The use of molecular testing to diagnose ambiguous melanocytic lesions

The Skin Cancer Prevention Working Group (SCPWG) is committed to ensuring patient's ready access to optimal tools for diagnosing melanoma at earliest stages. Ample data shows that diagnosis by histopathology is effective in identifying benign versus malignant nevi with consistency.

However, diagnosis by histopathology also consistently reveals inherent limitations. Data shows that 15%, or approximately 300,000 skin lesions annually in the United States, are ambiguous or equivocal by histopathology. Of these ambiguous lesions, 17% to 37% result in diagnostic discordance. Despite decades of studies and volumes of literature dedicated to the subject, the medical community has failed to identify reliable, reproducible histopathologic criteria that distinguish benign from malignant in this subset of cases. Accurate distinction for these lesions is beyond the technical limitations of a microscope.

Within the standard scope of clinical practice, ambiguous or equivocal cases are often treated as malignant, resulting in unnecessary treatment and anxiety for many patients.

Molecular testing using gene expression profiling (GEP) has been shown to add objective information to clarify this subset of ambiguous lesions – with improved diagnostic accuracy to guide appropriate patient management such as surgery and follow-up frequency. The medical community has long sought ways to improve the accuracy of melanoma detection. The accurate and objective data from GEP testing translates into fewer indeterminant cases. A decrease in unnecessary follow up visits results in fewer surgical re-excision procedures and lower patient anxiety.

In its most recent guidelines for Cutaneous Melanoma, the National Comprehensive Cancer Network (NCCN) supports the use of ancillary tests, including GEP, for the diagnosis of ambiguous melanocytic lesions. Likewise, in its most recent Appropriate Use Criteria publication, the American Society of Dermatopathologists (ASDP) identified (6) clinical scenarios when GEP utilization is supported. Furthermore, in its most recent guideline recommendations, the American Academy of Dermatology (AAD) recommends that ancillary diagnostic molecular techniques, including GEP, may be used for equivocal melanocytic neoplasms. Finally, the Centers for Medicare and Medicaid Services (CMS) issued a coverage policy for GEP tests to diagnose melanoma in 2019.

In line with the NCCN, the ASDP, the AAD, and CMS, the SCPWG formally advocates utilization of GEP tests to distinguish between benign and malignant nevi – now that they have been shown to improve key diagnosis parameters compared to histopathology alone.

April 30, 2022