Australian Functional Genomics Network

Expression of Interest Form for Model System Study

**Application Procedure:** Candidate genes with unresolved variants are proposed by the clinical team and prioritised by the Clinical Review Committee (CRC) on the criteria of the clinical need and likelihood that the genetic variant is disease-causing. AFGN registered researchers are invited to submit a research proposal to resolve the pathogenic attributes of the variant/s. Lead applicants must be affiliated with an Australian public or private institution that is able to formulate a funding agreement with the AFGN.

**Project Scope:** Project proposals should focus primarily on the generation of a model organism/system to generate data that enables critical assessment of variant pathogenicity and/or establish a novel gene-disease relationship. Funds will not be provided for extended mechanistic investigations through this Stream unless the disease is associated with a novel disease mechanism.

**Selection process:** Applications will be reviewed at the monthly SRC meeting.

Applications will be evaluated against**:**

1. Quality of the experimental strategy
2. Adequacy of experimental controls
3. Ability to assess functional consequence of the variant
4. Appropriateness of the proposed cost/timeline
5. Capability of the investigator/s to deliver the project

Applicants will be notified of the outcome of their submission following the meeting. The SRC may request revision of the proposal or suggest collaboration between applicants. The SRC may choose to fund multiple projects that propose complimentary work.

**Project Funding:** Projects will be funded in “quanta” based on the scope of the project, the number of variants to be modelled and the organism or cell-based system used. Each quantum equates to $15,000 and a maximum of 4 quanta (total of $60,000) can be applied for in any given Project submission. When applying for multiple quanta, the first quantum of funding will be awarded for the generation of the model. Further quantum will be released for variant analysis upon successful model development.

**The AFGN invites expressions of interest from Australian laboratories undertaking functional genomics research to resolve variants in unmatched genes.**

**Please submit your application to** [functional.genomics@mcri.edu.au](mailto:functional.genomics@mcri.edu.au) **within four weeks of receiving your invitation.** You will receive a submission confirmation email.

We encourage researchers to utilise existing capabilities in their project design. **Phenomics Australia** is an NCRIS-funded initiative that offers and expertise in developing animal to assist project development. For more information, contact John Parisot at [j.parisot@therapeuticinnovation.com.au](mailto:j.parisot@therapeuticinnovation.com.au) or visit <https://phenomicsaustralia.org.au/>

**Gene-List:**

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| **GENE** | **DISEASE ASSOCIATION** | **INHERITANCE PATTERN** |
| [*IFT140*](https://redcap.mcri.edu.au/surveys/?s=Y7eMDP7waBVeTWG5&new) | **Skeletal ciliopathy** | **Autosomal recessive** |
| [*SCN4A*](https://redcap.mcri.edu.au/surveys/?s=PbCrWbibtpDUhv2x&new) | Congenital myopathy and inborn errors of metabolism disorder | Autosomal dominant |
| [*SCN2A*](https://redcap.mcri.edu.au/surveys/?s=DFSB9ASWJe5PUvGA&new) | Refactory epilepsy | Autosomal dominant |
| [*CHRNA2*](https://redcap.mcri.edu.au/surveys/?s=5Ft5FtzGbSwiceFp&new) | Focal epilepsy | Autosomal dominant |
| [*NTRK2*](https://redcap.mcri.edu.au/surveys/?s=pj6bnnQaAYpQUatk&new) | Epilepsy syndrome (developmental and epileptic encephalopathy) and obesity, hyperphagia, and developmental delay | Autosomal dominant |
| [*DYNC1H1*](https://redcap.mcri.edu.au/surveys/?s=MUu9x4yXprGEoFGy&new) | Genetic peripheral neuropathy (Charcot-Marie-Tooth disease), intellectual disability and, proximal spinal muscular atrophy | Autosomal dominant |
| [*NPRL3*](https://redcap.mcri.edu.au/surveys/?s=6Sx2f57r6nnhn8Vd&new) | Drug resistant focal epilepsy | Autosomal dominant |

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| APPLICANT DETAILS | |
| Name: |  |
| Institutional affiliation: |  |
| Position: |  |
| Department: |  |
| Email: |  |
| Phone: |  |
| Project Title: |  |

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| GENE NAME: | DATE: |
| PROJECT PROPOSAL (1-page)  Provide relevant background on the research question/s, and the experimental/work plan, including information of the model, experiment controls, the scope of analyses, and anticipated outcome. Describe how the functional consequence of the variant is related to the phenotype. | |
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| SAMPLES | | | |
| Are patient-specific samples required | YES | NO | |
| If yes – what type: |  | | |
| Outline the timeline and process of Human/Animal Research Ethics compliance (where appropriate): |  | | |
| If your application is successful, do you agree to acquire appropriate ethics approval from your institution prior to commencement? | YES | | NO |

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| DELIVERABLES and MILESTONES (1/3 page) |
| Outline the expected outcomes of your experimental strategy and associated milestones. |
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| DELIVERABLE | MILESTONE | DURATION (MONTHS) | DATE (MM/YY) |
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| BUDGET and BUDGET JUSTIFICATION (1/3 page) |
| Enumerate the budget items (salary, reagents, services, etc.) for this proposal.\*  The budget should be structured in quantum of $15K, up to a total of four quanta of $60K. |
| Total requested:  $15,000  $30,000  $45,000  $60,000 |

\* While the itemized budget is required for the assessment of the deliverables of the research, this catalyst grant (a grant-in-aid) is awarded with a one-line budget for personnel support and direct research cost. Please note that overheads are not eligible for acquittal under the funding rule.

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| BIOGRAPHY (1/3 page) |
| Provide a brief summary of evidence of your capability to deliver this project. |
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| REFERENCES (maximum five (5) relevant references) |
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