WHOLE EXOME SEQUENCING

Library Preparation Assays

CE IVD

G2M Whole Exome Sequencing Library Preparation Assay

Exons represent just 1–2% of the genome, they harbour up to 85% of disease-causing mutations, making WES a highly effective and efficient approach for identifying genetic variants associated with disease. It enables high-throughput detection of single nucleotide variants (SNVs), insertions/deletions (in-dels), and other coding-region alterations that underlie a wide range of genetic disorders. For many patients—especially those with rare or complex genetic conditions—the journey to a diagnosis can be long, expensive, and emotionally exhausting.

Conventional exome sequencing panels often require a trade-off between comprehensive genomic coverage and assay performance, leading to uneven coverage, higher duplication rates, ultimately impacting the sensitivity and reliability of variant detection.

Developed with a deep understanding of both clinical and research needs, G2M's Whole exome sequencing assay is designed with exceptional uniformity and high on-target efficiency with the panel content aligned with the latest curated genomic data for enhanced clinical relevance. The panel encompasses ~21,500 genes catering to various hereditary conditions and germline cancers.

Key Features

Comprehensive Genomic Content
- Covers >99% of clinically relevant protein-coding regions at >20X
- Includes updated gene content based on RefSeq, GENCODE, OMIM, ClinVar & HGMD

Exceptional Coverage Uniformity
- Minimizes dropouts in GC-rich or hard-to-capture exons
- Enables confident detection of heterozygous and low-frequency variants

High On-Target Rate
- Maximized hybridization efficiency ensures a high percentage of reads align to intended targets
- Reduces off-target noise, lowering sequencing waste and cost

Clinical-Grade Performance
- Validated for SNVs, indels, and CNV calling
- Applications in rare disease, oncology, reproductive health, and inherited disorders

Flexible and Scalable Workflow
- Compatible with major sequencing platforms such as Illumina, MGI, Element Biosciences (AVITI)

Key Application Areas

- Rare Genetic Disease Diagnosis
- Hereditary Cancer Risk Assessment
- Neuro-genetic Disorders
- Peadiatric and Neonatal Genetics
- Cardio-genetics
- Undiagnosed or Atypical Presentation
- Pharmacogenomics & Precision Medicine
- Carrier Screening & Reproductive Planning

Performance Specifications

Metric	Performance
On-Target Rate	>85%
Coverage Uniformity (≥ 0.2× mean)	>95% of targets
Mean Coverage at 100× Sequencing	>98% of targets covered at ≥20×
Duplication Rate	<10%
Fold 80 Base Penalty	1.25
Mapped Reads	>99%
Sensitivity for SNVs	>99.5%
Sensitivity for InDels	>98%
GC Bias Performance	Optimized for high- and low-GC regions
Panel Update Cycle	Biannually (aligned with RefSeq, ClinVar updates)
Validated platforms	Illumina, MGI, Element Biosciences (AVITI)

Quality Metrics

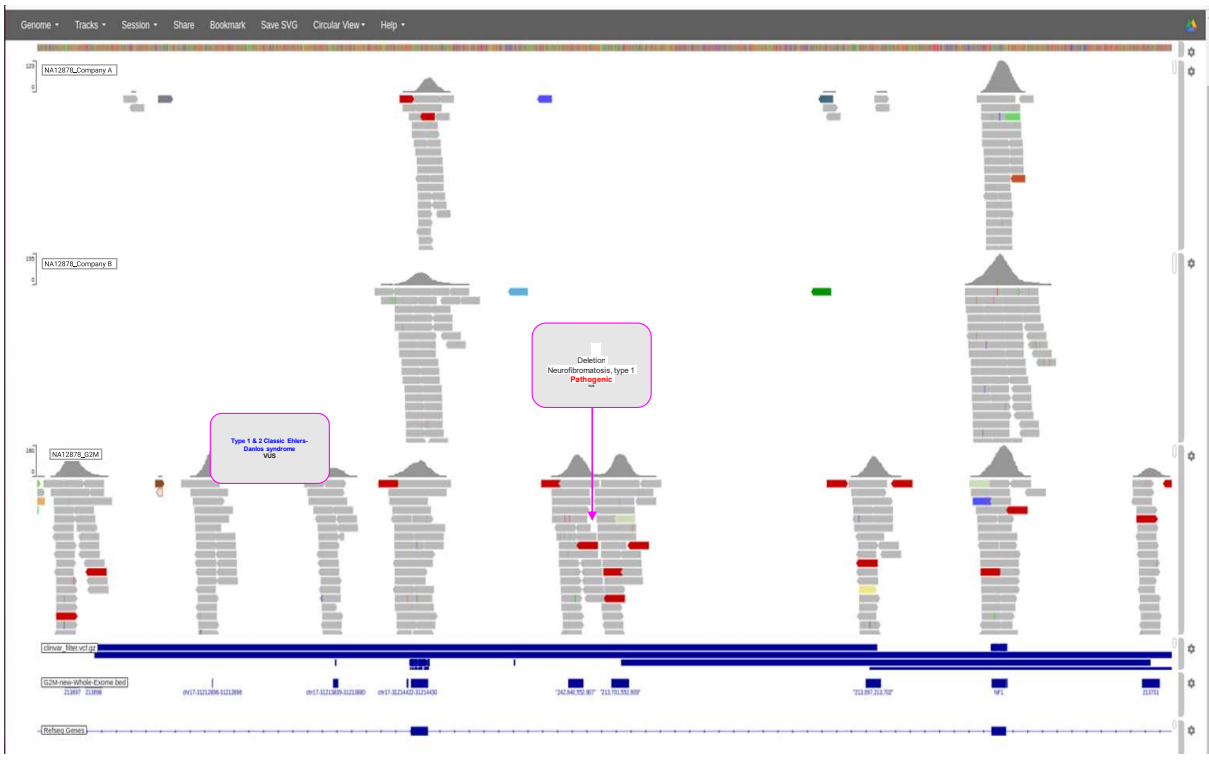


Fig 01: NF1 Gene Coverage Sequencing for Precise Variant Detection : High-depth sequencing of the NF1 gene demonstrates exceptional coverage, enabling precise detection of both pathogenic variants and variants of uncertain significance (VUS), including clinically relevant deletions. This comprehensive analysis supports accurate diagnosis of Neurofibromatosis Type 1 and related syndromes such as Type 1 & 2 Classic Ehlers-Danlos syndrome.

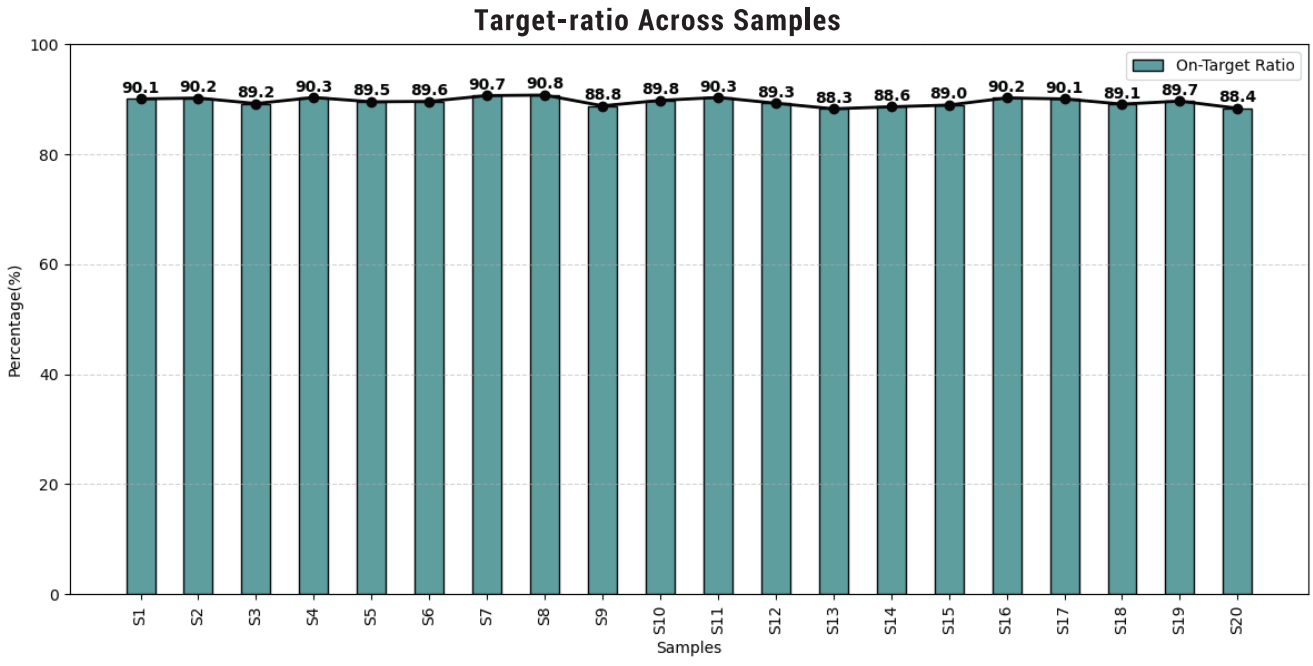


Fig 02: Comprehensive Gene Coverage Across Key Databases : The chart illustrates the high percentage of genes mapped to major clinical and phenotype databases - OMIM, ClinVar, and HPO ensuring robust integration of genetic information. Nearly 100% coverage in OMIM and HPO, along with over 90% in ClinVar, highlights the robustness of these databases for accurate gene interpretation and evidence based clinical insights.

Post-Sequencing Data Quality

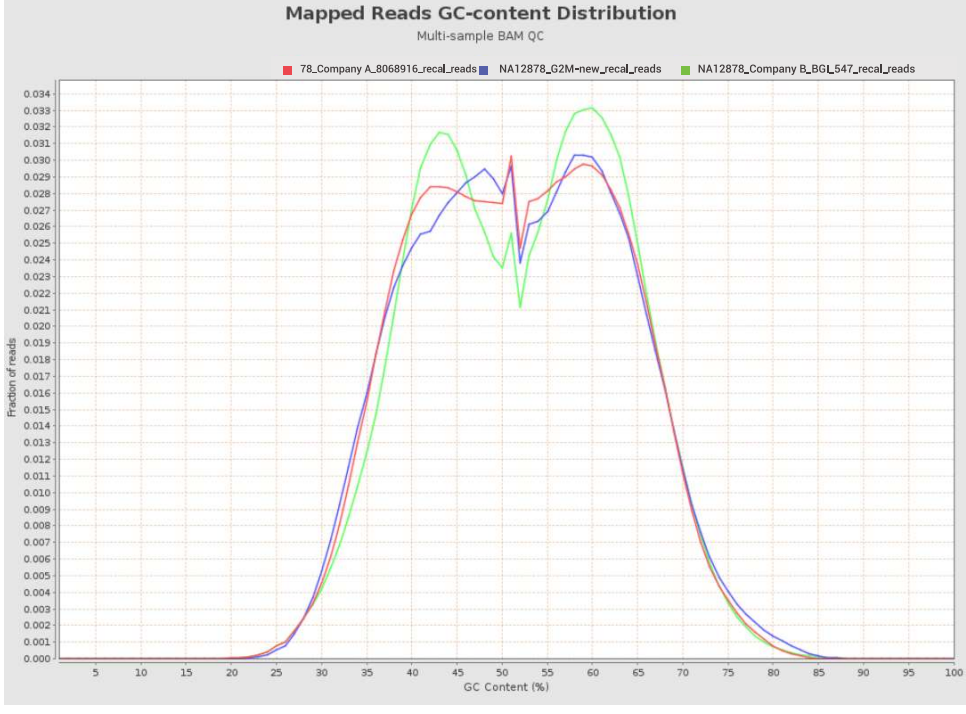


Fig 03: G2M Delivers Superior GC Coverage Uniformity Across Leading Platforms : GC content distribution across target regions for Company A (red), G2M (blue), and Company B (green) platforms reveals superior uniformity in G2M's coverage across varying GC percentages. This indicates reduced GC bias and more consistent hybridization efficiency, supporting reliable variant detection across challenging genomic landscapes.

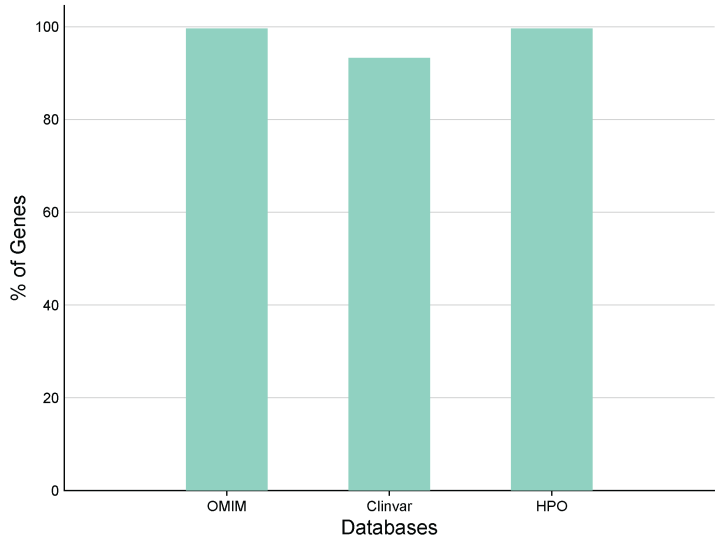


Fig 04: Comprehensive Gene Coverage Across Key Databases : The chart illustrates the high percentage of genes mapped to major clinical and phenotype databases - OMIM, ClinVar, and HPO ensuring robust integration of genetic information. Nearly 100% coverage in OMIM and HPO, along with over 90% in ClinVar, highlights the robustness of these databases for accurate gene interpretation and evidence based clinical insights.

Multi-Metric Comparison of Exome Performance

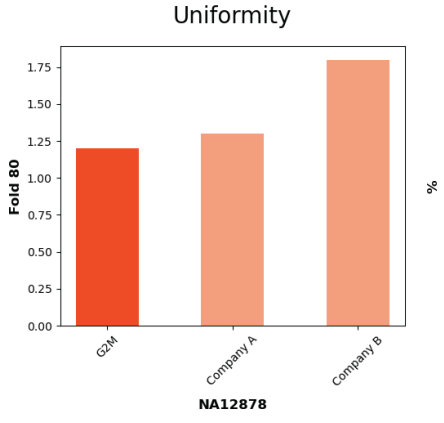


Fig 05: Exceptional Coverage Uniformity Validated by Low Fold 80 Metric : Fold 80 base penalty is the fold-over-mean coverage required to ensure that 80% of targeted bases are covered at or above the mean coverage depth. It is a key metric used to evaluate the uniformity of coverage in next-generation sequencing (NGS). A lower value indicates more uniform coverage—which means less over-sequencing is needed to achieve complete and reliable results. G2M shows <1.25 Fold 80 penalty value implying good capture design and hybridization efficiency than the competition.

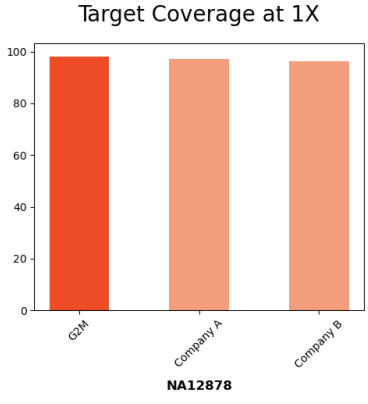
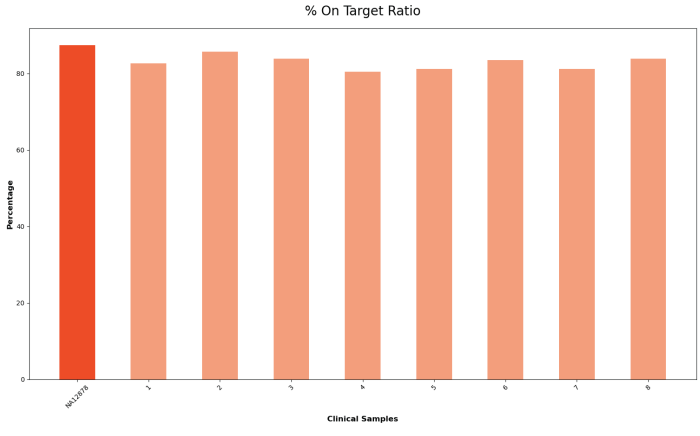


Fig 06: Efficient Whole Exome Sequencing with Ultra-Low Depth Coverage : G2M WES assay shows >98% coverage at a very low depth (1X)

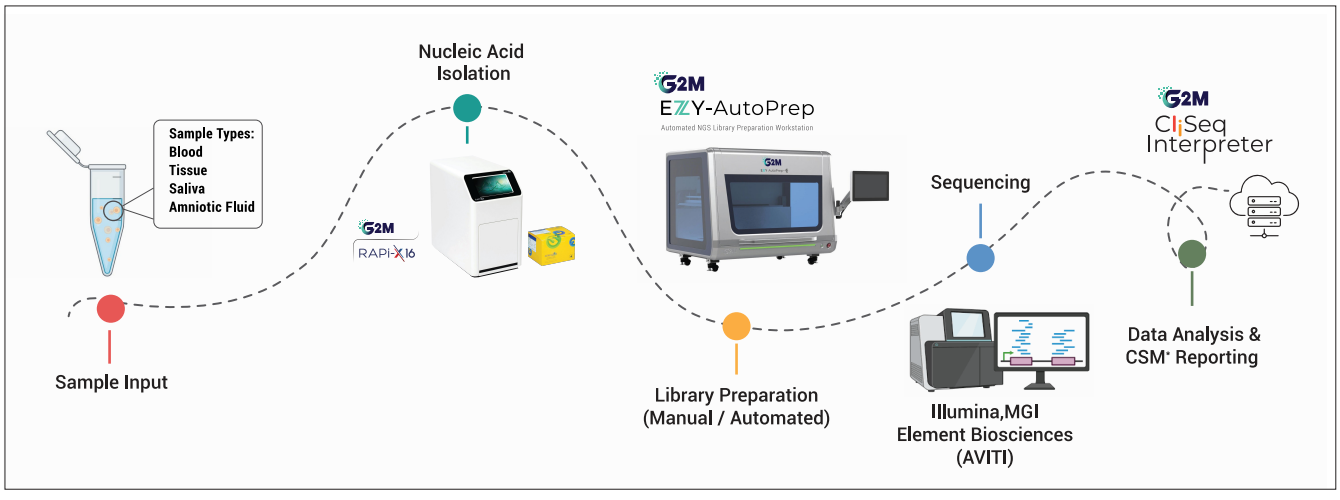
Fig 07: High On-Target Alignment Across Different Cancer Patient Samples : G2M WES assay shows an average on target ratio of 85% for both clinical and control samples (NA12878)



Panel Specifications

No of genes	~21,500
Inclusions	SNVs, Indels, CNVs, Mitochondrial genome
Target size	38.2 Mb
Probe region	49.8 Mb
Recommended coverage	150-180X
Data required (per sample)	6 GB
Pre-capture pooling	8
Methodology	Hybridization capture based target enrichment
Recommended sequencing mode	Paired-end 150 (PE 150)
Sample types	Blood, Saliva, Amniotic fluid, Chorionic Villus, Tissue
Starting sample input	50-500 ng Genomic DNA
Average library insert size	~300 bp

End-to-End G2M Workflow

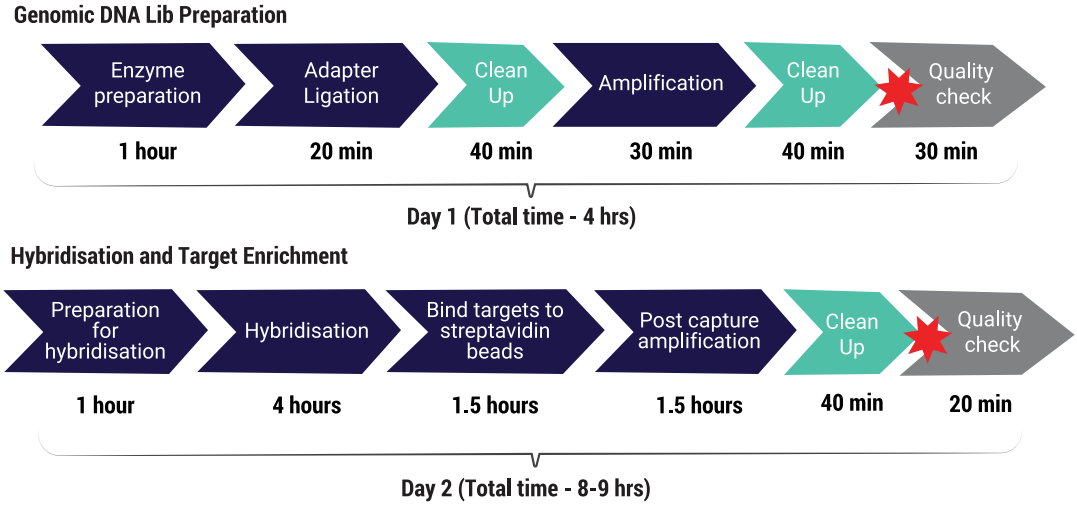


* CSM = Clinically Significant Mutations

G2M WES Assay Library Construction Workflow

The library preparation is based on Hybridization capture based target enrichment

Our Whole Exome Sequencing (WES) panels utilize a robust hybridization capture-based target enrichment approach to ensure high specificity and uniform coverage across the exome. This workflow enables the selective enrichment of coding regions from genomic DNA, allowing for comprehensive detection of single nucleotide variants (SNVs), insertions/deletions (indels), and other clinically relevant mutations. The method combines proven biochemical precision with optimized probe design, providing a reliable foundation for accurate and high-throughput sequencing results.



Data analysis & Reporting solution

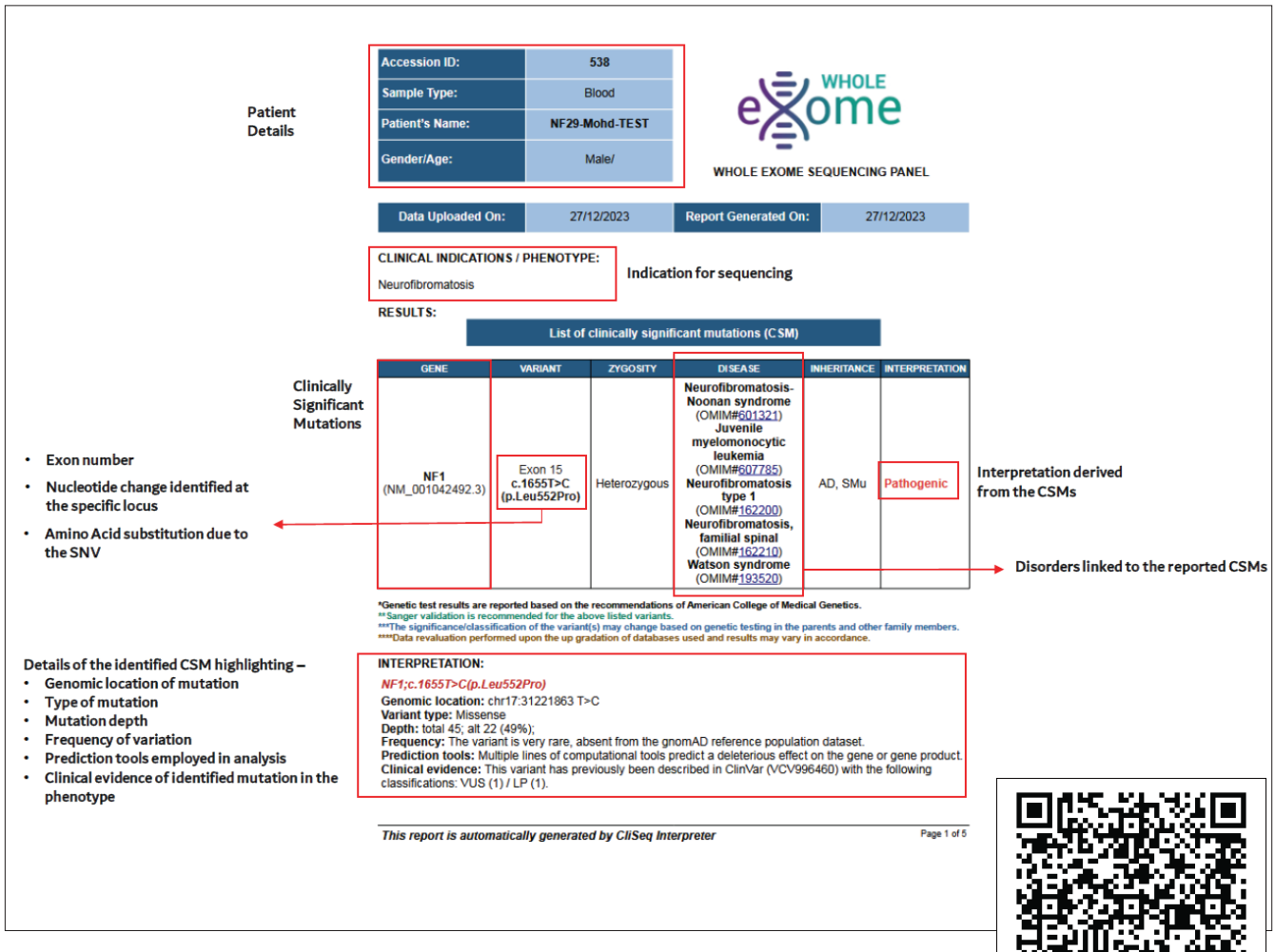
CliSeq Interpreter

Interpret and report relevant variants with Cliseq Interpreter Platform

The NGS data analysis is supported by combining guideline recommended variants with the analytical capability of G2M's Cliseq Interpeter Platform. Cliseq Interpreter is a cloud based NGS data analysis software which offers an unparallel platform performance designed to streamline and enhance the interpretation of complex biological data. Once Quality Check, Alignment, Variant calling, and annotations are achieved, the annotated VCF files and clinically significat mutations (CSM) report will be available to download.

*As per allele frequency & phenotypic indicator.

CSM Report



Scan for WES Sample Report

Automate your
NGS Libraries *with*

EZY-AutoPrep-48

Construct upto 48 sample libraries in one run

Ordering Information

Commercial Name	Cat No.
Clinical Exome Sequencing Expanded Panel (Whole Exome Sequencing)	G2MCES07001(WES)-ill; G2MCES07001(WES)-MG, G2MCES07001(WES)-TF

CE IVD | ISO 13485:2016 | ISO 9001:2015 | GMP Quality

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