***Application for a Connection to a Model Organism Researcher***

This application is intended for investigators who wish to link their gene discovery to a model organism researcher. Genes to be considered for such connections will fall into one of the following categories:

1. Novel gene supported by genetic evidence (mutations in the same gene in unrelated patients with a similar phenotype) and functional data is being rapidly sought prior to publication.
2. Candidate gene for a rare disease in a single patient/family/isolated population such that additional functional data is necessary to support disease-causation. Two types of candidate genes will be considered:
   1. Candidate gene has not previously been associated with disease; and,
   2. Novel mutation in known disease gene causing a very distinct disease secondary to a presumed alternate mechanism.
3. Known disease gene that is of therapeutic or biological interest to a unique Canadian population (e.g. Hutterite, First Nation, French Canadian, etc.).
   1. Therapeutic opportunities will include projects that propose to develop a model asset for drug screening or develop a model to evaluate efficacy of a treatment; and,
   2. Biological interest will include those genes published by a Canadian group for which further study has the potential to impact a new area of biology.

Each proposal will be reviewed by 3 members of the Clinical Advisory Committee (CAC) and evaluated based on the following:

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| **Evaluation Criteria** | **Category I** | **Category II** | **Category III** |
| Quality of the genetic data as disease-causing | + | + | NA |
| Disease severity and medical need | NA | + | + |
| Potential therapeutic tractability | NA | + | + |
| Impact on unique Canadian population | NA | + | + |
| Novelty of the implicated biological pathway | NA | + | + |

**Application Procedure:** Applications may be submitted by any investigator associated with a Canadian institution. Requests must not exceed 2 pages (11 pt font size, 0.75 inch margins) using the attached application. **Please submit: Two page application by email to: info@rare-diseases-catalyst-network.ca.** You will receive a submission confirmation email. Decision to proceed to a request for a connection will generally be available within 2 weeks. For further enquires please contact **info@rare-diseases-catalyst-network.ca**.

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| **INVESTIGATOR** | | | |
| **Name** | **Faculty Appointment & Institutional Affiliation** | **Email** | **Telephone** |
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| **SPECIFICATION** | |
| **Disease name:** | |
| **OMIM Number** (*if available for phenotype*): | |
| **Disease description** (*Please include details on the phenotype, such as degree of intellectual disability, for each affected individual. Ideally, attach a pedigree using NSGC nomenclature, specifying who was assessed clinically and at the molecular level)***:** | |
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| **Gene name:**  (*HGNC approved gene symbol*) | **Gene discovery project** (*if applicable*)**:** |
| **Category of Study** (*check one, see instructions*)  1  2a  2b  3a  3b  Unique Canadian population if 3a or 3b: | |
| **Genetic evidence that the gene or variant is disease-causing** *(Please describe variants using HGVS nomenclature at the cDNA and protein level validated using* [*https://mutalyzer.nl/name-checker*](https://mutalyzer.nl/name-checker)*. Provide filtering details if WES/WGS was used, the highest variant frequency from* [*http://exac.broadinstitute.org*](http://exac.broadinstitute.org)*, and prediction programs used. Discuss why other rare variants which could match inheritance were excluded from further consideration. Confirm that Sanger validation and segregation of the variant(s) in the family has occurred. Applications lacking sufficient details will be returned)* | |
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| **Disease severity and medical need** | |
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| **Potential therapeutic tractability** | |
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| **Role of the encoded protein and novelty of the implicated biological pathway if applicable.** | |
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| **Impact on unique Canadian population** *(if applicable)* | |
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| **Please provide information on the type and results of any validation studies performed to date** (*screening of additional families or functional* studies, *if applicable*) | |
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| **Additional details** (*relevant references, known Canadian experts*) | |
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| **REQUEST TO CONSORTIUM** |
| **Connection Requested** (*specify preferred model organism if known*) |
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| **Partner has already been identified** (*provide reasons for selecting partner, details of work done, why preferred model organism will be used)* |
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