Genes 2Me Whole Exome Sequencing
Panel Comparison data:

Whole exome sequencing (WES) has emerged as a transformative tool in the field of genomics, enabling comprehensive analysis of the protein-coding regions of the genome, or exome. This subset, comprising approximately 1-2% of the human genome, harbors the majority of known disease-related variants. With the advent of next-generation sequencing (NGS) technologies, WES offers a high-throughput, cost-effective approach to uncovering genetic variations associated with a wide range of diseases, including rare disorders and complex traits.

The utility of WES extends beyond clinical diagnostics; it facilitates insights into the underlying genetic mechanisms of diseases, guiding the development of targeted therapies and personalized medicine. Despite its potential, the implementation of WES presents several technical challenges, including sample preparation, library construction, sequencing accuracy, and bioinformatics analysis. These factors significantly influence the quality and interpretability of sequencing data, necessitating robust
methodologies and stringent quality control measures.

In this study, we aim to optimize a whole exome sequencing panel by addressing these challenges through innovative approaches in sample processing, sequencing technology selection, and data analysis. By evaluating the performance of our optimized WES panel in diverse clinical cohorts, we seek to demonstrate its efficacy in identifying pathogenic variants and enhancing our understanding of genetic disorders. Our findings will contribute to the ongoing efforts to refine WES methodologies and expand its application in both research and clinical settings.

Comparison of G2M Whole Exome Sequencing Panel with two of the commercially available Whole Exome panels manufactured by widely used companies.

G2M WES Panel uses hybridization-capture based workflow selection of the exonic region from the whole genome. Unlike overnight hybridization integrated in the majority of commercially available panels, G2M achieved hybridization within 4 hours reducing the turnaround time and enhancing rapid results.

A study was performed to compare performance of G2M WES panel with that of some of the commercially available, widely used panels. Libraries were prepared using Coriell NA12878 genomic DNA and then sequenced on Avity platform by Element Biosciences. The commercial panel used were labeled as “Company S’ and “Company T”

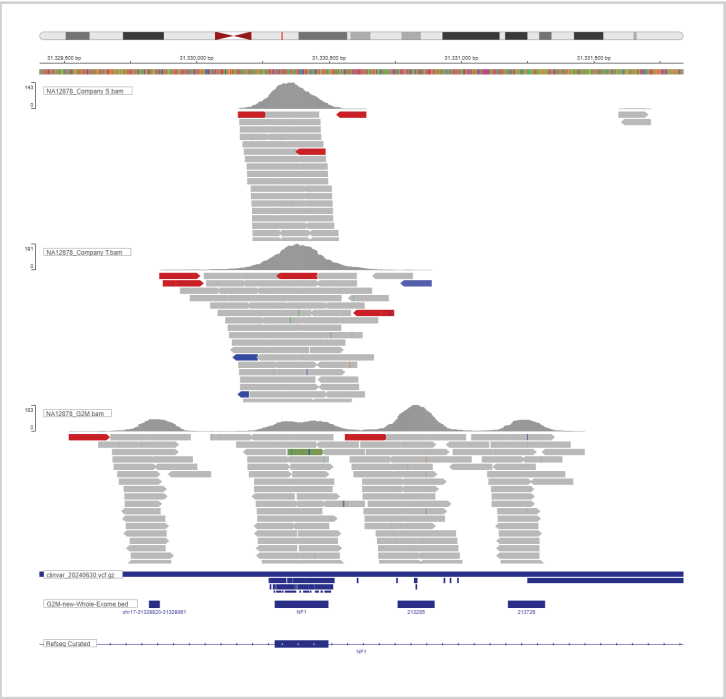
Better coverage for multiple genes:

When the alignment files generated from the sequencer were visualized on IGV, the G2M WES panel showed more coverage of target regions as compared to the competitor panels. Following figures represent performance of the G2M panel against the other commercially available panels.

1: NF-1 gene:

The NF1 gene, located on chromosome 17, encodes the protein neurofibromin, which acts as a tumor suppressor. Mutations in the NF1 gene can lead to neurofibromatosis type 1 (NF1), a genetic disorder characterized by the development of multiple benign tumors called neurofibromas, as well as skin changes, learning disabilities, and an increased risk of certain cancers.

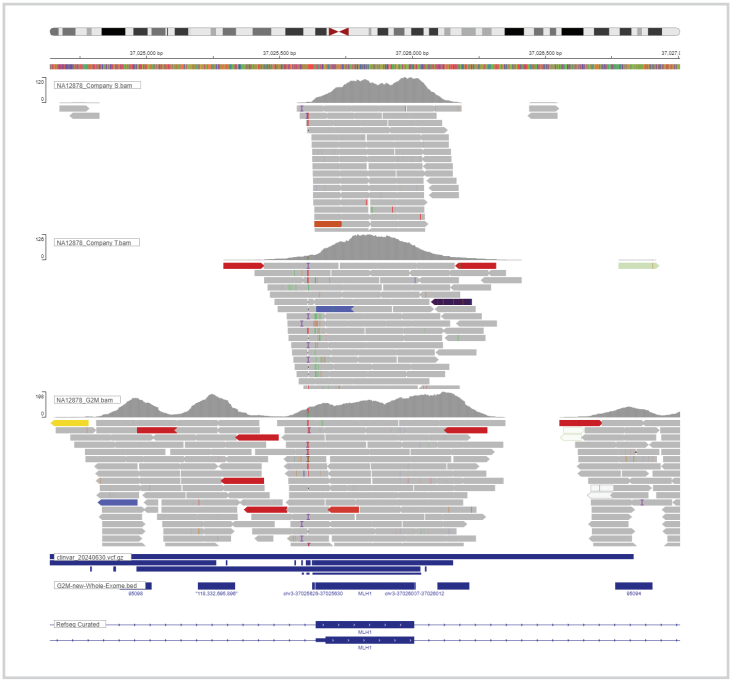
The following image clearly indicates a higher coverage of target regions using G2M WES panels as compared to those of Company S and Company T. The region which is targeted by G2M probes indicates a pathogenic allele of Nf-1 gene. This allele was not covered by the other two panels.



2: MLH-1 gene:

The MLH1 gene is a crucial component of the DNA mismatch repair (MMR) system, which helps maintain genomic stability by correcting errors that occur during DNA replication. Located on chromosome 3, mutations in the MLH1 gene can lead to a higher risk of certain cancers, particularly colorectal and endometrial cancers, often associated with Lynch syndrome (hereditary non-polyposis colorectal cancer).

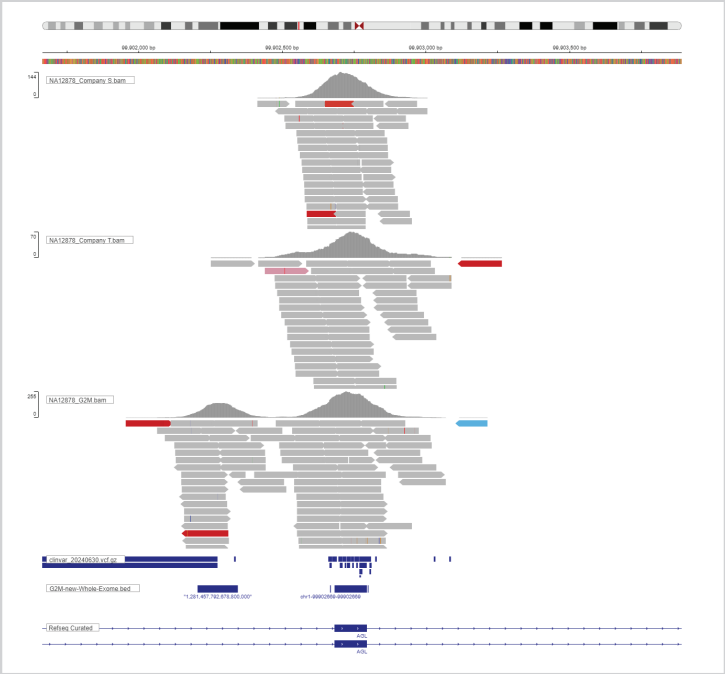
A higher coverage of MLH-1 gene in G2M WES generated data can be observed using the following image. The region targeted by G2M probes represent pathogenic allele from MLH-1 gene associated with Lynch Syndrome.



3: AGL gene:

The AGL gene encodes the enzyme amylo-1,6-glucosidase, which is involved in carbohydrate metabolism. This enzyme plays a key role in breaking down glycogen, the stored form of glucose, into usable sugar molecules. Mutations in the AGL gene can lead to glycogen storage diseases, particularly glycogen storage disease type III (Cori disease), which is characterized by the accumulation of abnormal glycogen in tissues.

A better coverage of AGL gene in G2M WES generated data can be observed using the following image. The region targeted by G2M probes represent pathogenic allele from AGL gene associated with Glycogen storage disease.



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