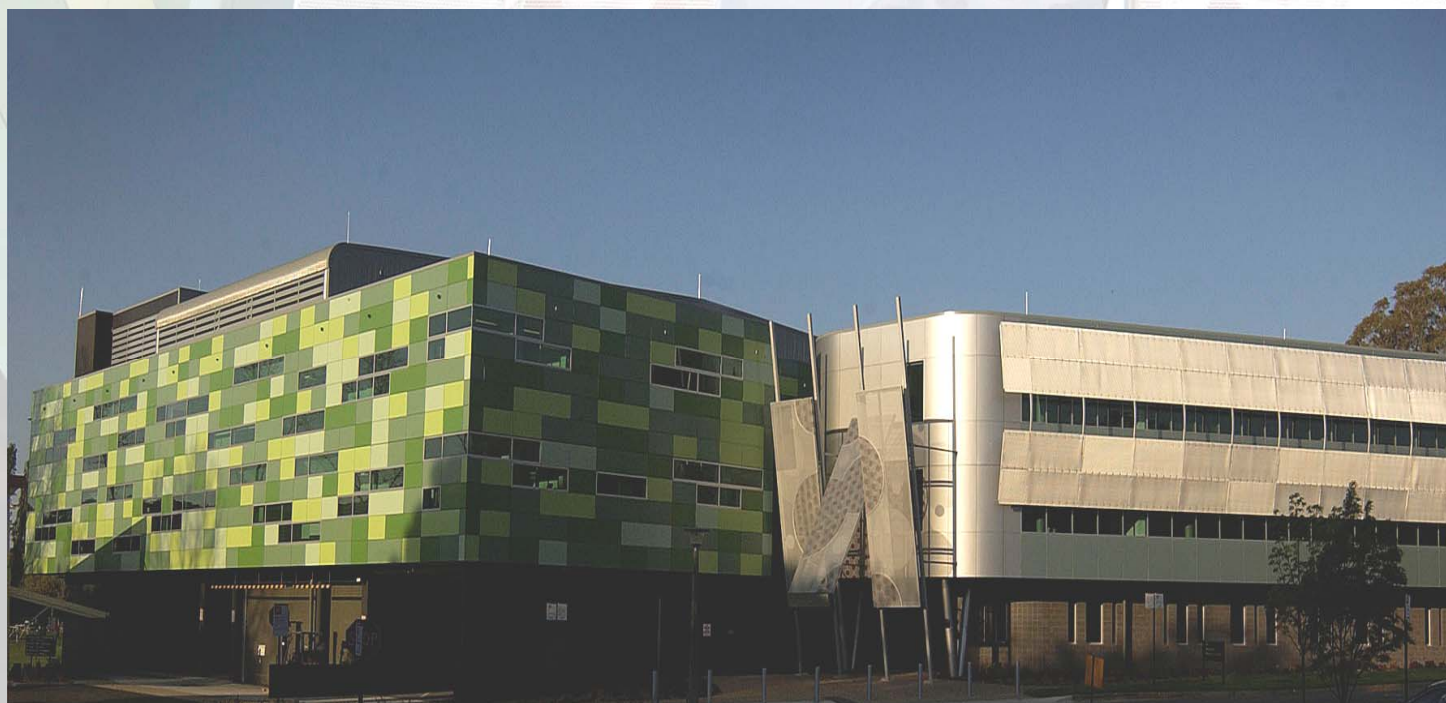


THE AUSTRALIAN PHENOMICS FACILITY



www.apf.edu.au



Genome-Phenome Discovery

THE APF IS YOUR LEADING ORGANIZATION OFFERING YOU ACCESS TO ONE OF THE MOST COMPREHENSIVE CENTRES FOR RESEARCHING HUMAN AND ANIMAL HEALTH VIA THE ELUCIDATION OF THE GENOME—PHENOME CODE.

OUR TEAM WILL HELP FAST TRACK YOUR RESEARCH TO THE INTERNATIONAL STAGE BY PRODUCING PEDIGREES OF MICE WITH INFORMATIVE GENE ALTERATIONS FOR UNDERSTANDING CAUSAL RELATIONSHIPS BETWEEN GENES AND DISEASE.

The Process of Genome-Phenome Discovery

Pedigrees of ENU Gene Variant Mice

Genome-wide chemical mutagenesis of mouse spermatogonia with Ethyl-Nitrosourea (ENU) allows a high fraction of mammalian genes to be altered without any prior assumptions or knowledge about their physiological roles. The point mutations produced by ENU yield a large range of gene variants that reveal novel phenotypic correlations of genes as well as different functional roles not revealed by gene knockout models.

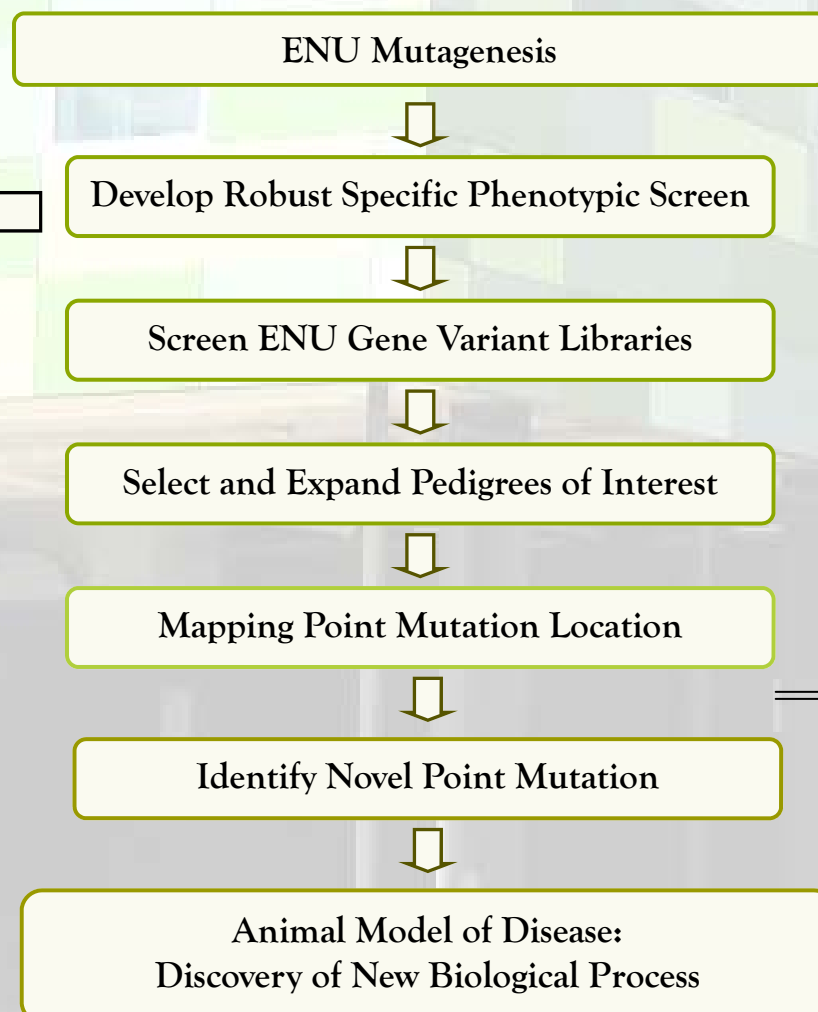
Researchers work with the Scientific Programs team to custom-design ENU-gene variant pedigrees of mice and develop robust phenotypic screens for the identification of new gene-variant mouse models of disease in their area of interest.

The APF provides the expertise and scale to advance your research programme through:

- Access to large-scale collections of ENU gene-variant pedigrees for the detection of genetic alterations for new recessive or dominant traits. Types of ENU-gene variant pedigrees:
 - Pedigrees founded on inbred background, e.g. C57BL/6
 - Pedigrees on a sensitised background, e.g. transgenic or knockout
- Expertise in project management, screen and library design, phenotyping equipment and pathology.
- Researchers have access via the internet to a custom built *Musterer* database that maintains and manages all data relating to their ENU-gene variant mouse pedigrees including breeding, genotype data and test results.

Examples of Screens:

- Immune development
- Autoimmunity: Type-1 Diabetes, Lupus
- Humoral Immunity
- Development
- Childhood deafness
- Cancer
- Obesity
- Infertility
- Lactation
- Neurological
- GI tract
- Infectious diseases



Comprehensive Genotyping & Mapping Services

The Genotyping and Mapping team provides personalised and complete project services that include accurate high-throughput genotyping, mapping and gene identification of your gene variant strains.

- **Sample Tracking:**
 - Mouse samples and DNA preparation performed in-house and tracked by bar coding LIMS on our *Musterer* database
- **Genome Wide Mapping:**
 - Advice on mapping strategies suited to pedigree structure
 - Use of Amplifluor fluorescent SNP-based assay and Affymetrix 5K Mouse SNP chip
 - Fine Mapping Custom Primer Design: New marker primer design to narrow the chromosomal linkage
 - Comprehensive Analysis and interpretation of mapping data
- **Gene Identification:**
 - Experienced, personalised service: advice on resequencing methodologies.
 - High throughput primer design, gene resequencing and analysis of data for final identification of causal mutations.

Design Mapping Strategy



Genome-wide scan to determine linkage



Linkage Confirmation and Fine Mapping



Interpretation of Fine Mapping Data



Sequence Candidate Gene(s) in Interval



Identification of Causal Mutation



Custom Design Genotyping Assay for Causal Mutation

“THE APF PROVIDES
THE EXPERTISE AND
SCALE TO ADVANCE
YOUR RESEARCH”

NHMRC Australian Phenome Bank

The NHMRC Phenome Bank is a central database of genetically modified mice, both live and cryopreserved, that are held in Australia.

An internet-accessible database, the Phenome Bank allows you to search for murine strains carrying mutations in particular genes, strains with transgenic alterations or mice with particular phenotypes.

The database relies on researchers contributing their mouse strain information. To do so, please provide your strain information to the database curator, Dr Stuart Read (stuart.read@anu.edu.au)

To access the database please visit: <http://pb.apf.edu.au>

Mouse Sperm Cryopreservation Service

To complement the Phenome Bank, we offer **free of charge** sperm cryopreservation from all mouse strains distributable to other researchers.

- Sperm are cryopreserved from multiple mice and duplicate samples are stored at both the ANU and Monash University storage facilities.
- The information of cryopreserved strains are entered onto the NHMRC Australian Phenome Bank.
- The depositing researcher/institution maintains ownership of the strain and may impose MTAs with requesting scientists.
- Researchers may also pay for the cryopreservation and block listing on the database.
- The APF also offers strain rederivation by in vitro fertilization (IVF) and intra-cytoplasmic sperm injection (ICSI) through Monash University.

To find out more about how the APF can help expand your research prospects, please contact us through the details shown below, or visit us online at

www.apf.edu.au

The Australian Phenomics Facility

117 Garran Road
Acton ACT 0200
Australia

Contact:

**Anusha Subramaniam
Scientific Programs**

P: +612 6125 9168

F: +612 6125 1381

E: anusha.subramaniam@anu.edu.au