Many studies have documented suboptimal accuracy and reproducibility in the diagnosis of melanocytic lesions when histopathology is used alone. Adjunctive methods that provide objective and reliable data have been sought to distinguish melanoma from nevi.

A single Melanoma Diagnostic Score (MDS) has been clinically validated based upon the expression of the gene signature is reported to ordering pathologists and used as an adjunctive diagnostic tool (Table 1).

STUDY DESIGN

Objective

Quantify the impact of a novel molecular diagnostic test on diagnosis and treatment recommendations made by dermatopathologists attempting to differentiate malignant from benign melanocytic tumors.

Endpoints

The percentage change in diagnosis and intended treatment recommendations.

Studies are underway to further characterize the larger cohort of cases of diagnostically challenging melanocytic lesions and personalized medical treatment.

Methods

- Representative sections of difficult to diagnose melanocytic lesions encountered during routine dermatopathology practice were submitted to a clinical laboratory for gene expression testing by qRT-PCR.
- An MDS was calculated based upon measured gene expression, and the score was reported to the submitting dermatopathologist.
- Changes in the pre- and post-test questionnaires were calculated for diagnostically challenging cases.
- For treatment recommendations, only the most severe, or invasive, recommendation selected on the survey was considered. Sentinel lymph node biopsy and/or other evaluation for evidence of metastasis was considered the most invasive and further treatment necessary was considered the least invasive.

RESULTS

- This study provides prospective evidence of the clinical utility of a novel molecular assay capable of differentiating malignant melanoma from benign melanocytic lesions.
- When the MDS was available as part of a comprehensive evaluation of diagnostically challenging cases, indeterminate diagnoses were reduced by 42.7% and changes in treatment recommendations were observed in 49.1% of cases.

Table 2. Baseline characteristics of the total population of cases evaluated for study inclusion (N=1,695), as well as the diagnostically challenging subset of interest (N=216).

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<thead>
<tr>
<th>Characteristic</th>
<th>Statistic/Category</th>
<th>Total Population</th>
<th>Diagnostically Challenging</th>
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Figure 1 details the changes in treatment recommendations made for diagnostically challenging cases according to the MDS result.

- In 35.4% of cases receiving a benign MDS result, recommendations were downgraded to less invasive treatment.
- In 41.8% of cases receiving a malignant MDS result, recommendations were upgraded to a more invasive treatment.

CONCLUSIONS

- The change in indeterminate diagnoses along with a change in confidence for benign or malignant diagnoses resulted in an overall decrease in the number of diagnostically challenging cases identified within the total population after testing.
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- Figure 2 details the changes in treatment recommendations made for diagnostically challenging cases according to the MDS result.