Key publication summary

MyPath Melanoma

Clinical validity

PUBLICATION	SUMMARY
Clinical validation of a gene expression signature that differentiates benign nevi from malignant melanoma Clarke L, et al. *Journal of Cutaneous Pathology*. 2015.	The initial development and validation of MyPath Melanoma included 437 archived cases with at least two dermatopathologists agreeing on the final diagnosis. This validation established sensitivity of 90% and specificity of 91% in differentiating benign from malignant lesions across a broad range of histopathologic subtypes.
An independent validation of a gene expression signature to differentiate malignant melanoma from benign melanocytic nevi Clarke L, et al. *Cancer*. 2016.	In the second validation, 736 prospectively submitted cases with at least three dermatopathologists agreeing on the final diagnosis confirmed high performance metrics (sensitivity of 91.5% and specificity of 92.5%). A minimum tumor volume of > 10% was established.
Diagnostic distinction of malignant melanoma and benign nevi by a gene expression signature and correlation to clinical outcomes Ko J, et al. *Cancer Epidemiology, Biomarkers & Prevention*. 2017.	An additional validation study confirmed the accuracy of MyPath Melanoma in 182 lesions determined to be malignant melanoma or benign nevi based on clinical outcomes (distant metastasis or 6+ year event free follow-up). Assay performance was determined to be sensitivity of 93.9% and specificity of 96.2%.
Gene expression signature as an ancillary method in the diagnosis of desmoplastic melanoma Clarke L, et al. *Human Pathology*. 2017.	Fifty melanocytic neoplasms with a desmoplastic component demonstrated positive results are confirmatory of melanoma (specificity of 100%) but negative results do not completely exclude the possibility of malignancy (sensitivity of 80%). The performance of MyPath Melanoma in desmoplastic lesions is similar to aCGH (n=9).
Correlation of melanoma gene expression score with clinical outcomes on a series of melanocytic lesions Ko J, et al. *Human Pathology*. 2019.	In a study of 127 samples taken from the first validation cohort, MyPath Melanoma was found to have a sensitivity of 100% in detecting metastatic melanoma based on comparison to clinical outcomes and patient follow-up data. Of the 48 cases with benign MyPath Melanoma GEP scores, no evidence of malignancy was identified during the follow-up period (mean follow up of 30 months).
Clinical validity of a gene expression signature in diagnostically uncertain neoplasms Clarke L, et al. *Personalized Medicine*. 2020.	In a retrospective study in the intended use population of diagnostically ambiguous melanocytic lesions (n=181), MyPath Melanoma had a sensitivity of 90.4% and specificity of 95.5%. Performance in this study was established by comparing MyPath Melanoma results to clinical outcomes.

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MyPath Melanoma

Clinical utility

PUBLICATION	SUMMARY	
The influence of a gene expression signature on the diagnosis and recommended treatment of melanocytic tumors by dermatopathologists Cockerell C, et al. *Medicine*. 2016.	In 218 prospectively tested melanocytic lesions considered to be diagnostically ambiguous by histopathology, there was a 57% increase in definitive diagnoses following MyPath Melanoma testing.	
The influence of a gene-expression signature on the treatment of diagnostically challenging melanocytic lesions Cockerell C, et al. *Personalized Medicine*. 2017.	Evaluation of 77 prospectively tested lesions considered to be diagnostically ambiguous, identified a 72.3% reduction in re-excisions following a benign MyPath Melanoma result.	
Clinical use of a diagnostic gene expression signature for melanocytic neoplasms Tschen J, et al. *Cutis*. 2021.	In a prospective clinical outcome study, 25 patients with diagnostically ambiguous lesions and benign MyPath Melanoma results were selected. All patients were clinically managed as having benign nevi with 88% foregoing re-excision entirely. No adverse events, including recurrence or metastasis, were observed in any patient over the follow-up period (mean time of 38.5 months) confirming patient management can be safely aligned to MyPath Melanoma test results.	
A physician's guide to the use of gene expression profile ancillary diagnostic testing for cutaneous melanocytic neoplasms Marks E, et al. *JCAD*. 2023.	In this clinical use guide, dermatologists and dermatopathologists substantiate previous clinical utility studies by detailing real-world scenarios in which MyPath Melanoma testing aids in rendering a more definitive diagnosis and guides patient treatment.	
A clinical impact study of dermatologists' use of diagnostic gene expression profile testing to guide patient management Witkowski A, et al. *Melanoma Management*. 2024.	In patients with diagnostically ambiguous melanocytic lesions, dermatologists align both surgical management and follow-up frequency with MyPath Melanoma test results compared to baseline with no GEP results provided. Surgical management and follow-up frequency were increased with malignant GEP results and decreased with benign GEP results.	

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