GWS Reanalysis Policy
Division of Genome Diagnostics

The Division of Genome Diagnostics at BC Children's and BC Women's Hospitals (C&W) provides clinical genetic testing for the province of British Columbia, including a rare disease genome-wide sequencing (GWS) clinical pilot. This policy outlines the criteria and process for reanalysis of previously generated GWS data.

Eligibility:

Reanalysis of GWS data may be requested 2 years after the original report following updated consent and clinical review of the patient and must include current clinical and phenotypic information relevant to genomic interpretation.

Reanalysis requested less than 2 years after the initial report will generally be considered only when there has been a significant change in patient's clinical presentation or a compelling, time-sensitive indication for establishing a molecular diagnosis. Such requests must demonstrate substantial additional clinical utility beyond the current management plan and a reasonable likelihood of influencing near-term medical, reproductive, prognostic, or other healthcare decisions. Potential future utility alone is not normally sufficient justification for early reanalysis.

Analysis Scope:

During the initial GWS analysis, validated variants (e.g., SNVs, small insertions/deletions, etc.) are identified across the entire genomic sequence, without restriction. Filtering and prioritization identify variants associated with the clinical phenotype of the patient based on the gene-disease knowledge at the time of analysis.

Currently, reanalysis involves re-filtering and prioritization of the initial identified variants, using current clinical, phenotypic, and gene-disease information. Only the validated variant types identified in the original analysis are assessed.

In some circumstances, reanalysis may not be possible due to limitations of the original data, software, reference files, or validated pipelines.

Turnaround Time:

Routine reanalysis: 12 weeks.

STAT reanalysis is not available.

Expedited reanalysis: 6 weeks.

- Requests involving compelling, time-sensitive clinical circumstances may be considered for expedited review following consultation with the laboratory; however, expedited turnaround times cannot be guaranteed.