Doctor's Note — Clinical Trial Summary

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TRIAL

Frequency and Clinical Phenotype of BAP1 Hereditary Predisposition Syndrome

NCT ID: NCT04792463 Phase: N/A Sponsor: Mohamed Abdel-Rahman Status: Recruiting

SUMMARY

This research will have a significant impact on the overall management of those cancer patients and their family members who are at risk for hereditary cancer due to germline inactivation of BAP1. Our study will ultimately facilitate the development of novel screening, prevention and treatment strategies for these individuals with the syndrome. Because the vast majority of UM develop in pre-existing nevi, characterization of individuals at high risk for development of UM will allow closer screening and earlier intervention which would improve the treatment outcome not only for retaining vision but also for overall survival. Similarly in patients with germline BAP1 mutation CM develops in premalignant atypical melanocytic lesions and careful follow up of these patients will improve the outcome of their disease. In addition this study could have impact on the management of patients with personal and/or family history of several other cancers reported in patients with germline BAP1 mutation such as mesothelioma, renal cell carcinoma, cholangiocarcinoma, hepatocellular carcinoma, meningioma and basal cell carcinoma.

KEY ELIGIBILITY CRITERIA

- Patients who meet any of the following criteria:
- 1. Personal history of one cancer reported in BAP1 cancer predisposition syndrome and family history of at least two 1st or 2nd degree relatives with cancer reported in hereditary BAP1 cancer predisposition syndrome such as UM, CM, mesothelioma, RCC, cholangiocarcinoma, meningioma and hepatocellular carcinoma.
- 2. Any patient with personal history of at least 2 cancers reported in hereditary BAP1 cancer predisposition syndrome.
- 3. Any subject (affected or unaffected) with a documented BAP1 pathogenic/ likely pathogenic variant.
- 4. Any patient with a cancer reported in BAP1 and a germline variant of uncertain significance.
- 5. At risk relatives of a patient with documented BAP1 mutation.
- * Study material including consent forms are currently only available in English so non-English speaking subjects are excluding

ENROLLMENT CONTACT

Mohamed H Abdel-Rahman, MD, PhD
The Ohio State University Wexner Medical Center
Columbus, Ohio, United States
Phone: 614-292-1396
Email: Mohamed.Abdel-Rahman@osumc.edu

Total sites: 1 | 1 currently recruiting