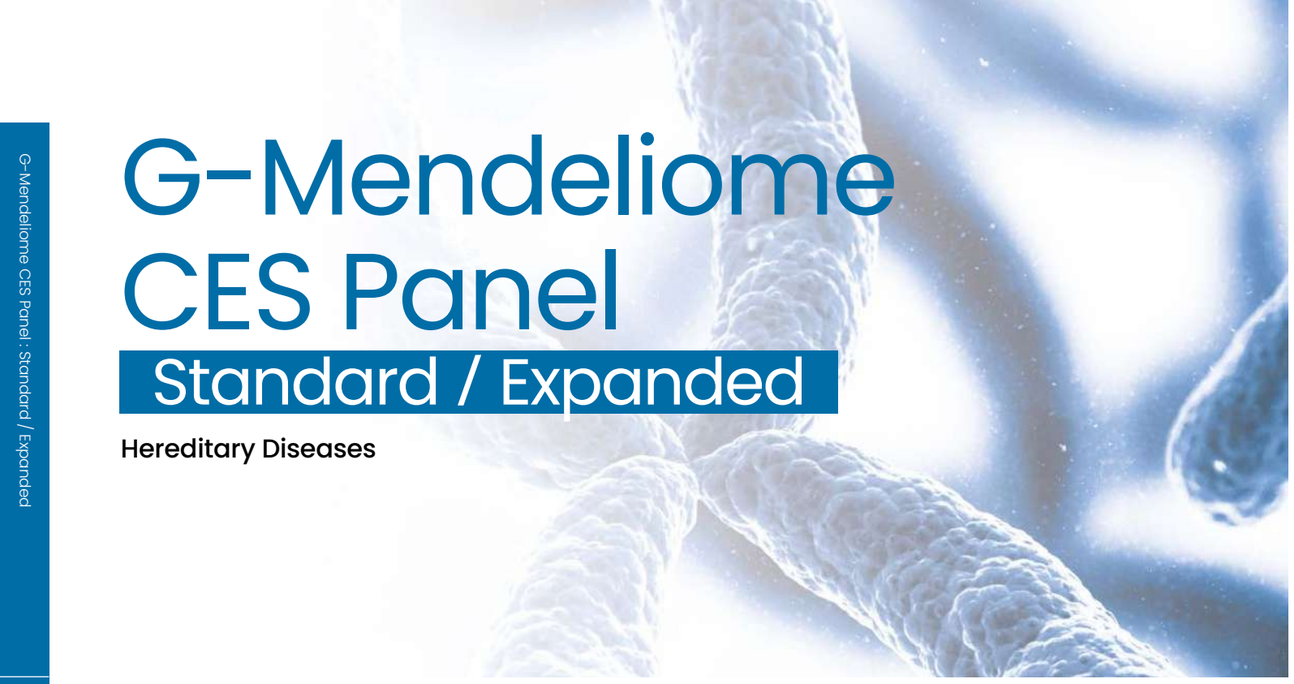


G-Mendeliome CES Panel

Standard / Expanded

Hereditary Diseases

DESCRIPTION

The G-Mendeliome CES Panel has overcome the limitations of analyzing clinical diseases with whole exome sequencing. By selectively targeting the clinically significant genes, the panel enables comprehensive analysis with the most effective sequencing throughput.

KEY FEATURES

1. Comprehensive genomic profiling of a variety of genetic diseases	Includes 7,000 genes associated with clinically significant genetic diseases
2. A wide range of target regions	Includes all clinically significant regions that are not covered from competitor panels
3. Cost-effective analysis	Able to provide accurate analysis with reduced sequencing costs compared to WES

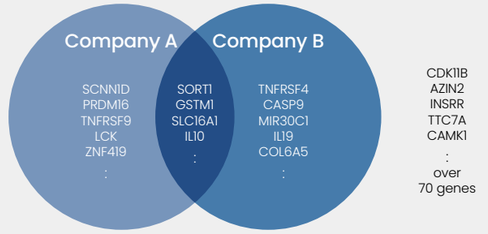
SPECIFICATION

Gene count*	5,517 / 7,563 genes
Covered region	CDS, hotspots, Mitochondrial genome
Target size	13.8 / 19.6 Mb
Mutation type	SNV, Indel, CNV
Sample type	Blood (> 50 ng of fragmented DNA)
Platform	All sequencers from Illumina, Thermo Fisher, MGI, PacBio, and Oxford Nanopore
Bioinformatics pipeline	Primary, Secondary and Tertiary analysis result (FASTQ to VCF, VCF to Clinical report)

PANEL PERFORMANCE

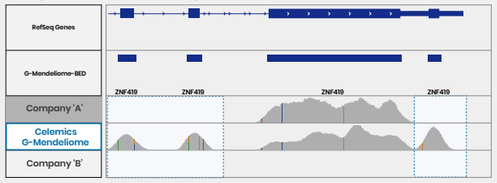
	Celemics	Company A	Company B
On-Target Read Ratio	82.8%	65.9%	80.8%

G-Mendeliome CES: Company A + Company B + over 70 genes

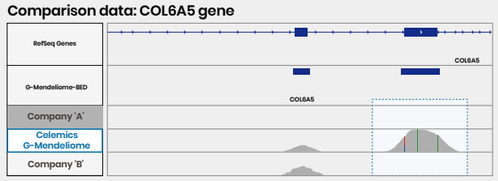


The CES panel was developed from the needs of GC Genome, the largest clinical NGS service provider in Korea, to solve the problem of poor diagnosis and high costs

Comparison data : ZNF419 gene



Comparison data: COL6A5 gene



PACKAGE COMPOSITION

Package name	Compositions		
Target Enrichment	Target capture Probe	-	
Standard	Target Enrichment reagents	Library prep Kit	-
All-In-One			Beads / Polymerase

Package option	Options	
Pooling method	Single Reaction	Pre-capture Pooling
Library Preparation kits	Standard Kit	EP-kit
Hybridization Enhancer	Included	Not included

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
	Pulmonary hypertension
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
Immunology	Antibody deficiencies
	Autoinflammatory disorders
	Combined T/B cell deficiencies
	Complement deficiencies
	Defects in intrinsic and innate immunity
	Immune dysregulation
	Phagocytic defects

Category	Related Diseases
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	Bartter syndrome
	Ciliopathies
	Diabetes insipidus
	Hemolytic uremic syndrome
	Hypokalemia
	Hypomagnesemia
	Hypophosphatemic rickets
	Nephrolithiasis
	Nephrotic syndrome/Focal glomerulonephrosis
	Pseudohypoaldosteronism
	Renal malformation
	Renal tubular acidosis
Neurology	Autism
	Movement disorders
	Neurodegenerative disorders
	Neuromuscular disorders
	Neuropathies and related disorders
	Seizures and Brain abnormalities
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
	Arthrogryposes
	Cleft lip palate
	Craniosynostosis
	Exostosis
	Facial dysostosis
	Macrocephaly/Overgrowth syndrome
	Osteopetrosis
	Short stature syndrome
	Skeletal dysplasia