



Roadmap to integrate Indigenous genome assemblies into a national instance of the international human reference genome resource.

The human genome reference sequence provides the spatial coordinates that underpin all human genomic and epigenomic research. Managed by the GRC, it is the foundation of much of modern biomedical research and, increasingly, clinical practice. Over the next few years, it will be transformed from its current linear form to a graphical representation of the diversity of human ancestry. This change will address current inequity and lack of inclusion resulting from primarily European representation.

NCIG (with NHMRC funding) is creating genome assemblies from Indigenous Australians required for their inclusion in this enterprise. This proposal is to develop a roadmap for how the outcomes of this research can be developed for incorporation in the pan-human reference graph.

The project will address quality and data/metadata standards, data curation, methodological and analytical, hosting and Indigenous governance requirements, validation and documentation systems, proficiency testing, staffing, training and education requirements, participant involvement, accessibility, national coordination and international engagement.

Start date

24 July 2019

Expected completion date

21 October 2019

Investment by ARDC

\$49,999

Co-investment partners

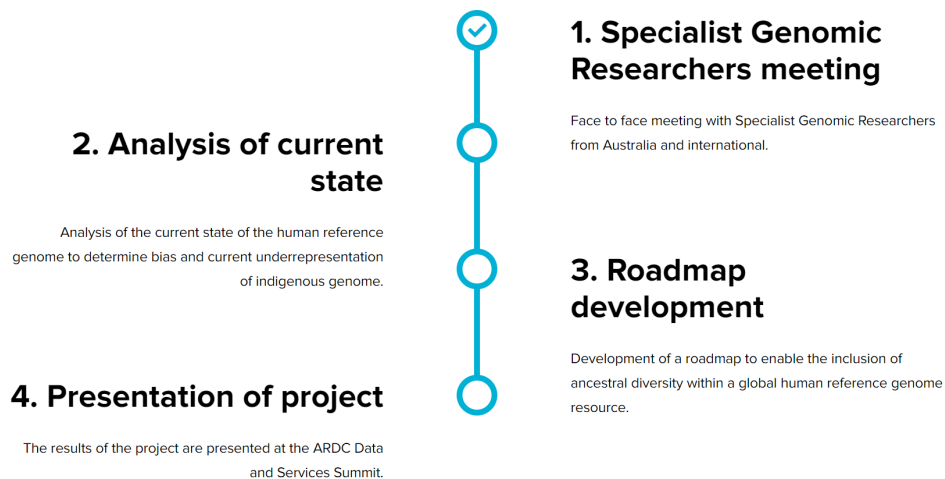
[National Centre for Indigenous Genomics, ANU](#)

[Australian National University](#)

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Lead node



Core features



Needs analysis

What is needed to enable these indigenous genome assemblies to be incorporated into the evolving human genome reference resource?



Roadmap created

The project identified that the requirements to ensure Indigenous inclusion are far greater than anticipated. The resulting roadmap identifies how this critical unmet need can be addressed.

Who is this project for?

- Peak bodies
- Research organisations
- Infrastructure providers
- Commercial infrastructure providers
- Government (state and commonwealth)



What does this project enable?

This project identified the need for an Australian reference genome resource to underpin future developments in important areas of biomedical research and to enable the transformative impact of precision medicine. The project has raised awareness of the need for this resource among contributors, collaborators and consumers, and as part of NCIG's ongoing engagement program, among Aboriginal and Torres Strait Islander communities and organisations.

More generally, the human reference genome is the resource central for all of genomics and much of biomedical research. It is a unifying resource that binds biomedical research to clinical practice. Its role in biomedical discoveries is increasingly important as the direct study of human biology underpinning health and wellbeing replaces much of work for which model systems have historically been required.



Handy resources


- [FAIR assessment](#)
- [Final Report](#)
- [Presentation](#)
- [National Centre for Indigenous Genomics](#) – ANI College of Health and Medicine
- [Repatriation](#): bringing home the manggu samples
- [NCIG-genomebrowse](#)




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