Rare disease

Nevoid malignant melanoma in an albino woman

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Summary

Albinism is a disorder of hypopigmentation affecting the skin, hair and eyes. Ultraviolet light induced cutaneous tumours are common in patients with albinism due to reduced or absent protection from melanin, with squamous cell carcinoma being the most common. Although non-melanomatous skin cancers are more frequent in patients with albinism, dysplastic nevi and melanoma present a greater diagnostic challenge in this group because of their hypopigmented appearance. Here the authors report a case of nevoid malignant melanoma in a patient who had type 1A oculocutaneous albinism.

BACKGROUND

Oculocutaneous albinism is a genetically heterogeneous group of disorders in which there is generalised decreased or absent melanin pigment in the eyes, hair and skin.¹ At least 10 forms of this condition have been identified, each presumably resulting from a different biochemical mechanism blocking the synthesis of melanin.² In type 1A, the classic type, the defect is a complete absence of tyrosinase activity in melanocytes.³ The tyrosinase gene is present on chromosome 11q14–q21.^{4 5 6} Inheritance is autosomal recessive in type 1A. Skin disorders include accelerated photo aging and an increased incidence of keratosis,

squamous cell carcinoma and basal cell carcinoma.^{7 8} Malignant melanoma develops occasionally.⁹ Nevoid melanoma is a rare variant of melanoma characterised by deceptive morphological features reminiscent of a benign melanocytic nevus. Here we report the case of an albino woman with nevoid malignant melanoma.

CASE PRESENTATION

Our patient was a 32-year-old Iranian albino woman who had a pink nevus on her back since her girlhood. Recently she noticed some enlargement, elevation and trauma-induced ulceration. She was admitted to the dermatology

Figure 1 Nevoid melanoma: the tumoural cells show minimal pleomorphism (H&E ×40).

department. On physical examination albinism, nystagmus and a 7×5 mm crusted lesion on her back were observed. The lesion was a firm, pink, dome-shaped tumour nearly 7 mm at its greatest diameter with an ulcerated centre. Routine laboratory studies and biochemical tests were normal. The lesion was resected and the specimen was sent to the surgical pathology department for histological examination. Sections were stained with H&E and Fontana Mason. Histologically, the sections showed a tumour composed of slightly pleomorphic nevus-like cells demonstrating mild irregular nuclei with one prominent eosinophilic nucleolus. There was no melanin in the cytoplasm of the neoplastic cells. Atypical mitotic figures were present. The epidermis was ulcerated, but there was no prominent intraepidermal component. There was no maturation at the base of the lesion (figure 1) and the tumour was less than 1 mm thick. These findings were consistent with a nevoid malignant melanoma. Immunohistochemical analysis for S100 and HMB-45 confirmed the diagnosis.

DIFFERENTIAL DIAGNOSIS

Nevoid melanoma superficially resembles a nevus as regards cell type, symmetry and lack of a prominent intraepidermal component. These tumours may even show maturation of deep cells, although this is often impaired. The diagnosis can be made by the presence of, often deep, dermal mitosis and cells in the base with prominent nucleoli. A subtle pleomorphism is often present.¹⁰

TREATMENT

The patient subsequently underwent a wide local resection which revealed scar tissue but no residual tumour. A cranial MRI, chest CT scan and abdominal sonography revealed no abnormalities. According to tumour, node, metastasis (TNM) staging, the tumour was T3a N0 (II_A), so the patient did not require any treatment.

OUTCOME AND FOLLOW-UP

The patient is currently being followed closely and shows no evidence of disease 1 year later.

DISCUSSION

Approximately 1% of all malignant tumours and 3% of malignant tumours of the skin are malignant melanomas.¹¹ Albinism is an autosomal recessive disorder of melanin synthesis that affect the skin, eyes and hair. Melanocytes are normally distributed in the eye and skin, but normal amounts of melanin are not synthesised.¹² The most frequently encountered tumour is squamous cell carcinoma, with basal cell carcinoma seen less often.¹² Malignant melanoma is very rare in oculocutaneous albinism,¹² but this low incidence is unexplained.¹² The present case was a albino woman with malignant melanoma of nevoid type. The term nevoid malignant melanoma is used to describe rare nodular malignant melanoma that may escape detection in routine histological sections due to lack of a prominent intraepidermal component, slight pleomorphism and

evidence of partial maturation with descent into the dermis. Nevoid melanoma mimics ordinary compound or intradermal melanocytic nevi. Furthermore, melanomatous skin cancer presents a greater diagnostic challenge in albino patients because of their hypopigmented appearance. In our patient because of the rarity of association of oculocutaneous albinism with malignant melanoma, especially the nevoid type, further investigation in addition to histopathological examination under light microscopy was carried out. Immunohistochemistry analysis for S100 and HMB-45 demonstrated the melanocytic nature of the neoplasm.

Learning points

- ▶ Melanocytic tumours are rare in albino patients and their diagnosis is difficult.
- ▶ Nevoid melanoma is an uncommon subtype of malignant melanoma and because of its deceptive morphological features is usually misdiagnosed as melanocytic nevus.
- ▶ Some studies have shown a better than expected prognosis for nevoid melanoma, but this has not been confirmed and sufficient follow-up is needed.
- ▶ We believe the association of nevoid melanoma with oculocutaneous albinism in this case merited attention.

Competing interests None.

Patient consent Obtained.

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