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Summary of research

Genetics, eye, hair, colour, skin cancer

Application Lay Summary:

1a: Skin cancers are very common and potentially deadly diseases. A number of factors that determine the individual risk to developing melanoma. Exposure to sunlight is an important risk factor, yet its effects depend on individual, genetically inherited characteristics, such as the skin, hair and eye colour as well as the skin type. These traits, important to physical appearance are the quintessential heritable and have long been empirically used, among other traits, to determine family relationship and identical twins.

This research will find: 1) genes that influence physical appearance traits that are risk factors to skin cancer (eye, hair colour), 2) genes that determine skin type. Results of this research will improve our understanding of the mechanisms of how these traits are controlled and potentially how they can contribute to skin cancer. This research may lead to personalised recommendation to avoid skin damage or cancer in certain individuals.

1b: This research fits with the UK Biobank's stated objectives to "improve the prevention, diagnosis, and the promotion of health throughout society". The results of this work will contribute to better skin cancer prevention and will promote to safer exposures to sunlight and other environmental factors.

1c: This research will use a statistical association test of gene differences (GWAs) to address current gaps in knowledge about molecular mechanisms behind the three traits. This research will undertake: 1) statistical association analyses between phenotypes (requested phenotypes as specified) and genotypes, and 2) statistical modelling of interactions between external factors and some of these phenotypes (e.g. sun exposure, using "time spent outdoors as a proxy" and skin type).

1d: We anticipate that phenotypic values will be available for most of the UK Biobank participating subjects (minimum number of individuals for any of the requested phenotypes is 498,425), but since genotype availability is partial, at the time of application, we anticipate that genotypes will be the limiting factor. This limitation will be of decreasing importance since more UK Biobank genotypes will become available in the near future.

We anticipate being able to run an exceptionally well powered analyses, at the current stage, with most of the 152,729 individuals for whom genotypes are available and have overlapping phenotypic information.