MelaNostrum New Proposal Form ***(version May 2017)***

Investigators who would like to propose a new research project, please complete the form and send it back to Dr. Maria Teresa Landi (landim@mail.nih.gov)

I. Project Title

Evaluation of *SLC45A2* variant rs250417 and *MC1R* variants as modifying factors for the development of melanoma in *CDKN2A* mutation carriers from Mediterranean countries.

Date: May 25th 2017

II. Project Group Investigators

Project proposal: Miriam Potrony and Susana Puig

All Melanostrum members interested who have and wish to share *SLC45A2* variant rs250417 and *MC1R* variants in *CDKN2A* mutation carriers (affected and non-affected individuals).
If *SLC45A2* variant rs250417 and *MC1R* variants data is not available for *CDKN2A* carriers, but DNA is available, we can genotype and sequence them in Barcelona.

III. Background

MC1R is the master regulator of human pigmentation and is a highly polymorphic gene. Some *MC1R* variants are associated with the red hair color phenotype. *MC1R* variants are also associated with an increased risk to develop melanoma. Furthermore *MC1R* variants can modulate the risk to develop melanoma in patients with *CDKN2A* mutation.
SLC45A2 SNPs have been associated with the development of melanoma and also phenotypic traits.
In the recent meeting in Athens we have seen that *SLC45A2* variant rs250417 is more frequent in the Mediterranean population than *MC1R* variants.

IV. Specific Aims

To evaluate the role of *SLC45A2* variant rs250417 together the presence of *MC1R* variants as modifying factors for the development of melanoma in *CDKN2A* mutation carriers from Mediterranean countries.

V. Methods

1. Recruit phenotypic information, *MC1R* variants and *SLC45A2* variant rs250417, from patients and non-affected with mutation in *CDKN2A.* An excel file with the necessary information will be sent to any group willing to participate.
2. If *MC1R* variants and *SLC45A2* variant rs250417 is not available from the *CDKN2A* carriers, DNA can be sent to Barcelona and we can genotype the variant and sequence *MC1R.*
3. Perform statistical analysis.
4. Manuscript Draft

VI. Materials or variables needed from the study PIs

Phenotypic data + *MC1R* variants + *SLC45A2* variant rs250417 from CDKN2A-mutated individuals (affected and non-affected)
If *MC1R* variants or *SLC45A2* variant rs250417 is not available, DNA can be sent to Barcelona and we can analyze it.

VII. Time line

Answer in principle on willingness to participate in the project- June 2017.
Sending data and DNA (if necessary) to Barcelona – October 2017.
End of data and DNA collection - October 2017
Assessing *MC1R* variants or *SLC45A2* variant rs250417 – December 2017
Prepare a report with the results to share with every group participating – February 2018
Paper Drafting – March/April 2018

VIII. Funding Sources and Declaration of Conflict of Interests. *To ensure full transparency and to protect collaborating study PIs, MelaNostrum requires the Project Leaders to disclose any circumstances that could give rise to a potential conflict of interests related to the proposed project activity in particular, or to melanoma in general, including but not limited to funding sources, employment and consulting, board membership and investment interests within the last 5 years.*

Each group's funding for its own sequencing/genotyping
Teresa Landi's funding for Melanostrum sequencing /genotyping
No conflict of interest

IX. Other remarks (e.g. dissemination plan, etc)

Publication time
Paper submission – May/June 2018

Authorship
The study won't interfere with Melanostrum and each group publications plans.

Proposed authorship to be discussed based on the number of families included by site of Melanostrum PI and additional data provided. First author: Miriam Potrony (first co-authors to be discussed). Last author: Susana Puig (others to be discussed). All PI accepting to participate will be co-authors of the paper, according to the number of samples included other authors from each group can be included. Order according to the number of samples included.