

Quickly

Uncovering

Important

Clinical

Knowledge



# ANALYSIS

QUICK Analysis is Greenwood's *free* NGS-reflex analysis that rapidly screens the full exome data for pathogenic variants when panel results are negative.



### What is the QUICK Analysis?

With the **QUICK Analysis**, GGC screens whole exome data for pathogenic variants when panel results are normal.

### How is it done?

All NGS testing at GGC is run on an exome backbone, so whole exome sequence is available for every patient who has a panel. We have designed a stringent filtering system to "QUICKly" sort and review variants that are highly likely to explain the patient's phenotype. Only pathogenic or likely pathogenic alterations will be reported. Secondary findings genes are excluded from this analysis unless indicated by the patient's phenotype.

### What is the benefit of the QUICK Analysis?

The **QUICK Analysis** increases the yield of cost-effective gene panel tests. By screening variants based on molecular criteria first and reviewing the phenotype fit second, QUICK is good at picking up new or poorly characterized genes. While the **QUICK Analysis** is not as thorough as a whole exome analysis, we have found it effective in expediting a diagnosis for a growing number of patients. Depending on the patient's clinical features and the initial testing, we have identified the causative variant in 10-50% of cases analyzed by **QUICK Analysis**.

### What can I expect to get in the results?

Variants reviewed during the **QUICK Analysis** must be rare in the general population and meet our quality thresholds regarding depth of sequence coverage and allele frequency. Variants must fall into one of the following categories:

- Previously reported in the Human Gene Mutation Database (HGMD)
- Causes a frame shift, stop gain, start loss or stop loss effect on the encoded protein
- Expected to affect a splice site at the +/- 1 or 2 position
- Compound heterozygous variant in a gene associated with an autosomal recessive disorder

*A complete secondary findings analysis is not included as part of the QUICK Analysis; however, a variant, in one of these genes may be reported if it is a good match for the patient's phenotype.*