (Figure 2C). Although S100 is frequently used in melanoma immunohistochemistry, strong SOX10 positivity was considered sufficient in this case.

The Breslow thickness was 8.1 mm, the mitotic count was 4 mitoses/mm², and there was no ulceration. One important finding was that perineural invasion was noted, explaining the patient's deep-seated neurological symptoms. The lesion was completely excised with a peripheral margin of 4.7 mm, and a deep margin of 12 mm. Molecular testing yielded no pathogenic variants in the BRAF, NRAS, or KIT genes. This finding signifies a limitation in

the application of currently available targeted therapies, such as BRAF and MEK inhibitors, which are generally employed in mutation-positive melanoma cases. Following the findings, a diagnosis of desmoplastic melanoma, classified as pT4a, was established. Staging imaging, which included PET-CT and MRI of the brain, indicated no evidence of metastatic disease.

Management: The management plan was discussed at a regional specialist skin cancer multidisciplinary meeting, where a wide local excision and sentinel lymph node biopsy were recommended. The results from the sentinel lymph node biopsy showed no signs of metastatic melanoma. Clinical evaluation and cross-sectional imaging staged the condition as Stage IIB. After a thorough discussion with the patient, it was decided that surveillance would be prioritised over adjuvant immunotherapy, given the potential risk of exacerbating peripheral neuropathy with immunotherapy. The follow-up plans entail a combination of clinical and radiological assessments. A comprehensive skin examination is scheduled every three months for the first three years, followed by six-month intervals in the fourth and fifth years. Additionally, PET-CT scans and brain MRIs will be performed every 6 months for the first 3 years to monitor the patient's status. The lesion on her right lateral thigh was confirmed to be basal cell carcinoma and has been surgically excised.

Discussion

Desmoplastic melanoma (DM) accounts for fewer than 4% of melanomas but carries distinct prognostic and therapeutic implications that extend beyond its diagnostic difficulty.³ In this case, the lesion's amelanotic, scar-like appearance contributed to delayed recognition, reflected in its substantial Breslow thickness (>8 mm) at diagnosis. While ultrasound demonstrated dermal and subcutaneous involvement without deeper structural extension, definitive risk stratification relied on histopathological assessment. The presence of spindle cell morphology with marked desmoplasia, combined with perineural invasion, identifies a phenotype associated with increased local recurrence risk due to neurotropic spread.³

Importantly, this tumour was classified as mixed desmoplastic melanoma. Unlike pure DM, which is characterised by low sentinel lymph node positivity rates (approximately 5–10%) but higher rates of local recurrence, mixed DM demonstrates

nodal metastatic behaviour more comparable to conventional melanoma. This distinction is clinically significant and supports consideration of sentinel lymph node biopsy in surgical planning. The combination of high Breslow thickness and perineural invasion further heightens recurrence risk and justifies multidisciplinary discussion regarding adjuvant strategies.⁶

Although molecular testing was negative for BRAF, NRAS, and KIT mutations, limiting the role of targeted therapies, DM is recognised to have a high tumour mutational burden and has shown favourable responses to immune checkpoint inhibitors in advanced disease. Additionally, adjuvant radiotherapy may reduce local recurrence in high-risk cases, particularly where perineural invasion is present, and should be considered in selected patients.⁷

Early multidisciplinary team involvement was therefore critical in balancing decisions regarding

wide local excision margins, sentinel lymph node biopsy, potential adjuvant radiotherapy, and systemic therapy pathways. The co-existing basal cell carcinoma was incidental and most likely reflects cumulative ultraviolet damage rather than biological association, but underscores the importance of comprehensive full-skin examination in patients presenting with suspicious lesions.⁸

Ethics statement

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Author contributions

PV was responsible for the writing and submission of the manuscript, while SK contributed to both the writing process and the clinical care of the patient. JA also assisted with the care of the patient, and BL provided the histopathological case slides and essential diagnostic support.

Learning points

Key learning points can be derived from this case. Hypertrophic scars on the body should be closely examined, particularly if they exhibit unusual characteristics, as they are likely to be atypical. It is important to recognise that desmoplastic melanoma often presents as an amelanotic spindle cell lesion, which can create diagnostic ambiguity with other soft tissue tumours.

The prognosis for this type of melanoma is significantly influenced by factors such as Breslow thickness, perineural invasion, and margin status. Although molecular profiling is valuable, actionable mutations in desmoplastic melanoma are relatively rare. A multidisciplinary team discussion is crucial for guiding both surgical and oncological management. Consequently, a thorough full skin examination is essential to exclude the possibility of concurrent skin cancers and identification of recurrences during the longitudinal monitoring.

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