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COMMENTARY

Collaborative efforts in studying Mediterranean melanoma families: A step towards precision medicine

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Collaborative efforts in the field of medical research play a pivotal role in advancing our understanding of rare diseases. The recent article by Pellegrini et al. focusing on melanoma susceptibility within Mediterranean populations serves as an exemplary model of the profound benefits that can be derived from such cooperative endeavours.[1] The study, led by the MelaNostrum Consortium, successfully bridges a significant gap in the existing body of melanoma research, making it a notable contribution to the field. In the realm of melanoma susceptibility research, a predominant bias has long prevailed, with most epidemiological studies mainly concentrating on fair-skinned individuals hailing from regions such as the United States, Australia and Northern Europe. Unfortunately, this focus has perpetuated the underrepresentation of Southern European populations, characterized by their unique genetic backgrounds, heightened UV exposure and a greater prevalence of dark-skinned individuals. The MelaNostrum Consortium, including centres from Greece, Italy and Spain, embarked on a mission to rectify this long-standing imbalance. This strategic shift is not only a laudable endeavour but also marks a significant step towards diversifying the knowledge base and enhancing the applicability of research findings to a broader spectrum of people. One of the study's most remarkable accomplishments lies in its comprehensive approach to data collection, including epidemiological, clinical and genetic data, with a particular focus on well-established high- and intermediate-penetrance genetic variants. The identification of pathogenic/likely pathogenic CDKN2A variants in 13.8% of the studied families stands as a significant finding, in tandem with recent Italian experiences in melanoma genetics.[2] Furthermore, the study's revelation regarding the most robust predictors of melanoma risk, including criteria such as the presence of ≥2 multiple primary melanoma cases, >3 affected members within a family and the occurrence of pancreatic cancer within the family, confirms the recommendations currently guiding genetic counselling referral in clinical practice.[3] These discoveries serve as the steppingstones for further investigation in this field. Notably, despite the geographical proximity of Greece, Italy and Spain, they exhibit distinct historical and genetic variations, highlighting the need for further research stratification based on ethnic backgrounds with different genetic landscapes. Italy, for instance, possesses a rich genetic tapestry shaped by millennia of history, including Neolithic migrations, like the settlement of Sardinia and more recent 'Barbarian invasions' in central and northern Italy.[4] Recognizing and delving into these intricacies can empower researchers to identify genetic markers that transcend nationality, catering to specific subpopulations. This approach, in turn, can facilitate heightened precision in risk assessment and the development of precisely targeted preventive strategies. Moreover, expanding the consortium to include more Mediterranean centres with similar melanoma demographics becomes a crucial proposal. This extension would enhance the study's overall robustness by encompassing the diverse range of genetic backgrounds found in different regions within these nations. Ultimately, this comprehensive approach propels us closer to the forefront of precision medicine and personalized interventions for cancer prevention and treatment.

CONFLICT OF INTEREST STATEMENT

None.

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DATA AVAILABILITY STATEMENT

Data sharing not applicable to this article as no datasets were generated or analysed during the current study.

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