Targeted NGS Panel or Exome?

Focus on Clinical Exome Sequencing!

G-Mendeliome CES Panel

Utilizing one broad exome panel has raised many issues of poor coverage

Most whole exome sequencing panels often cover less of the whole exome than required, with data indicating that 50 percent of exons have lower than 30x average coverage.
Due to the low-coverage regions, WES can miss critically important variant regions.

Variant calling and clinical interpretation still requires extensive expertise and time

Clinical exome sequencing data analysis still demands both technical expertise and significant time expenditure. Clinical interpretation of various detected variants is very challenging as it involves a comprehensive understanding of variants, drugs, guidelines and etc.

"Streamline both targeted NGS panel and WES panel use and simplify your germline mutation profiling test with **G-Mendeliome Clinical Exome Sequencing panel**, consistently yielding excellent coverage over the desired target regions"

Diagnose rare disease with CES/DES panel

G-Mendeliome Clinical Exome Sequencing Panel, CES (called Diagnostics Exome Sequencing, DES panel at GC Genome) was developed from the needs of **GC Genome** (https://www.gc-genome.com), the largest clinical NGS service provider in Korea.

CES solved the problem of poor diagnostics resulting from the large number of genes omitted from the inherited disease panels of Companies A and B (Figure 1) and reduced costs by removing areas from both panels irrelevant to diagnosis.

GC Genome previously used the panels of both Company A and B, but have since replaced their workflow using the G-Mendeliome CES Panel.

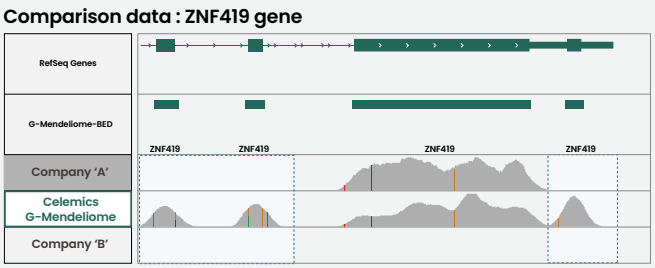
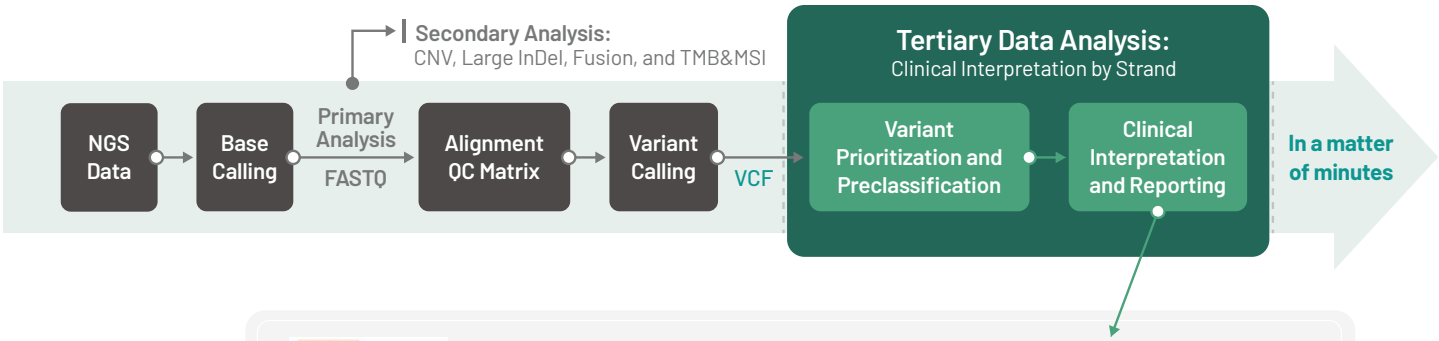


Figure 1. Clinically significant regions of interest absent from competitor's products

Figure 2. Customer Testimonial of GC Genome

Celemics Analysis Service (CAS) Workflow



Celemics' CES Panel is bundled with a free-of-charge CAS Secondary Report and powerful, cost-effective Clinical Report, both optimized for your real-life clinical requirements.

KEY FEATURES

1. Comprehensive genomics profiling of a variety of genetic diseases	Includes 7,513 genes associated with clinically significant genetic diseases
2. A wide range of target regions	Includes all clinically significant regions that are not covered by competing panels
3. Cost-effective analysis	Able to provide accurate analysis with lower sequencing cost compared to that of WES via CAS

SPECIFICATIONS

1. Gene count	7,513 genes
2. Covered region	CDS, intronic hotspots, mitochondrial genome
3. Target size	19.6 Mb
4. Multiplexing	4-plex enrichment
5. Sample type	50 ng of gDNA
6. Platform	All sequencers of illumina
7. Bioinformatics pipeline	Primary, secondary, and tertiary analysis via CAS (Fastq to annotated VCF) Clinical Report via Strand (VCF to clinical report)

PUBLICATION

MDPI diagnostics

Incidental Severe Fatty Degeneration of the Erector Spinae in a Patient with L5–S1 Disc Extrusion Diagnosed with Limb-Girdle Muscular Dystrophy R2 Dysferin-Related

Kim et al, Diagnostics 2020, 10, 530; doi:10.3390/diagnostics10080530

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