ORIGINAL ARTICLE

A clinical, histologic, and follow-up study of genital melanosis in men and women

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Background: Genital melanosis may clinically mimic melanoma. Little is known about the potential risk for genital and nongenital melanoma in these patients.

Objective: In this retrospective cohort study, we analyzed clinical and histologic data from patients with genital melanosis to better characterize these lesions and the risk they confer for genital and nongenital melanoma.

Methods: In all, 41 patients were identified for a retrospective chart review and histologic analysis.

Results: Genital melanosis can clinically mimic melanoma but the typical age of onset is younger than for genital melanoma. A majority of lesions were found to stabilize or regress over time. Five patients were found to have a history of melanoma, only 1 of which was in the genital region. Lesions from these patients were more likely to show melanocytes with suprabasal movement (P = .0101) and to have a higher melanocyte count ($P < .0462$).

Limitations: We present a relatively small cohort of patients with an average follow-up of only 30.5 months.

Conclusion: Patients with genital melanosis, and in particular those with any level of histologic atypia in the genital melanosis lesion, may require careful total body skin examinations for the possibility of melanoma in any body site. (J Am Acad Dermatol http://dx.doi.org/10.1016/j.jaad.2016.11.003.)

Key words: genital melanoma; genital melanosis; melanosis; penile melanoma; Sox10; vulvar melanoma.

Genital melanosis is a relatively infrequent condition with an estimated incidence of 0.01% of dermatologic patients.[1,2] Current literature on the topic is limited. The clinical appearance may be strikingly atypical with large multifocal and irregular dark macular pigmentation, often rendering clinical distinction from genital melanoma difficult without adequate histologic sampling. However, little is known about whether genital melanosis confers any risk for the development of genital melanoma or melanoma of other sites.

In light of these many unanswered questions regarding genital melanosis, we review our experience involving genital melanosis in 41 patients. We report the range of clinical and histologic findings in these cases to help clinicians better differentiate this entity from genital melanoma. We also review the melanoma history in these patients to better define the relationship between genital melanosis and melanoma. We believe this information can help to better guide clinicians regarding optimal management and follow-up recommendations for such patients.

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METHODS

After institutional review board approval, 41 patients seen over a 10-year period were included for review and a retrospective chart review was conducted through the Enterprise Data Warehouse system at Northwestern Memorial Hospital. Inclusion criteria included patients with a pigmented patch (≥1.0 cm) or multiple coalescent macules that added up to 1.0 cm or more in size that had an associated biopsy specimen. On histology, melanosis was defined by a lack of nested melanocytes with or without an increased number of single melanocytes within the epidermis but not meeting criteria for melanoma.

Demographic information, clinical history, and clinical presentations were recorded for each patient. Lesions were placed into 2 categories based on size: those additively greater than or equal to 1.5 cm or those less than 1.5 cm in size. Of the 41 patients included in the study, 35 had tissue available for histologic analysis. Three histologic features were assessed in all of the cases: melanocytic cell count/mm^2 in the epidermis using a Sox10 immunostain, nuclear atypia, and location of the cells in the epidermis. Five of the patients had multiple biopsies performed on their lesions. As in the other samples, the area with the highest melanocytic Sox10 count/mm^2 was used in the final statistical analysis.

All statistical analyses were generated using software (SAS, Version 9, SAS Institute Inc, Cary, NC). Fisher exact test was used to analyze relationships between categorical variables and a *t* test for means was used for continuous variables. A *P* value of less than .05 was considered statistically significant.

CAPSULE SUMMARY

- Genital melanosis is a benign entity that can clinically mimic melanoma.
- Genital melanosis is unlikely to present imminent risk for genital melanoma but may be associated with melanoma at other sites.
- Despite concerning clinical features, most patients with genital melanosis require only close clinical follow-up.

RESULTS

Clinical characteristics of the 41 patients included in our study are shown in Table I. Detailed clinical information was available for 38 cases. In 17 of these cases, the lesion was described as irregular in shape and color. Of these cases, 22 included lesions with at least 2 different colors, including shades of brown and black, areas of blue or gray discoloration, and areas of hyperpigmentation intermixed with areas of hypopigmentation. Twelve of the lesions had a documented size of greater than or equal to 1.5 cm. In all, 22 patients presented with multiple lesions whereas 19 patients had a single lesion. There was no significant correlation between clinical presentation and histologic parameters assessed. We also assessed patients for a history of inflammatory dermatoses or human papillomavirus infection, as these conditions have been postulated to play a role in genital melanosis, and found no correlation between these conditions and any histologic or clinical parameters and no evidence that any specific conditions are associated with melanosis. Clinical features in these patients are shown in Table II.

Of patients assessed for personal and family history, 7 of 32 patients had a family history of melanoma and 5 of 34 patients had a personal history of melanoma. Clinical and histologic characteristics of patients with a history of melanoma are shown in Table III. Only 1 patient in the entire cohort was found to have both genital melanosis and genital melanoma. There were no significant differences in clinical features between patients with and without a history of melanoma yet a correlation was found between a personal history of melanoma and 2 of the 3 measured histologic parameters: suprabasal versus nonsuprabasal location of melanocytes and melanocyte count via Sox10. Patients with a history of melanoma had a greater tendency for more suprabasal location of melanocytes within the area of genital melanosis ($P = .0101$). The average Sox10 count was 53.6 in patients with a history of melanoma, which was much greater than the average of 35.5 in patients without a personal history of melanoma ($P < .0462$). Of the 3 cases that demonstrated melanocyte nuclear atypia, 2 were from patients with a history of melanoma. This relationship approached statistical significance ($P = .0657$). This was in spite of the fact that, in 4 of the 5 cases, the melanoma was not even in the genital region. A summary of histologic findings is shown in Table IV.

Follow-up information was available for 30 of the 41 patients included in the study and none of the patients with follow-up were found to develop genital melanoma, although 1 patient developed genital melanosis 10 years after a diagnosis of genital melanoma. The average amount of follow-up time was 30.5 months with a range between 3 and 115 months of follow-up. Detailed dermatologic