



Sector submission to National Consultation on the Roadmap and Implementation Plan for the MRFF Genomics Health Futures Mission

April 2021

The Genomics Health Futures Mission (GHFM) is investing \$500 million over 10 years in genomics research under the Medical Research Future Fund (MRFF). It will improve testing and diagnosis for many diseases, help personalise treatment options to better target and improve health outcomes, and reduce unnecessary interventions and associated health costs

Questions -

1. What is your name?

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2. What is your email address?

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3. What is your organisation?

Vision 2020 Australia

4. Residential state or territory

Victoria

5. Are these priority areas for investment identified in the Implementation Plan the most effective way for delivering on the Missions goals and aims? (300 words)

Vision 2020 Australia would like to see the Genomics Mission provide more support for research that can support the roll out of genomic treatments in areas such as eye disease, where significant data collection has occurred and the field is able to now use genomics therapeutically.

In addition to its state of readiness, supporting such research in the area of eye disease has the potential to have a significant impact on health status by altering the course of conditions such as glaucoma and inherited retinal diseases (IRDs), which are one of the current leading causes of permanent vision loss in Australia.

These are areas where development of novel early-stage therapeutics (Priority area 2.3) are strongly advanced internationally with more than 30 ongoing gene therapy clinical trials for IRDs for example, but none in Australia. Australia has strong clinical and research expertise in vision science and therefore a high potential of being part of the ongoing transformative status of genomics eye research worldwide.

Supporting research that can underpin such therapeutic use can not only enhance outcomes for those patients, but inform the broader therapeutic application of genomics in other priority fields.

6. Are there existing research activities which could be utilised to contribute to the Genomics Mission Roadmap and/or Implementation Plan aims and priority areas for investment. How can these be leveraged? (200 words)

- Australian Inherited Retinal Disease Registry (AIRDR): national IRD patient database, which provides genetic screening and conclusive genetic reports (Priority area 1.1)
- Several research groups nationally are already working on the development of novel gene therapy-based therapeutics for vision loss. Several of these studies are currently being hampered by the lack of national funding as these conditions are considered rare. This leads to a slow progression of these therapeutics, not because they are not effective, but because the lack of funding does not allow them to progress further.
- Building of an advanced viral vector manufacturing facility at the Westmead Health and Innovation Precinct. This ongoing and already funded project by the NSW Government, will greatly facilitate the translation of preclinical gene therapy data into clinical trials stages.
- Glaucoma, as one of the most heritable polygenic diseases, is suitable as a flagship for testing the implementation of polygenic risk scores and considerable progress to this translation has been made by Australian researchers. This will help address the major challenge of undiagnosed glaucoma.

7. Are the 'Evaluation approach and measures 'appropriate for assessing and monitoring progress towards the mission's goal and aims? (200 words)

Yes