IDS collaborative study on BAP1-deficient nevi: clinical and dermoscopic features

-Background and rationale:

BAP1-deficient nevi (BDN), also referred to as Wiesners nevi or Bapomas, are recently described melanocytic tumors with unique genetic profiles and unknown biological behaviors. While some authors consider these lesions to be variants of Spitz nevi, others think they represent distinct entities with some overlapping clinical and cytologic features.[1,2] The occurrence of multiple BDN have been associated with a familial tumor syndrome involving germline inactivating mutations in *BAP1*, a tumor suppressor gene located on chromosome 3p21.[3] BAP1-associated tumor syndrome predisposes to the development of various malignancies, including mesothelioma, uveal melanoma, and cutaneous melanoma. Sporadic cases of singular BDN without any syndromal associations have also been described.[3,4] Genetic analysis of both syndromal and sporadic BDN have demonstrated biallelic loss of *BAP1*, often combined with *BRAF V600E* mutations.[2,4] There are limited reports on the clinical characteristics of BDN.[3,5] Literature regarding the dermoscopic appearances of these lesions is almost completely lacking.[6]

-Study objectives:

The primary objective of this study is thus to correlate the clinical and dermoscopic morphologies associated with BDN. In doing so, we can improve the diagnosis of these lesions, as well as aid in the identification of individuals at increased risk for the development of systemic and cutaneous malignancies.

-Study Design and Procedures:

Because of the rarity of BDN, we will solicit help from the International Dermoscopy Society (IDS) to amass a comprehensive collection of clinical and dermoscopic images of BDN. Our objective is to create an international database and to thoroughly analyze these images in order to determine their common clinical and dermoscopic features.

-Data Collection Procedures:

We will be emailing all members of the International Dermoscopy Society a brief project description requesting de-identified retrospectively photographed clinical and

dermoscopic images of BDN with an accompanying e-survey requesting the following de-identified clinical information per case submitted: age at diagnosis, anatomic site fitzpatrick skin type, histopathologic diagnosis, results of any ansillary immunohistochemical and molecular studies, and personal or family history of melanoma. As in previous collaborative projects between IDS and MSKCC, we will request contact information (including: name, institution, country, e-mail) from each submitting physician.[7,8] This information will be used for authorship purposes. To reiterate, no identifying information or contact information will be requested for the patients that are being studied. The de-identified forms will be emailed to Dr Oriol Yelamos (yelamoso@mskcc.org) who will coordinate data collection.

For selection of cases from MSKCC, we will perform a Dataline search of surgical pathology biopsy records over the past 15 years to systematically identify cases of histopathologically proven WN. We will then collect associated retrospective clinical and dermoscopic images that have a pre-existing consent via our medical imaging archiving software (Dermagraphix). Only non-patient identifiable lesion images will be exported from Dermagraphix (software gives users the option to extract de-identified data). Immediately upon export, all images and their associated image data will be reviewed by MSKCC personnel to ensure that all data has been de-identified by the software. Each image will be coded with a unique study identification number. We will also be collecting the aforementioned clinical information for each inhouse WN case from the patients' medical records. Protected Healthcare Information (PHI) will not be disseminated to the collaborators and will only be available to the study personnel at MSKCC for quality assurance purposes. Dermoscopic images taken at initial biopsy will similarly be obtained from the patients' medical records (Mirror Database).

-References:

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