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A Rare Case of Primary Vulval Amelanotic Melanoma Involving the Urethra

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Key words: Amelanotic melanoma – AM – PVAM – Amelanotic melanoma vulva – Vulval melanoma – Gynecologic tumor – Vulvar cancer – Diagnostic procedure – CT – MRI

Abstract: A 60-year-old woman came to the Emergency Department complaining of a vaginal formation. The urologist suspected a urethral caruncle: the patient was discharged with vaginal oestrogen cream to relieve symptoms and a follow-up was suggested. After two months the patient returned to the Emergency Department since the mass was increasing in volume and complaining of dysuria and haematuria. Ultrasound, contrast-enhanced computed tomography, and contrast-enhanced magnetic resonance revealed a mass arising from the mucosa and involving the vulva and the urethra, suspicious of malignancy. We present a challenging diagnosis of an infiltrative and rapidly progressive primary vulval amelanotic melanoma with a complete imaging evaluation and a confirmed histological diagnosis.

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Introduction

Cancers of the vulva and vagina are uncommon neoplasms, with vulval cancers being more common than vaginal cancers; the most common subtype being squamous cell carcinoma (Gong et al., 2019).

Melanomas are even rarer tumours responsible for a diagnostic challenge. Primary vaginal amelanotic melanoma (PVAM) is a rare not pigmented subtype of melanoma that accounts for 0.4–27% of all cases. Patients with red hair, type I skin, freckles, a lack of nevi on the back, a sun-sensitive phenotype, or a history of AMs (amelanotic melanomas) are more prone to acquire them: the male/female ratio ranges between 0.5 and 4. Since tumour cells lack melanin colour, PVAM may resemble other frequent vaginal cancers with a better prognosis, creating a diagnostic challenge (Weinberg and Gomez-Martinez, 2019).

The most important prognostic factors include tumour size, depth of invasion, metastasis status of lymph nodes, and distal metastasis status (Muinonen-Martin et al., 2018).

Surgery is the mainstay of melanoma treatment: the resection might be extensive, but the recurrence is possible and frequent. Radiotherapy can be used as a neoadjuvant or adjuvant treatment to increase local control rates, while chemotherapy can shrink large masses before surgery (Pye et al., 2023).

This case report describes the imaging examination, especially CT (computed tomography) scans, and MRI (magnetic resonance imaging) in the diagnostic process of an unaware patient who experienced pain and vaginal discomfort.

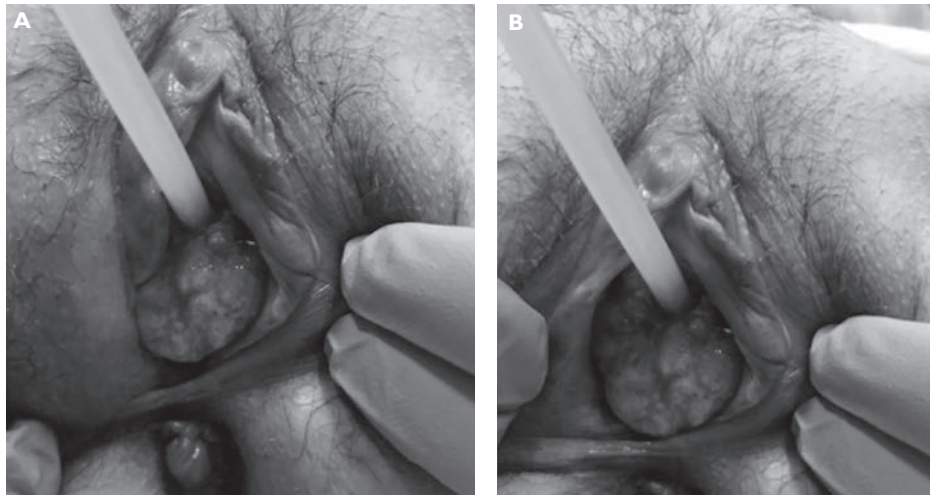

Figure 1 – Macroscopic image showing the vulvar formation with a round morphology, hard consistency, non-homogeneous colour, and appearance, not mobile, without superficial ulcerations.

Case report

A 60-year-old woman attended the Emergency Department with a spherical, growing formation between her vulva and urethra. The urologist suspected a urethral caruncle, so she returned home with a vaginal oestrogen lotion to relieve the discomfort; a follow-up was scheduled. Two months later, the mass had suddenly enlarged, causing her pain, embarrassment, dysuria, and haematuria, motivating her to return to the Emergency Department for a clinical examination (Figure 1).

The physician requested further imaging investigations (CT and MRI) to suspect a malignancy motivated by the high growth rate and the little mobility to the fascial plane.

An abdomen CT with contrast medium administration was performed to better localize the lesion and for staging (Figure 2).

A pelvic MRI with contrast medium administration was scheduled for the next day to characterize the lesion better and evaluate the local infiltration (Figure 3).

A few days following the MRI, the patient underwent a histological sample, which confirmed the suspicion: vulval amelanotic melanoma, a quickly progressing malignancy (Figure 4).

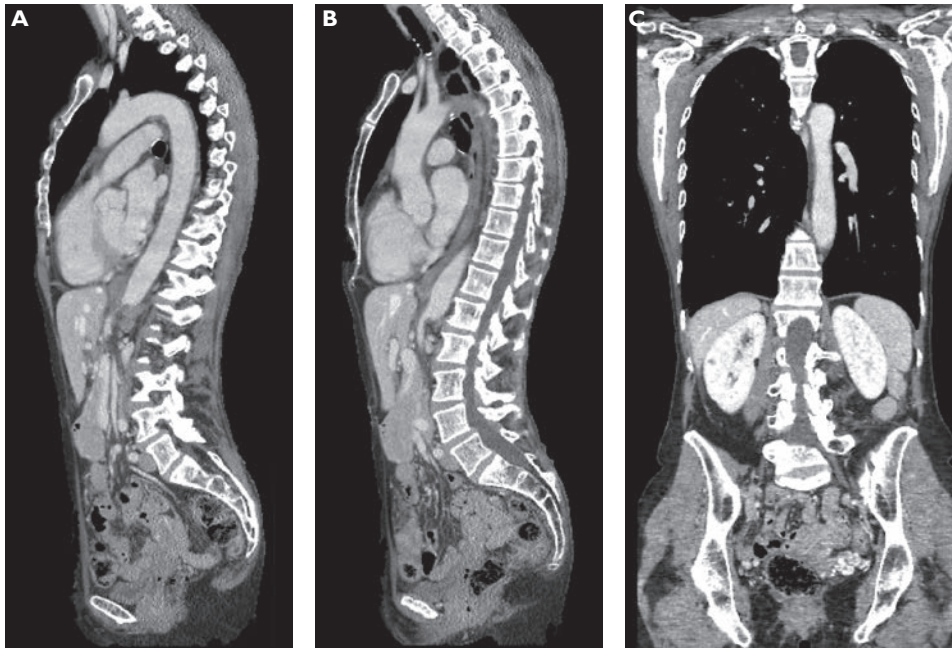

Figure 2 – Subsequent computed tomography scans in the sagittal (A and B) and coronal (C) planes in the venous phase showed a lesion of the vaginal pertaining with ill-defined margins and inhomogeneous contrast-enhancement without clear planes of cleavage with the surrounding perineal muscles; no distant secondary lesions were evident.

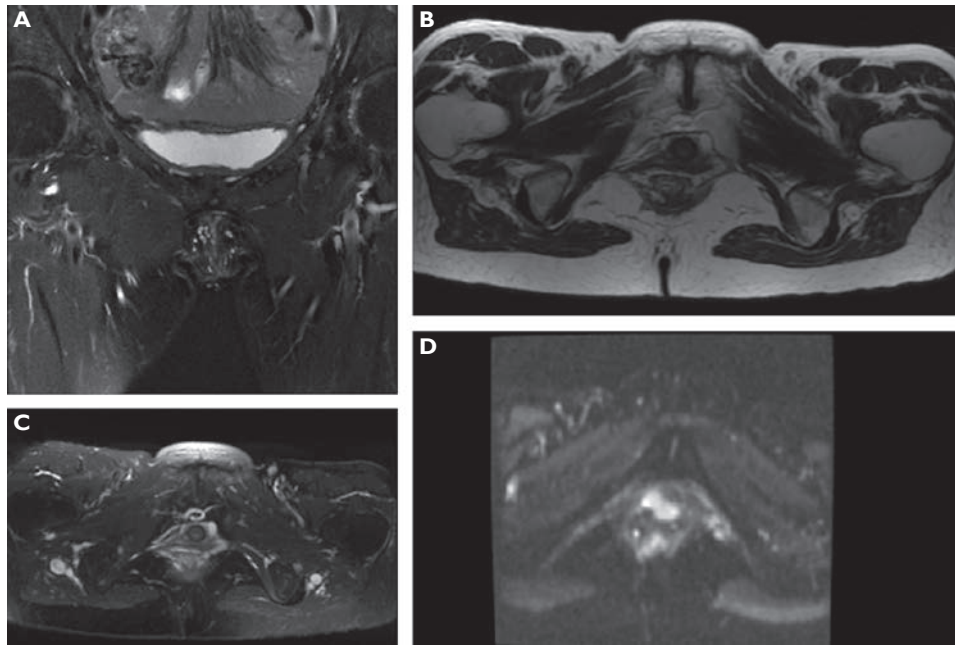

Figure 3 – Magnetic resonance imaging sequences: A) coronal T2 PROP fat-suppressed; B) axial T2; C) axial T1 fat-suppressed after contrasting medium administration; D) axial DWI (diffusion-weighted imaging) at high b-values with a small FOV (field of view). Examination revealed a lobulated, homogeneous lesion with intermediate SI (signal intensity) and maximum diameter of about 3.2 cm originating from the anterior wall of the vagina and extending and infiltrating the urethral meatus posteriorly and the levator muscles of the anus laterally.

Tumour cells were immunohistochemically categorized as positive for HMB-45, S-100, Melan-A, SOX-10, and p16. They also tested negative for cytokeratin's AE1-AE3, CD45 (LCA), and CD79a. Fontana-Masson staining did not detect melanin pigments (Wechter et al., 2004).

Immediately afterward the patient was started on radiotherapy and chemotherapy, to reduce the size of the tumour and promote survival. The patient underwent extensive surgery and had an MRI to evaluate the effects (Figure 5).

The patient is currently on immunotherapy and close follow-up. The early treatment of urethral melanoma is essential due to the tendency to early metastasis.

Discussion

Malignant melanoma is an aggressive, infamous malignancy with a wide variety of morphological and immune-histochemical expressions, frequently leading to an

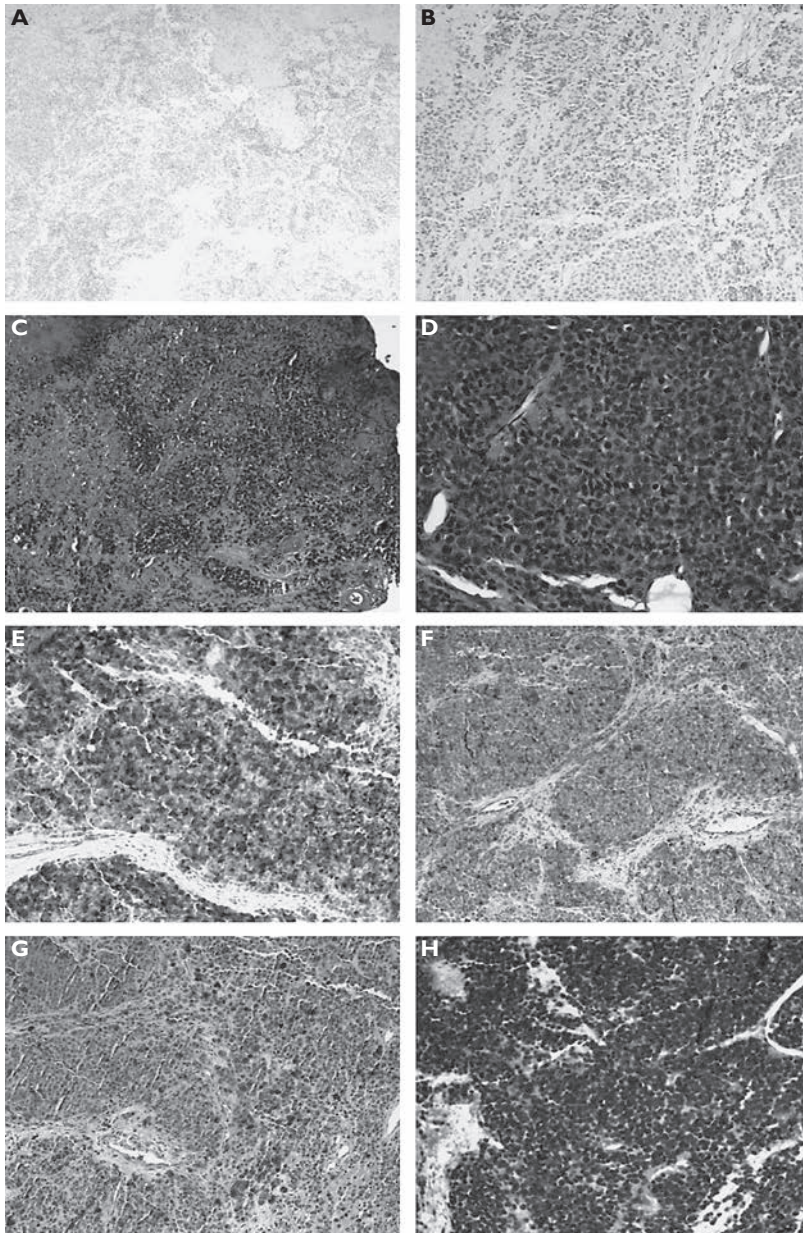

Figure 4 – Histological examination: A) cytokeratin 20 (CK 20) (4×); B) cytokeratin AE1/AE3 (CK) (10×); C) stained by haematoxylin-eosin (HE) (10×); D) stained by HE (20×); E) the antibody HMB45 (10×); F) the protein Melan-A (10×); G) proteins S100 (10×); H) gene SOX10 (10×). Sheets and nests of large pleomorphic epithelioid malignant melanocytic cells with prominent nucleoli and several atypical mitotic figures from the tumour. Melanin production can be focally present. Immunoreactivity for Melan-A, HMB45, S100, and SOX10 and negativity for CKAE1/AE3, CK8/18, CK20, p63, and GATA3 allows the differential diagnosis with high-grade urothelial carcinoma and confirms the diagnosis of melanoma.

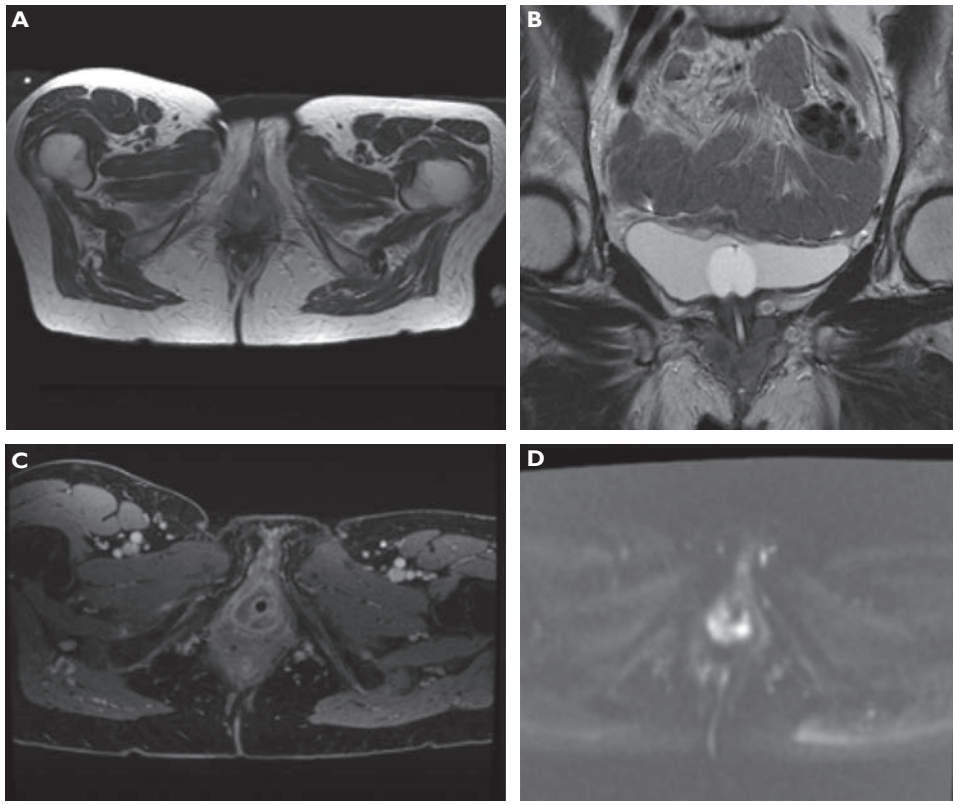

Figure 5 – Magnetic resonance imaging sequences: A) axial T2; B) coronal T2; C) axial T1 fat-suppressed performed after contrasting medium administration; D) axial DWI (diffusion-weighted imaging) at high b-values. Examination revealed an increase in the size of the known vaginal lesion; there were no substantial variations of the intensity-contrastographic characteristics and the infiltrative appearance on the urethral meatus and the levator ani muscles.

incorrect diagnosis. Amelanotic melanoma, with its diverse clinical presentations, lack of pigmentation, and various histological findings, has earned a reputation as a master masquerade within the melanoma group.

Amelanocytic melanoma of the vulva is a rare form characterized by a lack of pigmentation (amelanotic precisely). Typically, melanoma is associated with skin pigmentation due to melanin, which gives the lesion a dark or black colour. However, amelanocytic melanoma can present as a whitish, reddish, or the same shade as the surrounding skin, making its diagnosis more complex (Osama et al., 2022).

Amelanocytic melanoma in the vulva with infiltration of the urethra represents an even rarer condition that is difficult to diagnose, as in this case. It is paramount to emphasize that diagnosing amelanotic melanoma requires careful evaluation through

clinical examination, imaging, and histological and immunochemical examinations. It is crucial to include amelanotic melanoma in the differential diagnosis of cutaneous lesions of the vulva, as it can often be confused with other nonmelanocytic skin neoplasms or urological conditions such as an of a urethral caruncle (Maetzold and Takacs, 2022).

Suspicious symptoms of gynecological oncology include frequent bleeding after sexual intercourse, atypical vaginal discharge, dyspareunia, constipation, and persistent pelvic pain. Rapid and comprehensive diagnostics are necessary for vulvar melanoma, which spreads locally and mostly through the lymphatic system. Prognostic factors for carcinoma vulvar include tumour size, depth of invasion, lymph node status, and distal metastases (Filippetti and Pitocco, 2015).

A skin biopsy and immunochemical examination of tumour cells can help confirm the diagnosis of amelanotic melanoma. Melanoma staging, which includes the amount of infiltration into the urethra, is critical for treatment planning and determining the risk of metastatic metastasis. The disease's stage determines the treatment of amelanotic vulval melanoma and may include surgery, targeted medicines, immunotherapy, and chemotherapy. The patient must be followed by a multidisciplinary team consisting of a urologist, oncologist, and radiologist who can assure regular disease monitoring and targeted customized treatment (Oiso et al., 2010).

The diagnosis involves a thorough clinical and laboratory evaluation and CT and MRI scans with contrast medium. CT scans are effective for characterization and local staging. In contrast, MRI scanning is appropriate for identifying local infiltration given the rapid progression of these neoplasms necessitates surgical therapy, affecting prognosis and follow-up (Patil et al., 2021).

Amelanocytic melanoma of the vulva, particularly when associated with external urethral invasion, is an uncommon but hazardous condition that necessitates early detection and a comprehensive treatment strategy. Educating healthcare providers and patients on the importance of preventing, diagnosing, and treating cutaneous melanoma, especially amelanotic melanoma is critical.

Conclusion

This case emphasizes the necessity of detecting non-pigmented nodules on the vulva of older ladies as possibly malignant melanoma, and that a combination of diagnostic imaging and immunohistochemistry stains may be effective in determining the stage of the melanosomes in melanoma cells. Vulvar melanoma is a sporadic and aggressive tumour, even more so in its amelanotic variant. First- and second-level imaging examinations are useful in the characterization and oncologic staging, treatment, and follow-up.

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