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Collaboration and infrastructure key in improving survival rates from Melanoma

Melanoma in Australia represents a major, unsolved public health challenge. It remains one of the leading cancer killers, especially of young and middle-aged adults. This is partly because prognosis remains poor and largely unpredictable for all but early stage disease, and there are essentially no effective treatments for advanced disease. The underlying biology of the disease, and its lack of response to traditional cancer therapies is poorly understood. However recent studies of melanoma has provided proof of principle in identifying possible novel therapeutic targets for treating this disease through genomic analysis.

A project to create large-scale Systems Biology datasets to allow cell biology to be undertaken in a systematic and comprehensive manner was first proposed to Bioplatforms Australia (BPA) by a joint team from the Queensland Institute of Medical Research (QIMR), Westmead Millennium Institute (WMI) and Melanoma Institute Australia (MIA). With a view to ensuring the proposed framework data would be supported, in an enduring and accessible state, using multiple research communities with multiple research agendas, BPA undertook a broader consultation involving stakeholders from various institutions and States. Parties now involved in this project comprise a broad collaboration of leading Australian scientists and institutions including MIA, WMI, QIMR, University of Sydney, Western Australian Institute of Medical Research, Australian National University John Curtin School, Peter MacCallum Cancer Centre and others.

BPA led the working groups responsible for Communications and co-investment; Sampling, design, ethics and data; and Informatics (storage, accessibility and privacy issues). The management and accessibility of the project data is undertaken by a combination of BPA-embedded and non-embedded bioinformatics groups and is hosted on infrastructure supported by BPA.

“The tissue resource from this collaborative project has created the largest repository of melanoma tissue and linked data in the world. Data from this resource sets the standard for melanoma clinical and pathologic research, such as setting criteria for staging of the disease,” said BPA General Manager, Andrew Gilbert. “The project leverages and expands collaborations from a pre-existing NHMRC program grant and previous collaborative parties established under the International Cancer Genome Consortium program of the NHMRC.”

Project results were recently published in *Nature*. BPA’s support enabled the first high coverage whole genome sequencing study of a large cohort of melanomas from cutaneous (skin), acral (hands and feet) and mucosal sites. The data revealed mutational signatures of ultraviolet radiation (UVR) exposure were dominant in cutaneous melanomas, while complex structural rearrangements accounted for the majority of aberrations in acral and mucosal melanomas (areas typically not exposed to UVR). Understanding mutational processes and genomic drivers in all subtypes [including those attributable to ultraviolet radiation (UVR) and non-UVR] is key to further progress in melanoma epidemiology, prevention and targeted treatment globally.

Robust collaboration and enduring maintenance of accessible, searchable and extensive data are the cornerstones of the success of this project. “The datasets will have utility far beyond melanoma research, and will be of value to all basic and translational cancer researchers, cell and molecular biologists working outside cancer, and population geneticists,” said Gilbert.

Further information:

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