NGS Collaboration Proposal

for Korea Medical Group

LabGenomics Co., Ltd

Nov. 2019

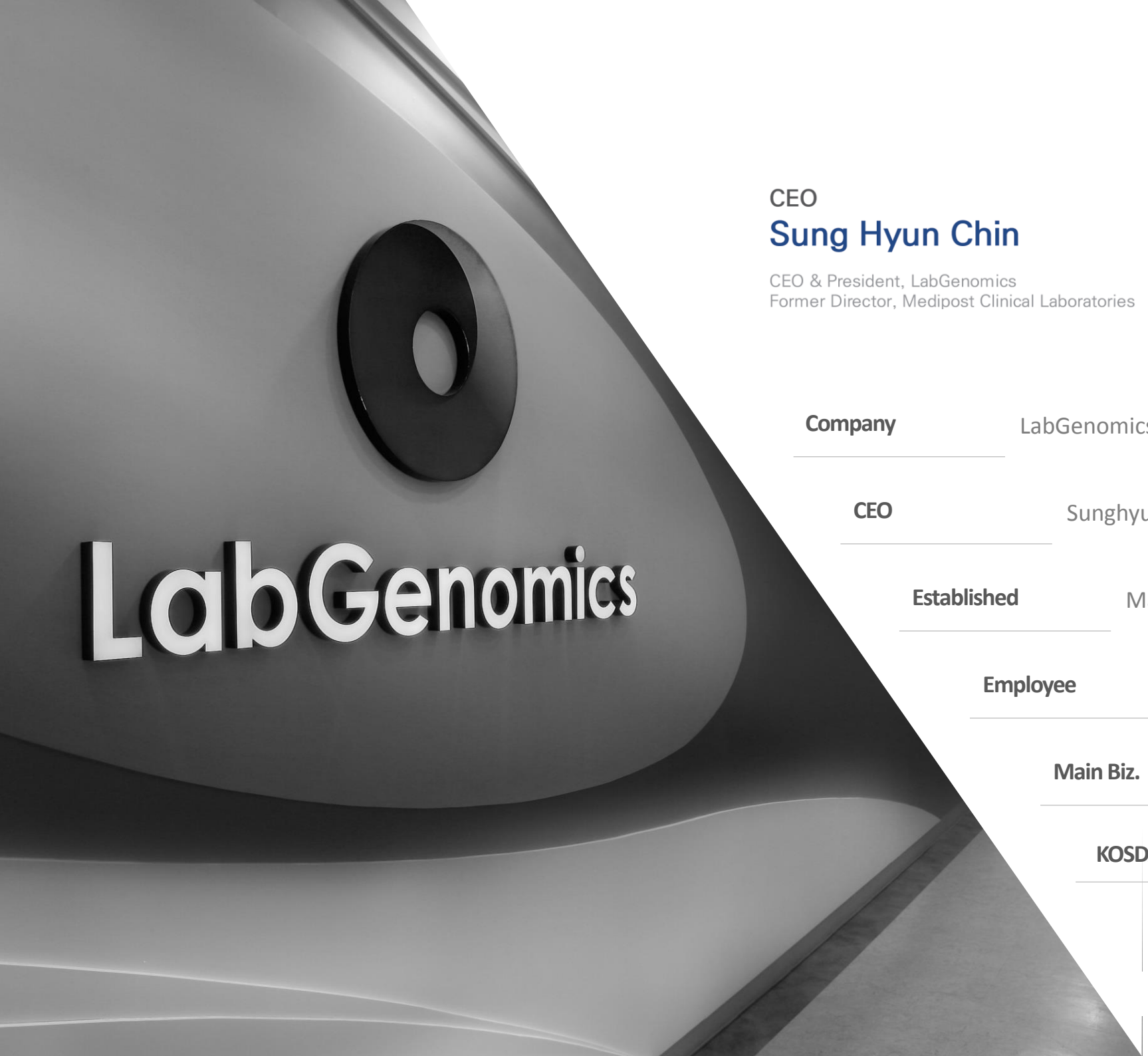
Who is LabGenomics

Introduction

Business Strategy

Network

Company Organization



CEO

Sung Hyun Chin

CEO & President, LabGenomics
Former Director, Medipost Clinical Laboratories

Company	LabGenomics Co.,Ltd
CEO	Sunghyun Chin
Established	March 2002
Employee	270 (As of 2019)
Main Biz.	IVD / PGS / NGS IVD Products
KOSDAQ	Listed on Korea Stock Market(Since 2014)

01 LabGenomics - healthcare company specialized in MDx

Quality of healthcare enhanced through innovation of MDx technology in a more accurate and efficient way

LabGenomics

Second-to-none in MDx

- Ranked No.1 in Korean MDx market
- Develop new MDx application by focusing on molecular & genetic research

Largest client network with hospitals

- More than 3,000 hospitals & clinics including 200 or more Maternity clinics
- Nation-wide sales networks. 20 branch offices across Korea

Realization of Personalized Medicine

- Early Detection & more accurate diagnosis
- Contribution to better life through high quality healthcare service

02 Robust Business & Medical Network

Research collaboration with global healthcare companies and major hospitals in Korea

Business Networks

- Genetic Analysis of LSD (Lysosomal Storage Diseases)
- Validation Study of Reagents
- ADHD Genetic Study
- Strategic Alliance in HealthCare Biz

Medical Networks

- SEOUL NATIONAL UNIVERSITY COLLEGE OF MEDICINE
- ASAN Medical Center
- SAMSUNG MEDICAL CENTER
- CHUNG-ANG UNIVERSITY HOSPITAL
- INJE UNIVERSITY PAIK HOSPITAL
- GANGNAM SEVERANCE HOSPITAL
- HANYANG UNIVERSITY MEDICAL CENTER
- KCCH
- 제일병원 CHEIL GENERAL HOSPITAL & WOMEN'S HEALTHCARE CENTER
- EMC 을지의료원 EULJI MEDICAL CENTER
- CHA Gangnam Medical Center, CHA University
- SCH SOON CHUN HYANG UNIVERSITY HOSPITAL SEOUL
- 아주대학교병원 Ajou University Hospital

03 The largest client network with hospitals (Domestic Site)

3,000 Hospitals

16 years

20 Branches

SEOUL NATIONAL UNIVERSITY COLLEGE OF MEDICINE

ASAN Medical Center

GIMS Gachon University Gil Medical Center International Medical Service

SAMSUNG MEDICAL CENTER

CHUNG-ANG UNIVERSITY HOSPITAL

INJE UNIVERSITY PAIK HOSPITAL

GANGNAM SEVERANCE HOSPITAL

HANYANG UNIVERSITY MEDICAL CENTER

SOON CHUN HYANG UNIVERSITY HOSPITAL SEOUL

CHA Gangnam Medical Center, CHA University

DONG-A UNIVERSITY HOSPITAL

04 International Network

Europe

Spain, Italy, Greece,

Middle East

UAE, Lebanon, Jordan,
Turkey

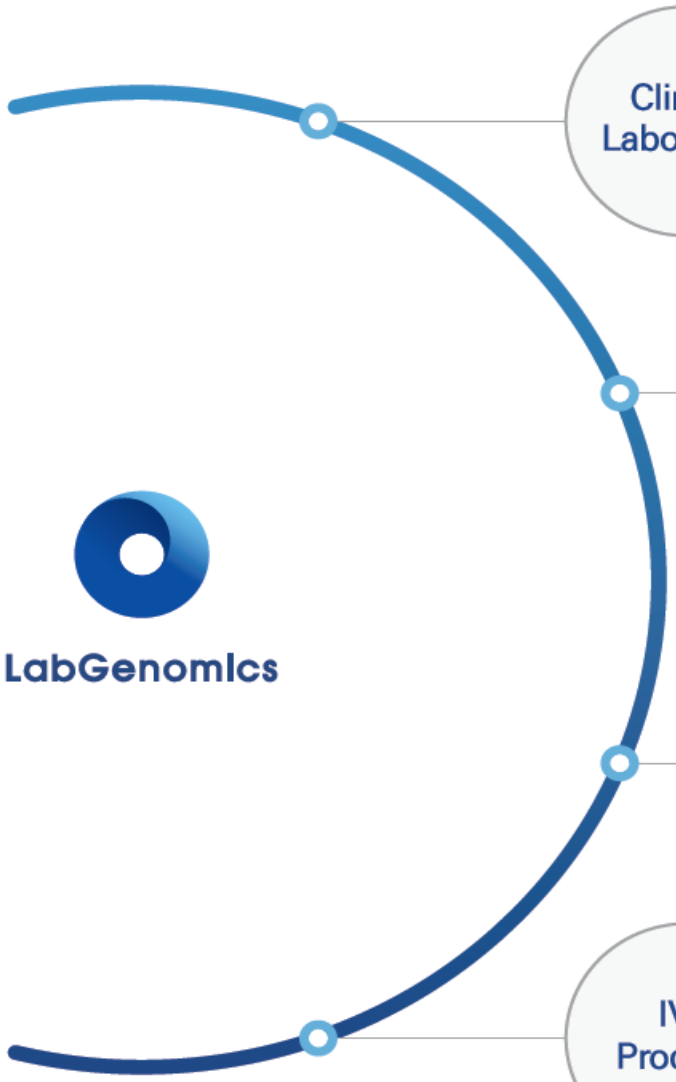
Asia

Vietnam, Thiland , China,

Korea

All over the country

05 Business Model



Clinical Laboratory

- Diagnostics Laboratory
- Molecular Diagnostics
- Pathology

NGS based diagnostics

- MomGuard(NIPT)
- EnfantGuard
- NobelGuard
- Cancer 4cast
- Cancer Scan

NGS Service (R&D)

- Whole Genome Sequencing (De *novo* Sequencing/Resequencing)
- Exome & Targeted Sequencing
- RNA sequencing
- Epigenome & Metagenome Sequencing

IVD Products

- PCR Kit (LabGun Sereis/LabGScan Series)
- DNA chip

Expansion into Dx products & NGS-based Services with higher growth potential

1. How it works

Genetic Service Workflow for 1st STEP

Patients Samples
(DNA or Buccal Swab(Epitherial tissue)

Overseas Area

Genetic Counseling

Sample Preparation

DNA Sequencing

Data Analysis(BI)

Korean Area

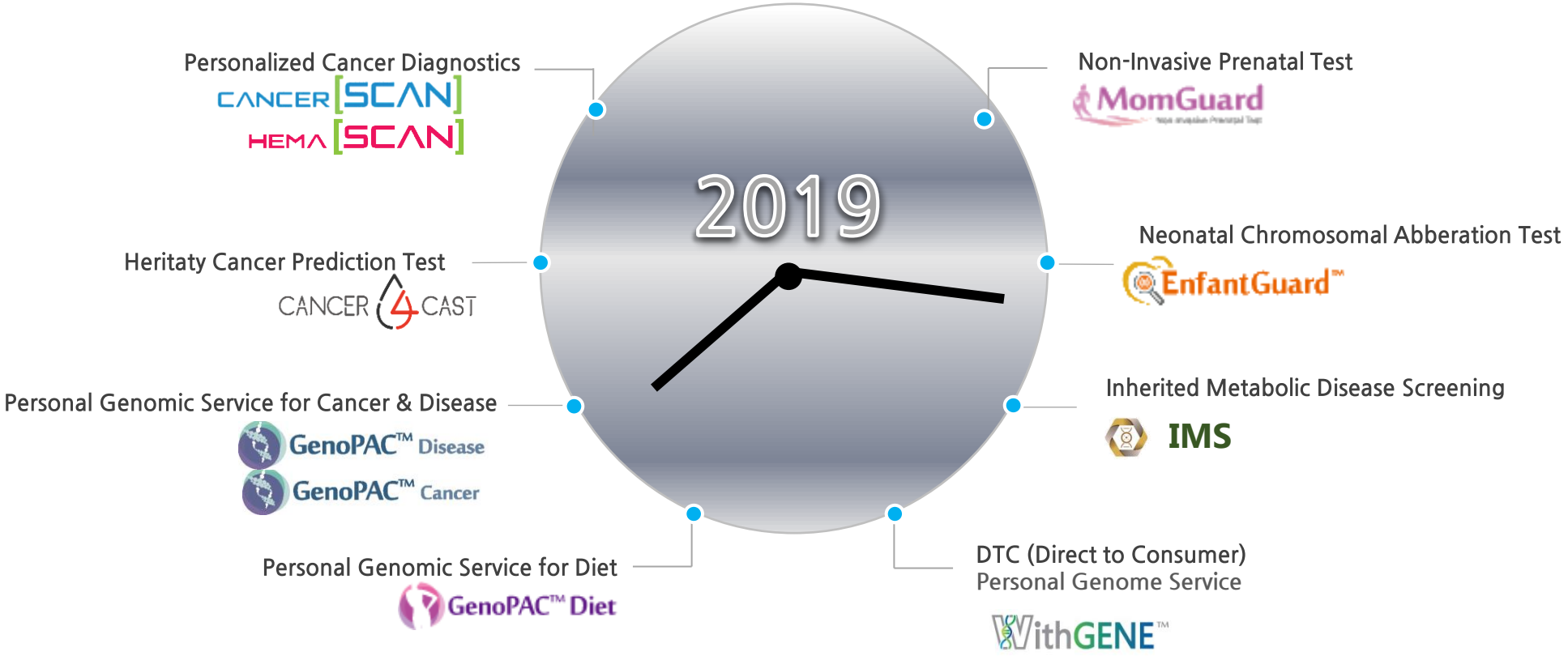
Test Report

2. NGS based Services

1) GenoPAC™ (Personal Genomic Service)
2) MomGuard™ (Non-Invasive Prenatal Test)
3) EnfantGuard™ (Newborn Screening Test)
4) BRCA 1,2 Test
5) CancerSCAN™ (Somatic Mutation Cancer Test)
6) Cancer4Cast™ (Hereditary Cancer Test)
7) IMS™ (Newborn/Child Inherited Metabolic Disease Test)

01 NGS Service Lineup

NGS Service Lineup through Whole Life Cycle



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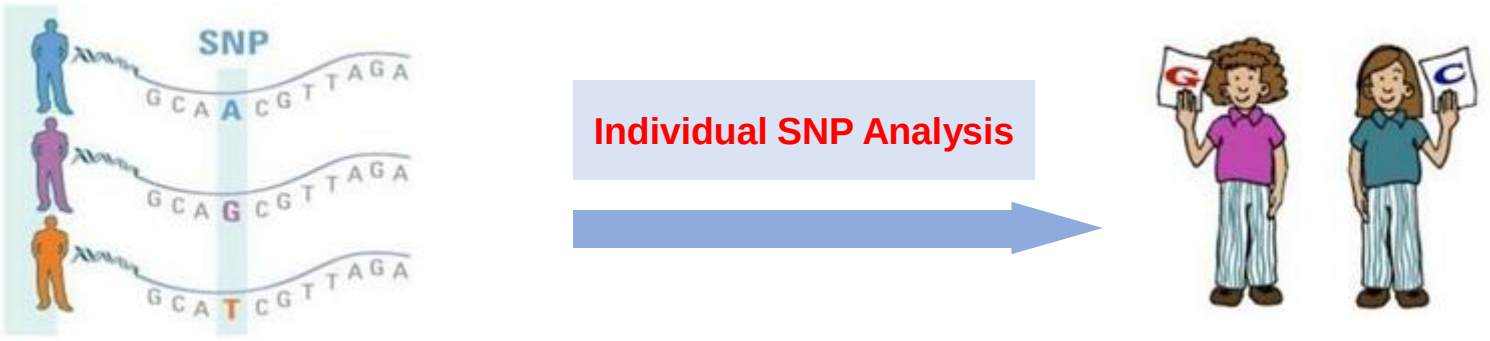
3. NGS based Services

1) GenoPAC™ (Personal Genomic Service)
2) MomGuard™ (Non-Invasive Prenatal Test)
3) EnfantGuard™ (Newborn Screening Test)
4) BRCA 1,2 Test
5) CancerSCAN™ (Somatic Mutation Cancer Test)
6) Cancer4Cast™ (Hereditary Cancer Test)
7) IMS™ (Newborn/Child Inherited Metabolic Disease Test)

1) GenoPAC™ Cancer Disease Personal Genomic Service



1. Individual with family history

In case family members are concerned about the high incidence of cancer or heredity disease you can be tested together, checked for risks and managed early.

2. Individual seeks for health checkup

A health checkup conducted at a hospital is a test to diagnose current problems on the blood by collecting blood. GenoPAC testing analyzes the unchanging genes from birth to predict cancers or diseases that
are genetically at risk, even if there are no current problems. Therefore, it is another form of healt h checkup in the sense that it confirms the genetic health that was born and not the present con dition.

3. Anyone interested in health care

If you are concerned about your health and wish to effectively manage your lifestyle, you can us e your personalized genetic tests to focus more on the areas of risk.

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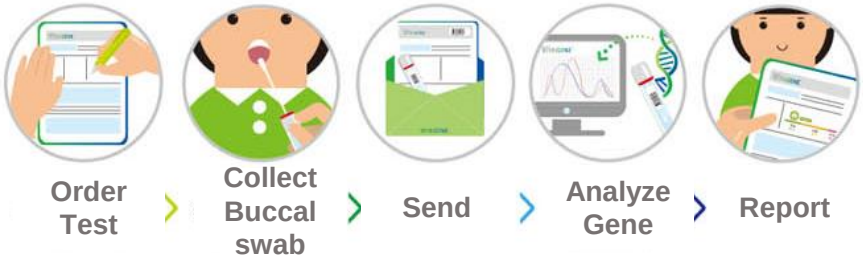
1) GenoPAC™ Cancer Disease Personal Genomic Service

Required Sample *: Buccal Swap*



Service Cancer/Disease LIST

GenoPAC Male Service

Cancer 11types	Disease 15types
Thyroid cancer	Hypertension
Esophageal cancer	Brain Aneurysm
Liver cancer	Stroke
Gastric cancer	Myocardial infarction
Colorectal cancer	Atrial fibrillation
Bladder cancer	Obesity
Renal cancer	Osteoarthritis
Lung cancer	Osteoporosis
Pancreatic cancer	Type II diabetes mellirus
Prostate cancer	Asthma
Testicular cancer	Chronic obstructive pulmonary disease
	Alzheimers disease
	Parkinson's disease
	Migraine
	Depression

GenoPAC Female Service

Cancer 12types	Disease 15types
Thyroid cancer	Hypertension
Esophageal cancer	Brain Aneurysm
Liver cancer	Stroke
Gastric cancer	Myocardial infarction
Colorectal cancer	Atrial fibrillation
Bladder cancer	Obesity
Renal cancer	Osteoarthritis
Lung cancer	Osteoporosis
Pancreatic cancer	Type II diabetes mellirus
Brest cancer	Asthma
Ovarian cancer	Chronic obstructive pulmonary disease
Endometrial cancer	Alzheimers disease
	Parkinson's disease
	Migraine
	Depression

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1) GenoPAC™ Cancer Disease Personal Genomic Service

Report Sample

GenoPAC™

Cancer/Disease Report

[Total / Cancer 11 + Disease 15]

Collection	년	월	일
Submitted	년	월	일
Report	년	월	일

검체 접수번호 Sample ID	Sample	의뢰기관명 Institution	Sample
담당의사명 Name of Doctor		차트번호 Chart No.	
소속/연락처 Dep./Phone No.		검사대상자명 Name of Patient	
주소 Address		생년월일 Date of Birth	

검사정도관리결과 Quality Control

DNA 정도관리 DNA quality	유전자형 정도관리 Genotyping quality	분석 정도관리 Analysis quality
Pass	Pass	Pass

요약결과 Result Summary

Disease	Gene	Risk Allele	Ref. Allele	Your Allele	Result	Relative Risk
Liver Cancer	STAT4	G	T	G G	High Risk	2.01
	HLA-DRB1 - LOC107986589	A	G	A A		
	MTCO3P1 - LOC102725019	A	G	A G		
Pancreatic cancer	LOC105370243	G	T	G G	High Risk	1.76
	LINC01394 - LOC105374880	C	T	C C		
Prostate cancer	CASC8, CCAT2	G	T	G G	High Risk	1.9
	RFX6	T	C	T T		
Testis cancer	BAK1	G	A	A G	High Risk	1.56
	KITLG	G	A	G G		
Colorectal cancer	C5orf66	A	C	C A	Average Risk	1.18
	MYRF	G	T	T T		
	CASC8, CCAT2	G	T	G G		
Renal cell carcinoma	SCARB1	T	C	C T	Average Risk	1.19
	LOC105369705	G	A	G G		
Thyroid cancer	PTCSC3 - RN7SKP21	T	C	C C	Low Risk	0.76
	DIRC3	T	C	T T		
Gastric cancer	PSCA	T	C	C C	Low Risk	0.47
	PRKAA1	C	T	T T		
Bladder cancer	CASC11	T	G	G G	Low Risk	0.65

LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23-2 Genetic Research Laboratory: No. 7-2

Inspector : MJ OH Ph.D. Laboratory Director : SY KIM M.D., Ph.D.

Analysis Officer : HJ HU Ph.D. Laboratory Officer : DY CHO Ph.D.

www.labgenomics.co.kr

GenoPAC™

Sample ID 2018-04-22 99913

의뢰기관명 Institution	검사대상자명 Name of Patient	차트번호 Chart No.
Sample	Sample	Sample

요약결과 Result Summary

Disease	Gene	Risk Allele	Ref. Allele	Your Allele	Result	Relative Risk
Bladder Cancer	C20orf187	A	C	C C	Low Risk	0.65
Esophageal Cancer	HECTD4	A	G	G G	Low Risk	0.22
	ALS2CR12	A	G	G G		
Lung Cancer	TERT	G	A	A A	Low Risk	0.57
	BPTF	A	G	G A		
Cerebral aneurysm	CDKN2B-AS1	T	C	T T	High Risk	1.43
	RP1	A	G	A A		
Osteoarthritis	LOC101929163, BTNL2	T	C	C T	High Risk	1.45
	ALDH1A2	C	G	G C		
Migraine	PRDM16, LOC105378606	C	T	T C	High Risk	2.06
	MEF2D	C	T	T T		
Obesity	FTO	A	T	T T	Average Risk	0.84
	MC4R	C	T	T T		
	CLOCK	A	G	A A		
Hypertension	UMOD	A	G	A A	Average Risk	0.81
	FGF5	C	T	T T		
Stroke	PITX2 - MIR297	G	A	G A	Average Risk	0.99
	SPSB4	G	A	G G		
Atrial fibrillation	ZFHX3	T	C	C T	Average Risk	1.22
	PITX2 - MIR297	C	T	T C		
Chronic obstructive pulmonary disease	KRT18P51 - HHIP-AS1	T	C	T T	Average Risk	0.77
	FAM13A, LOC105377327	C	T	T T		
Asthma	HLA-DQB1	T	C	T C	Average Risk	1.03
	IKZF4	G	T	T G		
Osteoporosis	WHSC1L2P - SOST	T	C	T T	Average Risk	0.76
	MECOM	A	G	G G		

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omics

1) GenoPAC™ Cancer Disease Personal Genomic Service

Report Sample

GenoPAC™

Registration No. 2018-04-22 99913

의뢰기관명 Institution	검사대상자명 Name of Patient	차트번호 Chart No.
Sample	Sample	Sample

요약결과 Result Summary

Disease	Gene	Risk Allele	Ref. Allele	Your Allele	Result	Relative Risk
Alzheimer's disease	CLU	T	C	C T	Average Risk	1.08
	SORL1	T	C	T T		
Depressive disorder	CNTN5	C	T	C T	Average Risk	0.92
	KSR2	A	G	G G		
Diabetes mellitus type 2	LOC105375716, SLC30A8	C	T	C T	Low Risk	0.67
	KCNQ1	C	T	T T		
Myocardial infarction	AP3D1 - DOT1L	C	A	C A	Low Risk	0.81
	PLCL2	G	A	A G		
Parkinson's disease	MCCC1	G	A	G A	Low Risk	0.64
	HLA-DRA	G	A	A A		

LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23-2 | Genetic Research Laboratory: No. 7-2

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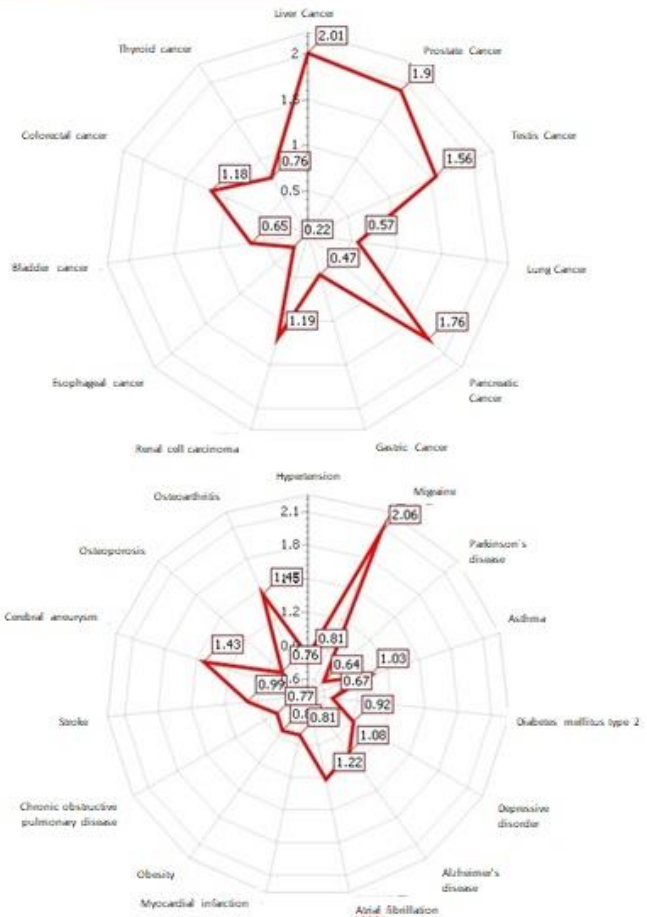
GenoPAC™

Registration No. 2018-04-22 99913

의뢰기관명 Institution	검사대상자명 Name of Patient	차트번호 Chart No.
Sample	Sample	Sample

요약결과 Result Summary



LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23-2 | Genetic Research Laboratory: No. 7-2

Inspector : MJ OH Ph.D. Laboratory Director : SY KIM M.D., Ph.D.

Analysis Officer : HJ HU Ph.D. Laboratory Officer : DY CHO Ph.D.

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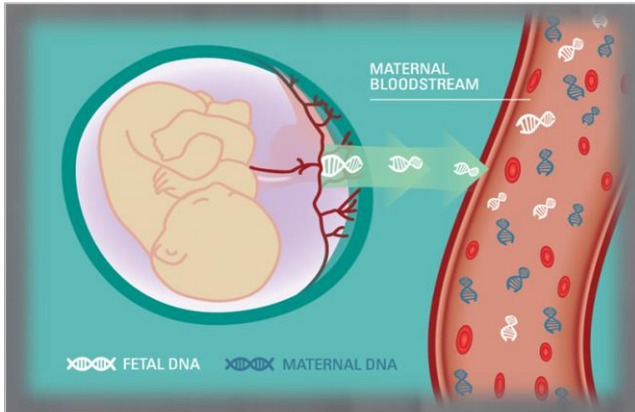
2) MomGuard™ Service (Non-Invasive Prenatal Test)

Provides an individualized assessment for the most common fetal aneuploidies and replaces conventional tests, quad screen, 1st-trimester screen and integrated screening.

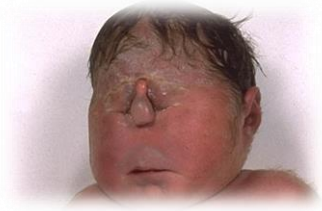


Cell Free DNA from the mother and fetus flows together in the maternal bloodstream

Focuses on cfDNA from the chromosomes of interest and this allows deeper analysis and ultimately yields more accurate results.

For most common chromosomal disorders:

Trisomy 21,
Down Syndrome



Trisomy 18,
Edwards Syndrome

Trisomy 13,
Patau Syndrome

LabGenomics

2) MomGuard™ Service (Non-Invasive Prenatal Test)

Type of Test	Test Items
MomGuard™ Standard (13990)	▪ T 21 / T 18 / T 13 ▪ Sex aneulpoidy(Tuner, Klinefelter, XXX syndromes) with fetal sex *(* In the case of twin, Sex anueploidy is not available.)*
MomGuard™ Lite (13998)	▪ T 21 / T 18 / T 13 only. ▪ ~~Sex aneulpoidy(Tuner, Klinefelter, XXX syndromes) with fetal sex~~
MomGuard™ Premium -Single (13996)	▪ T 21 / T 18 / T 13 / T9 / T16 / T22 ▪ Sex aneulpoidy(Tuner, Klinefelter, XXX syndromes) with fetal sex ▪ Microdeletion (Cri-du-chat, 1p36 deletion, DiGeorge, Jacobsen , Prader-Willi, Angelman)
MomGuard ™ Premium -Twin (13997)	▪ T 21 / T 18 / T 13 / T9 / T16 / T22 ▪ ~~Sex aneulpoidy(Tuner, Klinefelter, XXX syndromes) with fetal sex~~ ~~▪ Microdeletion (Cri-du-chat, 1p36 deletion, DiGeorge, Jacobsen , Prader-Willi, Angelman)~~

LabGenomics

2) MomGuard™ Service (Non-Invasive Prenatal Test)

Report Sample

MomGuard™
Non Invasive Prenatal Test

Standard Test Report

Registration No.	20180810-59002
Collection Date	2018-08-06
Receipt Date	2018-08-10
Analysis Date	2018-08-13
Report Date	2018-08-16

Department :	/	Organization :	Sample
Doctor :		Patient :	Sample
Specimen :	Whole blood	Chart No.:	
Address :		DOB :	1975-06-05
		Age/Gender :	43 / F

QR code

Patient Information				
	Number of Fetus	Single	Ultrasonographic Finding	
	Ultrasound Gestational Age	10W 1D	Prenatal Biochemical Screening findings	
	Height/Weight	165 cm / 65 kg		

Quality Control / Test Result

Cell free DNA quality	Sequencing quality	Fetal fraction	Standard Material Test result
Pass	Pass	Pass(15.28%)	Pass

INTERPRETATION

Low Risk - The chance of the baby having a chromosomal abnormality is very low.

RESULT DETAILS

Chromosome Aneuploidies		
Items (Disease Type)	Result	Risk Score
Trisomy 21 (Down Syndrome)	Low Risk	1/12435
Trisomy 18 (Edward Syndrome)	Low Risk	1/38045
Trisomy 13 (Patau Syndrome)	Low Risk	1/12737
Sex aneuploidy (mX, XXY, XXX)	Low Risk	1/5557, Male

Test Method

·cfDNA is isolated from maternal blood and sequenced using Next Generation Sequencing (NGS) technology. Sequencing data is analyzed using in-house bioinformatics pipeline to identify fetal aneuploidy in the tested chromosomes.
·This test provides a result only when a sample meets the quality threshold.

Test Purpose & Limitation

·This test is a screening test for T21, T18, T13 and sex chromosome aneuploidy under the consent of the mother.
·No analysis for sex chromosome aneuploidy is to be performed in case of twin.
·This test is highly accurate, but not diagnostic. Therefore, if a confirmatory test is required according to the test result or the clinical situation of the mother, an amniocentesis or CVS should be performed.

MomGuard Performance Table

	Sensitivity	Specificity
Chromosome 21	98.65%	99.94%
Chromosome 18	100%	99.98%
Chromosome 13	100%	99.97%

· Overall PPV = 0.8932, NPV=0.9999
· Overall False Positive (FP) % = 0.102
· Overall False Negative (FN) % = 0.009

LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23 | Genetic Research Laboratory: No. 7

Inspector : MJ OH Ph.D. Laboratory Director : SY KIM M.D, Ph.D.
Analysis Officer : HJ HU Ph.D. Laboratory Officer : DY CHO Ph.D.

LGT J101 1(E) Rev(2) 18.07.01

MomGuard™ + PLUS
Non Invasive Prenatal Test

Test Report

Registration No.	E123456789
Collection Date	2018 Y 05 M 13 D
Receipt Date	2018 Y 05 M 20 D
Analysis Date	2018 Y 05 M 22 D
Report Date	2018 Y 05 M 23 D

Department :	/	Organization :	Sample
Doctor :		Patient :	Sample
Sample ID:		Chart No.:	
Address :		DOB :	1977.06.25
		AGE/Gender :	40 / F

QR code

Patient Information				
	Number of Fetus	SINGLE	Ultrasonographic Finding	
	Ultrasound Gestational Age	15 W 4 D	Prenatal Biochemical Screening findings	
	Height/Weight	163 cm / 83 kg		

Quality Control / Test Result

Cell free DNA quality	Sequencing quality	Fetal fraction	Standard Material Test result
Pass	*Pass*	*Pass (12.06 %)*	*Pass*

INTERPRETATION

Trisomy 21 (Down Syndrome) : High Risk
Increased risk of Trisomy 21. Genetic counseling and confirmatory testing is required.

Chromosome Aneuploidies		
Tested condition	Result	Risk Score
Trisomy 21 (Down Syndrome)	High Risk	> 9/10
Trisomy 18 (Edward Syndrome)	Low Risk	<1/12,300
Trisomy 13 (Patau Syndrome)	Low Risk	<1/11,435
Trisomy 9	Low Risk	<1/10,000
Trisomy 16	Low Risk	<1/10,000
Trisomy 22	Low Risk	<1/10,000
Sex aneuploidy (mX, XXY, XXX)	Low Risk	<1/10,000

Chromosome Microdeletions	
Tested condition	Result
Cri-du-chat	Low Risk
1p36 deletion	Low Risk
DiGeorge(22q11.2 deletion)	Low Risk
Jacobsen	Low Risk
Prader-Willi/Angelman	Low Risk

Test Method

· cfDNA is isolated from maternal blood and sequenced using Next Generation Sequencing (NGS) technology. Sequencing data is analyzed using in-house bioinformatics pipeline to identify fetal aneuploidy in the tested chromosomes.
· This test provides a result only when a sample meets the quality threshold.

Test Purpose & Limitation

· This test is a screening test for T21, T18, and T13 with the following options under the consent of the mother: additional chromosomal aneuploidies (T9, T16, T22), sex chromosome aneuploidy and selected microdeletion syndromes.

· This test is highly accurate, but not diagnostic. Therefore, if a confirmatory test is required according to the test result or the clinical situation of the mother, an amniocentesis or CVS should be performed.

MomGuard Performance Table

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Chromosome 21	98.65%	99.94%
Chromosome 18	100%	99.98%
Chromosome 13	100%	99.97%

• Overall PPV = 0.8932, NPV=0.9999
• Overall False Positive (FP) % = 0.103
• Overall False Negative (FN) % = 0.009

LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23-2 | Genetic Research Laboratory: No. 7-2

Inspector : MJ OH Ph.D. Laboratory Director : SY KIM M.D., Ph.D.
Analysis Officer : HJ HU Ph.D. Laboratory Officer : DY CHO Ph.D.

LGT_J101 Rev(2) 18.05.01

enomics

3) EnfantGuard™ (Newborn Screening Test)

A Very effective test that can detect the presence of chromosomal abnormalities related to the developmental disorder of the newborn.

Ex) Down syndrome

Newborn screening test analyzes more than six million nucleotide sequences, enabling more precise detection of microdeletions or microduplications in specific areas associated with approximately **250 developmental disorders.**

Overall Process

Sample → Library → Sequencing → GeneBro® Bioinformatic Analysis → Result Delivery

- *CNABro®: EnfantGuard™ BI platform*

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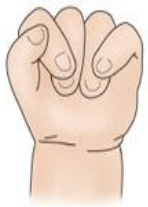
3) EnfantGuard™ (Newborn Screening Test)

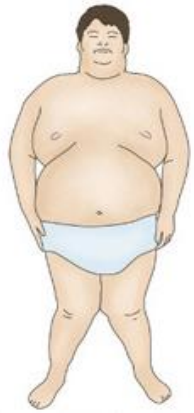
√ **Who needs this?**

: Newborn baby

: Child and adult willing to know about their chromosomal disorder







√ **Advantages**

: Quick and Safe

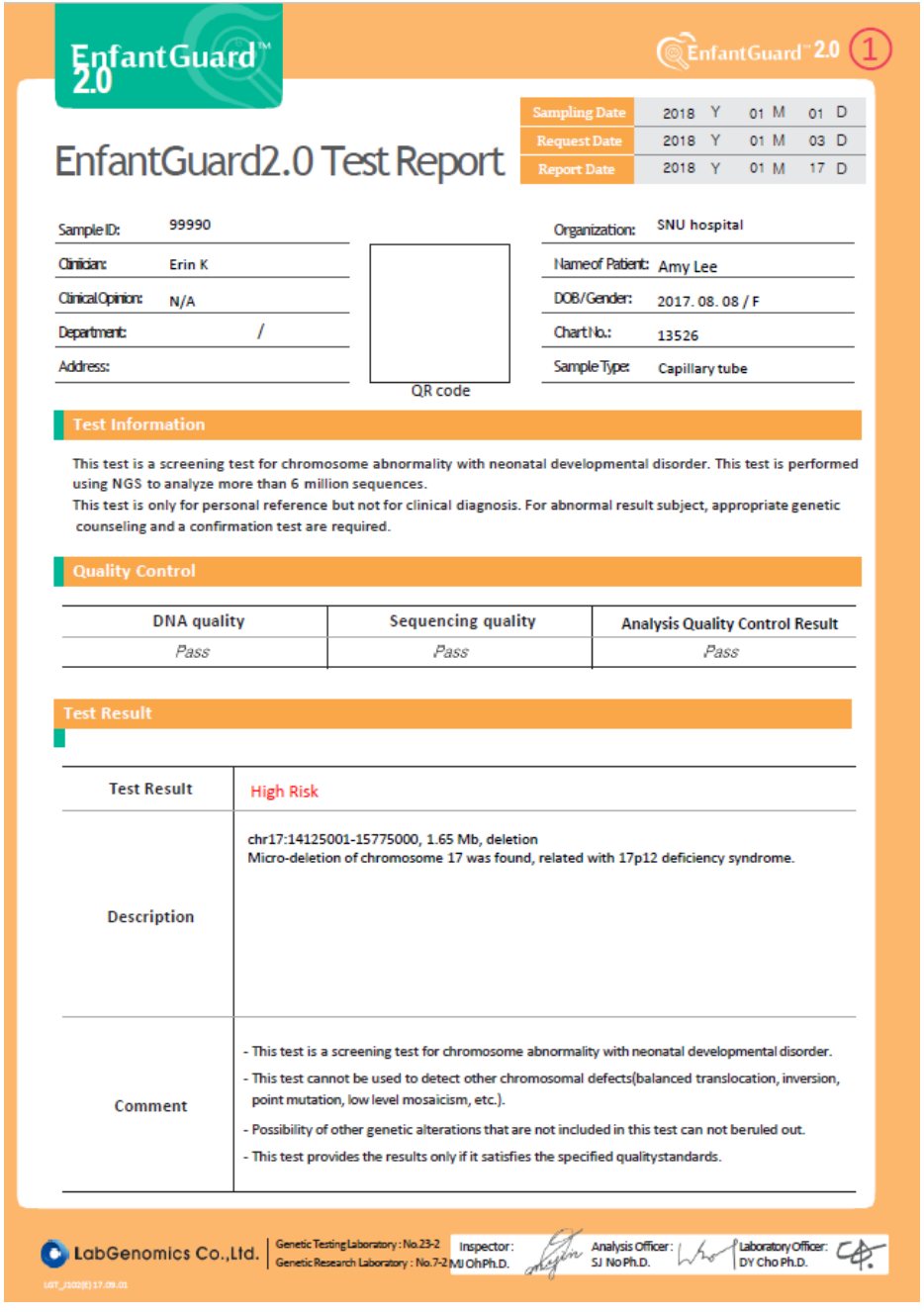
: High Accuracy

: Early Detection & Treatment

LabGenomics

3) EnfantGuard™ (Newborn Screening Test)

Report Sample



EnfantGuard 2.0 ①

EnfantGuard2.0 Test Report

Sampling Date	2018	Y	01	M	01	D
Request Date	2018	Y	01	M	03	D
Report Date	2018	Y	01	M	17	D

Sample ID:	99990	Organization:	SNU hospital
Clinician:	Erin K	Name of Patient:	Amy Lee
Clinical Opinion:	N/A	DOB/Gender:	2017. 08. 08 / F
Department:	/	Chart No.:	13526
Address:		Sample Type:	Capillary tube

QR code

Test Information

This test is a screening test for chromosome abnormality with neonatal developmental disorder. This test is performed using NGS to analyze more than 6 million sequences.
This test is only for personal reference but not for clinical diagnosis. For abnormal result subject, appropriate genetic counseling and a confirmation test are required.

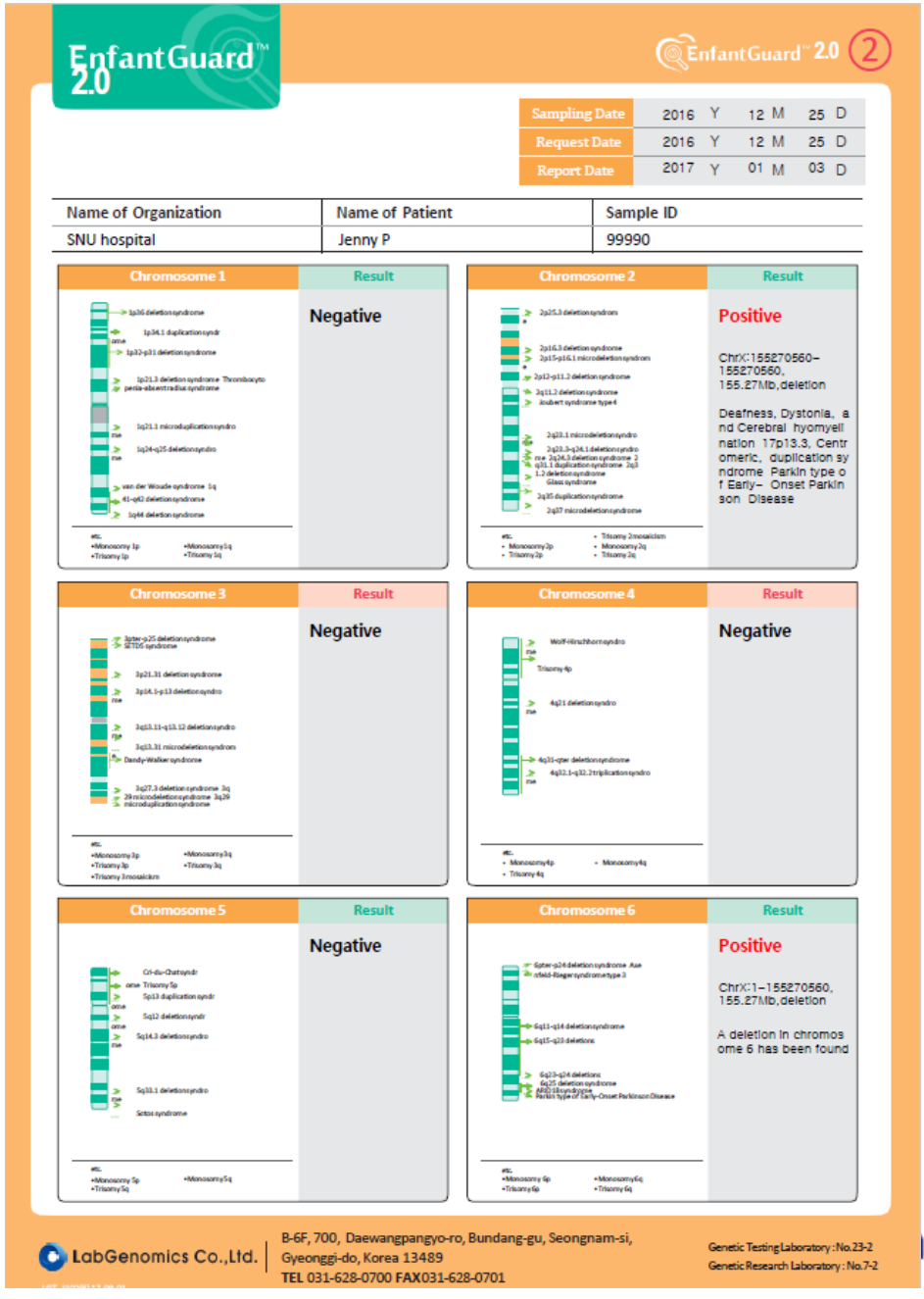
Quality Control

DNA quality	Sequencing quality	Analysis Quality Control Result
Pass	*Pass*	*Pass*

Test Result

Test Result	High Risk
Description	chr17:14125001-15775000, 1.65 Mb, deletion Micro-deletion of chromosome 17 was found, related with 17p12 deficiency syndrome.
Comment	- This test is a screening test for chromosome abnormality with neonatal developmental disorder. - This test cannot be used to detect other chromosomal defects(balanced translocation, inversion, point mutation, low level mosaicism, etc.). - Possibility of other genetic alterations that are not included in this test can not beruled out. - This test provides the results only if it satisfies the specified qualitystandards.

LabGenomics Co.,Ltd. | Genetic Testing Laboratory : No.23-2 Genetic Research Laboratory : No.7-2 | Inspector: MJ Oh Ph.D. | Analysis Officer: SJ No Ph.D. | Laboratory Officer: DY Cho Ph.D.



EnfantGuard 2.0 ②

Sampling Date	2016	Y	12	M	25	D
Request Date	2016	Y	12	M	25	D
Report Date	2017	Y	01	M	03	D

Name of Organization	Name of Patient	Sample ID
SNU hospital	Jenny P	99990

Chromosome	Result
Chromosome 1	Negative
Chromosome 2	Positive ChrX:155270560–155270560, 155.27Mb,deletion Deafness, Dystonia, and Cerebral hyomyelination 17p13.3, Centromeric, duplication syndrome Parkin type of Early–Onset Parkinson Disease
Chromosome 3	Negative
Chromosome 4	Negative
Chromosome 5	Negative
Chromosome 6	Positive ChrX:1–155270560, 155.27Mb,deletion A deletion in chromosome 6 has been found

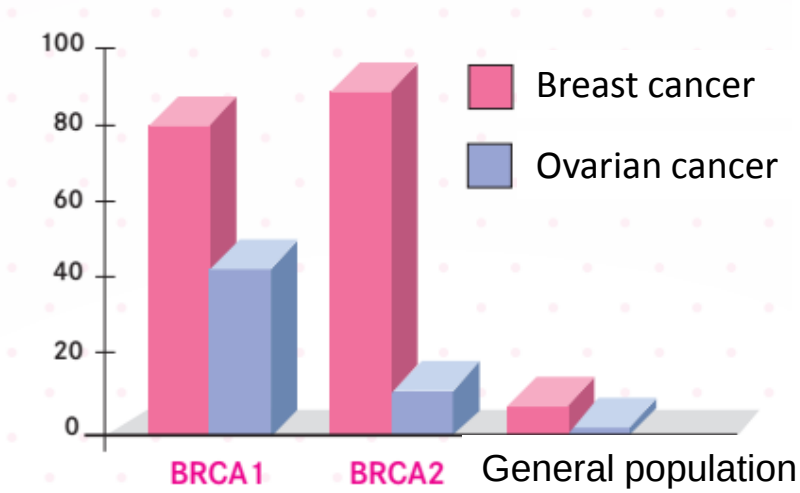
LabGenomics Co.,Ltd. | B-6F, 700, Daewangpangyo-ro, Bundang-gu, Seongnam-si, Gyeonggi-do, Korea 13489 TEL 031-628-0700 FAX031-628-0701 | Genetic Testing Laboratory : No.23-2 Genetic Research Laboratory : No.7-2

omics

4) BRCA 1,2 Test

NGS-based Panel

Cancer risk with BRCA1,2 mutation



Highly reliable sequence analysis

- Analysis by ACMG guideline
- Analysis based on reliable public DB and vast in-house variant DB
- Analysis of Review board consisting of experts
- In-house LIMS Review board System

Test	sample	Test method	TAT
BRCA1,2	Whole blood 3 ml	PCR & NGS	4 Weeks

Angelina Effect

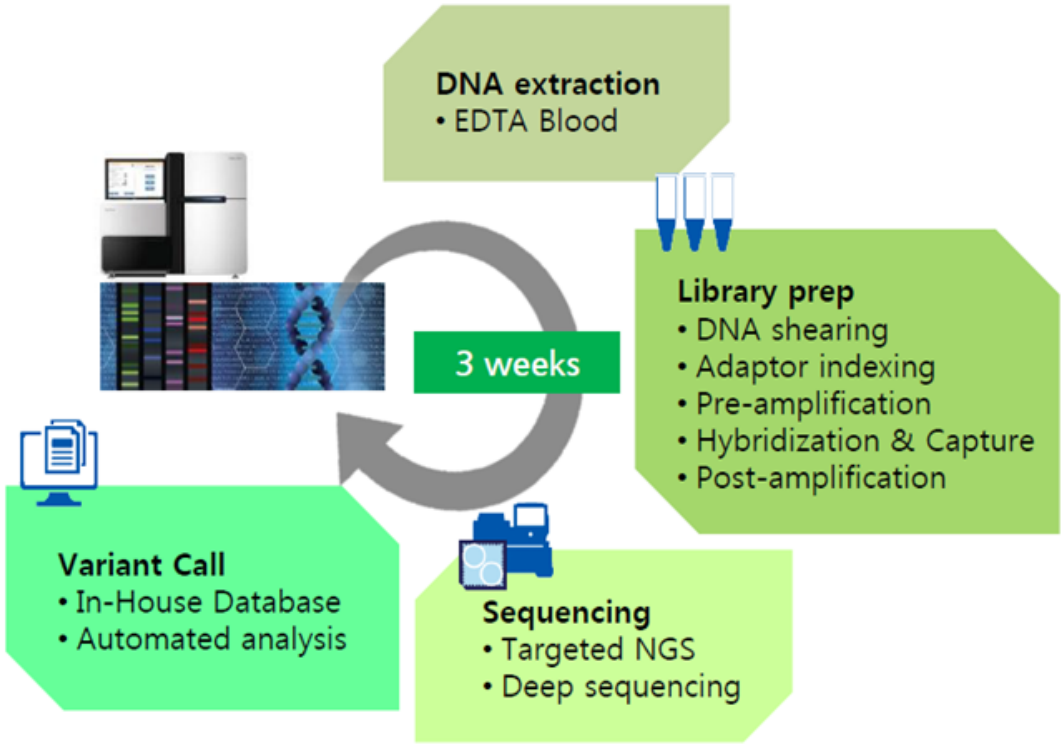
“ My doctors estimated that I had an 87% risk of breast cancer and a 50% risk of ovarian cancer.”

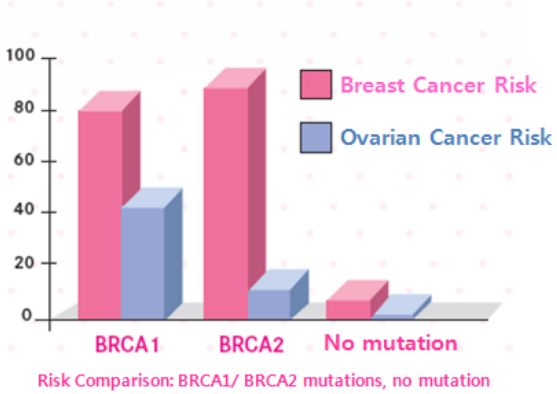
Tests for the breast cancer BRCA gene shot up by 64 percent following Jolie’s 2013 New York Times op-ed about her decision to have preventive mastectomy after genetic testing that revealed she carried the disease-fueling mutation. Test increases, with each test priced at approximately $3,000, are estimated to have cost the U.S. health care system at least $13.5 million in the two weeks following the disclosure. Increased testing rates were not accompanied by a corresponding increase in mastectomy rates, suggesting additional testing did not identify new BRCA mutations.

LabGenomics

4) BRCA 1,2 Test

The entire gene coding regions, as well as all flanking noncoding regions including ±25bp, of the BRCA1 and BRCA2 genes is analyzed by *NGS (next generation sequencing)* technology.





Risk Comparison: BRCA1/ BRCA2 mutations, no mutation

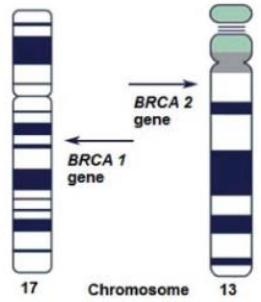


Figure 1:

The BRCA1 and BRCA2 genes in the corresponding chromosomes.

LabGenomics

4) BRCA 1,2 Test

Report Sample

BRCA1/BRCA2
GENETIC TEST REPORT

Collection Date	
Receipt Date	
Report Date	

Organization		SPECIMEN	
Name	SAMPLE	Specimen #	
Address		Specimen Type	

PHYSICIAN		PATIENT	
Name	SAMPLE	Name	
Contact Info		Date of Birth /Gender	
Clinical comment		Medical Chart #	

SUMMARY RESULT

[A p|P]athogenic variant[|s] [was|were] detected in the [BRCA2 gene|BRCA1 gene|BRCA1 and BRCA2 genes]

[VUS]
X variant[s] of uncertain significance (VUS) [was|were] detected.

[Comment]
This variant[s] [is|are] clinically significant and [is|are] associated with an increased cancer risk. In this case, genetic counseling and a BRCA1,2 genetic test of your family members are recommend. This result does not mean that you have a diagnosis of cancer or that you will definitely develop cancer in your lifetime. Your actual risk may be different based on other genetic and non-genetic factors.

LabGenomics Clinical Laboratories
Institution number : 41355709
Director of NGS Laboratory: Mijin Oh Ph.D.
Laboratory Medical Director: Dong Hee Seo M.D.

BRCA1/BRCA2
GENETIC TEST REPORT

Collection Date	
Receipt Date	
Report Date	

DETAILED RESULT

[Pathogenic variant]

Gene	Classification	Zygosity	Variant Detected	Amino Acid Change	dbSNP
BRCA2	Pathogenic	Hetero	c.5074G>C	p.Asp1692His	rs80187739

A pathogenic variant ([homozygous | heterozygous] c.XXXXXX (p.XxxXXXXXxx)) was detected in the [exon X| intron] of the BRCA2 gene ([NM_000059|NM_007294]).

This [nonsense | misssense | silent | insertion | deletion | framshift] mutation has been previously reported [once|twice|X times] in our database before (NM_00XXXX). Variants detected within each gene are reported and classified according to our internal criteria with reference to ACMG guidelines

[VUS]

Gene	Classification	Zygosity	Variant Detected	Amino Acid Change	dbSNP
BRCA2	VUS	Hetero	c.29C>A	p.Thr10Lys	rs1057519494

X VUS (BRCAX c.XXXXXX (p.XxxXXXxx); BRCAX c.XXXXXX (p.XxxXXXxx)) with unknown relationship with disease [was|were] detected

In addition, X benign variant[s] which [has|have] no impact on health [was|were] detected in the [BRCA1 gene|BRCA2 gene|BRCA1,2 genes].

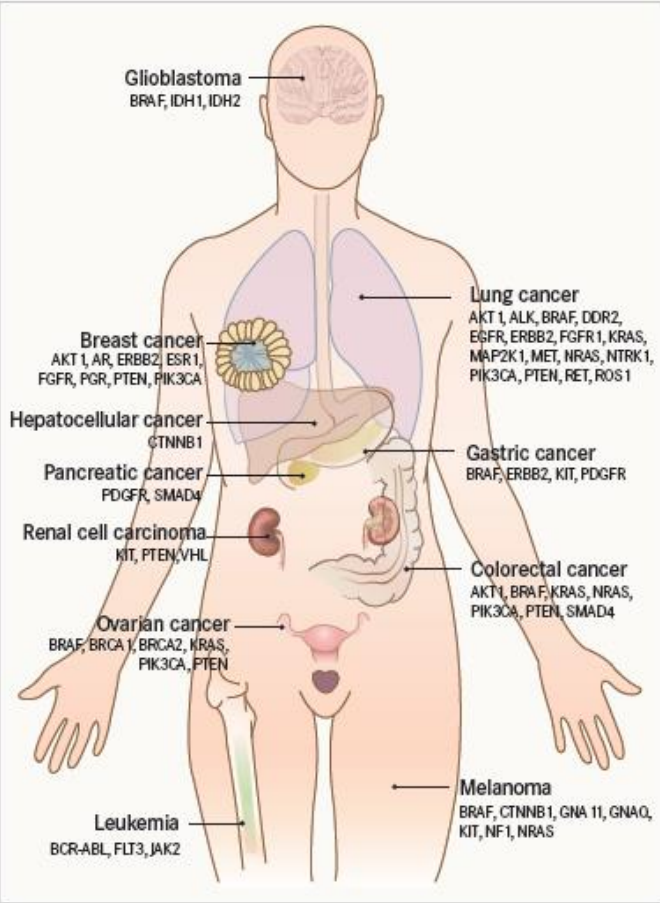
LabGenomics Clinical Laboratories
Institution number : 41355709
700 Daewangpangyo-ro, Korea Bio Park Bldg. 6F-B, Bundang-gu, Seongnam-si, Gyeonggi-do 463-400, Republic of Korea
Tel: +82-31-628-0702 Fax: +82-31-628-0701 e-mail: info@labgenomics.com

enomics

5) CancerSCAN™ (Somatic Mutation Cancer Test)

- Provides the cancer-related genetic variations with high precision using NGS technology
- Confirm the various genetic variations at once with a small amount of patient's specimen
- Targeted Anti-cancer drug information is provided.

Cancer genes & targeted agents



Gene	Drug
ALK	Crizotinib, Ceritinib
EGFR	Gefitinib Erlotinib Afatinib Osimertinib
BRAF	Dabrafenib, Trametinib, Vemurafenib
KRAS	Cetuximab, Panitumumab
ERBB2	Trastuzumab Lapatinib Neratinib

NGS-based personalized cancer treatment

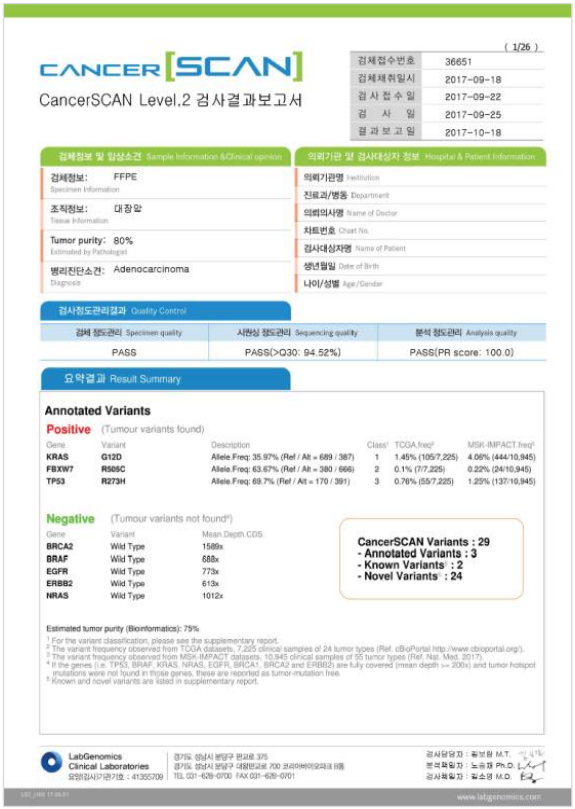
- Personalized anti-cancer treatment through analysis of patients' genetic information
- To design guidelines for cancer therapy by analyzing 77 genes (level 1) or 375 genes (level 2) as bio-markers
- Optimized to characterize SNVs, Indels, CNVs and selected gene fusions across tumor related marker genes
- Collaboration with Samsung Medical Center in R&D sponsored by Korean government
- Applied and validated to more than 7,000 Korean solid tumor patients

LabGenomics

5) CancerSCAN™ (Somatic Mutation Cancer Test)

Cancer Scan Report Example

1 Summarized Report


CANCER [SCAN]

CancerSCAN Level.2 검사결과보고서

(1/26)

검체접수번호	36651
검체채취일시	2017-09-18
검 사 접 수 일	2017-09-22
검 사 일	2017-09-25
결 과 보 고 일	2017-10-18

검체정보 및 임상소견 Sample Information &Clinical opinion

검체정보:	FFPE
조직정보:	대장암
Tumor purity:	80%
병리진단소견:	Adenocarcinoma

의뢰기관 및 검사대상자 정보 Hospital & Patient Information

의뢰기관명 Institution	
진료과/병동 Department	
의뢰의사명 Name of Doctor	
차트번호 Chart No.	
검사대상자명 Name of Patient	
생년월일 Date of Birth	
나이/성별 Age/Gender	

검사정보관리결과 Quality Control

검체 정도관리 Specimen quality	시퀀싱 정도관리 Sequencing quality	분석 정도관리 Analysis quality
PASS	PASS(>Q30: 94.52%)	PASS(PR score: 100.0)

요약결과 Result Summary

Annotated Variants

Positive (Tumour variants found)

Gene	Variant	Description	Class[2]	TCGA freq[2]	MSK-IMPACT freq[3]
KRAS	G12D	Allele.Freq: 35.97% (Ref / Alt = 689 / 387)	1	1.45% (105/7,225)	4.06% (444/10,945)
FBXW7	R505C	Allele.Freq: 63.67% (Ref / Alt = 380 / 666)	2	0.1% (7/7,225)	0.22% (24/10,945)
TP53	R273H	Allele.Freq: 69.7% (Ref / Alt = 170 / 391)	3	0.76% (55/7,225)	1.25% (137/10,945)

Negative (Tumour variants not found[4])

Gene	Variant	Mean.Depth.CDS
BRCA2	Wild Type	1589x
BRAF	Wild Type	688x
EGFR	Wild Type	773x
ERBB2	Wild Type	613x
NRAS	Wild Type	1012x

CancerSCAN Variants : 29
- Annotated Variants : 3
- Known Variants[5] : 2
- Novel Variants[5] : 24

Estimated tumor purity (Bioinformatics): 75%

[1] For the variant classification, please see the supplementary report.
[2] The variant frequency observed from TCGA datasets, 7,225 clinical samples of 24 tumor types (Ref. cBioPortal http://www.cbioportal.org/).
[3] The variant frequency observed from MSK-IMPACT datasets, 10,945 clinical samples of 55 tumor types (Ref. Nat. Med. 2017).
[4] If the genes (i.e. TP53, BRAF, KRAS, NRAS, EGFR, BRCA1, BRCA2 and ERBB2) are fully covered (mean depth >= 200x) and tumor hotspot mutations were not found in those genes, these are reported as tumor-mutation free.
[5] Known and novel variants are listed in supplementary report.

LabGenomics Clinical Laboratories

2 Variant annotation

Drug with Clinical significance


CANCER [SCAN]

(2/26)

검체접수번호 36651

의뢰기관명 Institution	검사대상자명 Name of Patient	차트번호 Chart No.

상세결과 I Result Detail - Part A

A.Variant Annotation[6] (Table view)

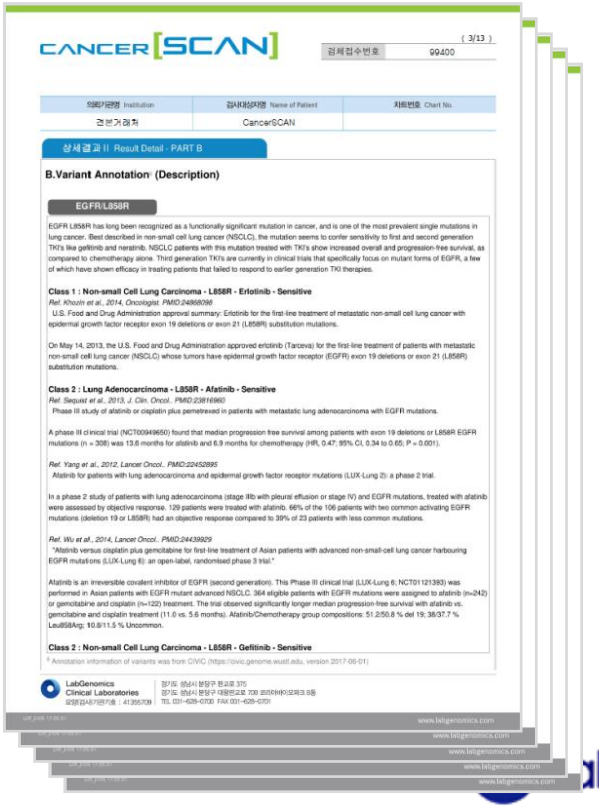
Variant	Disease	Class	CIVIC Variant	Drug	Clinical significance
KRAS/G12D	Colorectal Cancer	1	EXON 2 MUTATION	Cetuximab	Resistant
			MUTATION	Cetuximab	Resistant
			MUTATION	Panitumumab	Resistant
	Colorectal Cancer	2	MUTATION	Bevacizumab	Resistant
			EXON 2 MUTATION	Cetuximab	Resistant
			G12/G13	Cetuximab	Resistant
			MUTATION	Cetuximab	Sensitive
			MUTATION	Chemotherapy	Resistant
			EXON 2 MUTATION	EGFR Inhibitor	Resistant
			EXON 2 MUTATION	Irinotecan	Sensitive
			EXON 2 MUTATION	Regorafenib	Sensitive
			MUTATION	RO4987655	no info
			EXON 2 MUTATION	Selumetinib (AZD6244)	Sensitive
			MUTATION	Selumetinib (AZD6244)	Sensitive
			G12	(Prognostic)	no info
			G12D	(Prognostic)	Better Outcome
	Non-small Cell Lung Carcinoma	3	MUTATION	Docetaxel	Resistant
			MUTATION	Selumetinib (AZD6244)	Resistant
			MUTATION	(Predisposing)	no info
	Acute Leukemia	4	G12	(Diagnostic)	Positive
	Endometrial Cancer	4	MUTATION	Ridaforolimus	Sensitive
			MUTATION	Temsirolimus	Sensitive
	Hepatocellular Carcinoma	4	MUTATION	Retametinib	Sensitive
			MUTATION	Sorafenib	Sensitive

[6] Annotation information of variants was from CIVIC (https://civic.genome.wustl.edu, version 2017-09-01)

LabGenomics Clinical Laboratories

3 Description of variants

Reference information
Supplementary report


CANCER [SCAN]

(3/13)

검체접수번호 99400

의뢰기관명 Institution	검사대상자명 Name of Patient	차트번호 Chart No.
건보거래처	CancerSCAN	

상세결과 II Result Detail - PART B

B.Variant Annotation[6] (Description)

EGFR/L858R

EGFR L858R has long been recognized as a functionally significant mutation in cancer, and is one of the most prevalent single mutations in lung cancer. Best described in non-small cell lung cancer (NSCLC), the mutation seems to confer sensitivity to first and second generation TKI's like gefitinib and neratinib. NSCLC patients with this mutation treated with TKI's show increased overall and progression-free survival, as compared to chemotherapy alone. Third generation TKI's are currently in clinical trials that specifically focus on mutant forms of EGFR, a few of which have shown efficacy in treating patients that failed to respond to earlier generation TKI therapies.

Class 1 : Non-small Cell Lung Carcinoma - L858R - Erlotinib - Sensitive

Ref. Khozin et al., 2014, Oncologist. PMID:24860098

U.S. Food and Drug Administration approval summary: Erlotinib for the first-line treatment of metastatic non-small cell lung cancer with epidermal growth factor receptor exon 19 deletions or exon 21 (L858R) substitution mutations.

On May 14, 2013, the U.S. Food and Drug Administration approved erlotinib (Tarceva) for the first-line treatment of patients with metastatic non-small cell lung cancer (NSCLC) whose tumors have epidermal growth factor receptor (EGFR) exon 19 deletions or exon 21 (L858R) substitution mutations.

Class 2 : Lung Adenocarcinoma - L858R - Afatinib - Sensitive

Ref. Sequist et al., 2013, J. Clin. Oncol.. PMID:23816960

Phase III study of afatinib or cisplatin plus pemetrexed in patients with metastatic lung adenocarcinoma with EGFR mutations.

A phase III clinical trial (NCT00949650) found that median progression free survival among patients with exon 19 deletions or L858R EGFR mutations (n = 308) was 13.8 months for afatinib and 6.9 months for chemotherapy (HR, 0.47; 95% CI, 0.34 to 0.65; P < 0.001).

Ref. Yang et al., 2012, Lancet Oncol.. PMID:22452895

Afatinib for patients with lung adenocarcinoma and epidermal growth factor receptor mutations (LUX-Lung 2): a phase 2 trial.

In a phase 2 study of patients with lung adenocarcinoma (stage IIIb with pleural effusion or stage IV) and EGFR mutations, treated with afatinib were assessed by objective response. 129 patients were treated with afatinib. 66% of the 106 patients with two common activating EGFR mutations (deletion 19 or L858R) had an objective response compared to 39% of 23 patients with less common mutations.

Ref. Wu et al., 2014, Lancet Oncol.. PMID:24439929

"Afatinib versus cisplatin plus gemcitabine for first-line treatment of Asian patients with advanced non-small-cell lung cancer harbouring EGFR mutations (LUX-Lung 6): an open-label, randomised phase 3 trial."

Afatinib is an irreversible covalent inhibitor of EGFR (second generation). This Phase III clinical trial (LUX-Lung 6; NCT01121393) was performed in Asian patients with EGFR mutant advanced NSCLC. 364 eligible patients with EGFR mutations were assigned to afatinib (n=242) or gemcitabine and cisplatin (n=122) treatment. The trial observed significantly longer median progression-free survival with afatinib vs. gemcitabine and cisplatin treatment (11.0 vs. 5.6 months). Afatinib/Chemotherapy group compositions: 51.2/50.8 % del 19; 38/37.7 % Leu858Arg; 10.8/11.5 % Uncommon.

Class 2 : Non-small Cell Lung Carcinoma - L858R - Gefitinib - Sensitive

[6] Annotation information of variants was from CIVIC (https://civic.genome.wustl.edu, version 2017-09-01)

LabGenomics Clinical Laboratories

LabGenomics

5) CancerSCAN™ (Somatic Mutation Cancer Test)

Cancer Scan Report Example

⇒ **Cancer :** Colorectal Cancer
⇒ **Sample :** FFPE

요약결과 Result Summary

Annotated Variants

Positive (Tumour variants found)

Gene	Variant	Description	Class[1]	TCGA.freq[2]	MSK-IMPACT.freq[3]
KRAS	**G12D**	Allele.Freq: 13.42% (Ref / Alt = 787 / 122)	1	1.45% (105/7,225)	4.06% (444/10,945)
PIK3CA	**E542K**	Allele.Freq: 34.97% (Ref / Alt = 502 / 270)	2	1.4% (101/7,225)	1.97% (216/10,945)
FBXW7	**W365***	Allele.Freq: 12.64% (Ref / Alt = 1285 / 186)	2	.	0.02% (2/10,945)

Negative (Tumour variants not found[4])

Gene	Variant	Mean.Depth.CDS
ALK	Wild Type	982x
BRAF	Wild Type	531x
BRCA1	Wild Type	1,148x
BRCA2	Wild Type	959x
ERBB2	Wild Type	815x
IDH1	Wild Type	997x
IDH2	Wild Type	1,054x
KIT	Wild Type	1,054x
MYC	Wild Type	1,082x
MYCN	Wild Type	908x
NRAS	Wild Type	1,204x
PDGFRA	Wild Type	485x

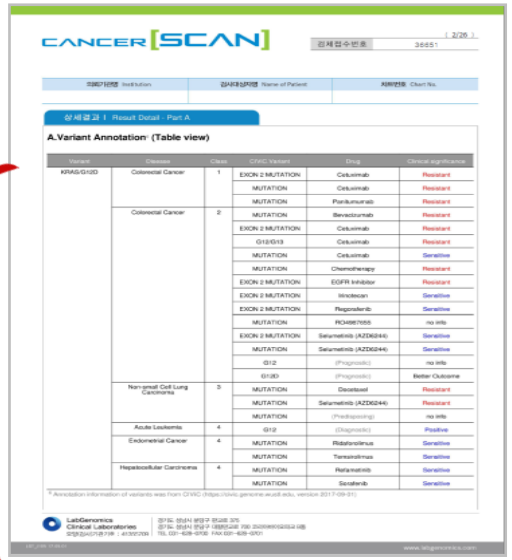
Estimated tumor purity (Bioinformatics): 13%

CancerSCAN Variants : 21
- Annotated Variants : 3
- Known Variants[5] : 9
- Novel Variants[5] : 9

LabGenomics

5) CancerSCAN™ (Somatic Mutation Cancer Test)

Cancer Scan Report Example


CANCER [SCAN]

A.Variant Annotation (Table view)

상세결과 I Result Detail - Part A

A.Variant Annotation® (Table view)

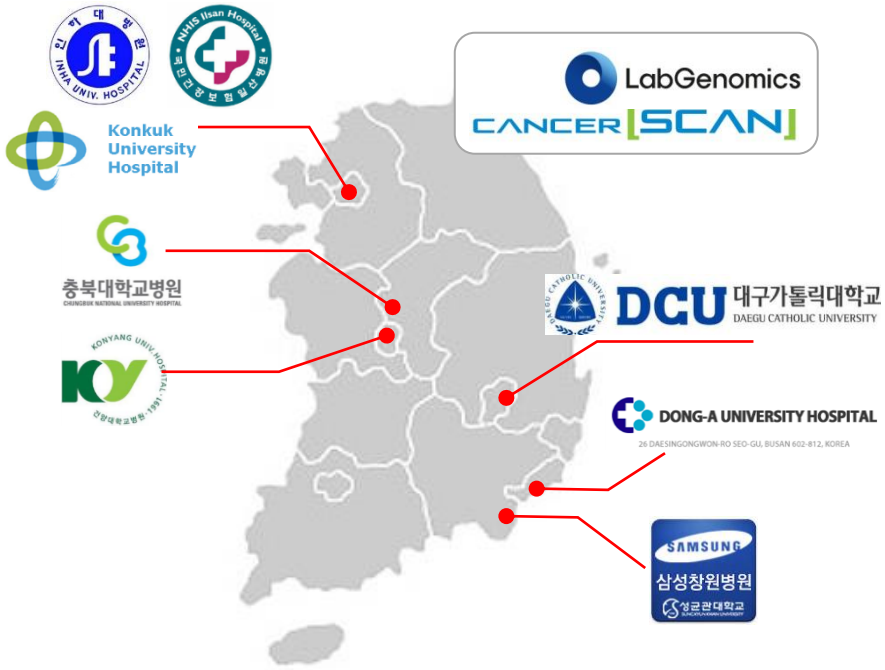
Variant	Disease	Class	CIViC.Variant	Drug	Clinical.significance
KRAS/G12D	Colorectal Cancer	1	EXON 2 MUTATION	Cetuximab	Resistant
			MUTATION	Cetuximab	Resistant
			MUTATION	Panitumumab	Resistant
	Colorectal Cancer	2	MUTATION	Bevacizumab	Resistant
			EXON 2 MUTATION	Cetuximab	Resistant
			G12/G13	Cetuximab	Resistant
			MUTATION	Chemotherapy	Resistant
			EXON 2 MUTATION	EGFR Inhibitor	Resistant
			EXON 2 MUTATION	Irinotecan	Sensitive
			EXON 2 MUTATION	Selumetinib (AZD6244)	Sensitive
			G12D	(Prognostic)	Better Outcome
	Non-small Cell Lung Carcinoma	3	MUTATION	Docetaxel	Resistant
			MUTATION	Selumetinib (AZD6244)	Resistant
	Acute Leukemia	4	G12	(Diagnostic)	Positive
	Hepatocellular Carcinoma	4	MUTATION	Refametinib	Sensitive
			MUTATION	Sorafenib	Sensitive
	Lung Adenocarcinoma	4	MUTATION	(Prognostic)	Poor Outcome
	Lung Cancer	4	G12D	Gefitinib	Resistant
			G12D	(Diagnostic)	Positive

LabGenomics

5) CancerSCAN™ (Somatic Mutation Cancer Test)

- Approved for National Reimbursement Program of NGS-panel cancer analysis service
- KFDA approval for CancerSCAN™ CDx kit (ongoing)

National Reimbursement Program



within Top 5 players nation-wide

CancerSCAN CDx kit (KFDA)

CDx kit	Components	Software

Gene	Variant	Disease	Drug
ALK	Fusion	NSCLC	Crizotinib
EGFR	E19del, L858R	NSCLC	Erlotinib Gefitinib Afatinib
EGFR	T790M	NSCLC	Osimertinib
ERBB2	Amp.	Breast cancer	Trastuzumab

Much more diseases and drugs will be added

LabGenomics

6) Cancer4Cast™ (Hereditary Cancer Test)

Designed to analyze 36 genes associated with Breast, Ovarian, Colorectal, Endometrial, Melanoma, Pancreatic, Gastric, Prostate, and Lung cancers

10-15% of most cancers in women and men are due to inherited genetic mutations. [1)]

Assay Genes	APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, SMAD4, STK11, TP53, VHL etc.

10-15% Hereditary

5-15% Familial

70-80% Sporadic

Hereditary cancer is caused by an inherited genetic mutation.
It is typical to see a recurring pattern of cancer across two to three generations—like multiple individuals diagnosed with the same type of cancer(s) and individuals diagnosed with cancer much younger than average.

1) Tung N, Battelli C, Allen B, et al. Frequency of mutations in individuals with breast cancer referred for BRCA1 and BRCA2 testing using next-generation sequencing with a 25-gene panel. Cancer. January 2015;121(1):25-33.

LabGenomics

6) Cancer4Cast™ (Hereditary Cancer Test)

NGS-based Cancer Predisposition Panel

CANCER 4 CAST

Cancer4Cast™ does not miss critical variants

36 cancer predictive gene mutations can be scanned at once	
Breast cancer Ovarian cancer	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MLH1, MRE11A, MSH2, NBN, TP53, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D, STK11
Colon cancer etc.	APC, BMPR1A, CDK4, CDKN2A, EPCAM, MEN1, MSH6, MUTYH, PMS2, POLD1, POLE, RET, SDHC, SDHD, SMAD4, VHL,SDHB
Test candidate	- Two or more people with cancer in the family. - Cancer diagnostics before 40 years old. - Several types of cancer patients are in the Family
Benefits	- Prediction of cancer incidence risk - Being proactive in cancer risk management

Highly reliable sequence analysis

- Analysis by ACMG guideline
- Analysis based on DB of genetic variations and diseases such as Clinvar, KMD etc.
- Analysis of Review board consisting of experts of Laboratory medicine, Molecular genetics and Bioinformatics
- In-house LIMS Review board System

- *ACMG: Standard and guidelines of the interpretation of sequence variants. Genetics In Medicine. 2015*
- *Clinvar: (NCBI) Genetic DB*
- *KMD: Korea genetic variation DB from Korea National Institute of Health , Organization for rare diseases*

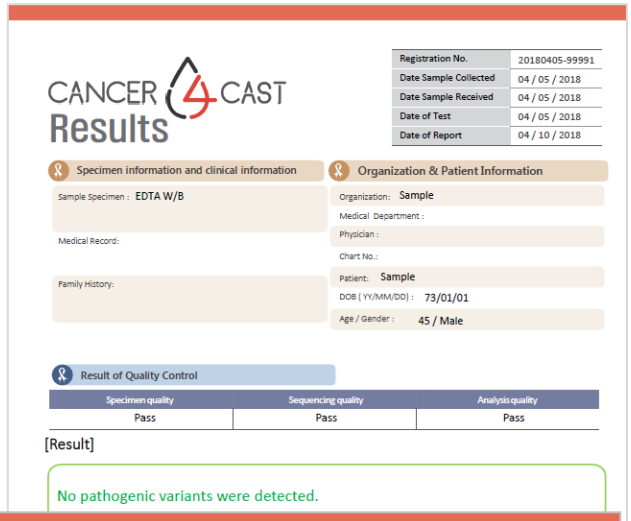
LabGenomics

6) Cancer4Cast™ (Hereditary Cancer Test)

Cancer4cast Report

1. **Summarized Report**
2. **Variant annotation**

The function of the gene with mutation
Health care guidelines based on cancer risk



CANCER 4 CAST
Results

Registration No.	20180405-99991
Date Sample Collected	04 / 05 / 2018
Date Sample Received	04 / 05 / 2018
Date of Test	04 / 05 / 2018
Date of Report	04 / 10 / 2018

Specimen information and clinical information

Sample Specimen : EDTA W/B

Medical Record:

Family History:

Organization & Patient Information

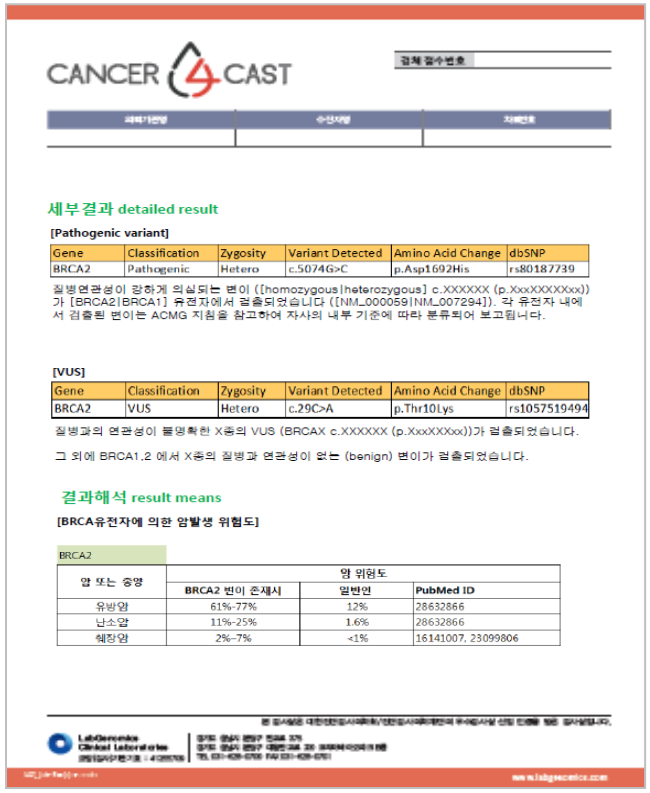
Organization: Sample
Medical Department :
Physician :
Chart No.:
Patient: Sample
DOB (YY/MM/DD) : 73/01/01
Age / Gender : 45 / Male

Result of Quality Control

Specimen quality	Sequencing quality	Analysis quality
Pass	Pass	Pass

[Result]

No pathogenic variants were detected.



CANCER 4 CAST

검체 접수번호

세부결과 detailed result

[Pathogenic variant]

Gene	Classification	Zygosity	Variant Detected	Amino Acid Change	dbSNP
BRCA2	Pathogenic	Hetero	c.5074G>C	p.Asp1692His	rs80187739

질병연관성이 강하게 의심되는 변이 ([homozygous|heterozygous] c.XXXXXX (p.XxxXXXXXxx))가 [BRCA2|BRCA1] 유전자에서 검출되었습니다 ([NM_000059|NM_007294]). 각 유전자 내에서 검출된 변이는 ACMG 지침을 참고하여 자사의 내부 기준에 따라 분류되어 보고됩니다.

[VUS]

Gene	Classification	Zygosity	Variant Detected	Amino Acid Change	dbSNP
BRCA2	VUS	Hetero	c.29C>A	p.Thr10Lys	rs1057519494

질병과의 연관성이 불명확한 X종의 VUS (BRCAX c.XXXXXX (p.XxxXXXxx))가 검출되었습니다.

그 외에 BRCA1,2 에서 X종의 질병과 연관성이 없는 (benign) 변이가 검출되었습니다.

결과해석 result means

[BRCA유전자에 의한 암발생 위험도]

BRCA2

암 또는 증명	암 위험도		
	BRCA2 변이 존재시	일반인	PubMed ID
유방암	61%-77%	12%	28632866
난소암	11%-25%	1.6%	28632866
췌장암	2%-7%	<1%	16141007, 23099806

CANCER 4 CAST

검체 접수번호

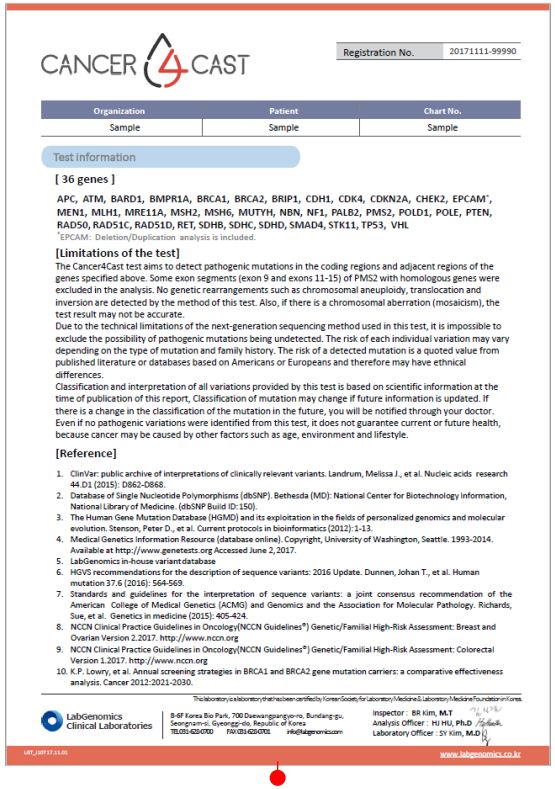
결과해석 result means

[BRCA 유전자 기능]

BRCA2 유전자는 세포가 너무 빠르게 성장하거나 분열되는 것을 막음으로써 종양발생을 억제하는 단백질을 만드는데 관여합니다. 이 단백질은 손상된 DNA의 복구를 도와 유전체의 안정성을 유지하는데 중요한 역할을 합니다. 또한 다른 유전자의 활동을 조절하고 배아 발생에 필수적인 역할을 합니다. BRCA1 유전자에 돌연변이가 발생하면 단백질이 올바르게 만들어지지 않거나 혹은 이들 기능이 저하되어 손상된 DNA가 올바르게 복구되지 않게 됩니다. 이로 인해 세포들은 추가적인 유전적 변화를 일으킴으로써 유방암을 비롯한 여러 종류의 암을 유발하게 됩니다.

[병원성 변이에 대한 암 위험도 관리지침]

- 18세부터 정기적인 유방 자가 체크를 하도록 합니다.
- 25세 부터 시작하여 매 6개월 ~1년 마다 유방암 검진을 시행합니다.
- 유방 스크리닝 검사
  <25~29세> 가족력을 고려하여 매년 유방 MRI 촬영 혹은 MRI 가 불가능한 경우 유방 X선 검사 (mammogram)
  <30~75세> 매년 유방 X선 검사 혹은 유방 MRI 촬영
  <75세 이상> 개인별 상황을 고려하여 판단
- 유방암으로 치료를 받은 BRCA 유전자 변이가 있는 여성의 경우, 남은 유방 조직에 대하여 매년 유방 X선 검사 혹은 유방 MRI 를 통해 지속적으로 관리합니다.
- 예방 차원에서의 유방절제술 (Mastectomy)을 고려할 수 있습니다.
- 출산을 완료한 여성의 경우 예방 차원에서의 난관 난소 절제술 (Salpingo-oopherectomy) 고려할 수 있습니다.(35~40 세)
- 예방 차원에서의 난관 난소 절제술을 선택하지 않은 경우, 난소암 스크리닝을 위해 검사의 민감도 및 특이도 면에 충분하지는 않지만 자궁과 난소 경질(transvaginal) 초음파 검사 및 CA-125 혈액 검사 시행을 고려할 수 있습니다.
- 위험도를 줄이기 위한 약물 사용을 고려할 수 있습니다. (tamoxifen, raloxifene, 경구용 피임약 등)
- 남성의 경우 35세부터 유방 자가 체크, 매년 유방검사, 45세 부터 BRCA1/BRCA2 보인자에 대한 전립선암 스크리닝 검사를 고려할 수 있습니다.
- 췌장암과 흑색종에 대한 구체적인 스크리닝 가이드라인은 없으나 가족력을 고려하여 개별적인 스크리닝을 고려할 수 있습니다.
- 가족 및 친척들에게 유전성 암 가능성을 알리고, 위험도 평가방법 및 관리에 대해 안내합니다.
- 가임기에 있는 검사자의 경우 산전진단이나 착상전 유전자 검사와 같은 추가관리를 고려할 수 있습니다.
- BRCA2 나 본 검사에 포함된 다른 유전자에서 이중대립 돌연변이(biallelic mutation)가 나타난 경우, 상염색체 열성 질환아의 출산 가능성이 있습니다. 이 경우 배우자의 보인자 검사를 통해 출산과 관련된 결정에 참고하시기 바랍니다.



CANCER 4 CAST

Registration No.	20171111-99990

Organization	Patient	Chart No.
Sample	Sample	Sample

Test information

[36 genes]

APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM*, MEN1, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51C, RAD51D, RET, SDHB, SDHC, SDHD, SMAD4, STK11, TP53, VHL

*EPCAM: Deletion/Duplication analysis is included.

[Limitations of the test]

The Cancer4Cast test aims to detect pathogenic mutations in the coding regions and adjacent regions of the genes specified above. Some exon segments (exon 9 and exons 11-15) of PMS2 with homologous genes were excluded in the analysis. No genetic rearrangements such as chromosomal aneuploidy, translocation and inversion are detected by the method of this test. Also, if there is a chromosomal aberration (mosaicism), the test result may not be accurate.
Due to the technical limitations of the next-generation sequencing method used in this test, it is impossible to exclude the possibility of pathogenic mutations being undetected. The risk of each individual variation may vary depending on the type of mutation and family history. The risk of a detected mutation is a quoted value from published literature or databases based on Americans or Europeans and therefore may have ethnical differences.
Classification and interpretation of all variations provided by this test is based on scientific information at the time of publication of this report, Classification of mutation may change if future information is updated. If there is a change in the classification of the mutation in the future, you will be notified through your doctor. Even if no pathogenic variations were identified from this test, it does not guarantee current or future health, because cancer may be caused by other factors such as age, environment and lifestyle.

[Reference]

1. ClinVar: public archive of interpretations of clinically relevant variants. Landrum, Melissa J., et al. Nucleic acids research 44.D1 (2015): D862-D868.
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3. The Human Gene Mutation Database (HGMD) and its exploitation in the fields of personalized genomics and molecular evolution. Stenson, Peter D., et al. Current protocols in bioinformatics (2012):1-13.
4. Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1993-2014. Available at http://www.genetests.org Accessed June 2, 2017.
5. LabGenomics in-house variant database
6. HGVS recommendations for the description of sequence variants: 2016 Update. Dunnen, Johan T., et al. Human mutation 37.6 (2016): 564-569.
7. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics (ACMG) and Genomics and the Association for Molecular Pathology. Richards, Sue, et al. Genetics in medicine (2015): 405-424.
8. NCCN Clinical Practice Guidelines in Oncology(NCCN Guidelines®) Genetic/Familial High-Risk Assessment: Breast and Ovarian Version 2.2017. http://www.nccn.org
9. NCCN Clinical Practice Guidelines in Oncology(NCCN Guidelines®) Genetic/Familial High-Risk Assessment: Colorectal Version 1.2017. http://www.nccn.org
10. K.P. Lowry, et al. Annual screening strategies in BRCA1 and BRCA2 gene mutation carriers: a comparative effectiveness analysis. Cancer 2012:2021-2030.

LabGenomics Clinical Laboratories
B-6F Korea Bio Park, 700 Daewangpangyo-ro, Bundang-gu, Seongnam-si, Gyeonggi-do, Republic of Korea
TEL:031-628-0700 FAX:031-628-0701 info@labgenomics.com
Inspector : BR Kim, M.T
Analysis Officer : Hi HU, Ph.D
Laboratory Officer : SY Kim, M.D
www.labgenomics.co.kr

3. **Supplementary Information**
   Description of the test
   Method of the test
   Limitation of the test

LabGenomics

6) Cancer4Cast™ (Hereditary Cancer Test)

Who have to consider CANCER 4CAST™

✓**If a hereditary cancer is suspected**

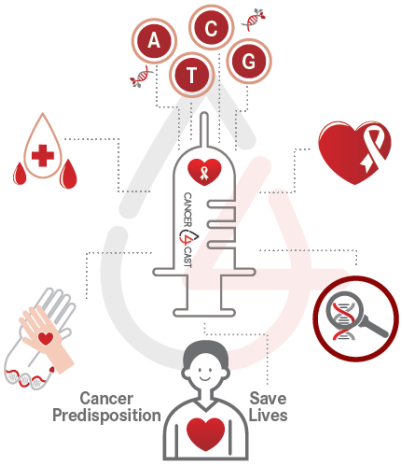
- Family or relatives have hereditary cancer (Breast / Ovarian/ Colorectal Caner) patients
- Family or relatives have cancer at an early age

✓**If you want to have an accurate cancer genetic testing**

✓**If you want to regularly monitor your health, maintain recommended checkups and adopt measures to reduce your risk of developing cancer**

Test Guideline

- **Specimen : EDTA blood 3.0ml**
- **Method : Next Generation Sequencing + Specific cancer panel**
- **Turnaround time : 3 weeks**
- **Insurance coverage available in Korea (나-598-1가 / CB00100B) **



LabGenomics

7) IMS™ (Inherited Metabolic Disease Test)

A Very effective test that can detect the presence of chromosomal abnormalities related to the inherited metabolic disorder of the newborn/child.

IMS test enables more precise detection of 21 inherited metabolic disease listed below

No	Disease	Genes
1	Gaucher's disease	GBA
2	Fabry disease	GLA
3	Pompe's disease	GAA
4	Hunter's syndrome	IDS
5	Hurler syndrome	IDUA
6	Sanfilippo syndrome A	SGSH
7	Sanfilippo syndrome B	NAGLU
8	Sanfilippo syndrome C	HGSNAT
9	Sanfilippo syndrome D	GNS
10	Morquio syndrome A	GALNS
11	Morquio syndrome B	GLB1
12	Maroteaux-Lamy syndrome	ARSB
13	Sly's syndrome	GUSB
14	Wilson's disease	ATP7B
15	Glycogen storage disease Type I-A	G6PC
16	Glycogen storage disease Type I-B	SLC37A4
17	Glycogen storage disease Type III	AGL
18	Glycogen storage disease Type IV	GBE1
19	Glycogen storage disease Type V	PYGM
20	Glycogen storage disease Type VI	PYGL
21	Glycogen storage disease Type VII	PFKM

LabGenomics

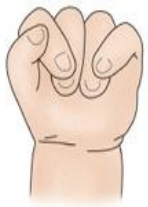
7) IMS™ (Inherited Metabolic Disease Test)

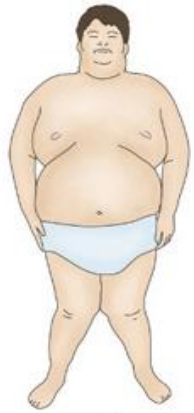
√ **Who needs this?**

: Newborn baby

: Child and adult willing to know about their metabolic disorder







√ **Advantages**

: Quick and Safe

: High Accuracy

: Early Detection & Treatment

LabGenomics

7) IMS™ (Inherited Metabolic Disease Test)

Report Sample

IMS Test Report

Inherited Metabolic disease Screening Test

Registration No.	20181227-99990
Receipt Date	2018-12-27
Analysis Date	2018-12-27
Report Date	2019-01-21

Specimen information & Clinical opinion

Specimen: Blood

Specimen collection date : 2018-12-27

Clinical opinion: 임상소견

Family history: 가족력

Organization & Patient information

Organization: 본사 Test

Patient: 진료과 / 진료과

Chart No.: 담당의

DOB: 12345

Age/Gender: IMS

Department: 2010-05-04

Doctor: 8 / M

Address: 성남시 분당구 삼평동 694-1번지 코리아바이오파크 B-6

Quality Control

Specimen quality	Sequencing quality	Analysis quality
Pass	Pass	Pass

Test Result

Pathogenic mutations associated with Glycogen storage disease Ib was detected.

[Comment]

A pathogenic mutation known to be associated with Glycogen storage disease Ib was detected in the SLC37A4 gene.
Genetic counseling and confirmation tests are required for accurated diagnosis.
And a mutation known to be associated with Pompe's disease and Glycogen storage disease VII were detected in the GAA and PFKM gene.
Pompe's disease and Glycogen storage disease VII is an autosomal recessive disorder. A carrier of the recessive disorder usually have no disease-related symptoms (asymptomatic) or may appear very mild even when present. However, in the future, if the client meets a spouse with the same mutation and gives birth, there is a 25% chance of having a baby with the disease. Therefore, we recommend a genetic test for a spouse or a baby in the future. This is a screening test for genetic metabolic disorders. Genetic counseling and confirmation tests are required for accurate diagnosis.

LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23 | Genetic Research Laboratory: No. 7

Medical Doctor : M.D., Ph.D.
Analysis officer : Hj Hu PH.D
Laboratory officer : MJ Oh PH.D

IMS Test Report

Inherited Metabolic disease Screening Test

Registration No. 20181227-99990

Organization	Patient	Chart No.
본사 Test	IMS	12345

Disease Type	Disease	Gene	Result	Declaration
Lysosomal storage disorders	Gaucher's disease	GBA	Not detected	Normal
	Fabry's disease	GLA	Not detected	Normal
	Pompe's disease	GAA	Not detected	Normal
	Hurler syndrome	IDUA	Not detected	Normal
	Hunter's syndrome	IDS	Not detected	Normal
	Sanfilippo syndrome A	SGSH	Not detected	Normal
	Sanfilippo syndrome B	NAGLU	Not detected	Normal
	Sanfilippo syndrome C	HGSNAT	Not detected	Normal
	Sanfilippo syndrome D	GNS	Not detected	Normal
	Morquio syndrome A	GALNS	Not detected	Normal
	Morquio syndrome B	GLB1	Not detected	Normal
	Maroteaux-Lamy syndrome	ARSB	Not detected	Normal
	Sly syndrome	GUSB	Not detected	Normal
Copper metabolic disorder	Wilson's disease	ATP7B	Not detected	Normal
Glycogen Storage Diseases	Glycogen storage disease Ia	G6PC	Not detected	Normal
	Glycogen storage disease Ib	SLC37A4	Not detected	Normal
	Glycogen storage disease III	AGL	Not detected	Normal
	Glycogen storage disease IV	GBE1	Not detected	Normal
	Glycogen storage disease V	PYGM	Not detected	Normal
	Glycogen storage disease VI	PYGL	Not detected	Normal
	Glycogen storage disease VII	PFKM	Not detected	Normal

LabGenomics Co.,Ltd. | Genetic Testing Laboratory: No. 23 | Genetic Research Laboratory: No. 7

Medical Doctor : M.D., Ph.D.
Analysis officer : Hj Hu PH.D
Laboratory officer : MJ Oh PH.D

LabGenomics

4. PCR Solution

01 LabGscan™ FRAXA PCR kit

Intended Use

The LabGscan™ FRAXA PCR kit is an *in vitro* diagnostic test, based on PCR technology, for the amplification and detection of CGG repeats in the 5'-untranslated region (5'-UTR) of FMR1 (Fragile X mental retardation 1) gene. The Kit aids to diagnose fragile-X syndrome and other fragile-X associated disorders such as Fragile X-associated primary ovarian insufficiency (FXPOI) and Fragile X-associated tremor/ataxia syndrome (FXTAS) and to identify carriers for fragile X syndrome.

BackGround

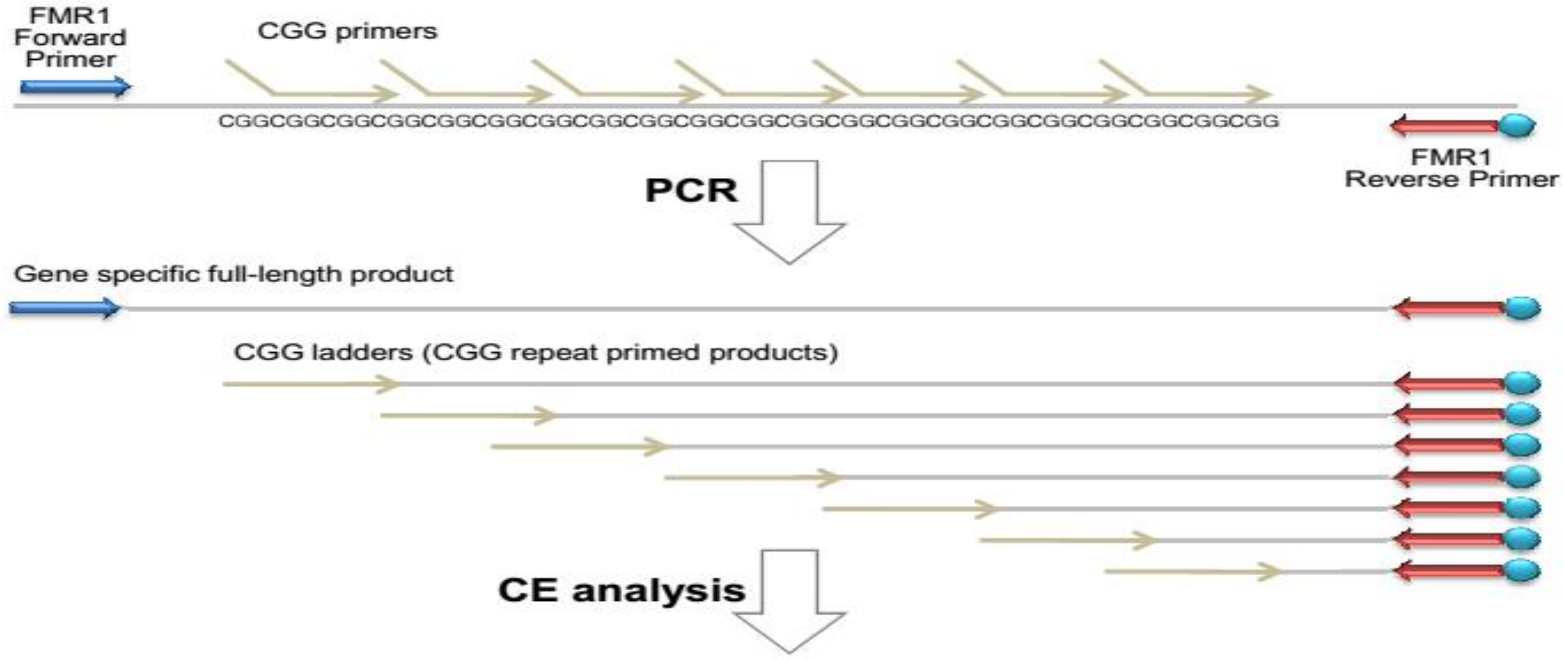
Fragile X syndrome is the most common inherited cause of intellectual disability with an estimated prevalence of 1 in 4000 to 6000 males.

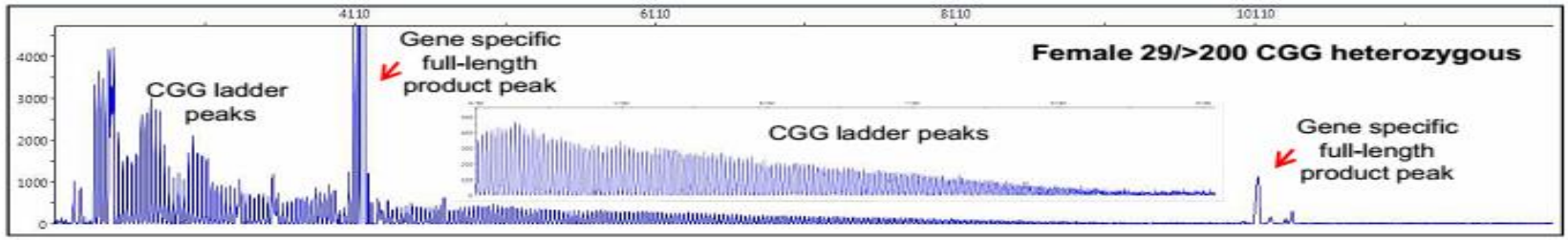
- Mainly caused by a large expansion(‘full mutation’) in a CGG repeat tract in thee 5-UTR gene located in X chromosome.
- Males with full expansion mutations have Fragile X symptoms.
- Females with a full expansion mutation may or may not have the symptom or may be mildly affected.

02 LabGscan™ FRAXA PCR kit

Principle

Based on the triplet repeat primed PCR (TP-PCR)



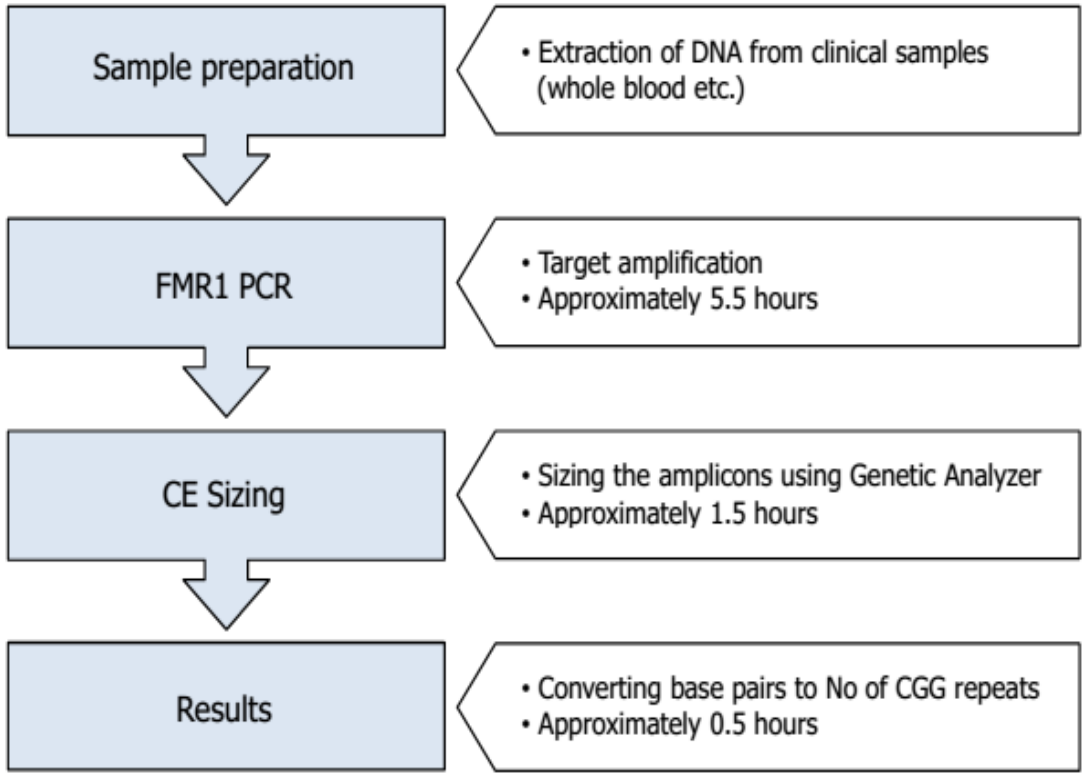


LabGenomics

03 PCR Solution

Procedure Overview

Enter emerging markets with competitive price and user friendly interface



LabGenomics

04 PCR Solution

Storage and Handling

- Store the reagents in a non-frost-free freezer in the dark at -15 to -25℃.
- The reagents will maintain performance through the expiration date indicated on the label when stored under the specified conditions.
- Avoid repeated thawing and freezing. If you expect to freeze-thaw the reagents more than 3 times, consider aliquoting the regents to minimize the number of freeze-thaw cycles.
- Allow reagents (except polymerase Mix) to thaw at room temperature before assay.
- Briefly vortex all reagents after thawing and spin down all reagents to collect the solutions at the bottom of the vials.
- Perform assay setup at room temperature (approximate range of 18-25℃)
- Place FMR1 Primer Mix, Polymerase Mix, and ROX 1000 Size Ladder on ice during the working steps.

05 PCR Solutions

PCR Kit products

Enter emerging markets with competitive price and user friendly interface

• Infectious Diseases

Sexually Transmitted Infection	- LabGun HPV
Mosquito-Related	- LabGun Dengue - LabGun rtZika

• Human Genetic Diseases

Fragile-X	- LabGscan FRAXA

LabGenomics

5. MDx Clinical Lab with Professionals

01 MDx Clinical Lab in LabGenomics

Automation equipment and information system

- Automatic equipment for quicker turn-around-time
- Quick and accurate report at your hands
- Convenient and efficient LIS and synchronized system

Professional research staff

- Established by diagnostics professors of Asan Medical Center and Samsung Medical Center
- Research and technology team organized with the staffs from Seoul National University Hospital and KAIST

Aggressive investment in R&D

- Development of new diagnostics methodology
- Development and production of new products

LabGenomics

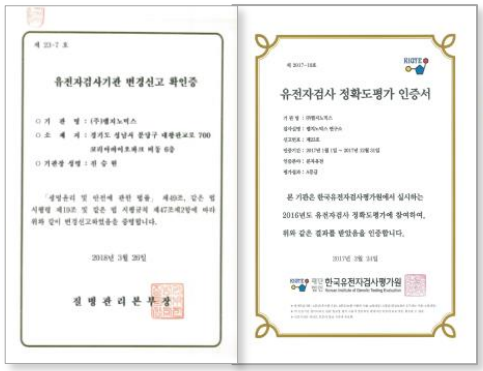
02 R&D Professionals(Ph.D) / R&D Professionals(MD)

Name	Degree	Major	School	Professional Experience	Research Area
DY Cho	Ph. D	Molecular Biology	KAIST	KRIBB	MDx, NGS
IK Shin	Ph. D	Molecular Biology	KAIST	KAIST Natural Science Institute Daewoong Pharma., ISUabgis	MDx, DNA Chip
MJ Oh	Ph. D	Biological Science	Seoul Nat'l Univ.	Seoul Nat'l Univ. Research Institute Nat'l Institute of agriculture	NGS R&D
HJ Hu	Ph. D	Computer Science	Georgia State Univ.	LG Research Park Integrated Research Center for Genome Polymorphism	Bio-informatics R&D
SJ Noh	Ph. D	Molecular Biology	KAIST	KRIBB NIH/NIDDK, Insilicogen	Bio-informatics R&D
DH Jang	Ph. D	Molecular Biology	Korea Univ.	Korea Centers for Disease Control & Prevention	MDx, PCR Kit
SH Lee	Ph. D	Bio informatics	UST	KRIBB	Bio-informatics R&D

Name	Degree	Professional Experience
DH Seo	MD	Seoul Nat'l University College of Medicine
SE Cho	MD	Ehwa University College of Medicine
HL Koo	MD	Yonsei Univeristy College of Medicine
SM Park	MD	Soon Chun Hyang University
HM Shin	MD	Chungbuk National University Hospital
SY Kim	MD	Kyung Hee University College of Medicine

03. Accredited Lab

Acquired A grade Accuracy



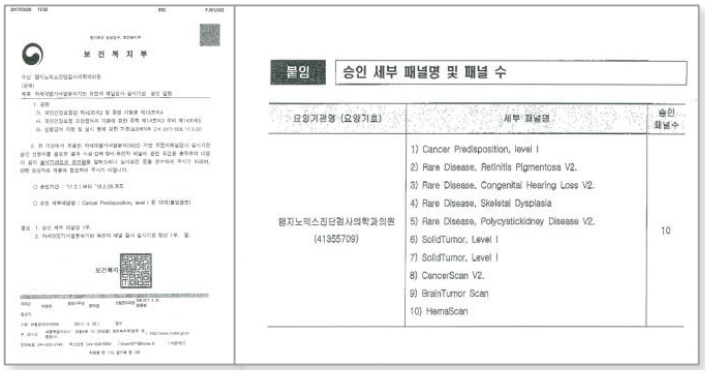
질병관리본부

KIGTE 재단법인 한국유전자검사평가원 Korean Institute of Genetic Testing Evaluation

Certified from the Korean Association of Quality Assurance for Clinical Laboratory, Korean Society of Laboratory Medicine and The Korean Society of Pathologists

Overseas Quality Management Certification

The first approval of NGS(Next Generation Sequencng) based solid cancer/ blood cancer/rare disease test in Korea



보건복지부

붙임 승인 세부 패널명 및 패널 수

요청기관명 (요양기호)	세부 패널명	승인 패널수
랩지노믹스진단검사의학과의원 (41355709)	1) Cancer Predisposition, level I 2) Rare Disease, Retinitis Pigmentosa V2. 3) Rare Disease, Congenital Hearing Loss V2. 4) Rare Disease, Skeletal Dysplasia 5) Rare Disease, Polycystickidney Disease V2. 6) SolidTumor, Level I 7) SolidTumor, Level I 8) CancerScan V2. 9) BrainTumor Scan 10) HemaScan	10

Participate in the quality management program of internationally accredited laboratories (CAP, ISO 13485, CE Certificate)

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6. Reference Price for the Service

1) Personal Genomic Service

Service Price

Price per Test		
Item	**Price**	**Cat #**
MomGuard Standard	160	13990
MomGuard Lite	150	13998
MomGuard Premium Single	210	13996
MomGuard Premium Twin	230	13997
BRCA1/2	350	13399
GenoPAC CD	175.5	13899/13898
GenoPAC Lite	50	13487
IMS	80	
BRCA1/2	310	13399
EnfantGuard	250	13974
Cancer4cast	420	13895

Other Price

	Product	Price per item
1	Buccal Swab Collection Kit	5.5
2	Safe Box	6.0
3	Collection Kit and Safe Box Pack	11.0 (discount)

✓ ***Report be offered by PDF file***

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2) PCR kits (Diagnostic kits)

Price per KIT						
MDx Product		Cat.No / Unit		Price	USD	
		50rxn	100rxn	Per Test	50rxn	100rxn
LabGun TM	Dengue	DG9001A	DG9001B	7	300	600
	HPV Real-time	HP9008A	HP9008B	7	450	900
	ZIKA Real-time	ZK9010A	ZK9010B	10	450	900
LabGScan ™	Avellino	AV9201A	AV9201B	10	450	900
	FRAXA	FX9202A	FX9202B	15	600	1200

**OEM is available for our customer(Those who would like to have the brand on their own)

LabGenomics

Thank you!

Contact

LabGenomics Co., Ltd

Philip Kim
Email: kyh@labgenomics.com
Phone: +82-31-628-0726

Website: http://www.labgenomics.co.kr/eng
Location: 4F PDC Building (Bldg C). Pangyoro 242, Bundang, Seongnam, 13487 South Korea