Funding Confirmatory Testing of Research Variants through a Clinical Laboratory

APPLICABILITY

This document applies to all healthcare providers who are requesting out-of-province funding for clinical confirmation of research variants through the Genetic Resource Centre (GRC).

Out-of-province testing will not be funded if the gene of interest can be tested through the Molecular Genetics Laboratory in Calgary or Edmonton. In these cases, in-house targeted testing should be requested to clinically confirm the research variant. The conditions outlined below also apply to in-house testing.

PURPOSE

This guideline provides information regarding when the GRC will fund targeted testing of research variants through an accredited clinical laboratory.

A number of Alberta patients participate in genetic research studies to help inform their medical management. When variants are identified through a research study, it is standard practice to perform targeted testing to confirm variants through an accredited clinical laboratory. Once a genetic test result is clinically confirmed, it can be included in the patient's medical record and can be incorporated into the patient's treatment plan.

GUIDELINE

The GRC is only able to fund clinical confirmation of research variants when ALL of the following conditions are met:

1) Using the ACMG variant interpretation guidelines[1], the variant is classified as pathogenic or likely pathogenic.
2) The variant position is known and documented using appropriate HGVS nomenclature (transcript must be provided).
3) Either:
   A) The read depth at the variant position is at least 25 and the variant allele frequency is at least 30%
   OR
   B) Sanger sequencing in the research setting confirmed the presence of the variant.
4) Clinically confirming the research variant will impact the patient's management, at-risk relatives, and/or family planning.

RESPONSIBILITY

Ordering healthcare providers and the GRC personnel are responsible for implementing this guideline.

REFERENCES

[1]Richards, S., Aziz, N., Bale, S., Bick, D., Das, S., Gastier-Foster, J., & Voelkerding, K. (2015). Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genetics in medicine*, *17*(5), 405.