Whole Exome Sequencing (WES)

NGS Panel

NGS Clinical Panels

The Genes2Me Whole Exome Sequencing (WES) Expanded NGS panel is a hybridization based solution for screening ~21500 clinically relevant genes (coding regions of the genome) for diseases associated with genetic mutations and mitochondrial genome. It covers all major germline mutations like SNV, CNV, and Indels with hotspots adding up to a target size of 38.2 Mb.

Comprehensive Panel: Provides uniform and deep coverage of exome

Low Input: Process compatible with low input quality compromised samples

Robust and Rapid Workflow: Hybridization enhancer technology and enzyme based library preparation enables quick turn around time.

CliSeq Interpreter: User friendly companion software for automated & cloud based analysis and reporting.

Whole Exome Sequencing (WES)

NGS Panel

Exome is a subset of the genome that covers sequences of all the exons, reflecting the protein-coding region of the genome. In humans, the exome is about 1% of the genome. Whole Exome Sequencing is a comprehensive DNA test to identify disease causing variants within the genpme. Advances in next-generation sequencing technologies have decreased the cost of sequencing per base pair about 10-fold, improved accuracy, and greatly increased the speed of generating sequence data. This improved accuracy has enabled development of WES at a faster and cheaper rate of variant identification. It is rapidly becoming a common molecular diagnostic test for individuals with genetic disorders.

No. of Genes	~21500
Covered region	Whole CDS, Hotspots, Mitochondrial Genome
Target size	38.2 Mb
Mutation type	SNVs/InDels/CNVs
Sample type	Blood/ZF/Tissue/CVS
Type of cancer	Germline & Somatic

The Genes 2Me OncoCheck Panel screens a range of cancer causing genes to identify mutations in DNA from FFPE and blood tissue. It provides comprehensive detail of the cancer and helps to decide the best course of treatment.

Specifications

- More than 80% of bases with ≥ Q30 quality score
- Recommended sequencing depth for Mendelian disorder/rare disease: ≥ 80-100x

Starting Material (DNA)	Library preparation time	Bioinformatics analysis	Databases used for Annotation
50-500 ng	1.5 days (including Target Capture & Enrichment) for manual process With **G2M Auto EzyPrep automated NGS Library preparation system:** Minimum Hands-on required	Within 24hrs (Raw data to CSM report)	ICGC, FusionDB, OncoDB, ClinVar, gnomAD, 1000Genome, dbSNP

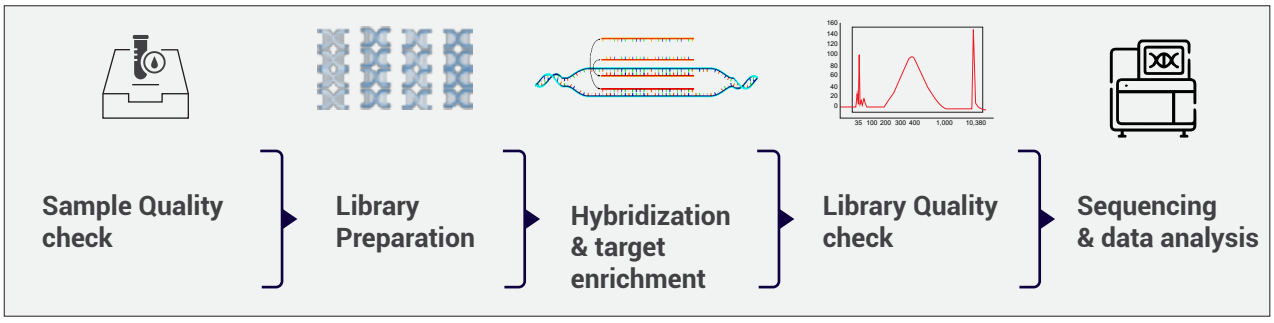
Whole Exome Panel Applications

- Biomarker discovery
- Drug target discovery
- Rare mutations discovery
- Low frequency mutations detection

Process Workflow

A. Platform Agnostic

Sequencing on multiple platforms (Thermo Ion-Torrent, Illumina, MGI and Element Biosciences)



B. Bioinformatics Solutions

Data Analysis and Interpretation using Genes 2Me Cliseq Interpreter software

RAW DATA (FASTQ) → **QUALITY CHECK** TRIMMING QC METRICES → **ALIGNMENT** (BAM) → **VARIANT CALLING** (VCF) → **GERMLINE DATABASE** → **ANNOTATION** (VCF) → **FILTER** As per Allele frequency & Phenotypic information → **CSM** (Clinical Signification Mutations) **REPORT**

Panel Performance

Features	Illumina	MGI	Thermo Fisher
Coverage uniformity	96%	96%	87%
Precision	94%	94%	87%
Reproducibility	97%	97%	93%
Sensitivity	94%	94%	87%
On Target Ratio	85-95 %	85-95%	80-85%

Performance Plots

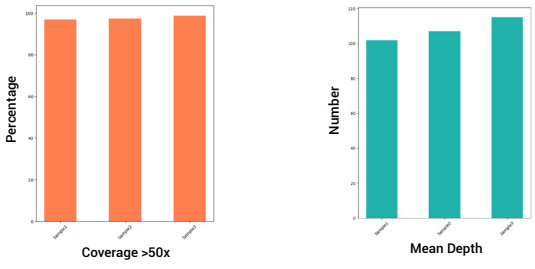


Figure 1: Mean Depth and Coverage (>50X) plot

Important Diseases Covered in Panel

Disease Class	List of Diseases
Cardiac disorders	Dyslipidemia, Aortopathy, Congenital heart defect, cardiovascular diseases, Long QT syndrome, Short QT syndrome, Brugada syndrome, Dolichoectasia,Hereditary hemorrhagic telangiectasia Xeroderma pigmentosum, Ichthyosis
Dermatological disorders	Ectodermal dysplasia, Albinism, Xeroderma pigmentosum, Ichthyosis
Endocrinological disorders	Pancreatitis, Premature ovarian failure, Adrenal hyperplasia, Hyperparathyroidism
Bone disorders	Arthrogryposis, Osteopetrosis, Cleft lip palate, Amelogenesis, Abnormal mineralization, High bone density disorders, imperfecta, Low bone density disorders
Immunological disorders	Immune dysregulation, Defects in intrinsic and innate immunity
Hepatological disorders	Polycystic liver disease, Cholestasis, Congenital hepatic fibrosis
Hematological disorders	Bleeding & Thrombotic disorder, Bone marrow failure, Anemia, Hereditary spherocytosis, Sitosterolemia
Metabolic disorders	Aminoacidopathies, Purine/Pyrimidine disorders, Creatine biosynthesis disorders
Eye disorders	Achromatopsia, Albinism, Bardet-Biedl syndrome, Cone-rod and cone dystrophy, Glaucoma, Hermansky-Pudlak syndrome, Microphthalmia/anophthalmia/coloboma spectrum, Oculomotor apraxia, Retinitis pigmentosa AD/AR, Vitreoretinopathy
Neurological disorders	Neuromuscular disorders, Autism, Seizures and Brain abnormalities, Neurodegenerative disorders syndrome, Kallmann syndrome, Leber congenital amaurosis, Meckel syndrome, Nephronophthisis
Oncological disorders	Hematological malignancy, Brain cancer, Colorectal cancer, Breast cancer, Ovarian cancer
Respiratory disorders	Bronchiectasis, Cystic fibrosis, Primary ciliary dyskinesia
Nephrological disorders	Alport syndrome, Bardet-Biedl syndrome, Bartter syndrome, Focal segmental glomerulosclerosis, Hypogonadotropic hypogonadism, Joubert
Connective tissue disorders	Ehlers-Danlos syndrome, Marfan syndrome, Ehlers-Danlos syndrome, Loeys-Dietz syndrome
Mitochondrial disorders	NARP, Chronic progressive external ophthalmoplegia, Neonatal mitochondrial hepatopathies, Mitochondrial encephalomyopathy, Kearns-Sayre syndrome, Leigh's syndrome, Myogastrointestinal encephalomyopathy

References

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- JAMA. 2014 Nov 12; 312(18): 1880–1887.

Ordering Details

Commercial Name	Cat No.	Pack Size
Whole Exome Sequencing (WES) Panel	G2MCES07001(WES)-ill	96T
	G2MCES07001(WES)-MG	96T
	G2MCES07001(WES)-TF	96T

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