Background

Diagnostic discordance in suspicious cutaneous melanocytic lesions has been well documented, and it is particularly prevalent among difficult-to-diagnose cases, for which clinical examination and histopathology may be insufficient to render a definitive diagnosis.1,2 A variety of diagnostic tools are available to differentiate difficult cases including dermoscopy, reflectance confocal microscopy (RCM) and ancillary studies including immunohistochemistry (IHC) and gene expression profile (GEP) tests.

The 35-GEP test is a clinically available ancillary tool that facilitates the diagnosis of melanocytic lesions with ambiguous histopathology. The test uses a set of gene expression arrays derived from an artificial intelligence-based approach to produce results of suggestive of benign neoplasm, intermediate (cannot rule out malignancy), or suggestive of malignant neoplasm. The 35-GEP has demonstrated a 99.1% sensitivity, 94.3% specificity, 93.6% positive predictive value (PPV) and 99.2% negative predictive value (NPV): 3.6% of samples received an intermediate result.3 Clinical utility has been demonstrated with the 35-GEP, showing an increase in diagnostic confidence and frequent adjustments in re-excision and management plans.4

Here we present a difficult case of a suspicious melanocytic lesion, using in vivo clinical imaging, immunostaining, and 35-GEP testing to inform the final diagnosis and management plan.

Case Presentation

An 85-year-old male with history of extensive sun damage and non-melanoma skin cancers presented with a 12mm diameter, irregular, multicolored skin lesion on the upper back near the superior thoracic spine (Figure 1A-B). Dermatoscopic examination revealed an atypical network, grey granules, and pigmentary structures adjacent to the periphery with central, structuralless areas (Figure 1C).

Figure 1. Lesion on the upper back of an 80-year-old male: A) Clinical photos. B) Zoom magnification of panel A. C) Dermatoscopic Imaging

Figure 2. Abnormalities detected by reflectance confocal microscopy (RCM)

A) Atypical cobblestone pattern in the epidermis. B) Loss of meshwork and bright cells infiltrating the dermis. C) Numerous spindle-shaped structures.

Figure 3. Histopathology: A-B) H&E: Discohesive nests of junctional melanocytes (yellow) and melanophages (black). C) S 100/MART-1 immunoperoxidase staining of melanocytes.

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Figure 3. Histopathology: A-B) H&E: Discohesive nests of junctional melanocytes (yellow) and melanophages (black). C) S 100/MART-1 immunoperoxidase staining of melanocytes.

Clinical Impression: Features characteristic of melanoma on sun-damaged skin

Dermoscopic shave biopsy performed and sent to the dermatopathologist for histologic workup

Acknowledgments & Disclosures

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3. The conflict between the clinical suspicions of melanoma and the ostensibly benign (though unusual) histopathology left the necessity for re-excision and the appropriate margins in question. Lacking definitive histologic features of melanoma, further diagnostic testing with the 35-GEP was implemented to achieve a more definitive diagnosis (Figure 4).

Conclusions

The malignant 35-GEP result supported the need to perform a local excision for an otherwise ambiguous case on a large lesion in an elderly patient.

The 35-GEP is a valuable ancillary tool to help achieve clinicopathologic correlation for melanocytic lesions with uncertain malignant potential (UMP) and discordance between clinical features, dermoscopy/RCM, and histopathology.

References


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