GC Genome

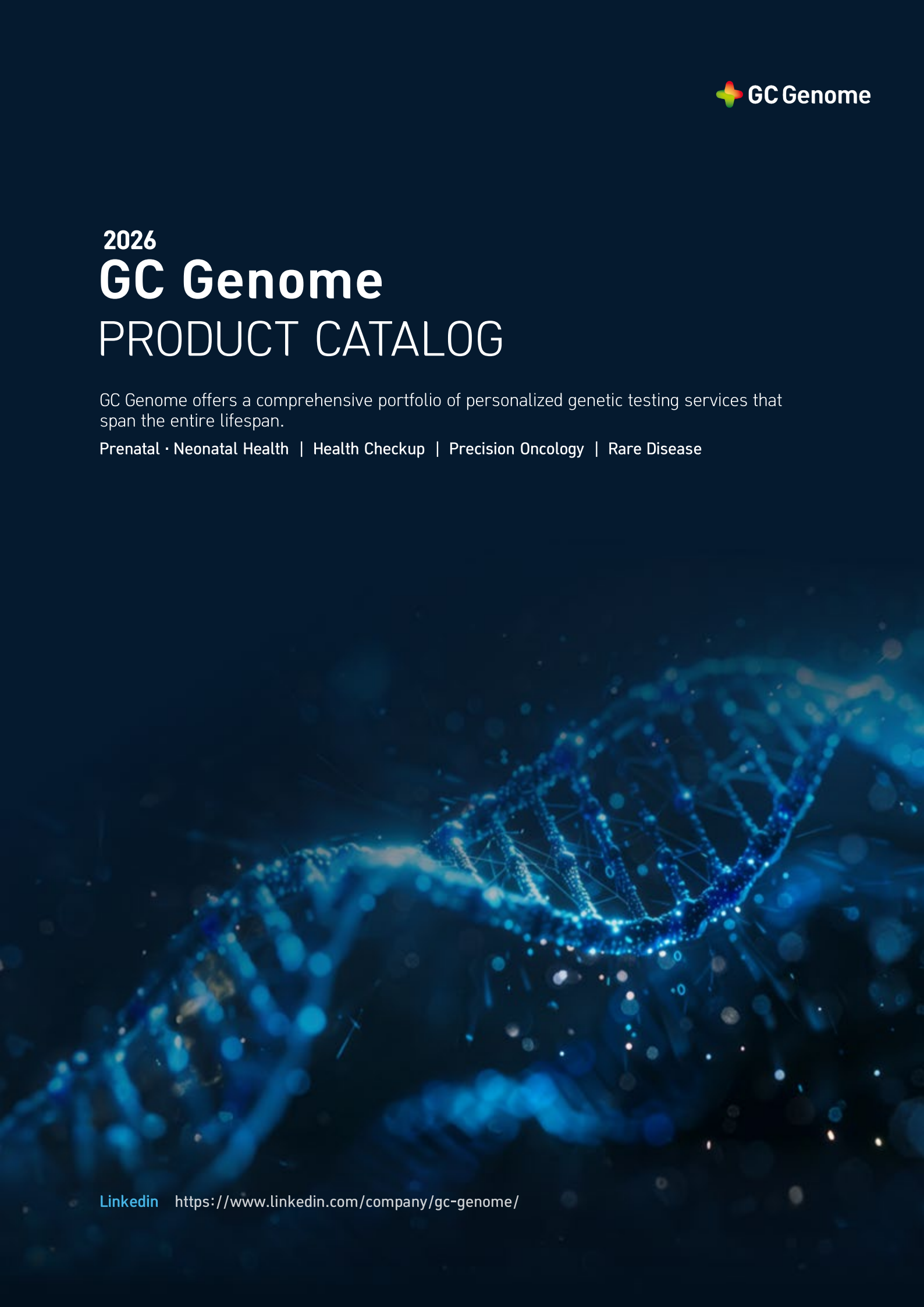
2026

GC Genome

PRODUCT CATALOG

GC Genome offers a comprehensive portfolio of personalized genetic testing services that span the entire lifespan.

Prenatal · Neonatal Health | Health Checkup | Precision Oncology | Rare Disease



Linkedin https://www.linkedin.com/company/gc-genome/

Korea's Leading
Clinical Genomics Company

1st

Published
Papers

150+

Countries with
Overseas Patent Filings

16+

Filed &
Registered Patents

160+

Continuous A-Level
Proficiency Test Ratings

10Y

CAP* Accreditation Maintained
*College of American Pathologists

10Y

Annual Genetic Testing
Capacity

200,0000+

Number of Genetic Testing Services

300+

Countries Served

20+

Partner Medical Institutions

900+

World-class Infrastructure

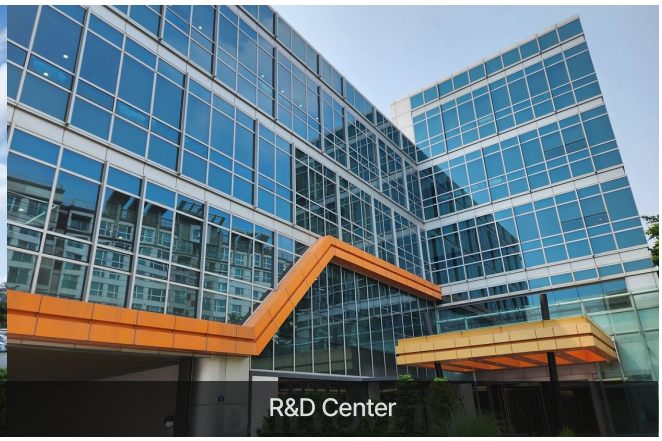


GC Genome, established in 2013, is a specialized clinical genomic analysis company dedicated to disease diagnosis and personalized treatment using genomic information.

By utilizing Next-Generation Sequencing (NGS), we provide fast and cost-effective testing services.

Our active R&D efforts at the Genomic Research Institute focus on developing new diagnostic tests and participating in national research projects.


Headquarter


R&D Center

Accreditation


COLLEGE of AMERICAN PATHOLOGISTS

CERTIFICATE OF ACCREDITATION

GC Genome
Laboratory
Yongin-si, South Korea
Chang Ahn Seol, MD

CAP Number: 8991762
AU-ID: 1746397

A

College of American Pathologists (CAP)

First non-medical institution for genetic testing in Korea to be accredited by CAP


Genetic Testing Evaluation Certificate

Institution : GC Genome
Laboratory : Genome Analysis Center
Lab ID : 252
Certification period : Jan. 1 to Dec. 31, 2026

The Korean Institute of Genetic Testing Evaluation hereby certifies that the aforementioned laboratory participated in the genetic testing proficiency evaluation and achieved the following results.

Dec 29, 2025

한국유전자검사평가원

Korean Institute of Genetic Testing Evaluation

Korea Institute of Genetic Testing Evaluation

The best grade(Level A) in accuracy of genetic test


임상시험검체분석기관 지정서

Ministry of Food and Drug Safety(MFDS)

Designated as a Clinical Trial Specimen Analysis Institution(GCLP)

Laboratory Infrastructure

Operating a comprehensive clinical genomics laboratory with over 140 instruments, including 13 NGS platforms, and an annual testing capacity exceeding 200,000 samples.

Core Sequencing Platform

Short-read Sequencing



Novaseq 6000Dx
illumina



Novaseq
illumina



Nextseq
illumina



Miseq
illumina

Long Read Sequencing



VEGA
PacBio

Alternative Sequencing Platforms



Ion Chef
Thermo Fisher



Ion S5
Thermo Fisher



Genexus
Thermo Fisher

Microarray



GCS 3000Dx
Thermo Fisher

Supporting Technologies

- Automated nucleic acid extraction systems
- High-throughput liquid handling platforms
- Clinical-grade sample tracking and LIMS
- Secure bioinformatics and data management infrastructure



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01
PART



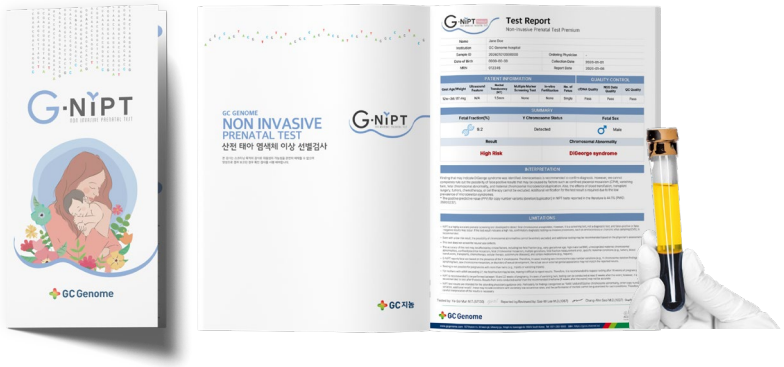
Prenatal · Neonatal Health

GC Genome provides genetic testing solutions to support safe pregnancy and early identification of chromosomal conditions, contributing to the healthy development of newborns.

Code O001 O002 O003

G-NIPT

Non-invasive prenatal screening test for fetal chromosomal abnormalities using cell-free fetal DNA



Representative Scientific Evidence & Publications	Front Genet. 2022 Nov 29:13:999587
Sample Report (including disease & gene)	

Introduction	G-NIPT® is a non-invasive prenatal screening test that analyzes cell-free fetal DNA present in maternal blood to assess the risk of fetal chromosomal aneuploidies. The test screens for common chromosomal abnormalities, including Down syndrome (trisomy 21), Edwards syndrome (trisomy 18), Patau syndrome (trisomy 13), and other selected chromosomal conditions. Key Result Components: Chromosomal abnormality results · Risk after NIPT · Fetal sex
Indication	G-NIPT is primarily used to screen for common chromosomal abnormalities. While G-NIPT is offered to all pregnant women as a primary screening tool, it is particularly recommended for: • Advanced Maternal Age: Women aged 35 or older at the time of delivery. • Abnormal Serum Screening: High-risk results from traditional biochemical screens (e.g., Quad or Integrated screen). • Ultrasound Abnormalities: Findings suggestive of chromosomal conditions, such as increased Nuchal Translucency (NT). • Personal/Family History: Previous pregnancy or family history of trisomy 21, 18, or 13.
Suitable for	All pregnant women from 10 weeks of gestation.

Technology	NGS		**Methods**		Low coverage WGS (lcWGS)	
G-NIPT Type	**Lite** O001		**Basic** O002		**Premium** O003	
No. of fetus	**Singleton**	**Twin**	**Singleton**	**Twin**	**Singleton**	**Twin**
Autosomal aneuploidies	T21,18,13		Chomosomoal Trisomy		Chomosomoal Trisomy & Monosomy	
Sexchromosomal aneuploidies	XO, XXX, XXY,XYY	Not Provided	XO, XXX, XXY,XYY	Not Provided	XO, XXX, XXY,XYY	Not Provided
CNV	Not Provided	Not Provided	Not Provided	Not Provided	140 types & >7Mb of CNVs	
Gender	Y chromosomal detected					

Specimen	(Recommended) cfDNA tube Whole Blood 10 ml or Plasma 1 ml X 2 vials
TAT	3~5 days
Limitation	• Screening test only; not intended for definitive diagnosis. • Low-level mosaicism, rare chromosomal abnormalities, or structural variants may not be detected. • Confirmatory diagnostic testing is required for positive results.

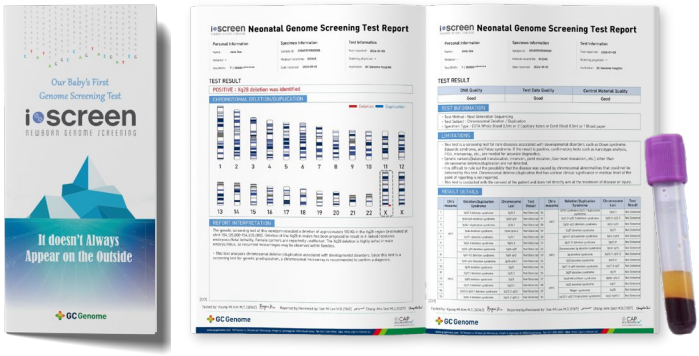
GC Genome

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

i screen NEWBORN GENOME SCREENING

Code ON035

Newborn chromosomal abnormality screening test using genetic analysis for early detection of congenital disorders



Sample Report
(including disease & gene)

Introduction	i-screen® is a screening test that analyzes approximately 170,000 regions across all 23 pairs of chromosomes to detect chromosomal deletions and duplications associated with congenital disorders. Large-scale chromosomal losses or gains related to conditions such as Down syndrome, Edwards syndrome, and Patau syndrome may result in characteristic clinical features including intellectual disability, developmental delay, and autism spectrum disorders. Key Result Components: Deletion/Duplication syndrome results		
Indication	• Genetic screening for chromosomal copy number variations associated with congenital and developmental disorders • Intended for newborns or pediatric infants requiring early genetic risk assessment • Supports early clinical evaluation of developmental delay and neurodevelopmental disorders		
Suitable for	Recommended within 6 months of birth		
Technology	NGS	**Methods**	Low coverage WGS (lcWGS)
Specimen	(Recommended) EDTA WB 0.5 ml or Blood Paper (G-Card)		
TAT	6 days		
Limitation	• Cannot detect balanced chromosomal rearrangements or single nucleotide variants • Small copy number changes below the resolution threshold may not be detected • Clinical correlation and confirmatory testing are required		



02
PART



Health Check-up

GC Genome provides comprehensive genetic health checkup services that assess cancer risk, disease susceptibility, and hereditary factors, supporting personalized health management based on individual genetic profiles.

Code ON203

ai-CANCERCH®

AI-based blood test for multi-cancer early screening.



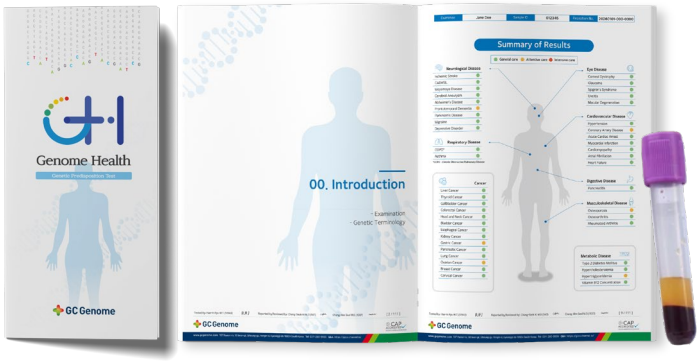
Representative Scientific Evidence & Publications	1) Nat Commun. 2023 Apr 10;14(1):2017. 2) Cancer Res. 2025 May 2;85(9):1696-1707.
Sample Report (including disease & gene)	

Introduction	ai-CANCERCH® is an AI-based blood test for multi-cancer early screening that analyzes cell-free DNA (cfDNA) using low-coverage whole-genome sequencing (lcWGS). By evaluating cancer-associated patterns derived from cell-free DNA (cfDNA), the test assesses the likelihood of current cancer presence across multiple cancer types. Powered by a proprietary deep-learning algorithm trained on a large clinical dataset, ai-CANCERCH® supports proactive cancer risk assessment as part of routine health checkups. For individuals with a positive or high-risk result, ai-CANCERCH® provides follow-up support to facilitate appropriate next steps in clinical evaluation. This includes support for follow-up monitoring tests and financial assistance for confirmatory diagnostic examinations, helping ensure timely and comprehensive clinical assessment. ai-CANCERCH® is designed to assess cancer-associated signals across multiple cancer types, including but not limited to: colon cancer, lung cancer, esophageal cancer, liver cancer, ovarian cancer, pancreatic cancer, bile duct cancer, breast cancer, gastric cancer, and head and neck cancer. **Key Result Components:** Overall results; CANCEHRCH® Score (0~100); methTOO result; Summary of results; Cancer risk; Interpretation of detailed results; Guidline
Indication	• Blood-based multi-cancer early screening test for assessing the likelihood of current cancer presence • Intended for asymptomatic adults seeking proactive cancer risk assessment as part of routine health checkups • Supports identification of cancer-associated signals across multiple cancer types using cfDNA analysis • Provides supplementary information to guide further clinical evaluation when cancer risk is indicated
Technology	NGS + AI
Methods	Low coverage WGS (lcWGS) & Liquid Biopsy
Specimen	(Recommended) Plasma 1 ml X 2 vials or cfDNA tube Whole Blood 10 ml (collected within 5days)
TAT	14 days
Limitation	• Screening test only; not intended for definitive cancer diagnosis • Does not localize tumors or determine cancer stage • Certain cancer types, early-stage cancers with low tumor DNA shedding, or non-shedding tumors may not be detected • False-positive or false-negative results may occur; results should be interpreted in conjunction with clinical findings • Positive results require confirmatory diagnostic testing using standard clinical methods

Code ON155 ON156

Genome Health

Comprehensive genetic screening test for inherited disease risk assessment.



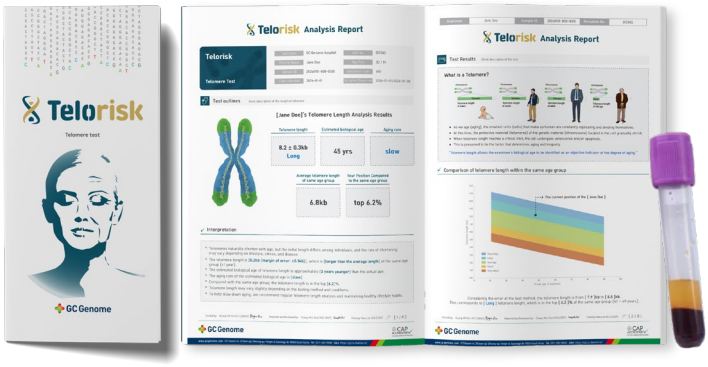
Sample Report
(including disease & gene)

Introduction	Genome Health® is a genetic screening test that analyzes single nucleotide variants (SNVs) identified from DNA in blood to assess individual disease susceptibility. The test provides personalized health management insights based on genetic risk factors. Genome Health evaluates 44 disease categories for males and 45 disease categories for females, supporting proactive and preventive healthcare planning. **Key Result Components**: Genetic Terminology; Summary of Results; Results by Disease; Summary of Genes Requiring Intensive Care; How to interpret result; Interpretation by Disease		
Indication	• Genetic screening to assess individual susceptibility to common multifactorial diseases • Intended for adults seeking proactive and preventive health management • Supports personalized health planning based on inherited genetic risk factors		
Technology	NGS & PCR	**Methods**	SNP genotyping
Type	Male ON155	Female ON156	
Specimen	(Recommended) EDTA WB 3.0 ml or Buccal swab		
TAT	14 days		
Limitation	• Evaluates genetic susceptibility only; does not diagnose disease • Environmental and lifestyle factors are not directly measured • Results should be interpreted in conjunction with clinical information		

Telorisk

Code OS099

Telomere-based genetic test for biological aging and health risk assessment.



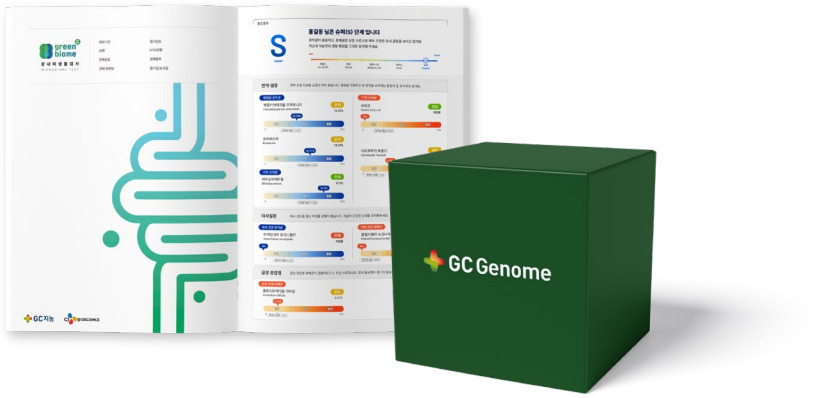
Sample Report
(including disease & gene)

Introduction	Telorisk is a genetic test that measures telomere length using quantitative polymerase chain reaction (qPCR) by calculating the ratio of telomere DNA to a reference gene. Telomere length naturally shortens with age, and the rate of shortening may vary depending on lifestyle and environmental factors. Based on established scientific evidence, the test provides insights into biological aging and supports personalized health management. **Key Result Components**: Telomere length; Estimated biological age; Aging rate; Position compared to the same age group; Predicted telomere length
Indication	• Assessment of biological aging based on telomere length measurement • Intended for adults seeking insights into aging-related health status and long-term wellness planning • Supports evaluation of telomere dynamics in relation to lifestyle and environmental factors • Provides supplementary information for personalized health management as part of routine health checkups
Technology	qPCR
Specimen	EDTA WB 3.0 ml
TAT	7 days
Limitation	• Telomere length is a surrogate biomarker and does not represent a direct measure of disease • Inter-individual variability may affect result interpretation • Results do not predict specific diseases or clinical outcomes • Not intended for diagnosis, prognosis, or treatment decision-making

greenbiome

Code NEW

Targeted microbiome test based on PCR technology.

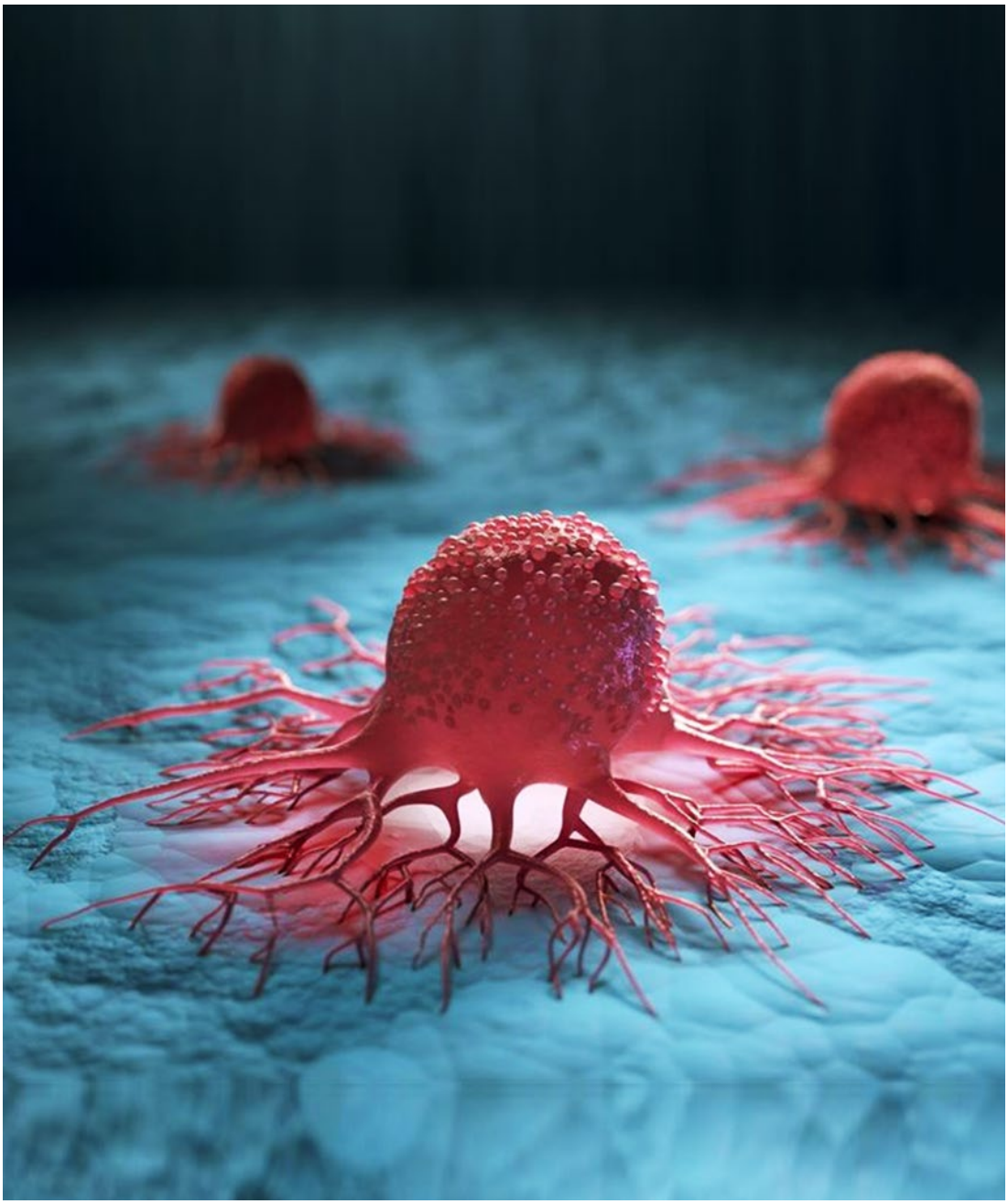


Sample Report
(including disease & gene)

Introduction	Greenbiome is a microbiome analysis test that utilizes using quantitative polymerase chain reaction (qPCR)to analyze microorganisms present in the human gut. The test evaluates the balance and composition of intestinal microbiota, including beneficial and harmful bacteria, and their associations with health and disease. **Key Result Components** Gut Microbiota Balance (Beneficial vs. Harmful Bacteria) Microbial Balance Related to Immune and Inflammation Status Microbial Balance Associated with Metabolic Health Conditions Microbial Balance Related to Acute Gut Infection Personalized Microbiome Solutions
Indication	• Gut microbiome analysis to evaluate the balance and composition of key intestinal microorganisms • Intended for individuals seeking insights into gut health, metabolic balance, and immune-related status • Supports assessment of microbiome patterns associated with digestive health, inflammation, and metabolic conditions • Provides supportive information for personalized lifestyle and nutritional management
Technology	qPCR
Specimen	Stool
TAT	14 days
Limitation	• Results reflect microbiome composition at the time of sampling and may vary over time • Does not diagnose active disease, infection, or gastrointestinal disorders • Microbiome profiles may be influenced by diet, medication, probiotics, and lifestyle factors • Results should be interpreted in conjunction with clinical findings and other health information



03
PART



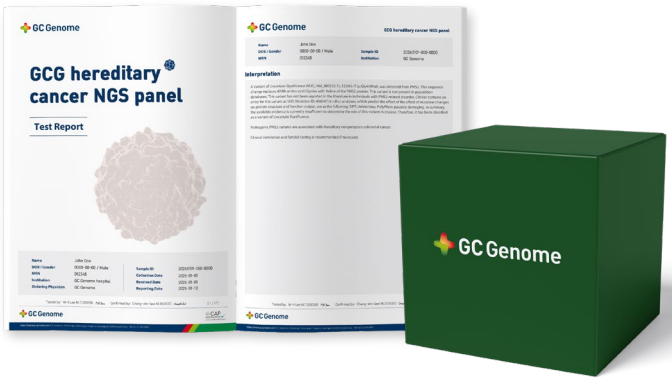
Precision Oncology

GC Genome provides comprehensive genomic testing to support precision cancer care, enabling informed decisions for diagnosis, treatment selection, and prognosis assessment.

GreenPlan® Hereditary

Code ON001 ON040

Genetic test for identifying inherited cancer predisposition across multiple cancer types.



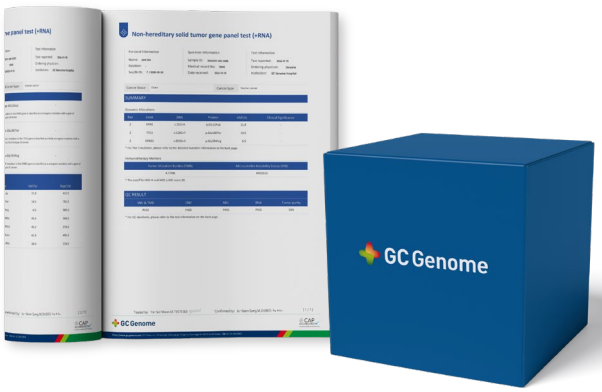
Sample Report
(including disease & gene)

Introduction	GreenPlan® Hereditary is a genetic screening test that analyzes inherited genetic variants associated with hereditary cancers. Hereditary cancers are caused by germline variants passed from parents to offspring and are associated with a significantly higher lifetime cancer risk compared to sporadic cancers.The panel supports comprehensive hereditary cancer risk assessment, including dedicated panels for women's cancers. Key Result Components: Gene, DNA Change, AA Change, Zygosity, OMIN Disease, Inherit, Class Result details and related diseases, Relevant disease information		
Indication	• Genetic screening for inherited cancer predisposition associated with germline variants • Intended for individuals with a personal or family history suggestive of hereditary cancer • Supports comprehensive hereditary cancer risk assessment and identification of individuals at increased lifetime cancer risk • Provides clinically relevant genetic information to support genetic counseling and risk management strategies		
Technology	NGS	Methods	Targeted Panel
Type	Woman's cancer panel ON001		Comprehensive cancer panel ON040
Specimen	EDTA WB 3.0 ml		
TAT	12 days		
Limitation	• Does not detect all hereditary cancer–associated variants • Variants of uncertain significance (VUS) may be identified • A negative result does not eliminate hereditary cancer risk • Results should be interpreted in conjunction with personal and family history and with genetic counseling		

GreenPlan® Solid tumor TBx LBx

Code ON198 ON199 ON200

Genetic profiling test including RNA (or without RNA) analysis for solid tumors.



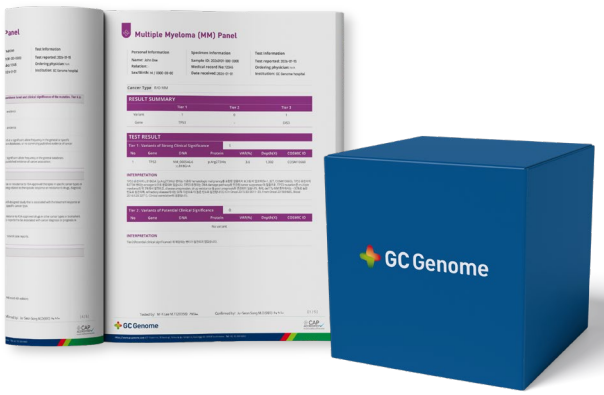
Sample Report
(including disease & gene)

Introduction	GreenPlan® TBx is a comprehensive tumor genomic profiling test for guiding treatment decisions in malignant solid tumors. The test analyzes DNA and RNA extracted from tumor tissue or FFPE specimens to evaluate genomic alterations across approximately 500 cancer-related genes. The test detects single nucleotide variants (SNVs), small insertions and deletions, copy number variations, and gene fusions, and reports tumor mutational burden (TMB) and microsatellite instability (MSI) status. Clinically actionable variants corresponding to Tier 1 and Tier 2 are reported. (RNA-included and RNA-excluded options available) **Key Result Components**: Tier, Gene, DNA, Protein, VAF, Clinical Significance, Immunotherapy Markers Variant Detail, Total variant list			
Indication	• Comprehensive tumor genomic profiling for malignant solid tumors • Intended for patients with advanced, recurrent, or refractory solid cancers where molecular profiling may support treatment decision-making • Supports identification of clinically actionable genomic alterations, including SNVs, small insertions and deletions, copy number variations, and gene fusions • Provides tumor mutational burden (TMB) and microsatellite instability (MSI) status to support consideration of targeted and immunotherapy options • Applicable to a broad range of solid tumors using tumor tissue or FFPE specimens			
Technology	NGS	**Methods**	Targeted Panel	
Type	**Tumor biopsy**	incl. RNA (DNA+RNA) ON199	excl. RNA (DNA only) ON200	
	Liquid biopsy	Liquid biopsy ON198		
Specimen	• TBx : FFPE 8~16 Slides and Mark lesion area on 1 H&E slide (Tumor cellularity≥30% required) • LBx : cfDNA tube Whole Blood 20 ml			
TAT	14 days			
Limitation	• Limited to genes and genomic alterations included in the panel • Low tumor content, poor DNA/RNA quality, or degraded FFPE samples may affect test performance • Not all detected variants are clinically actionable • Only Tier 1 and Tier 2 variants are reported; Tier 3 and Tier 4 variants are not included in the clinical report • Results should be interpreted in conjunction with clinical, pathological, and radiological findings			

GreenPlan® Hema

Code ON064 ON065 ON082 ON083 ON104

Genetic diagnostic test for hematologic cancer patients, including assessments of treatment options and prognosis



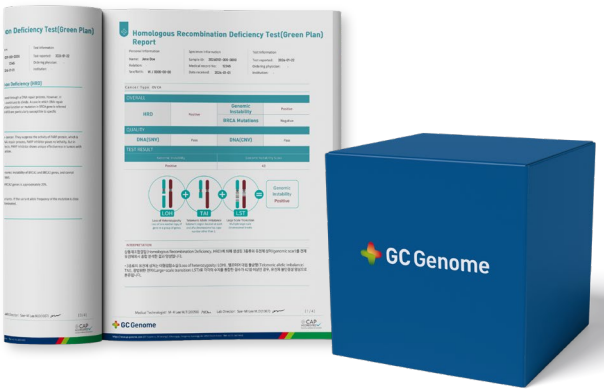
Sample Report
(including disease & gene)

Introduction	GreenPlan® Hema is a hematologic malignancy genomic test designed for diagnosis, prognostic evaluation, and treatment planning. By analyzing DNA extracted from bone marrow or peripheral blood, it detects mutations across a comprehensive panel of genes associated with hematologic cancers. The test identifies SNVs and small indels with a limit of detection (LoD) of approximately 5%, supporting clinical assessment for diseases including multiple myeloma, lymphoma, MDS/MPN, ALL, and AML. Key Result Components: Tier, Gene, DNA, Protein, VAF, Depth, COSMIC ID
Indication	• Genomic analysis for hematologic malignancies to support diagnosis, prognostic assessment, and treatment planning • Intended for patients with suspected or confirmed hematologic cancers, including acute myeloid leukemia (AML), acute lymphoblastic leukemia (ALL), myelodysplastic syndromes/myeloproliferative neoplasms (MDS/MPN), lymphoma, and multiple myeloma • Supports identification of clinically relevant genomic alterations, including single nucleotide variants (SNVs) and small insertions and deletions • Applicable to bone marrow or peripheral blood specimens as part of comprehensive hematologic evaluation
Technology	NGS — **Methods** — Targeted Panel
Type	AML panel ON064 / MDS/MPN panel ON065 / ALL panel ON082 / Lymphoma panel ON083 / MM panel ON104
Specimen	EDTA WB 3.0 ml or EDTA BM 3.0 ml
TAT	14 days
Limitation	• Detection sensitivity may be limited for low-frequency variants • Structural variants and copy number alterations may not be fully captured • Results may be affected by sample quality and tumor burden • Not all detected variants are clinically actionable • Results should be interpreted in conjunction with clinical, hematologic, and pathological findings

GreenPlan® HRD

Code ON140

Genomic test to assess homologous recombination deficiency for therapy guidance.



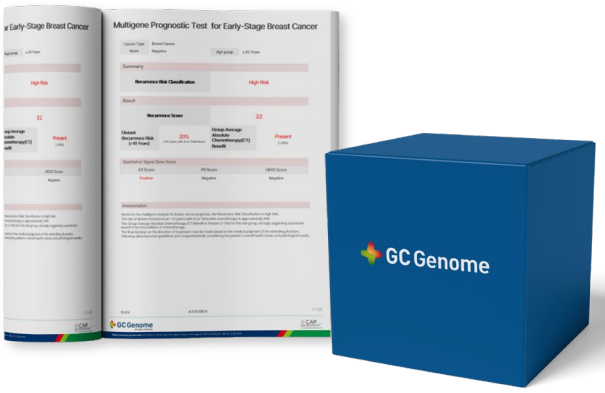
Sample Report
(including disease & gene)

Introduction	GreenPlan® HRD is a genomic test designed to assess homologous recombination deficiency (HRD), a condition in which DNA damage repair is impaired due to defects in homologous recombination pathways, including BRCA gene abnormalities. Tumors with HRD characteristics may show increased sensitivity to specific targeted therapies, such as PARP inhibitors, supporting treatment decision-making. **Key Result Components**: HRD status; Genomic Instability; Genomic Instability Score; BRCA Mutations		
Indication	• Genomic assessment of homologous recombination deficiency (HRD) in ovarian cancer tumor tissue • Intended for patients with ovarian cancer in which defects in DNA damage repair pathways may inform therapeutic decision-making • Supports evaluation of genomic instability and BRCA-related mutations associated with homologous recombination defects in ovarian cancer • Provides HRD-related genomic information to support consideration of targeted therapies, including PARP inhibitors, in ovarian cancer management		
Technology	NGS	**Methods**	Targeted Panel
Specimen	FFPE 8~16 Slides and Mark lesion area on 1 H&E slide (Tumor cellularity ≥ 30% required)		
TAT	21 days		
Limitation	• HRD status alone does not guarantee response to PARP inhibitors or other targeted therapies • Results should be interpreted in conjunction with clinical stage, histopathological findings, and prior treatment history • Not all DNA repair defects or mechanisms of genomic instability relevant to ovarian cancer may be captured • Test performance may be affected by tumor purity and sample quality, particularly in FFPE specimens		

Code NEW

GreenPlan® BC

A gene expression–based test for assessing early-stage breast cancer recurrence risk and the potential benefit of adjuvant chemotherapy.



Sample Report
(including disease & gene)

Introduction	GreenPlan® BC test is a prognostic test that analyzes the expression of 21 genes to calculate a Recurrence Score, predicting recurrence risk and helping determine whether chemotherapy will provide added benefit for early-stage breast cancer patients. **Key Result Components**: Recurrence risk classification; Quantitative Recurrence Score (0–100); Distant Recurrens Risk; Predicted chemotherapy benefit		
Indication	• Women with early-stage breast cancer who are hormone receptor–positive (HR+; estrogen receptor–positive [ER+] and/or progesterone receptor–positive [PR+]), HER2-negative, and lymph node–negative (pN0) or with limited lymph node involvement (pN1) • Supports assessment of recurrence risk through quantitative analysis of tumor gene expression • Provides information to support clinical decision-making regarding the potential benefit of adjuvant chemotherapy		
Technology	qRT-PCR	**Methods**	21-gene expression analysis
Specimen	FFPE 8~16 Slides and Mark lesion area on 1 H&E slide (Tumor cellularity≥30% required)		
TAT	7 days		
Limitation	• The GreenPlan® BC Test is not intended to diagnose a clinical condition. • The GreenPlan® BC Test is validated for use with formalin-fixed, paraffin-embedded (FFPE) breast cancer tumor tissue. The performance of the test with other specimen types has not been evaluated. • The GreenPlan® BC Test is not intended for use in a normal population. No reference or expected values have been established for a normal population. • The GreenPlan® BC Test is not intended for self-testing or for near-patient testing.		

04
PART



Rare Disease

GC Genome provides advanced genomic testing to support the diagnosis of a wide range of hereditary rare diseases.

Whole Exome Sequencing

Code ON201

Exome sequencing test for the genetic diagnosis of rare diseases.



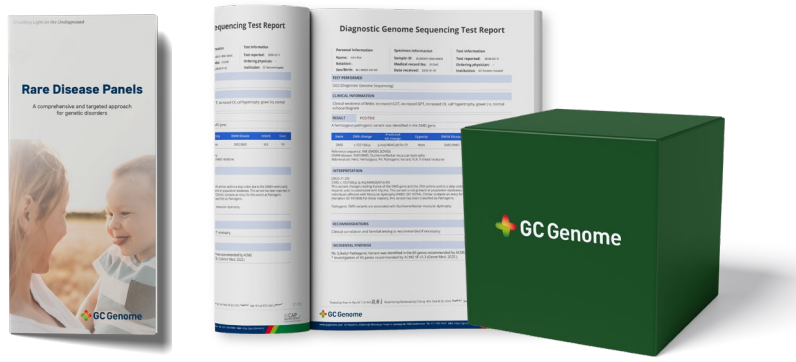
Sample Report
(including disease & gene)

Introduction	Whole Exome Sequencing (WES) is a genetic test that analyzes the coding regions (exons) of all protein-coding genes, which account for approximately 2% of the human genome, as well as mitochondrial DNA. Using next-generation sequencing, WES enables comprehensive variant analysis and supports the diagnosis of approximately 30–40% of rare genetic diseases. **Key Result Components**: Gene, DNA change, Predicted AA change, Zygosity, OMIM Disease, Inherit, Class, Interpretation, Incidental findings		
Indication	• Genetic analysis for the diagnosis of suspected rare or inherited diseases • Intended for patients with unexplained clinical phenotypes or suspected monogenic disorders • Supports identification of pathogenic variants within protein-coding regions of the genome • Applicable as a first-tier or second-tier test in rare disease evaluation and clinical genetics		
Technology	NGS	**Methods**	WES
Specimen	(Recommended) EDTA WB 3.0 ml or Blood Paper (G-Card)		
TAT	21 days		
Limitation	• Non-coding regions of the genome are not analyzed • Structural variants, copy number changes, and repeat expansions may not be fully detected • Variants of uncertain significance (VUS) may be identified • Diagnostic yield may vary depending on clinical phenotype and case complexity • Results should be interpreted in conjunction with clinical findings and genetic counseling		

Diagnostic Genome Sequencing

Code ON127

Whole genome sequencing test for comprehensive genetic analysis.



Sample Report
(including disease & gene)

Introduction	Diagnostic Genome Sequencing (DGS) is a whole genome sequencing–based test designed for comprehensive genetic diagnosis of suspected inherited disorders. The test is applied as a first-tier test when rapid diagnosis is required or as a second-tier test when prior genetic testing fails to identify causative variants. DGS supports trio-based genome analysis to identify de novo and recessive mutations and enables the detection of disease-causing variants in familial cases that are difficult to resolve with conventional testing. In addition, the test detects large-scale genomic alterations, including deletions, duplications, and complex genomic rearrangements. **Key Result Components**: Gene, DNA change, Predicted AA change, Zygosity, Inherit, OMIM Disease, Class, Interpretation, Incidental findings		
Indication	• Comprehensive genome-wide genetic analysis for the diagnosis of suspected rare or inherited disorders • Intended for patients requiring rapid genetic diagnosis or for cases unresolved by prior genetic testing • Supports detection of single nucleotide variants, insertions and deletions, and large-scale genomic alterations, including deletions, duplications, and complex rearrangements • Applicable to trio-based analysis for identification of de novo and recessive variants in familial cases		
Technology	NGS	Methods	WGS
Specimen	EDTA WB 3.0 ml		
TAT	35 days		
Limitation	• Interpretation may be challenging due to the complexity and volume of genomic data • Variants of uncertain significance (VUS) and incidental findings may be identified • Not all genetic variants or mechanisms may be fully captured • Diagnostic yield may vary depending on clinical presentation and case complexity • Results require expert clinical and genetic interpretation and should be correlated with clinical findings		

GC-REP™

Code NEW

Advanced sequencing test for accurate detection of repeat expansion–related genetic disorders.

Coming soon

Sample Report (including disease & gene)	Coming soon

Introduction	GC-REP™ is a targeted long-read sequencing test that utilizes PacBio PureTarget™ technology to detect and characterize pathogenic repeat expansions associated with genetic and neurological disorders. By selectively enriching repeat expansion loci and applying long-read sequencing, the test enables accurate sizing and sequence characterization of expanded repeats, including complex or GC-rich regions that are difficult to resolve using conventional short-read sequencing. Key Result Components: To be determined		
Indication	• Targeted genetic analysis for detection and characterization of pathogenic repeat expansion mutations • Intended for patients with suspected genetic or neurological disorders caused by known repeat expansion loci • Supports accurate sizing and structural characterization of expanded repeats to aid genetic diagnosis • Applicable as a complementary test when repeat expansion disorders are suspected or when short-read sequencing results are inconclusive		
Technology	NGS (Long-read sequencing, PacBio)	Methods	SMART sequencing
Specimen	EDTA WB 3.0 ml		
TAT	35~60 days		
Limitation	• Limited to repeat expansion regions targeted by the test design • Does not provide genome-wide variant information • Novel, atypical, or non-targeted repeat loci may not be detected • Results should be interpreted in conjunction with clinical findings and other genetic test results		

Code NEW

Long Read WGS

Long-Read whole genome sequencing to support advanced genetic diagnosis in complex cases.

Coming soon

Sample Report (including disease & gene)	Coming soon

Introduction	Long-Read Whole Genome Sequencing is a comprehensive genomic analysis test that utilizes PacBio long-read sequencing technology to analyze the entire human genome with high continuity and accuracy. By generating long sequencing reads, the test enables improved resolution of complex genomic regions and supports accurate detection of structural variants, repeat expansions, and complex genomic rearrangements that are difficult to identify using conventional short-read sequencing. This test supports advanced genetic diagnosis of rare and neurological disorders. Key Result Components: To be determined		
Indication	• Comprehensive genome-wide genetic analysis for patients with suspected rare or inherited disorders • Intended for cases in which short-read sequencing is insufficient to resolve complex genomic variants • Supports detection of single nucleotide variants, structural variants, repeat expansions, complex genomic rearrangements, and methylation • Applicable to patients with unexplained clinical phenotypes, particularly in neurological and genetic disorders • Supports advanced genetic diagnosis as part of a comprehensive clinical evaluation		
Technology	NGS (Long-read sequencing, PacBio)	**Methods**	SMART sequencing
Specimen	EDTA WB 3.0 ml		
TAT	60 days		
Limitation	• Higher cost and longer turnaround time compared to short-read sequencing approaches • Not all detected variants may be clinically interpretable • Interpretation of complex genomic variants requires expert clinical and genetic correlation • Clinical utility may vary depending on phenotype, indication, and disease context		

Code OT038

Family Test

Targeted confirmation of a known familial genetic variant.



Sample Report
(including disease & gene)

Introduction	Family Test is a targeted genetic confirmation test designed to evaluate the presence or absence of a specific familial variant identified in a proband. Using targeted sequencing, the test supports segregation analysis within family members and helps clarify the inheritance pattern of a previously detected genetic variant. Key Result Components: Familial variant detection		
Indication	• Confirmation of a known pathogenic or likely pathogenic variant identified in an affected family member • Intended for family members of patients with a detected disease-causing genetic variant • Supports segregation analysis to assess variant inheritance within a family • Applicable as a follow-up test after WES, DGS, or other comprehensive genetic testing		
Technology	Sanger sequencing	**Methods**	Targeted Sanger Sequencing of Familial Variant
Specimen	EDTA WB 3.0 ml		
TAT	60 days		
Limitation	• Limited to analysis of the specific variant tested • Does not provide comprehensive or genome-wide variant analysis • Novel or additional variants outside the targeted region will not be detected • Results should be interpreted in conjunction with the proband's genetic findings and clinical information		



Linked-in

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