

National Call for Expressions of Interest for production, characterisation and therapeutic testing of preclinical models of human disease

Opportunity

Support for the production and phenotyping of preclinical models and their use for the development of new therapies is available. This opportunity is jointly funded by the Medical Research Future Fund (MRFF) through the Phenomics Translation Initiative (PTI) and the National Collaborative Research Infrastructure Strategy (NCRIS) through Phenomics Australia (PA).

Support is available for:

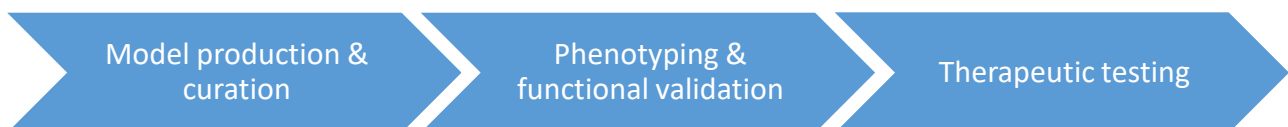
- Production of gene-edited mouse strains, somatic human and mouse cell lines carrying allele(s) identified in patient(s) and / or
- Functional validation of alleles to prove causality to disease of interest (phenotyping of model) and / or
- Preclinical testing of potential therapeutics to specifically target the pathogenic pathway

Support is intended to complement other funding. Applicants should outline the resources they have to move the project forward towards an outcome (i.e. other funding / support or publication).

Goals of the MRFF-Accelerated Research-Phenomics Translation Initiative (PTI) project

1. Understanding which rare alleles contribute to disease.
2. Defining how alleles lead to disease mechanisms.
3. Development and testing of therapies that target the pathways affected by the alleles.
4. Identifying novel diagnostics and better treatment options for patients with diseases that are currently chronic, debilitating and incurable.

This national partnership between Phenomics Australia and the Phenomics Translation Initiative offers the establishment of new experimental capability and expanded capacity for the accelerated discovery of disease-causing alleles and pathogenic mechanisms, the identification of therapeutic targets, and the development of new therapies and diagnostic assays.



Expressions of interest are invited to be submitted through an online application: [Link to application](https://anu.au1.qualtrics.com/jfe/form/SV_40BFrorluwX52e1), or found here: https://anu.au1.qualtrics.com/jfe/form/SV_40BFrorluwX52e1

Submission close date: 16/06/2021

Eligibility and Selection Criteria: Projects will be assessed according to the following criteria:

1. Project proposals aligned with any stage of the Phenogenomics pipeline for the production and/or phenotyping of cell- (somatic human) or mouse-based models, and/or therapeutic testing;
2. Projects must be focused on a human gene allele(s) highly likely to be causative in human disease;
3. Technical feasibility.

For more information on partners or for enquiries:

Phenomics Translation Initiative: <https://jcsmr.anu.edu.au/research/centres/pti>

Phenomics Australia: <https://phenomicsaustralia.org.au/>

Enquiries:

- phenogenomics@anu.edu.au or Miranda Liu, PTI Project Coordinator, 02 6125 5794
- contact@australianphenomics.org.au or Jim Hennessy, Phenomics Australia, COO, 02 6125 7735

Assessment Process

MRFF- and NCRIS-funded support will be allocated to projects for specific services following assessment and recommendation by the ANU PTI Advisory Committee, PTI Project Management Office, and Phenomics Australia.

All applications will be handled and assessed confidentially. PTI Advisory Committee members are required to declare conflicts of interest. This committee is chaired by the Director of The John Curtin School of Medical Research.

Please note that only high-level feedback will be provided to applicants following the Committee's decisions and applicants will not have the opportunity to respond.

Prior to the materials being delivered a Material Transfer Agreement will need to be established between the researcher and the institution that provides the service.

Production of mouse lines will take place through one of the following four Phenomics Australia-supported national Genome Engineering Service Nodes:

- Phenogenomics Targeting Facility (PTF), [Australian Phenomics Facility](#), Australian National University
- [South Australian Genome Editing \(SAGE\)](#), South Australian Health & Medical Research Institute / University of Adelaide
- [Melbourne Advanced Genome Editing Centre \(MAGEC\)](#), Walter and Eliza Hall Institute of Medical Research
- [Monash Genome Modification Platform \(MGMP\)](#), Monash University

Cell lines can be produced at either these two facilities:

- [Phenogenomics Targeting Facility](#), Australian National University
- [Monash Genome Modification Platform](#), Monash University

Functional validation (phenotyping) and therapeutic testing can be conducted through Phenomics Australia Service Nodes and the Australian Phenomics Facility at The Australian National University.

Further rounds are intended to be held through 2021. Support is available for researchers with proposals at any stage of the Phenogenomics pipeline.

Phenomics Australia and Phenomics Translation Initiative are funded through Medical Research Future Fund (Department of Health), National Collaborative Research Infrastructure Strategy (Department of Education).



Costs and Contribution

Production of a gene-edited mouse/cell model carrying gene allele identified in patient(s)

Support is provided to produce and supply mouse strains and/or cell lines carrying the gene allele of interest. A co-contribution will be required to support associated production costs.

Indicative contribution fees*:

- | | |
|--|-----------|
| • Somatic human cell lines (excludes hiPSC and mES cells) - Knockout | \$5,000 |
| • Somatic human cell lines (excludes hiPSC and mES cells) – Point Mutant | \$7,000 |
| • Simple modification of mouse or cell line model - Knockout | \$5,000 |
| • Simple modification of mouse or cell line model – Point Mutant | \$6,000 |
| • Complex modification of mouse or cell line model: | \$12,500+ |

* These indicative user fees apply to academic NFP projects. Actual fees charged will depend on specific production requirements and where production will be conducted.

NB: All mouse lines will be sperm cryopreserved and archived in the Australian Phenome Bank will be publicly available and distributed by the [Australian Phenome Bank](#), once an initial embargo period expires.



Functional validation of gene allele to prove causality to disease of interest (phenotyping of model)

- Support for phenotyping mouse strains modelling human disease: up to \$20,000
 - Includes consumables and staff time if PTI staff perform work. (Details of supported phenotyping assays will be determined via further discussion with applicant)
 - A co-contribution will be required in the form of animal service/ facility costs
- Support is for phenotyping to show causation not to collect sufficient data for publication of grant application



Testing of therapeutics in models that can specifically address the pathway that has been disrupted

- Support the phenotyping up to \$20,000 as above, using therapeutics
 - Does not support the cost of purchasing or preparation of the therapeutics
 - Includes consumables and staff time if PTI staff perform work. (Details of supported phenotyping assays will be determined via further discussion with applicant)
 - A co-contribution will be required in the form of animal service/ facility costs
- Support is for phenotyping to show an affect of the therapeutic on phenotype characterised above not to collect sufficient data for publication of grant application

As a research project progresses through the pipeline, from production to phenotyping and therapeutic testing, researchers work with the PTI Project Management Team to request additional support from the PTI Advisory Committee. Successful applicants will be required to co-contribute to costs.