

Celemics Clinical Exome Sequencing Panel

Focused On Sequencing of
Clinically Important Genes
Which Associated With
Various Genetic Diseases

Key Features

- Comprehensive genome profiling of a variety of genetic diseases
- Cost-effectiveness by compact target size compared to WES

Celemics Clinical Exome Sequencing

Celemics' G-Mendeliome Clinical Exome Sequencing (CES) Panel is designed to overcome the limitations of analyzing clinical diseases with conventional whole-exome sequencing. The CES panels aim to only include clinically significant regions specifically related to inherited diseases.

Targeted sequencing of clinically important genes is a valuable approach for diagnosing and managing a wide range of genetic diseases, including inherited conditions, cancer predisposition syndromes, and other genetic disorders. It allows for efficient and focused analysis, increasing the likelihood of finding disease-causing variants and providing patients and healthcare providers with actionable information for medical decision-making.



Specification

Panel	Clinical Exome Sequencing - Standard	Clinical Exome Sequencing – Expanded
Gene Count	5,516	7,563
Covered region	CDS, Hotspots, Mitochondrial genome	
Target size	13.8 Mb	19.6 Mb
Mutation type	SNV, Indel, CNV	
Sample type (Amount)	Blood (>50 ng of fragmented DNA)	
Platform	All sequencers from Illumina, Thermo Fisher, and MGI	
Bioinformatics Support	<ol style="list-style-type: none"> 1. Primary Analysis: FASTQ to annotated VCF 2. Secondary Analysis: CNV, Large Indel 3. Tertiary Analysis: Clinical interpretation, Visualization Curation 	

*Gene Add-On Service: Genes can be added by customer's request.

G-Mendeliome Clinical Exome Sequencing (CES) Panel

Comprehensive Genomic Profiling of a Variety of Genetic Diseases

CES panel includes wide range of 7,000 genes associated with clinically significant genetic diseases, even including all clinically significant regions that are not covered by competitors panels.

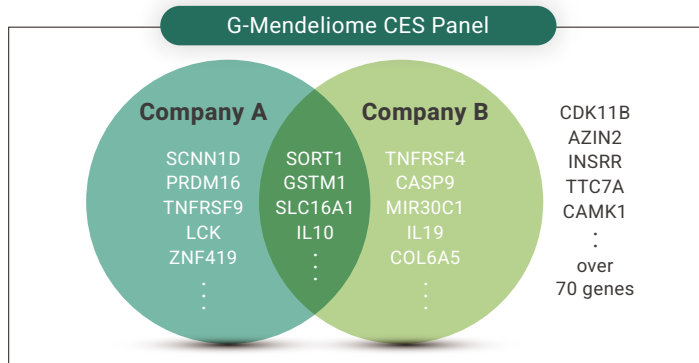


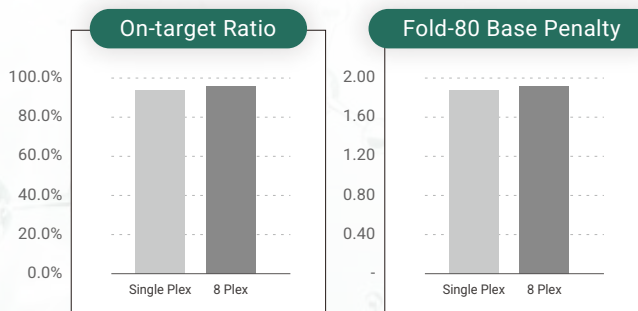
Figure 1. G-Mendeliome CES Panel. The G-Mendeliome CES panel, developed in collaboration with GC Genome, the largest clinical NGS service provider in South Korea, to solve the problems of poor diagnostic rates and high costs. This panel includes a wide range of genes, including those relevant to clinical testing and of significant medical importance. It encompasses many genes that clinicians find interesting and valuable in medical practice.

Comparison data for Hard-to-Capture region between Celemics G-Mendeliome and competitor panels



- Through the in-house manufacturing and probe design system of Celemics, customers can request the sequencing of homolog regions as well as hare-to capture regions
- The comparison data showed obtained sequencing reads for ZNF419 and COL6A5 genes, which are commonly masked in other panels.

High Effective Sequencing for Clinical-associated Genes



Sequencing Platform (Mean depth: 100X)	NextSeq 500/550 High Output v2.5	NovaSeq S4	MGI PE150-FCL
Clinical Exome	30 Samples	750 Samples	135 Samples
Whole Exome	17 Samples	428 Samples	77 Samples

The G-Mendeliome CES panel offers a more efficient solution to analyze clinical associated biomarkers in terms of key metrics such as the on-target ratio and fold-80 base penalty, even during multiplexing. Compact size of G-Mendeliome CES Panel compared to whole exome sequencing allows more cost-effective analysis.

List of Diseases Assessed by G-Mendeliome CES Panel

Category	Related Diseases
Cardiology	Aortopathy and connective tissue disorders
	Arrhythmia
	Cardiomyopathy
	Congenital heart defect
	Dyslipidemia
	Other cardiovascular diseases
Dermatology	Adams-Oliver syndrome
	Albinism
	Cardiofaciocutaneous syndrome
	Cutis laxa
	Dyskeratosis congenita
	Ectodermal dysplasia
	Ehlers-Danlos syndrome
	Epidermolysis bullosa
	Hereditary acrodermatitis enteropathica
	Hermansky-Pudlak syndrome
	Hypotrichosis
	Ichthyosis
	Neurofibromatosis
	Pachyonychia congenita
	Palmoplantar keratoderma
	Progeria and Progeroid Syndromes
	Skin cancer
	Tuberous sclerosis
	Waardenburg syndrome
	Xeroderma pigmentosum
Endocrinology	Adrenal hyperplasia
	Diabetes
	Hyperinsulinism
	Hyperparathyroidism
	Hypothyroidism
	Kallmann syndrome
	Multiple endocrine neoplasia
	Obesity
	Pancreatitis
	Premature ovarian failure
ENT	Hearing loss
GI/Hepatology	Cholestasis
	Congenital diarrhea
	Congenital hepatic fibrosis
	Gastrointestinal atresia
	Hirschsprung disease
	Polycystic liver disease
Hematology	Anemia
	Bleeding&Thrombotic disorder
	Bone marrow failure
	Congenital neutropenia
	Hemochromatosis
	RBC membrane disorder

List of Diseases Assessed by G-Mendeliome CES Panel

Category	Related Diseases
Immunology	Antibody deficiencies
	Autoinflammatory disorders
	Combined T/B cell deficiencies
	Complement deficiencies
	Defects in intrinsic and innate immunity
	Immune dysregulation
	Phagocytic defects
Metabolism	Aminoacidopathies
	Carbohydrate disorders
	Congenital disorders of glycosylation
	Creatine biosynthesis disorders
	Fatty acid oxidation defects
	Lipodystrophy
	Lysosomal storage disorders
	Organic acidemias
	Peroxisomal disorders
	Porphyria
	Purine/Pyrimidine metabolism disorders
	Pyruvate metabolism and tricarboxylic acid cycle defects
	Urea cycle disorders
	Nephrology
Ciliopathies	
Diabetes insipidus	
Hemolytic uremic syndrome	
Hypokalemia	
Hypomagnesemia	
Hypophosphatemic rickets	
Nephrolithiasis	
Nephrotic syndrome/Focal glomerulonephrosis	
Pseudohypoaldosteronism	
Renal malformation	
Renal tubular acidosis	
Neurology	
	Movement disorders
	Neurodegenerative disorders
	Neuromuscular disorders
	Neuropathies and related disorders
	Seizures and Brain abnormalities

List of Diseases Assessed by G-Mendeliome CES Panel

Category	Related Diseases
Oncology	Breast and gynecological cancer
	Colorectal cancer
	Endocrine cancer
	Gastrointestinal cancer
	Hematologic malignancy
	Lung cancer
	Nervous system/brain cancer
	Pancreatic cancer
	Prostate cancer
	Renal cancer
	Sarcoma
	Skin cancer
Ophthalmology	Albinism
	Cataract/Ectopia lentis
	Corneal dystrophy
	Glaucoma
	Microphthalmia/Anophthalmia
	Nystagmus
	Ophthalmoplegia/Oculomotor apraxia
	Optic atrophy
	Retinal dystrophy
	Retinoblastoma
Pulmonology	Bronchiectasis
	Central hypoventilation/Apnea
	Cystic fibrosis
	Cystic lung disease
	Hermansky-Pudlak syndrome
	Interstitial lung disease
	Primary ciliary dyskinesia
	Surfactant dysfunction
Skeletal disorders	Amelogenesis imperfecta
	Arthrogyposes
	Cleft lip palate
	Craniosynostosis
	Exostosis
	Facial dysostosis
	Macrocephaly/Overgrowth syndrome
	Osteopetrosis
	Short stature syndrome

Order Information | Clinical Exome Panel

Target Enrichment Kit

Cat. No.	Product	Product Unit(Hyb-rxn)
CEXL.HI.ES16/96	Clinical Exome Panel; Singleplex; Illumina	16/96
CEXM.HI.EM2/4/8/16	Clinical Exome Panel; Multiplex; Illumina	2/4/8/16
CEXL.HT.ES16/96	Clinical Exome Panel; Singleplex; Ion Torrent	16/96
CEXM.HT.EM2/4/8/16	Clinical Exome Panel; Multiplex; Ion Torrent	2/4/8/16
CEXL.HM.ES16/96	Clinical Exome Panel; Singleplex; MGI	16/96
CEXM.HM.EM2/4/8/16	Clinical Exome Panel; Multiplex; MGI	2/4/8/16

Library Prep Kit

Cat. No.	Product	Product Unit(Sample)
ILUY016/032/048/096	Illumina; Library Prep Kit; UDI; Short Adapter	16/32/48/96
IEUY016/032/048/096	Illumina; Enzymatic Library Prep Kit; UDI; Short Adapter	16/32/48/96
TLIP016/032/048/096	Ion Torrent; Library Prep Kit; IonXpress	16/32/48/96
TEIP016/032/048/096	Ion Torrent; Enzymatic Library Prep Kit; IonXpress	16/32/48/96
MLAT016/032/048/096	MGI; Library Prep Kit; MGI Single Index	16/32/48/96
MEAT016/032/048/096	MGI; Enzymatic Library Prep Kit; MGI Single Index	16/32/48/96

* For Illumina Prep Kits, more adapter and index options are available. Please inquiry to our sales team.

Accessories

Cat. No.	Product	Product Unit
CMPF-M-500	CLM Polymerase	5 mL
CMSB-M-200/00A	CeleMag™ Streptavidin Bead	2/10 mL
CMCB-M-050/500	CeleMag™ Clean-up Bead	5/50 mL
CNMS-R-16/96	CeleNM™ Bead	16/96 rxn

* For CeleNM™ Bead, more unit option is available. Please inquiry to our sales team.

Celemics HQ

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