# **SAMPLE REPORT**

Genome Health Premium (M)

Institution Hospital A
Examinee Asian-male
Date of receipt 2023-02-21

Reception No. 20230221-171-0030 Age/Sex 1993 / Male

# **CONTENTS**

# 00. Introduction

- Examination
- Genetic Terminology

# 01. Results

- Summary of Results
- Results by Disease
- Summary of Genes Requiring Intensive Care

# 02. Interpretation

- How to interpret result
- Interpretation by Disease

# 03. Appendix

- Testing Method & Limitations
- References

Tested by M.T. Lee Myeong-geun (20058)

19Klee

Confirmed by M.D. Seol Chang-an (1037)

Charz Aln Se

Confirmed by M.D. Gi Chang-seok (547)



[1/107]





# 00. Introduction

- Examination - Genetic Terminology

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



[2/107]





# Introduction

Genome Health is GC Genome's gene analysis and disease prediction service that identifies risk alleles related to major cancer types and general diseases and provides efficient prevention and management solutions.

- Using the latest gene analysis techniques, Genome Health examines genetic variants that may increase the risk of diseases.
- Genome Health analyzes previous studies and provides customized health care information.
- The purpose of this examination is not for the diagnosis of diseases. For the diagnosis and treatment of a disease, you must consult your physician.

% The results of the contents may be updated according to the latest research papers. This report is based on Genome Health v2.0(internal standard v2203).

# Limitation

- **Genome Health** Genome Health should not be used for medical diagnosis.
  - Genome Health does not include information on environmental factors.
  - Genome Health does not examine all variants related to diseases.
  - The more detected risk alleles do not necessarily mean that you will develop the corresponding disease.

#### GC Genome complies with the Personal Information Protection Act.

All personal information handled by GC Genome is collected, retained, and processed in compliance with the Act on Promotion of Information and Communications Network Utilization and Information Protection, Etc., and the Personal Information Protection Act, or with the consent of data subjects. In addition, we have a personal information processing policy in place to protect the personal information, rights, and interests of data subjects and resolve relevant problems swiftly and smoothly.

Information on the processing of personal information by service is posted on each service website so that the subject of information can check it

Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



[3/107]





# **Genetic Terminology**

The **cell** is the basic structural unit of humans.

A human consists of innumerable cells, reaching dozens of trillions, functioning as an organism.

Genetic information is contained in the cells, which are the basic structural unit of your body, and is passed down to the next generation through genes.

Humans have approximately 20,000 genes contained in 23 pairs of chromosomes, and also have 3 billion pairs (6 billion) of DNA sequences. Each human cell contains approximately 2 meters of DNA in a double helix structure.

**DNA sequences** may change depending on genetic and environmental factors, which are referred to as **genetic variants**.

Such genetic variants determine the characteristics of your physical appearance and the **risk of diseases**.

\*Nucleobase: One of the molecular substances composing DNA, expressed with adenine, thymine, guanine, and cytosine.

Tested by M.T. Lee Myeong-geun (20058)

MKlee

Confirmed by M.D. Seol Chang-an (1037)

Charz Aln Sec

Confirmed by M.D. Gi Chang-seok (547)

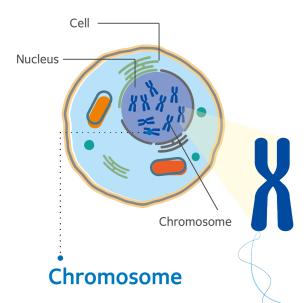
Bli

[4/107]





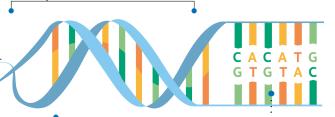
# **Genetic Terminology**



Gene

Genes consisting of DNA contain genetic information. Genes are passed down to the next generation. It is known that humans have over 20,000 genes.

A chromosome consisting of DNA and protein carries genetic information in the form of genes, existing in the nucleus in a thread-like twisted structure. Humans have 23 pairs (46 in total) of chromosomes, two chromosomes of the same shape and size each. because chromosomes are inherited from each parent.



### DNA

It is the minimum unit of storing human genetic information. DNA is composed of a sugar, a phosphate group, and a nucleobase, generally expressed in base pairs (bp). Each human cell contains approximately 2 meters of DNA in a double helix structure. The human genome has approximately 3 billion base pairs of DNA. Approximately 98 to 99% of human DNA consists of random sequences with no information, which are called [noncoding DNA sequence] Sequences containing genetic information are called [genes].

### Nucleobase •

Nucleobases composing DNA are expressed with adenine (A), thymine (T), guanine (G), and cytosine (C), connecting the two strands of DNA.

Adenine **Thymine** Guanine Cytosine



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chazalas Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



[5/107]

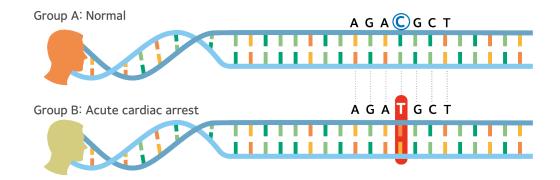




# Genetic Terminology

#### **Genetic Variant**

Genetic variant is the difference in DNA among individuals, which may be inherited or result from external factors. Genetic variant may determine your physical appearance or increase the risk of diseases, or may have no effect at all.



#### Genotype

Genotype refers to genetic information that determines the characteristics of an organism. If the same genetic information in a specific location has a single pair of chromosomes, it is called homozygous.

If genetic information is different from each other, it is called heterozygous. Inheritance is divided into dominant inheritance and recessive inheritance according to the inheritance type of genetic information increasing the risk of disease, which determine the phenotypes.

- Homozygous: A combination of the same genetic information
- Heterozygous: A combination of different genetic information

#### Risk Allele

Some genotypes affect the risk of disease and some do not. Genotypes increasing the risk of disease are called risk alleles.

Confirmed by M.D. Gi Chang-seok (547)

	Group A	Group B	Group C
Dominant Inheritance [e.g., Acute Cardiac Arrest] Expressed even if a single allele exists	a A a		A
Genotype result	Homozygous of normal allele	Homozygous of risk allele	Heterozygous
Phenotype result	Normal	2 risk alleles of acute cardiac arrest	1 risk allele of acute cardiac arrest
Recessive Inheritance [e.g., Liver Cancer]  Expressed only if a pair of alleles exist (Not expressed if only a single allele exists)		a a a	A
Genotype result	Homozygous of normal allele	Homozygous of risk allele	Heterozygous
Phenotype result	Normal	1 risk allele of Liver Cancer	Normal

\*The images above are for your understanding, and are not relevant to the actual genetic variants of the diseases.

The images above are for your understanding, and are not relevant to the actual genetic variants of the disease

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

[6/107]



Tested by M.T. Lee Myeong-geun (20058) / MK/Lee



# 01. Results

- Summary of Results
  - Results by Disease
- Summary of Genes Requiring Intensive Care

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



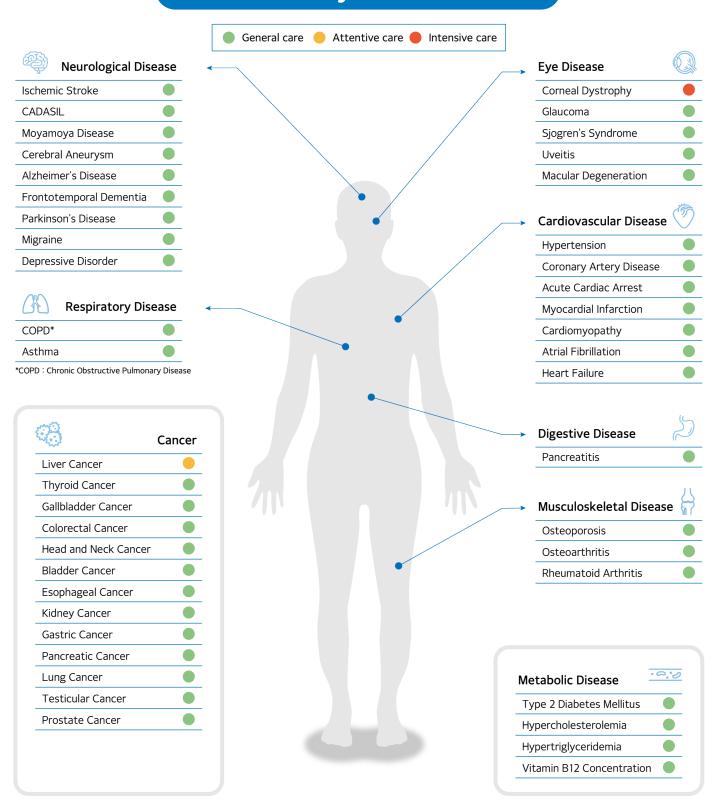
[7/107]





Sample ID 20230221-171-0030 Examinee Asian-male 14443 Reception No.

# **Summary of Results**



Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



[8/107]





# **Results by Disease**

The examination results are shown in 3 levels that show different degrees of risk of disease resulting from relevant risk alleles.

	General care	Attentive care	Intensive care
A degree of risk of disease resulting from risk alleles	Moderate	Relatively high	High
Type of care	Maintain your healthy dietary habits and lifestyle	Improve your dietary habits and lifestyle	Actively improve your dietary habits and lifestyle and consult your physician regularly

# Cancer Patient Asian-male's Cancer Examination Results

Attentive care Liver Cancer	General Thyroid Cancer	Gallbladder Care	General Colorectal Cancer
General Head and Neck Cancer	General Bladder Cancer	General care Cancer	General care Kidney Cancer
General Care Cancer	General Pancreatic Cancer	General care Lung Cancer	General Testicular Cancer
General Prostate Cancer			

Tested by M.T. Lee Myeong-geun (20058) / MK/Lee

Confirmed by M.D. Seol Chang-an (1037) دامایمالی Confirmed by M.D. Gi Chang-seok (547)



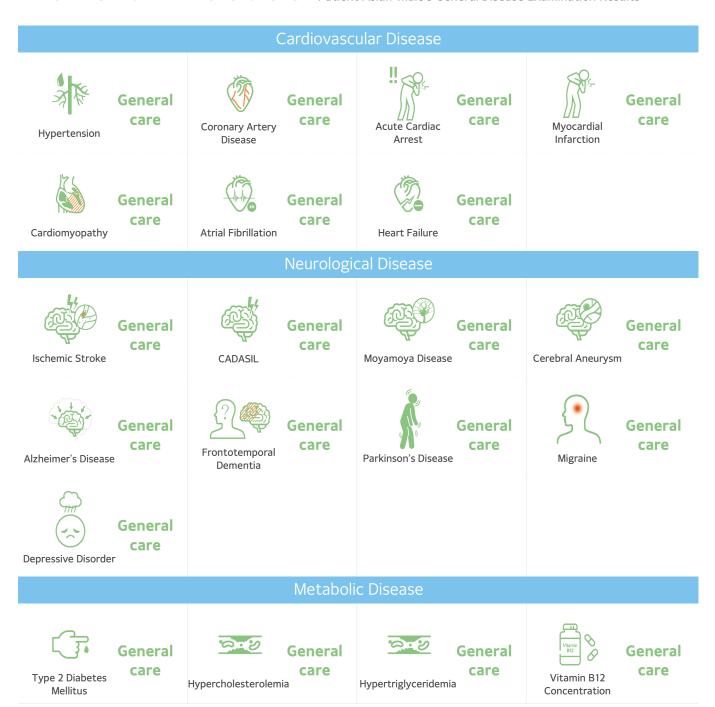
[9/107]





# **Results by Disease**

# General Disease Patient Asian-male's General Disease Examination Results



Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[10/107]





# **Results by Disease**

# General Disease Patient Asian-male's General Disease Examination Results



Tested by M.T. Lee Myeong-geun (20058) / MK/Lee

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[11/107]





Examinee Sample ID Reception No. Asian-male 14443 20230221-171-0030

# **Summary of Genes Requiring Intensive Care**

Genetic variants requiring intensive care are detected.

The following genetic variants are particularly relevant to the diseases. Genetic variants requiring intensive care are not detected, but some diseases may develop due to other genetic variants or your lifestyle. You need to manage your health to prevent diseases.

Disease	Gene (variant)	Your genotype	Risk allele	Disease	Gene (variant)	Your genotype	Risk allele		
16	CDH1	GG	Not detected	الساا	BRCA1 c.302-2A>C	AA			
	p.Gly239Arg			AP.	BRCA2 p.Arg2494Ter	CC	Not detected		
Gastric Cancer	CDH1 p.Arg335Ter	CC		Prostate Cancer	BRCA2 p.Lys467Ter	AA			
ii So	KCNH2		Not		LMNA p.Arg541His	GG			
Acute Cardiac	p.His492Tyr	CC	detected	(E)	PKP2 p.Arg355Ter	CC	Not		
Arrest	TNNI3		Not	Hoart Failure	PKP2 p.Arg651Ter	СС	detected		
Cardiomyopathy	p.Arg145Gln	GG	detected	Tical ciralial c	PTPN11 p.Asn308Asp	AA			
600%	NOTCH3 p.Arg75Pro	GG	Not detected	detected					
	NOTCH3 p.Arg544Cys	СС				RNF213 p.Arg4810Lys	GG	Not detected	
CADASIL	NOTCH3 p.Arg640Cys	СС			Moyamoya Disease				
	<b>GRN</b> p.Ala9Asp	СС	Not	Not			PCSK9	CC	
<b>{?(((((((((((((</b>	<b>GRN</b> c.708+1G>A	GG			S	p.Ala26ArgfsTer18	CC	Not	
Frontotemporal Dementia	MAPT p.Pro693Leu	СС	detected	Hyper Cholesterolemia	PCSK9	GG	detected		
Demenda	<b>MAPT</b> c.2091+16C>T	СС		Cholesterolenna	p.Glu32Lys	00			
	PRSS1	AA	Not detected		TGFBI p.Arg124Cys	СС			
	p.Asn29lle					TGFBI p.Arg124His	GG		
	PRSS1 p.Arg116Cys	СС				TGFBI p.Pro501Thr	CA	detected	
Pancreatitis	SPINK1					Corneal Dystrophy	TGFBI p.Arg555Trp	СС	
	c.194+2T>C	TT			TGFBI p.Arg555Gln	GG			

Tested by M.T. Lee Myeong-geun (20058) / MK/Lee

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[12/107]





# 02. Interpretation

- How to interpret result - Interpretation by Disease

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)

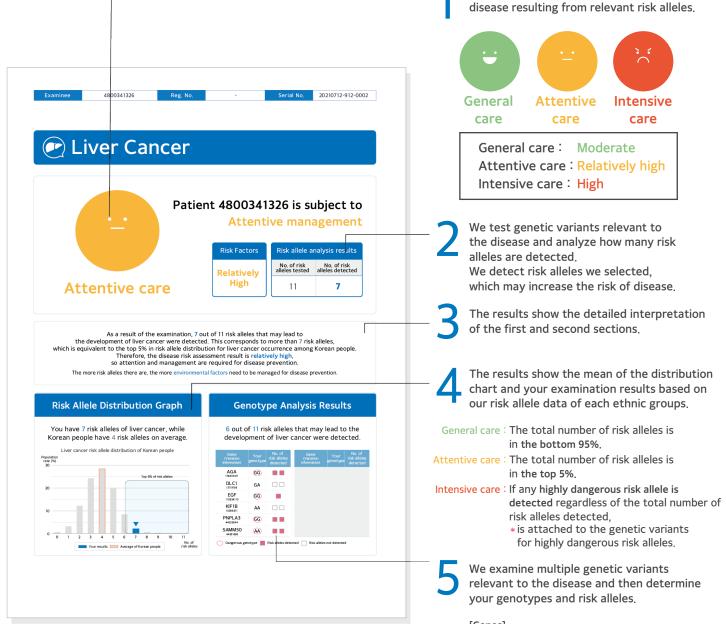


[13/107]





# How to interpret result



[Genes]

Genes subject to examination and genetic variant identification numbers [My genotype]

The results show a degree of risk of

Your identified genotype. If increasing the risk of disease, it is marked with .

[Risk allele detection results]

Risk alleles detected : Risk alleles not detected

The sum of the number of ■and □ means the total number of risk alleles examined

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sol

Confirmed by M.D. Gi Chang-seok (547)



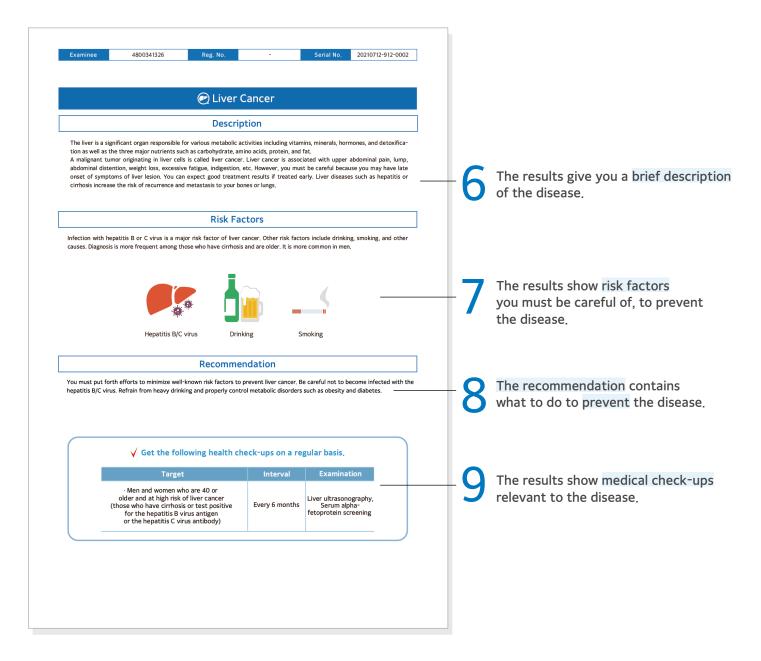
[14/107]





Examinee Sample ID 14443 Reception No. 20230221-171-0030 Asian-male

# How to interpret result



Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[15/107]





# Interpretation by Disease



See [How to interpret result] to understand the Interpretation by Disease more easily.

Genome Health Genetic Testing checks for specific risk alleles and genetic variants. You may develop a certain disease or have other variants even though this report does not show significant results.

Tested by M.T. Lee Myeong-geun (20058) MKJu

Confirmed by M.D. Seol Chang-an (1037) Chazalas Chazalas

Confirmed by M.D. Gi Chang-seok (547)



[16/107]





Sample ID Reception No. Examinee Asian-male 14443 20230221-171-0030





## Patient Asian-male is subject to Attentive care

Risk of Disease Relatively high

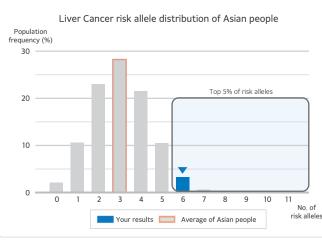
Risk allele analysis results	
No. of risk alleles tested	No. of risk alleles detected
11	6

As a result of the examination, 6 out of 11 risk alleles that may lead to the development of Liver Cancer were detected. This corresponds to more than 6 risk alleles, which is equivalent to the top 5% in risk allele distribution for Liver Cancer among Asian people. Therefore, the disease risk assessment result is relatively high. Close attention and management are required to prevent the disease.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

#### **Risk Allele Distribution Graph**

You have 6 risk alleles of Liver Cancer, while Asian people have 3 risk alleles on average.



### **Genotype Analysis Results**

6 out of 11 risk alleles that may lead to the development of Liver Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[17/107]







#### **Description**

The liver is a significant organ responsible for various metabolic activities including vitamins, minerals, hormones, and detoxification as well as the three major nutrients such as carbohydrate, protein, and fat. A malignant tumor originating in liver cells is called liver cancer. Liver cancer is associated with upper abdominal pain, lump, abdominal distention, weight loss, excessive fatigue, indigestion, etc. However, you must be careful because you may have late onset of symptoms of liver lesion. You can expect good treatment results if treated early. Liver diseases such as hepatitis or cirrhosis increase the risk of recurrence and metastasis to your bones or lungs.

#### **Risk Factors**

Infection with hepatitis B or C virus is a major risk factor of liver cancer. Other risk factors include drinking, smoking, and other causes. Diagnosis is more frequent among those who have cirrhosis and are older. It is more common in men.



#### Recommendation

You must put forth efforts to minimize well-known risk factors to prevent liver cancer. Be careful not to become infected with the hepatitis B/C virus. Refrain from heavy drinking and properly control metabolic disorders such as obesity and diabetes.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
<ul> <li>Men and women who are 40 or older and at high risk of liver cancer (those who have cirrhosis or test positive for the hepatitis B virus antigen or the hepatitis C virus antibody)</li> </ul>	Every 6 months	Liver ultrasound, Serum alpha- fetoprotein screening

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[18/107]





Sample ID Reception No. Examinee Asian-male 14443 20230221-171-0030





## Patient Asian-male is subject to General care

Risk of Disease Moderate

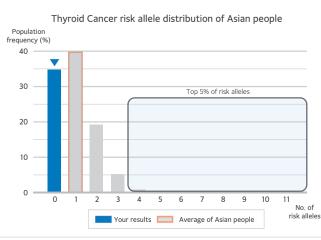
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
11	0	

As a result of the examination, 0 out of 11 risk alleles that may lead to the development of Thyroid Cancer were detected. This corresponds to less than 4 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Thyroid Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

#### **Risk Allele Distribution Graph**

You have **0** risk alleles of Thyroid Cancer, while Asian people have 1 risk alleles on average.



## **Genotype Analysis Results**

0 out of 11 risk alleles that may lead to the development of Thyroid Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[19/107]





## ( Thyroid Cancer

#### **Description**

The thyroid is a butterfly-shaped endocrine organ located below the thyroid cartilage that protrudes in the front of the neck. The thyroid is involved in metabolism by producing and storing the thyroid hormones that regulate body temperature, metabolism, and growth and releasing the hormones into the bloodstream when necessary. Lumps that develop in the thyroid are called thyroid nodules. 5~10% of these nodules are found to be cancerous. Unlike benign nodules, thyroid cancer increases in size and invades the surrounding tissues or may cause lymph node metastases and remote metastasis. In many cases, patients with thyroid cancer do not experience any symptoms. In the case of advanced cancer, the patient may find a mass in the neck and the tumor may invade the nerves of the vocal cords, which causes vocal changes. If the tumor grows larger, the patient may experience difficulty swallowing or shortness of breath.

#### **Risk Factors**

The most well-known risk factor is radiation therapy to the area of the neck. The risk of thyroid cancer increases as the patient is exposed to a radiation accident or radiation at younger ages and is exposed to a higher degree of radiation. Patients with a family history of thyroid cancer at are higher risk. According to recent reports, obesity was found to increase the risk of thyroid cancer.



Radiation therapy history and a degree of exposure to radiation



Family history



Obesity

#### Recommendation

Be careful for your neck not to be exposed to excessive radiation. In particular, children should not be exposed to radiation, especially in their heads and necks.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Those who have a family history of medullary thyroid cancer	-	RET proto-oncogene testing

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[20/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030



# (a) Gallbladder Cancer



## Patient Asian-male is subject to General care

Risk of Disease Moderate

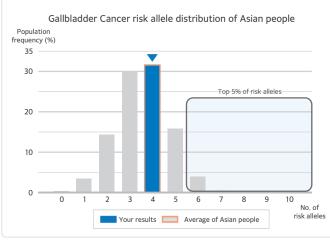
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
10	4	

As a result of the examination, 4 out of 10 risk alleles that may lead to the development of Gallbladder Cancer were detected. This corresponds to less than 6 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Gallbladder Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 4 risk alleles of Gallbladder Cancer, while Asian people have 4 risk alleles on average.



### **Genotype Analysis Results**

4 out of 10 risk alleles that may lead to the development of Gallbladder Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[21/107]





# (a) Gallbladder Cancer

#### **Description**

The gallbladder, located beneath the liver, is an organ where bile produced in the liver is temporarily stored. The gallbladder helps in the digestion and absorption of fats. Cancer that develops in the gallbladder is called gallbladder cancer. The biliary tract sends bile produced in the liver to the duodenum. Cancer that develops in the biliary tract is called bile duct cancer. Patients with gallbladder cancer or bile duct cancer do not experience any specific symptoms. Therefore the early diagnosis is very difficult. Patients with early-stage gallbladder/bile duct cancer who come to the hospital because of a stomachache or an abnormal finding of a liver function test are sometimes misdiagnosed with gallstones. Nonspecific symptoms include weight loss, fatigue, loss of appetite, nausea, vomiting, upper abdominal/epigastric pain, and jaundice, and are sometimes accompanied by the duodenum and colonic obstruction.

#### **Risk Factors**

The mechanism of the onset of gallbladder cancer and bile duct cancer is not known. It is deemed that both environmental factors and genetic factors are relevant to the diseases. Some medical conditions such as gallstones and chronic cholecystitis are known to have significant effects on the onset of gallbladder/bile duct cancer, but its process is unclear.



#### Recommendation

There is no clear guideline or check-up standard which may help in the prevention of gallbladder cancer or bile duct cancer. There is no early diagnosis method recommended. However, you should avoid risk factors in your daily life.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Men and women who are 50 or older	Every year	Blood test, Abdominal ultrasound

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[22/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030

# Colorectal Cancer



## Patient Asian-male is subject to General care

Risk of Disease Moderate

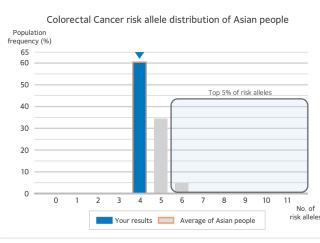
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
11	4	

As a result of the examination, 4 out of 11 risk alleles that may lead to the development of Colorectal Cancer were detected. This corresponds to less than 6 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Colorectal Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

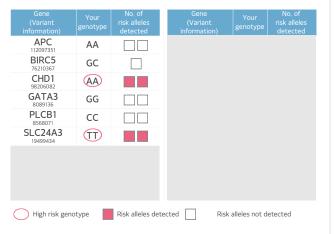
## **Risk Allele Distribution Graph**

You have 4 risk alleles of Colorectal Cancer, while Asian people have 4 risk alleles on average.



### **Genotype Analysis Results**

4 out of 11 risk alleles that may lead to the development of Colorectal Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[23/107]





# Colorectal Cancer

#### **Description**

The large intestine is a long digestive organ starting from the small intestine and connected to the anus. The large intestine is involved in the storage and excretion of food through digestion and absorbs water. The large intestine is divided into the vermiform appendix, cecum, colon, rectum, and anal canal. A malignant tumor that develops in the cecum or the colon among these sections is called colorectal cancer. Patients with early-stage colorectal cancer generally do not experience any symptoms. Colorectal cancer is likely to have advanced already when symptoms appear. Common signs and symptoms of colorectal cancer include changes in a person's bowel habits, diarrhea, constipation, rectal tenesmus, blood in the stool, mucous stool, stomachache, abdominal distention, fatigue, loss of appetite, indigestion, and abdominal mass (lump in the abdomen). Even though a patient undergoes radical proctocolectomy, cancer is likely to recur and there is a high risk of metastasis. Patients should get regular check-ups proactively for 5 years after having an operation known to reduce the likelihood of recurrence.

#### **Risk Factors**

Risk factors of colorectal cancer include those 50 or older, high intake of red meat and processed meats, obesity, alcohol use, genetic factors (hereditary colorectal cancer, hereditary colonic polyps, etc.), and relevant medical conditions such as adenomatous colonic polyps and chronic inflammatory bowel disease.









Red meat and processed meats

Obesity

Colorectal adenomatous polyps

Chronic inflammatory bowel disease

#### Recommendation

Increase your physical activity through regular exercise, avoid alcohol use, and get regular check-ups. Consume a diet high in fiber and calcium and reduce the intake of red meat (beef, pork).

### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Men and women who are 50 or older	Every year	If there are any abnormal findings in a fecal occult blood test, get a colonoscopy. (If a colonoscopy is not available, a double-contrast barium enema may be provided selectively.)

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[24/107]





Sample ID Reception No. Examinee Asian-male 14443 20230221-171-0030

# 1 Head and Neck Cancer



## Patient Asian-male is subject to General care

Risk of Disease Moderate

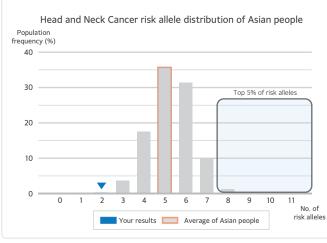
Risk allele analysis results			
No. of risk alleles tested alleles detected			
11	2		

As a result of the examination, 2 out of 11 risk alleles that may lead to the development of Head and Neck Cancer were detected. This corresponds to less than 8 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Head and Neck Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

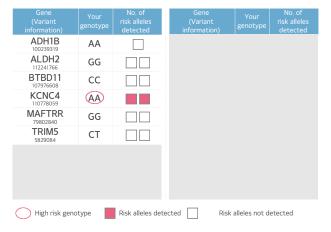
# **Risk Allele Distribution Graph**

You have 2 risk alleles of Head and Neck Cancer, while Asian people have 5 risk alleles on average.



## **Genotype Analysis Results**

2 out of 11 risk alleles that may lead to the development of Head and Neck Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[25/107]





# Head and Neck Cancer

#### **Description**

Head and neck cancer refers to any malignant tumor that develops in the face, nose, neck, oral cavity, larynx, pharynx, salivary glands, and thyroid, except the brain and eyes. Cancer that develops in the oral cavity, the larynx involved in producing sound, and the pharynx is called oral cancer, laryngeal cancer, and pharyngeal cancer, respectively. Pharyngeal cancer is divided into nasopharyngeal cancer, oropharyngeal cancer, and hypopharyngeal cancer, depending on the location of the lesion. Thyroid cancer is included in head and neck cancer in a comprehensive sense. Signs and symptoms vary depending on the location of the lesion. However, common symptoms include voice changes, difficulty swallowing, shortness of breath, and foreign body sensation in the neck, since the relevant organs are involved in speaking, breathing, and swallowing. It is recommended that you undergo a head and neck cancer test if you have any symptoms.

#### **Risk Factors**

Risk factors include smoking, drinking, and viral infections. Smoking increases the risk of head and neck cancer in the organs exposed to smoke, while tobacco smoke containing carcinogens seeps into the lungs through the oral cavity, pharynx, and larynx. Drinking also increases the risk of hypopharyngeal cancer and laryngeal cancer. Other risk factors include gastroesophageal reflux disease, esophagopathy, radiation, UV light, and deficiencies of vitamins and iron.



#### Recommendation

To prevent head and neck cancer, which is caused by a variety of factors, it is recommended that you quit smoking, avoid (or reduce) alcohol use, and receive the HPV vaccine. Eat healthy and/or organic foods and maintain a healthy lifestyle.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Those who have nonspecific symptoms and are associated with any risk factor	Regular medical consultations and check-ups recommended	Get regular check-ups and see an otolaryngologist when you have any symptoms.

Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[26/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030





### Patient Asian-male is subject to General care

Risk of Disease Moderate

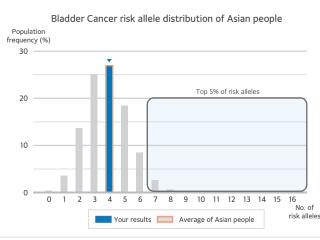
Risk allele analysis results			
No. of risk alleles tested No. of risk alleles detected			
16	4		

As a result of the examination, 4 out of 16 risk alleles that may lead to the development of Bladder Cancer were detected. This corresponds to less than 7 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Bladder Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

#### **Risk Allele Distribution Graph**

You have 4 risk alleles of Bladder Cancer, while Asian people have 4 risk alleles on average.



## **Genotype Analysis Results**

4 out of 16 risk alleles that may lead to the development of Bladder Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[27/107]





# Bladder Cancer

#### **Description**

Bladder cancer is a malignant tumor that develops in the urinary bladder. Bladder cancer is mostly epithelial tumors arising from epithelial cells. Depending on the progression, bladder cancer is also divided into non -muscular bladder cancer, which is limited to the bladder mucosa or the lower mucous membrane, and myofascial bladder cancer and metastatic bladder cancer in which bladder cancer invaded the muscle layer. One of the most common symptoms of bladder cancer is visually identifiable blood in the urine without pain. Other signs and symptoms include frequent urination, pain with urination, and exigent urinary incontinence. When bladder cancer has already advanced, patients may experience weight loss and other symptoms in the metastasis site such as bone pain resulting from bone metastasis. Patients with advanced bladder cancer also may find a lump in the lower abdomen or have flank pain due to hydronephrosis.

#### **Risk Factors**

Risk factors of bladder cancer include old age, smoking, exposure to aromatic amines, phenacetin, cyclophosphamide, chronic urinary tract infection, and radiation therapy to the pelvis.



#### Recommendation

The most important and effective way to prevent bladder cancer is not to smoke and to avoid secondhand smoke. Aromatic amines should be handled safely. Eating plenty of fruits and vegetables high in antioxidants and phytochemicals helps in the prevention of bladder cancer.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Those who are associated with risk factors	Regular medical consultations recommended	Urine test, Urine cytology, Cystoscopy

Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[28/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030

# Esophageal Cancer



## Patient Asian-male is subject to General care

Risk of Disease Moderate

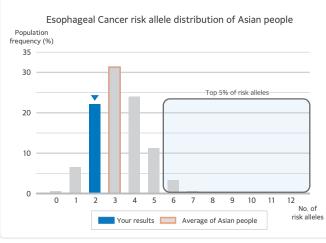
Risk allele analysis results			
No. of risk alleles tested alleles detected			
12	2		

As a result of the examination, 2 out of 12 risk alleles that may lead to the development of Esophageal Cancer were detected. This corresponds to less than 6 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Esophageal Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

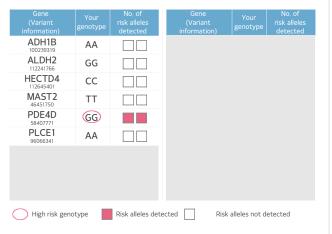
#### **Risk Allele Distribution Graph**

You have 2 risk alleles of Esophageal Cancer, while Asian people have 3 risk alleles on average.



#### **Genotype Analysis Results**

2 out of 12 risk alleles that may lead to the development of Esophageal Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[29/107]





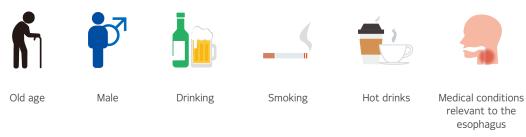
## Esophageal Cancer

#### **Description**

Esophageal cancer is cancer arising from the esophagus. Esophageal cancer is divided into cervical esophageal cancer, thoracic esophageal cancer, and gastroesophageal junction cancer, depending on the location of the lesion. One of the most common symptoms is difficulty in swallowing. In the case of advanced esophageal cancer, patients commonly experience weight loss due to undernutrition. Patients may suffer pain, but it is also common not to feel any pain.

#### **Risk Factors**

Esophageal cancer is common in men over the age of 60. Drinking, smoking, obesity, and frequent consumption of hot drinks (65°C or over) increase the risk of esophageal cancer. Some medical conditions such as gastroesophageal reflux disease, Barrett's esophagus, esophageal achalasia, esophagus damage, and strictures are also known to increase the risk of esophageal cancer.



#### Recommendation

Prevention includes stopping smoking and drinking and eating a healthy diet. If you are diagnosed with an esophageal disease such as gastroesophageal reflux disease or Barrett's esophagus, get proper treatment and regular check-ups for the prevention and early diagnosis of esophageal cancer.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Those who have any symptoms or are suspected to have esophageal cancer	Regular check -ups recommended	Biopsy when any abnormal findings are found during medical consultation or endoscopy, computed tomography (CT), or endoscopic ultrasound

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[30/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030





## Patient Asian-male is subject to General care

Risk of Disease Moderate

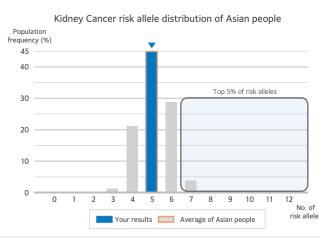
Risk allele analysis results				
No. of risk alleles tested alleles detected				
12	5			

As a result of the examination, 5 out of 12 risk alleles that may lead to the development of Kidney Cancer were detected. This corresponds to less than 7 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Kidney Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

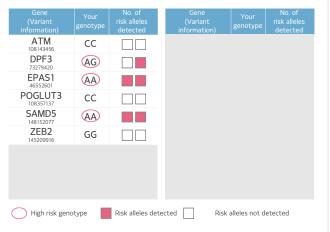
#### **Risk Allele Distribution Graph**

You have 5 risk alleles of Kidney Cancer, while Asian people have 5 risk alleles on average.



## **Genotype Analysis Results**

5 out of 12 risk alleles that may lead to the development of Kidney Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[31/107]





# Kidney Cancer

#### **Description**

Kidney cancer is a malignant tumor that starts in the kidneys. Kidney cancer is divided into renal pelvic cancer and renal cell cancer, depending on the location of the lesion. Generally, kidney cancer refers to renal cell cancer which is a malignant tumor. 85~90% of tumors arising from the kidneys are renal cell cancer. In many cases, kidney cancer does not cause any symptoms. As kidney cancer becomes more advanced or spreads to other organs, it results in flank pain, blood in the urine, and a mass (or a lump) in the flank or the upper abdomen. Only 10~15% of cases experience all these symptoms. Other signs and symptoms include weight loss, fatigue, loss of appetite, and fever.

#### **Risk Factors**

The causes of renal cell cancer are not clearly known. However, environmental factors, lifestyles, underlying renal diseases, and genetic factors are considered to be associated with the disease. Environmental factors and lifestyle factors include smoking, obesity, hypertension, and dietary habits (high intake of animal fat and high-calorie foods).



#### Recommendation

Reduce the intake of animal fat and increase the consumption of fruits and vegetables. Stopping smoking, general health management such as regular exercise, and maintaining a healthy weight help in the prevention of kidney cancer. Hypertension is a risk factor for renal cell cancer, so proper blood pressure control is required.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Men and women who are 40 or older	Every year	Abdominal ultrasound Those who have medical conditions that increase the risk of renal cell cancer, such as long-term hemodialysis, or those who have relevant genetic factors such as a family history of Von Hippel-Lindau syndrome need regular check -ups.

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[32/107]





Sample ID Reception No. Examinee Asian-male 14443 20230221-171-0030

# Gastric Cancer



### Patient Asian-male is subject to General care

Risk of Disease Moderate

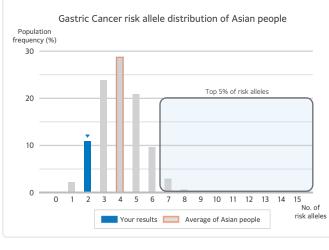
Risk allele analysis results				
No. of risk alleles tested alleles detected				
15	2			

As a result of the examination, 2 out of 15 risk alleles that may lead to the development of Gastric Cancer were detected. This corresponds to less than 7 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Gastric Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

#### **Risk Allele Distribution Graph**

You have 2 risk alleles of Gastric Cancer, while Asian people have 4 risk alleles on average.



### **Genotype Analysis Results**

2 out of 15 risk alleles that may lead to the development of Gastric Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[33/107]





# (>) Gastric Cancer

#### Description

The stomach, a digestive organ located between the esophagus and the duodenum, stores and digests food from the esophagus and passes the food to the small intestine. Gastric cancer is a malignant tumor that develops in the stomach. Early symptoms may include indigestion, heartburn, upper abdominal pain or discomfort, nausea, weight loss, loss of appetite, and fatigue. In the case of advanced gastric cancer, the patient may experience vomiting since the part between the stomach and the duodenum narrows or is blocked. Other signs may include vomiting blood, blood in the stool, abdominal lump, and hepatomegaly. However, many patients with early gastric cancer do not experience any symptoms and find their condition by accident. Therefore, it is important to undergo gastroscopy on a regular basis.

#### **Risk Factors**

It is known that gastric cancer is associated with genetic factors (familial adenomatous polyposis, hereditary diffuse gastric cancer, etc.) and some environmental factors such as dietary habits (salt-rich foods, burnt foods, and smoked foods), smoking, drinking, and ionizing radiation. Helicobacter pylori infection (40~60% of patients with gastric cancer test positive for Helicobacter pylori), the patient's history of stomach surgery, and his/her family history are associated with a higher risk of gastric cancer.



#### Recommendation

Eat plenty of fresh vegetables and fruits. Try not to eat salt-rich foods, burnt meat or fish, and smoked foods, Do not smoke because the risk of gastric cancer has been found to increase 1.5 to 2.5 times for smokers. Lastly, do not consume alcohol since long-term alcohol use may increase the risk of gastric cancer.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of CDH1 and other gastric cancer related genes.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Men and women who are 40 or older	Every 2 years	Gastroscopy (If gastroscopy is not available, you may get an upper gastrointestinal series instead.)

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[34/107]





Sample ID Reception No. Examinee Asian-male 14443 20230221-171-0030





## Patient Asian-male is subject to General care

Risk of Disease Moderate

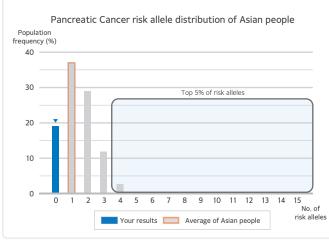
Risk allele analysis results			
No. of risk alleles tested alleles detected			
15	0		

As a result of the examination, 0 out of 15 risk alleles that may lead to the development of Pancreatic Cancer were detected. This corresponds to less than 4 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Pancreatic Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have **0** risk alleles of Pancreatic Cancer, while Asian people have 1 risk alleles on average.



### **Genotype Analysis Results**

0 out of 15 risk alleles that may lead to the development of Pancreatic Cancer were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information	Your genotype	No. of risk alleles detected
ABLIM2 8107943	GG				
CLPTM1L 1322087	GG				
DOK2 21766881	TT				
DPP6 153625843	CC				
ITGA3 48154668	GG				
LINC01173 235615197	CA				
NR5A2 200007432	GG				
PKN1 14574897	CC				
High risk geno	otype	Risk alleles det	ected	Risk alleles not de	etected

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[35/107]







#### **Description**

The pancreas, which is located behind the stomach and connected to the duodenum, secretes digestive enzymes and hormones. Insulin and glucagon, involved in blood sugar control, are also secreted from the pancreas. Pancreatic cancer is a malignant tumor arising from the pancreas. Tumors mainly develop in exocrine cells. Pancreatic cancer is one of the tumors with poor progress after treatment. The most common symptoms of pancreatic cancer include stomachache and weight loss. It may be accompanied by jaundice or indigestion and diabetes may occur or worsen.

#### **Risk Factors**

It is considered that both environmental factors and genetic factors are involved in the onset of pancreatic cancer. Among the genetic factors, KRAS gene variation is particularly important. KRAS variant is detected in more than 90% of pancreatic cancers, making it the most frequent genetic abnormality in all carcinomas. Environmental factors include smoking, obesity, diabetes, chronic pancreatitis, familial pancreatic cancer, age, drinking, dietary habits, and chemicals.









Diabetes

Chronic pancreatitis Familial pancreatic cancer

Chemicals

#### Recommendation

There is no clear pancreatic cancer prevention guideline. The best way to prevent pancreatic cancer is to avoid risk factors in your daily life. In particular, smoking is an important risk factor, so you must quit smoking. Avoid high-fat and high-calorie foods and eat plenty of fruits and vegetables in order to maintain a healthy weight which is basic to health. Those who have diabetes or chronic pancreatitis are known to be associated with the risk of pancreatic cancer must control the risk factors to the greatest extent possible through constant treatment.

#### ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· A family history of pancreatic cancer, 70 or older, smoking for a long time, or a medical history of chronic pancreatitis	Every year	Abdominal ultrasound, Abdominal CT

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[36/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030





## Patient Asian-male is subject to General care

Risk of Disease Moderate

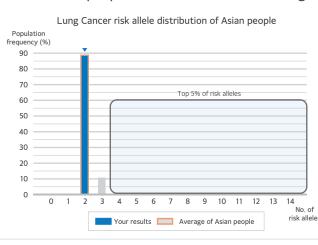
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
14	2	

As a result of the examination, 2 out of 14 risk alleles that may lead to the development of Lung Cancer were detected. This corresponds to less than 4 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Lung Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 2 risk alleles of Lung Cancer, while Asian people have 2 risk alleles on average.



## **Genotype Analysis Results**

2 out of 14 risk alleles that may lead to the development of Lung Cancer were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[37/107]





## Lung Cancer

## **Description**

Lung cancer is a malignant tumor arising in the lung, divided into non-small-cell lung carcinoma and small-cell lung carcinoma. Small-cell lung carcinoma is known to be more malignant. In many cases, lung cancer has already spread to other organs or the other lung through lymphatic vessels or blood vessels when the cancer is found. Lung cancer has a high risk of recurrence and metastasis and shows low recovery rates. As a result, there is a high risk of death. Therefore, prevention is the most important against lung cancer. Patients with lung cancer should manage their health constantly even after treatment.

#### Risk Factors

Smoking is the main contributor to lung cancer. There is more than 10 times the risk of lung cancer for smokers than nonsmokers. Other risk factors known to increase the risk of lung cancer include secondhand smoke, asbestos, radioisotopes (e.g., radon), heavy metals in polluted air, and fine dust. Pulmonary diseases and genetic factors (family history) are also known to be associated with the onset of lung cancer.



#### Recommendation

The most important prevention method of lung cancer is the avoidance of smoking. In addition, you need to avoid or reduce environmental factors to the greatest extent possible. Lastly, you should strengthen your immune system through a balanced diet, which is important in preventing all types of cancer.

## ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Men and women 54~74 who are high risk for lung cancer	Every 2 years	Low-dose chest CT

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[38/107]







# Testicular Cancer



## Patient Asian-male is subject to General care

Risk of Disease Moderate

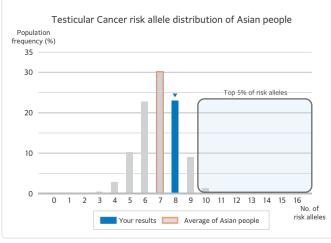
Risk allele analysis results		
No. of risk alleles tested alleles detected		
16	8	

As a result of the examination, 8 out of 16 risk alleles that may lead to the development of Testicular Cancer were detected. This corresponds to less than 10 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Testicular Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 8 risk alleles of Testicular Cancer, while Asian people have 7 risk alleles on average.



## **Genotype Analysis Results**

8 out of 16 risk alleles that may lead to the development of Testicular Cancer were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
ACTL8 18097445	GG				
C12orf50 88379520	AA				
DMRT1 845516	AA				
DMRT1 878563	AA				
KITLG 88953561	TT				
KITLG 88953959	TT				
KITLG 88955469	TT				
NELL1 20906640	AA				
High risk gend	otype	Risk alleles dete	ected Ri	isk alleles not de	etected

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[39/107]





# ( Testicular Cancer

## **Description**

Testicular cancer is a malignant tumor that develops in the testis. The onset of testicular cancer most commonly occurs in young males in their 30s or 40s and toddlers before 4 years old. Sometimes testicular cancer is found by accident due to scrotal trauma. Patients with testicular cancer may find a slowly growing painless hard lump in the testis. Other symptoms include testicular atrophy and acute pain resulting from bleeding or infarction in the testis. Some cases are accompanied by breast enlargement.

#### **Risk Factors**

Causes of testicular cancer are divided into congenital causes and acquired causes. Congenital causes include cryptorchidism, relevant medical history, and family history, Acquired causes include trauma, mother administered with female sex hormones during pregnancy, exposure to chemicals that may cause testicular atrophy, and infection with the mumps virus.











Cryptorchidism

Medical history

Family history

Chemicals

Viral infection

#### Recommendation

There is no specific way to prevent testicular cancer. The earlier the disease is found, the better the treatment results.

## ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
-	-	Males should conduct a testicular self-examination every month from adolescence. If the scrotum increases in size or lump or edema is found, diagnostic imaging and a serum tumor marker test help in diagnosis.

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[40/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

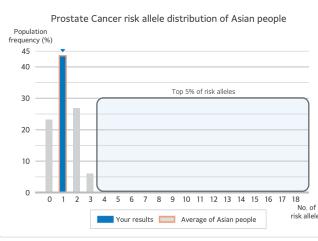
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
18	1	

As a result of the examination, 1 out of 18 risk alleles that may lead to the development of Prostate Cancer were detected. This corresponds to less than 4 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Prostate Cancer occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 1 risk alleles of Prostate Cancer, while Asian people have 1 risk alleles on average.



## **Genotype Analysis Results**

1 out of 18 risk alleles that may lead to the development of Prostate Cancer were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
* BRCA1 41256280	AA				
* BRCA2 32907014	AA				
* BRCA2 32930609	CC				
CASC19 128208369	TT				
CASC8 128485038	GG				
CCAT2 128413305	TG				
HOXB13 46805705	GG				
PCAT1 128077146	GG				
PCAT1 128131809	GG				
High risk genotype Risk alleles detected Risk alleles not detected  Gene requiring intensive care (PV): Even a single variant may greatly increase the risk of Prostate Cancer.					

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[41/107]





# Prostate Cancer

## Description

The prostate, a chestnut-sized organ that is a part of the male reproductive system and located below the bladder, produces and secretes sperm. Prostate cancer is a medical condition in which the cells of the prostate divide and grow abnormally and consequently become a malignant tumor. The tumor may invade nearby tissues without being confined to the prostate or may spread to other organs through blood vessels or lymphatic vessels. Prostate cancer may initially cause no symptoms. In later stages, patients experience a variety of urinary problems (nocturia, frequent urination, urinary hesitancy, etc.). Symptoms of more advanced prostate cancer include hydronephrosis (the swelling of a kidney due to the blockage of the ureter), renal failure, bone pain due to metastasis (including backache and sciatic neuralgia), and distal femur or spinal fractures.

#### **Risk Factors**

Risk factors of prostate cancer include old age (sharply increasing at age 50 and older), race (least common among Asian men), hereditary factors, a family history of prostate cancer, male sex hormones, diabetes, obesity, westernized high-calorie diet (increased intake of animal fat), and infection (urinary tract infection such as chronic prostatitis).









Family history

Male sex hormones

Chemicals

Infection

#### Recommendation

A healthy diet is important. Avoid high-calorie foods and fatty meat and eat plenty of fiber-rich foods, fresh fruits and vegetables, unpolished or less polished grains (wheat, rye, etc.), and beans. Maintain a healthy weight. Walk or exercise enough to more than five times a week, for more than 30 minutes each time.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of BRCA1, BRCA2 and other prostate cancer related genes.

## ✓ Get the following health check-ups on a regular basis.

Target	Intervals	Examinations
· Men who are 50 or older	Every year	Prostate specific antigen (PSA) test, Prostate ultrasound

Men with a BRCA gene family history (family history of breast cancer or prostate cancer), less than 5% of the total population, have a report that active early screening from the age of 45 lowers the mortality rate. Therefore, men with a family history of breast cancer or prostate cancer should actively undergo prostate screening and genetic testing, However, regular prostate cancer screening is not recommended for those aged 75 or older and with no symptoms.

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Challenge





[42/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030





## Patient Asian-male is subject to General care

Risk of Disease Moderate

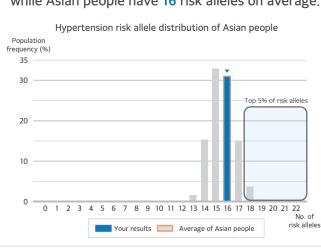
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
22	16	

As a result of the examination, 16 out of 22 risk alleles that may lead to the development of Hypertension were detected. This corresponds to less than 18 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Hypertension occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

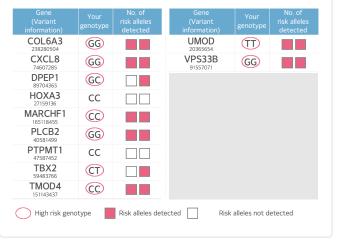
## **Risk Allele Distribution Graph**

You have 16 risk alleles of Hypertension, while Asian people have 16 risk alleles on average.



## **Genotype Analysis Results**

16 out of 22 risk alleles that may lead to the development of Hypertension were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[43/107]





# **Hypertension**

## **Description**

Hypertension is a medical condition in which the blood pressure in the arteries is higher than the normal level. Generally, hypertension is diagnosed when an adult over 18 has systolic blood pressure exceeding 140mmHg or diastolic blood pressure over 90mmHg. Hypertension may be caused by various reasons. Primary (essential) hypertension is defined as high blood pressure with no specific medical condition identified as a cause of high blood pressure, whereas secondary hypertension is hypertension due to an identifiable medical condition. No clear cause of primary hypertension has been identified. It is known to result from a complex interaction of various factors. About 90~95% of cases are primary hypertension. Hypertension typically does not cause clear symptoms. Long-term high blood pressure, however, may cause complications such as coronary artery disease and cerebrovascular disease.

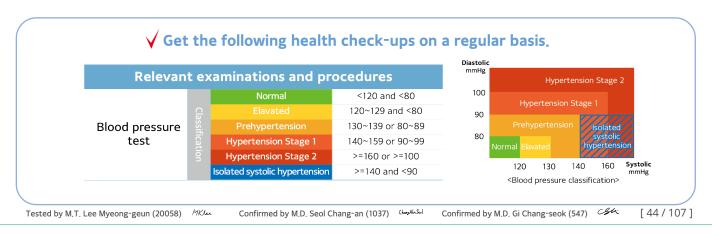
#### **Risk Factors**

Hypertension is caused by a combination of various factors. Hypertension is common in those who have a family history of hypertension. This medical condition may be caused by obesity, lack of exercise, and other lifestyle factors such as excess salt in the diet. Other risk factors include alcohol, smoking, and diabetes.



#### Recommendation

Healthy dietary habits are important in the prevention of hypertension. Avoid high salt intake and eat a low -sodium diet. Reduce fat intake and eat vegetables and fruits containing potassium. Prevent obesity by maintaining a healthy weight. Work out for around 30 minutes, 3 to 4 times a week. Reducing alcohol use, quitting smoking, and managing stress are required to prevent hypertension.









# Coronary Artery Disease



## Patient Asian-male is subject to General care

Risk of Disease Moderate

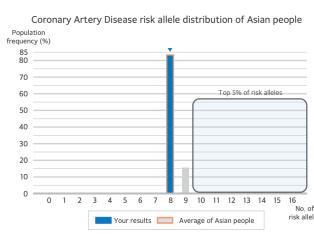
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
16	8	

As a result of the examination, 8 out of 16 risk alleles that may lead to the development of Coronary Artery Disease were detected. This corresponds to less than 10 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Coronary Artery Disease occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 8 risk alleles of Coronary Artery Disease, while Asian people have 8 risk alleles on average.



## **Genotype Analysis Results**

8 out of 16 risk alleles that may lead to the development of Coronary Artery Disease were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information	Your genotype	No. of risk alleles detected
APOA5 116661392	GG				
COL6A3 238280504	GG				
CXCL8 74607285	GG				
LPA 161013013	AA				
MARCHF1 165118455	CC				
PLCB2 40581499	GG				
TMOD4 151143437	CC				
VPS33B 91557071	GG				
High risk geno	otype	Risk alleles det	ected	Risk alleles not d	etected

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[45/107]





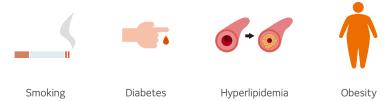
# Coronary Artery Disease

## **Description**

The heart pumps blood to transport oxygen and nutrients to the body. The blood vessels located on the surface of the heart are called the coronary arteries. The coronary arteries transport blood to the heart muscle whenever the heart beats. Coronary artery disease is a medical condition that involves the reduction of the blood flow to the heart muscle due to the narrowing of coronary arteries. Common types include angina pectoris and myocardial infarction. A common symptom is chest pain, but sometimes no symptoms are present. One of the major causes is arteriosclerosis which is the hardening and loss of elasticity of the blood vessels due to the buildup of fatty plaque. It is important to manage hyperlipidemia to prevent coronary artery disease.

#### **Risk Factors**

There are various risk factors of coronary artery disease. Common risk factors include age, smoking, diabetes, hyperlipidemia, hypertension, and a family history of the early onset of coronary artery disease.



#### Recommendation

To prevent coronary artery disease, it is important to maintain a healthy lifestyle that reduces blood cholesterol levels. Reduce the intake of saturated fat and high-cholesterol foods and eat plant-based foods such as fruits and vegetables. Regular exercise helps in the prevention of arteriosclerosis. You need to sufficiently warm up before working out.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Electrocardiography, Echocardiography, Coronary angiography, Exercise stress test

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[46/107]







# 📆 Acute Cardiac Arrest



## Patient Asian-male is subject to General care

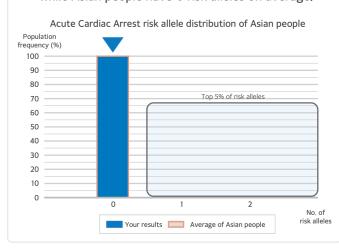
Risk of Disease Moderate

Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
2	0	

As a result of the examination, 0 out of 2 risk alleles that may lead to the development of Acute Cardiac Arrest were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Acute Cardiac Arrest occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

# **Risk Allele Distribution Graph** You have **0** risk alleles of Acute Cardiac Arrest, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

0 out of 2 risk alleles that may lead to the development of Acute Cardiac Arrest were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Saol

Confirmed by M.D. Gi Chang-seok (547)



[47/107]





# Acute Cardiac Arrest

## **Description**

Acute cardiac arrest is a condition in which the heart of a person who looks healthy suddenly fails. In South Korea, approximately 30,000 cases occur each year. Once acute cardiac arrest occurs, blood fails to flow into the brain and other major organs. If not treated within minutes, it leads to death. Ventricular fibrillation, a type of arrhythmia, may cause acute cardiac arrest. If you have a genetic disorder related to arrhythmia, the risk of acute cardiac arrest increases more. The KCNH2 gene is associated with Long QT syndrome that may lead to acute cardiac arrest. After cardiac impulse, the heart's electrical system recharges for the next heartbeat. If you have Long QT syndrome, it requires more time than healthy people, which may cause abnormally fast arrhythmia and acute cardiac arrest.

#### **Risk Factors**

Symptoms of arrhythmia as well as ventricular fibrillation are one of the most common causes of acute cardiac arrest. You must be more cautious of acute cardiac arrest if you have any genetic disorders causing arrhythmia, such as Long QT syndrome and a family history of arrhythmia. In addition, underlying medical conditions such as coronary artery disease and hypertension may cause acute cardiac arrest. Other risk factors include drugs, alcohol, and smoking.

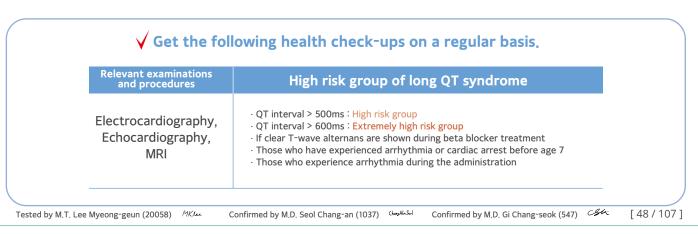


#### Recommendation

If emergency treatment is not performed as soon as acute cardiac arrest occurs, it may lead to death. Therefore, prevention is of utmost importance. It is important to maintain a healthy weight and not to smoke in order to prevent diseases that may lead to acute cardiac arrest, such as coronary artery disease and hypertension. If you have ever been diagnosed with Long QT syndrome or are in a high-risk group, you may prevent acute cardiac arrest through beta blocker treatment or an implantable cardioverter defibrillator after consulting your physician.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of KCNH2 and other acute cardiac arrest related genes.







Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030



# Myocardial Infarction



## Patient Asian-male is subject to General care

Risk of Disease Moderate

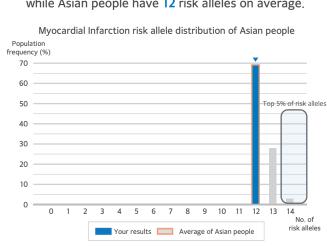
Risk allele an	alysis results
No. of risk alleles tested	No. of risk alleles detected
14	12

As a result of the examination, 12 out of 14 risk alleles that may lead to the development of Myocardial Infarction were detected. This corresponds to less than 14 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Myocardial Infarction occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 12 risk alleles of Myocardial Infarction, while Asian people have 12 risk alleles on average.



## **Genotype Analysis Results**

12 out of 14 risk alleles that may lead to the development of Myocardial Infarction were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[49/107]





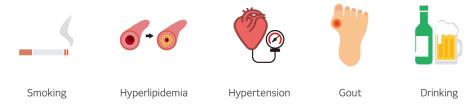
# **(f)** Myocardial Infarction

## **Description**

Myocardial infarction is a medical condition in which the coronary arteries supplying blood to the heart muscle are suddenly blocked due to various reasons such as a thrombus or fast contraction of blood vessels. Oxygen and nutrients stop being supplied to the heart, which consequently leads to the necrosis of the heart muscle. Part of the heart muscle dies permanently and loses its function. The heart becomes unable to pump effectively, which may lead to heart failure or sudden death. The most common symptom is chest pain in the epigastrium or the center of the chest. Other symptoms include shortness of breath, a cold sweat, and vomiting.

#### Risk Factors

The fundamental cause of myocardial infarction is arteriosclerosis. Major risk factors of myocardial infarction include smoking, hyperlipidemia, hypertension, and diabetes which may cause arteriosclerosis. It is important to manage these risk factors thoroughly in preventing myocardial infarction.



#### Recommendation

The most important thing is to prevent arteriosclerosis that causes myocardial infarction. The disease is associated with dietary habits. Eat a low-fat, low-sodium diet and have fresh vegetables and fruits. Work out for at least 30 minutes every day. Quit smoking and reduce alcohol use. If chest pain accompanied by a cold sweat continues for over 30 minutes, it is likely to be a sign of myocardial infarction. In such a case, you must immediately go to the E.R.



#### Relevant examinations and procedures

Electrocardiography, Cardiac enzyme test, Echocardiography, Coronary angiography, CT, MRI

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) (املامالمومها

Confirmed by M.D. Gi Chang-seok (547)



[50/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

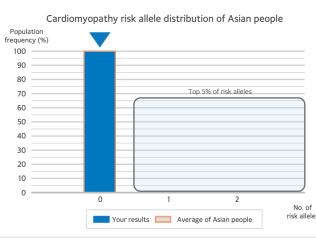
Risk allele an	alysis results		
No. of risk alleles tested alleles detected			
2	0		

As a result of the examination, 0 out of 2 risk alleles that may lead to the development of Cardiomyopathy were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Cardiomyopathy occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have **0** risk alleles of Cardiomyopathy, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

0 out of 2 risk alleles that may lead to the development of Cardiomyopathy were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Saol

Confirmed by M.D. Gi Chang-seok (547)



[51/107]





# ( Cardiomyopathy

## **Description**

Cardiomyopathy is a medical condition that affects the heart muscle and causes heart muscle dysfunction. Secondary heart muscle problems resulting from hypertension or coronary artery disease are not included in cardiomyopathy. Types of cardiomyopathy include dilated cardiomyopathy, hypertrophic cardiomyopathy, and restrictive cardiomyopathy. In dilated cardiomyopathy, the ventricles abnormally become enlarged, which consequently results in pump dysfunction of the heart. In hypertrophic cardiomyopathy, the walls of the ventricles thicken and stiffen. In restrictive cardiomyopathy, the walls of the ventricles stiffen, which causes a problem in the blood flow to the heart. Common symptoms include fatigue and shortness of breath during exercise. These symptoms typically occur slowly. Sometimes patients do not experience any symptoms for several months to several years. TNNI3 is associated with dilated cardiomyopathy.

#### **Risk Factors**

Common causes of cardiomyopathy include family history, inheritance, myocarditis due to a viral infection, and myocardial damage. Other risk factors include myocarditis resulting from the immune mechanism, obesity, and alcohol. Often, the cause remains unknown.







Inheritance

Myocarditis resulting from a viral infection

### Recommendation

It is important to have healthy dietary habits such as a low-sodium diet and to maintain a healthy weight through moderate exercise. Alcohol may be a cause of cardiomyopathy. Avoid heavy alcohol use.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of TNNI3 and other cardiomyopathy related genes.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Echocardiography, Coronary angiography, MRI, Chest X-ray

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[52/107]





# Atrial Fibrillation



## Patient Asian-male is subject to General care

Risk of Disease **Moderate** 

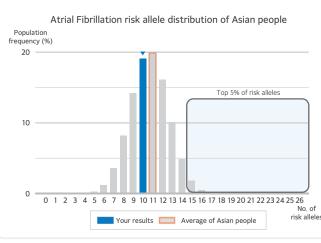
Risk allele an	alysis results			
No. of risk alleles tested alleles detected				
26 10				

As a result of the examination, 10 out of 26 risk alleles that may lead to the development of Atrial Fibrillation were detected. This corresponds to less than 15 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Atrial Fibrillation occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 10 risk alleles of Atrial Fibrillation, while Asian people have 11 risk alleles on average.



## **Genotype Analysis Results**

10 out of 26 risk alleles that may lead to the development of Atrial Fibrillation were detected.

Gene (Variant information)		No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
ASAH1 17913970	CT		PRRX1 170569317	GG	
CAV1 116191301	AA		SH3PXD2A 105480387	AA	
HAND2 174447349	T		TBX5 114793240	GG	
KCNN3 154814353	GG		ZFHX3 73051620	GG	
NEURL1 105299611	TT				
NEURL1 105324774	CC				
NEURL1 105342672	AA				
PITX2 111718067	TC				
PPFIA4 203034906	AA				
203034906  High risk gen	_	Risk alleles det	ected Risk	alleles not d	etected

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[53/107]





# Atrial Fibrillation

## **Description**

The heart repeats contraction and relaxation as electrical stimuli made by self-beating electric cells are delivered to the heart muscle. An arrhythmia is a group of medical conditions in which the heartbeat is abnormal. Of these, atrial fibrillation is an abnormal heart rhythm characterized by the rapid beating of the atria, approximately 600 times per minute. In atrial fibrillation, the atria cannot contract normally and consequently fails to supply the blood to the ventricles properly. There are a variety of symptoms of atrial fibrillation, including heart palpitations, chest pressure, shortness of breath, and dizziness. Often patients with atrial fibrillation experience no symptoms at all.

### **Risk Factors**

Atrial fibrillation easily occurs in patients with heart disease such as cardiac valvular disease, coronary artery disease, and cardiomyopathy. Atrial fibrillation may develop even though you have no heart disease. Atrial fibrillation is more common in older people. It also may result from hypertension or diabetes. It is often caused at night or dawn after drinking.











Old age (65 or older)

Coronary artery disease

Cardiac valvular disorder

Drinking

Overeating

#### Recommendation

Avoid drinking, smoking, coffee, and overeating which may worsen atrial fibrillation. Work out regularly and eat a healthy diet to prevent hypertension and diabetes which may cause atrial fibrillation.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Electrocardiography, Echocardiography, 24-hour electrocardiography (Holter monitoring)

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) (املامالمومها

Confirmed by M.D. Gi Chang-seok (547)



[54/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

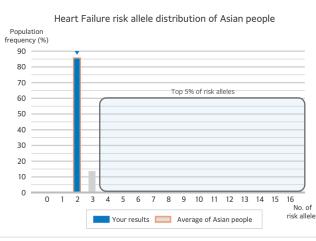
Risk allele an	alysis results
No. of risk alleles tested	No. of risk alleles detected
16	2

As a result of the examination, 2 out of 16 risk alleles that may lead to the development of Heart Failure were detected. This corresponds to less than 4 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Heart Failure occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 2 risk alleles of Heart Failure, while Asian people have 2 risk alleles on average.



## **Genotype Analysis Results**

2 out of 16 risk alleles that may lead to the development of Heart Failure were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[55/107]





# (2) Heart Failure

## **Description**

Heart failure is a condition caused by the failure of the heart's function, as a pump supporting the blood flow through the body, due to various heart diseases. When the function of the left-sided heart is degraded, the blood flow to the body reduces. As a result, patients experience fatigue, weakness, and shortness of breath due to fluid in the lungs. When the function of the right-sided heart is degraded, patients experience body anasarca. Heart failure may be accompanied by insomnia and indigestion as well as shortness of breath, chronic fatigue, and edema.

#### **Risk Factors**

The most common causes of heart failure are coronary artery disease, hypertension, and cardiovascular disease, Avoid heavy alcohol use, a high-sodium diet, and smoking which may cause medical conditions leading to heart failure. Other risk factors include excess stress, obesity, and lack of exercise.











Coronary artery disease

Hypertension

Drinking

High-sodium diet

Smoking

#### Recommendation

Eat a low-sodium, low-fat diet and avoid alcohol use and smoking which may increase the risk of cardiovascular disease that may cause heart failure. Excess stress may also cause heart failure. Manage your stress levels properly. If you have coronary artery disease, hypertension, or hyperlipidemia, you should have aggressive treatment to prevent heart failure.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of LMNA, PKP2, PTPN11 and other heart failure related genes.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Chest X-ray, Electrocardiography, Echocardiography

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[56/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030



# Ischemic Stroke



## Patient Asian-male is subject to General care

Risk of Disease Moderate

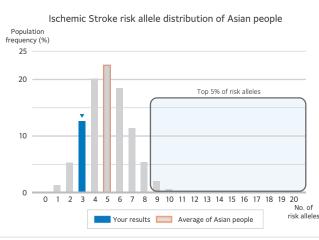
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
20	3	

As a result of the examination, 3 out of 20 risk alleles that may lead to the development of Ischemic Stroke were detected. This corresponds to less than 9 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Ischemic Stroke occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

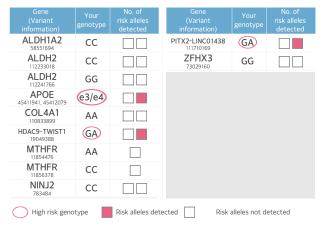
## **Risk Allele Distribution Graph**

You have 3 risk alleles of Ischemic Stroke, while Asian people have 5 risk alleles on average.



## **Genotype Analysis Results**

3 out of 20 risk alleles that may lead to the development of Ischemic Stroke were detected.



Tested by M.T. Lee Myeong-geun (20058) /9K/Lee

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[57/107]







## **Description**

A stroke is a medical condition in which a blocked or ruptured blood vessel of the brain causes brain damage and consequently leads to neurological disorders such as hemiplegia, language disorder, and disorder of consciousness. Strokes are largely divided into ischemic strokes and hemorrhagic strokes. An ischemic stroke is caused by the blockage of a blood vessel leading to brain damage. A hemorrhagic stroke is caused by a ruptured blood vessel of the brain which stops the blood supply, as a result, blood pools. Each blood vessel of the brain is responsible for different functions. A variety of symptoms may appear depending on which blood vessel is blocked or ruptured. Early symptoms include hemiplegia, language disorder, visual problems, dizziness, and severe headache.

#### **Risk Factors**

Medical conditions such as hypertension and diabetes greatly increase the risk of a stroke. Smoking and drinking are significant risk factors. If a blood vessel of the brain weakens, heavy alcohol use, severe stress, excessively vigorous physical activities, and overwork may cause a stroke.



#### Recommendation

Medical conditions such as hypertension, diabetes, and hyperlipidemia are the main causes of stroke. Reduce alcohol use and do not smoke to prevent a stroke. It is also helpful to maintain a healthy weight through steady exercise. A stroke easily recurs. If you have previously experienced a stroke, it is important to get treatment proactively and regular check-ups.



CT, MRI, Angiography, Carotid ultrasound, Echocardiography

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[58/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

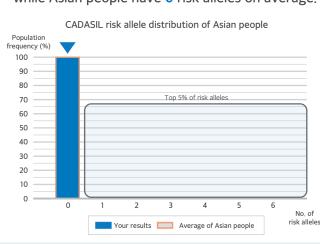
Risk allele an	alysis results			
No. of risk alleles tested alleles detected				
6	0			

As a result of the examination, 0 out of 6 risk alleles that may lead to the development of CADASIL were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for CADASIL occurrence among Asian people. Therefore, the disease risk assessment result is moderate.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 0 risk alleles of CADASIL, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

0 out of 6 risk alleles that may lead to the development of CADASIL were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[59/107]







## **Description**

CADASIL is an autosomal dominant hereditary disease characterized by migraine, recurrent stroke, and progressive dementia. It usually occurs between 40 and 50 years of age and shows a clear family history. CADASIL is caused by various mutations in the NOTCH3 gene on chromosome 19. It raises fine damage on the blood vessel wall which causes blood stream disorders and consequently leads to repetitive subcortical infarcts and vascular dementia. CADASIL can be diagnosed by identifying granular osmiophilic deposits on vascular smooth muscle cells or mutations in the NOTCH3 gene through genetic testing. CADASIL is a progressive medical condition that causes recurrent stroke and dementia. No specific treatment for CADASIL is available currently.

### **Risk Factors**

CADASIL is is an autosomal dominant hereditary disease caused by mutations in the NOTCH3 gene. If a young patient who does not have any risk factors of general stroke such as hypertension, diabetes, and smoking experiences any symptoms of ischemic stroke, CADASIL should be considered.





Inheritance

Mutations in the NOTCH3 gene

#### Recommendation

No fundamental prevention and treatment for CADASIL are available. Hypertension, diabetes, hypercholesterolemia, and vascular risk factors may worsen the symptoms. Such medical conditions need to be managed. Smokers must quit smoking since it increases the risk of stroke.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of NOTCH3 and other CADASIL related genes.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

MRI, CT, Skin biopsy, NOTCH3 genetic testing

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[60/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030



# Moyamoya Disease



## Patient Asian-male is subject to General care

Risk of Disease Moderate

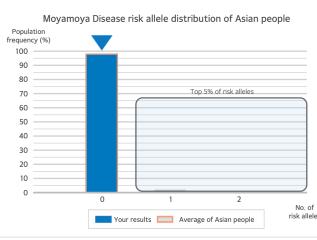
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
2	0	

As a result of the examination, 0 out of 2 risk alleles that may lead to the development of Moyamoya Disease were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Moyamoya Disease occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have **0** risk alleles of Moyamoya Disease, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

0 out of 2 risk alleles that may lead to the development of Moyamoya Disease were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Saol

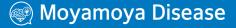
Confirmed by M.D. Gi Chang-seok (547)



[61/107]







## **Description**

Moyamoya disease is a hereditary cerebrovascular disease. A cerebral angiogram shows that the carotid artery and the major artery transporting blood to the brain narrows and consequently is blocked, or abnormal thin blood vessels grow from the skull base adjacent to the narrowed artery and form a vascular network. Common symptoms include cerebral ischemia, infarcts, cerebral hemorrhage, and headache. It may be accompanied by convulsions, fainting, and involuntary movements. Temporary symptoms of cerebral ischemia are common in children. Cerebral hemorrhage is more common in adults than in children. Moyamoya disease is common in the Far East such as Korea and Japan. The disease is more common in females and about 10~15% of cases are familial. Moyamoya disease is associated with the RNF213 gene on chromosome 17.

#### **Risk Factors**

Clear causes of the disease are unknown, but hereditary factors are the major cause of this disease. In particular, mutations in the RNF213 gene are known to be inherited as incomplete autosomal dominance. The autoimmune response due to infection may be a risk factor.





Inheritance

Mutations in RNF213 gene

#### Recommendation

No specific prevention is known yet. The disease is greatly affected by genetic factors. If you have a relevant family history, it is recommended that you get RNF213 genetic testing and magnetic resonance angiography (MRA). If hyperlipidemia or hypertension is accompanied, the risk of cerebrovascular stenosis may increase. It is advised to prevent hyperlipidemia and hypertension.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of RNF213 and other moyamoya disease related genes.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Magnetic resonance angiography (MRA), RNF213 genetic testing, Hyperlipidemia test

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[62/107]







# Cerebral Aneurysm



## Patient Asian-male is subject to General care

Risk of Disease Moderate

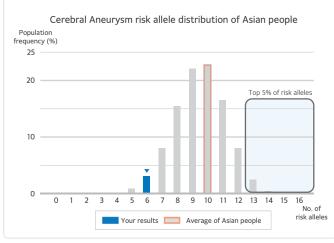
Risk allele an	alysis results
No. of risk alleles tested	No. of risk alleles detected
16	6

As a result of the examination, 6 out of 16 risk alleles that may lead to the development of Cerebral Aneurysm were detected. This corresponds to less than 13 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Cerebral Aneurysm occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 6 risk alleles of Cerebral Aneurysm, while Asian people have 10 risk alleles on average.



## **Genotype Analysis Results**

6 out of 16 risk alleles that may lead to the development of Cerebral Aneurysm were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information	Your genotype	No. of risk alleles detected
ALDH2 112241766	GG				
BET1L 203788	GG				
CDKN2B-AS1	CC				
CDKN2B-AS1 22083404	CC				
CDKN2B-AS1	CC				
SOX17 55437524	AA				
STARD13 33693837	GA				
STARD13 33704065	GA				
High risk geno	type	Risk alleles det	ected	Risk alleles not de	etected

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[63/107]







## **Description**

A cerebral aneurysm is a medical condition in which weakness in a cerebral artery causes a localized dilation or ballooning of the blood vessel. It typically occurs in the bifurcation where large blood vessels are located. Cerebral aneurysms are mostly less than 10 mm in size. Sometimes large aneurysms may occur. Cerebral aneurysms larger than 25mm in size are called giant aneurysms. The walls of cerebral arteries are very thin and more easily ruptured than normal blood vessels due to their structural characteristics. If a cerebral aneurysm ruptures, there is a possibility that cerebral hemorrhage will occur. No specific symptoms arise before a cerebral aneurysm ruptures. It may be accompanied by migraine, tension headaches, dizziness, etc.

### **Risk Factors**

Accurate causes are unknown. A cerebral aneurysm is likely to occur if you have a hereditary cerebrovascular disease such as moyamoya disease. It is recently known that an acquired cerebral aneurysm may occur in a part where high pressure is put on the blood vessels. Other risk factors include smoking, hypertension, arteriosclerosis, and old age.







Family history

Smoking

Hypertension

#### Recommendation

If you have a family history of cerebral aneurysms, get an examination proactively for prevention. Avoid lifestyle factors that may weaken the brain blood vessels, such as smoking, drinking, and stress. Moderate exercise and proper dietary habits help in the prevention of cerebral aneurysms.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Magnetic resonance angiography (MRA), RNF213 genetic testing

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[64/107]







# Alzheimer's Disease



## Patient Asian-male is subject to General care

Risk of Disease Moderate

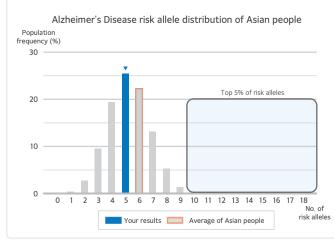
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
18	5	

As a result of the examination, 5 out of 18 risk alleles that may lead to the development of Alzheimer's Disease were detected. This corresponds to less than 10 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Alzheimer's Disease occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 5 risk alleles of Alzheimer's Disease, while Asian people have 6 risk alleles on average.



## **Genotype Analysis Results**

5 out of 18 risk alleles that may lead to the development of Alzheimer's Disease were detected.

Gene (Variant information)		No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
ABCA7 1063443	GA				
BCAM 45324138	GG				
BIN1 127892810	GG				
CD33 51727962	CC				
CNTNAP2 147634891	GG				
CR1 207692049	GG				
GAB2 78091150	TG				
INPP5D 234068476	CC				
NECTIN2 45382034	GA				

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[65/107]





# Alzheimer's Disease

## **Description**

Known as the most common brain disease that causes dementia. Alzheimer's disease is a neurodegenerative disorder characterized by the accumulation of abnormal proteins in the brain, such as amyloid β-protein and tau protein, leading to the gradual loss of brain cells. This is the cause of dementia in approximately 50~60% of all dementia patients. Alzheimer's disease, which characteristically develops and worsens gradually, causes memory loss in the initial phase, and subsequently, loss of cognitive functions, such as language skills and judgment.

### **Risk Factors**

The pathogenesis is not clear, but hereditary factors are the major cause of this disease while old age is another key factor. Hypertension, diabetes, and hyperlipidemia may directly or indirectly influence the occurrence of Alzheimer's disease. The risk of developing Alzheimer's disease increases if you have suffered a severe head injury in the past.







Family history

Head injury

#### Recommendation

It is important to maintain a healthy lifestyle to prevent Alzheimer's disease. Actively seek treatment for hypertension, diabetes, and heart disease, and refrain from smoking and heavy drinking. A diet rich in unsaturated fatty acids, antioxidants, and vitamins E, C, B, and D is recommended. Regular exercise according to the level of your physical ability is also helpful in disease prevention.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Physical examination, Neurological test, Psychological test, ADL(activities of daily living) test, Neuroimaging(MRI, CT, PET, etc.), Neuropsychological test

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[66/107]







# Frontotemporal Dementia



## Patient Asian-male is subject to General care

Risk of Disease Moderate

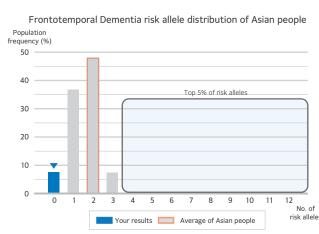
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
12	0	

As a result of the examination, 0 out of 12 risk alleles that may lead to the development of Frontotemporal Dementia were detected. This corresponds to less than 4 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Frontotemporal Dementia occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have **0** risk alleles of Frontotemporal Dementia, while Asian people have 2 risk alleles on average.



## **Genotype Analysis Results**

0 out of 12 risk alleles that may lead to the development of Frontotemporal Dementia were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
* GRN 42426558	CC				
* GRN 42428169	GG				
LOC730100 52600067	TT				
* MAPT 44087755	CC				
* MAPT 44087784	CC				
UNC13A 17752689	TT				
High risk genot	ensive care (P\		ected	Risk alleles not d	etected

Tested by M.T. Lee Myeong-geun (20058) /9K/Lee

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)

[67/107]





# Frontotemporal Dementia

## **Description**

Frontotemporal dementia is a degenerative brain disease caused by atrophy of the frontal and temporal lobes. Approximately 5% of the total dementia cases are frontotemporal dementia. In frontotemporal dementia, memory is relatively less affected than Alzheimer's disease. Symptoms vary depending on which part of the brain is damaged. Common symptoms include significant changes in personality, behavior, and language such as impulse control issues, reticence, and difficulties in understanding speech. These symptoms gradually progress. As dementia advances, more symptoms manifest. Frontotemporal dementia develops at relatively young ages, between the ages of 45 and 60.

### **Risk Factors**

The mechanism and cause of the disease were not clearly identified. However, it is known that intracellular protein dysfunction such as the tau protein causes brain cell damage. A family history of frontotemporal dementia is one of the significant risk factors. The prevalence of frontotemporal dementia increases with age.







Brain cell damage

**Epilepsy** 

#### Recommendation

Engaging in regular physical activities, maintaining a healthy weight, avoiding smoking, and reducing alcohol use are basic to the prevention of dementia. If you have any risk factors such as a family history or a head injury, it is recommended that you get regular examinations such as neurological test and neuroimaging.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of GRN, MAPT and other frontotemporal dementia related genes.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Physical examination, Neurological test, blood test, Neuroimaging(MRI, CT, PET, etc.)

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[68/107]







# Parkinson's Disease



## Patient Asian-male is subject to General care

Risk of Disease Moderate

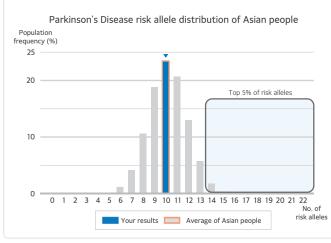
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
22	10	

As a result of the examination, 10 out of 22 risk alleles that may lead to the development of Parkinson's Disease were detected. This corresponds to less than 14 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Parkinson's Disease occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 10 risk alleles of Parkinson's Disease, while Asian people have 10 risk alleles on average.



## **Genotype Analysis Results**

10 out of 22 risk alleles that may lead to the development of Parkinson's Disease were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
FAM47E 77198986	CC		TMEM175 951947	TT	
GAK 852313	GG		TPM1 63346376	TT	
GCH1 55348869	GG				
MAPT 44865603	CT				
PARK16 205713378	CT				
RIT2 40673380	CC				
SLC2A13 40458384	AA				
SNCA 90626111	CC				
SNCA 90641340	GG				
High risk geno	otype	Risk alleles dete	ected Risk	alleles not d	etected

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[69/107]





# Parkinson's Disease

## **Description**

Parkinson's disease is a degenerative neurological disorder, caused by dopamine neuronal degeneration. The most common symptom is motor disturbances such as tremors, stiffness, and posture instability. Cognitive problems may also occur. Symptoms usually emerge slowly. In some patients, symptoms remain the same without worsening. Parkinson's disease is the second most common degenerative brain disease following Alzheimer's disease. The disease develops in 1% of those over 60 years of age. The risk of Parkinson's disease increases with age.

#### **Risk Factors**

Around 10% of Parkinson's disease cases are caused by genetic factors. Genetic factors are common in patients under 50 years old. Generally, the risk of the disease increases with age. Exposure to pesticides, heavy metals, carbon monoxide, etc. and, a history of a head injury may be linked with Parkinson's disease.







Old age

Inheritance

Exposure to toxins

#### Recommendation

Not many things are known for prevention. However, it is important to maintain a healthy weight and eat a balanced diet. Consistent and regular exercise may help in the prevention of Parkinson's disease.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Neurological test, Clinical symptoms, MRI, PET

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[70/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

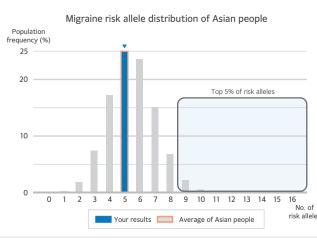
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
16	5	

As a result of the examination, 5 out of 16 risk alleles that may lead to the development of Migraine were detected. This corresponds to less than 9 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Migraine occurrence among Asian people. Therefore, the disease risk assessment result is moderate.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

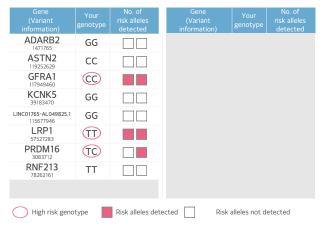
## **Risk Allele Distribution Graph**

You have 5 risk alleles of Migraine, while Asian people have 5 risk alleles on average.



## **Genotype Analysis Results**

5 out of 16 risk alleles that may lead to the development of Migraine were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[71/107]







## **Description**

Migraine is a headache that usually manifests on one side of the head. Specifically, pulsating pain continues on one side of the head for over a certain period, accompanied by nausea, vomiting, and sensitivity to light and sound. Migraines may manifest in all ages. However, prevalence rates vary depending on age and sex. Generally, people first experience migraines in their adolescence or youth. Migraines are more common among females than males.

#### **Risk Factors**

The onset of a migraine is greatly affected by family history. The most common environmental factor is stress. Some foods, especially red wine, affect migraines. In addition, cheese containing tyramine and caffeine in chocolate may trigger migraines. People are sensitive to different foods in terms of migraines. It is also reported that the female sex hormone estrogen has a considerable role in provoking migraines.







Family history

Stress

Foods (red wine, chocolate, cheese)

#### Recommendation

Various non-drug measures may alleviate migraine symptoms. Patients need to change their lifestyles through stress management, sleep management, and exercise. It is most important for the patient to understand and avoid the triggers of his/her migraines.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Clinical inquiry and examination

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[72/107]







## Depressive Disorder



## Patient Asian-male is subject to General care

Risk of Disease Moderate

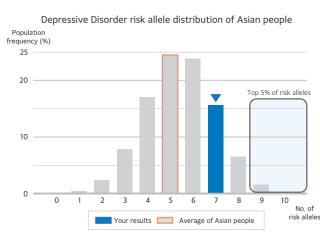
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
10	7	

As a result of the examination, 7 out of 10 risk alleles that may lead to the development of Depressive Disorder were detected. This corresponds to less than 9 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Depressive Disorder occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

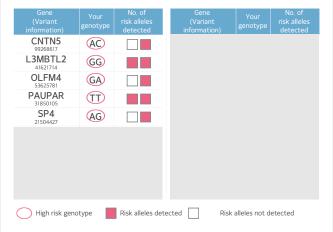
## **Risk Allele Distribution Graph**

You have 7 risk alleles of Depressive Disorder, while Asian people have 5 risk alleles on average.



## **Genotype Analysis Results**

7 out of 10 risk alleles that may lead to the development of Depressive Disorder were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[73/107]





## **a** Depressive Disorder

#### **Description**

Depressive disorder is a medical condition mainly characterized by a lack of motivation and low mood. This disease may degrade cognitive, mental, and physical functions, which may affect your daily life. A state of depression is not temporary but continues for several weeks. Many patients experience sleep disorders, loss of appetite, and weight loss. Around 15% of people experience the depressive disorder in their lives. However proper treatment helps patients with depressive disorder get better.

#### **Risk Factors**

There are a variety of causes of depressive disorder. Environmental factors such as stress and genetic factors affect the development of the depressive disorder. Changes in biorhythms due to abnormalities of hormones and neurotransmitters in the brain such as norepinephrine and serotonin may affect depressive disorder.









Changes in hormones

Inheritance

Old age

#### Recommendation

There is no proven prevention method. However, stress management and social support may help in the prevention of depressive disorder. The most important thing is to get and stick to proper treatment from experts when patients experience initial symptoms before the depressive disorder gets worse. Avoid alcohol use, smoking, illegal drugs, etc. which aggravate the depressive disorder. Research report that physical activities and exercises reduce depression. Therefore it is recommended that you engage in enjoyable exercise such as walking, jogging, and swimming.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Blood test for thyroid disease and vitamin deficiency, Neurological test

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[74/107]





## Type 2 Diabetes Mellitus



## Patient Asian-male is subject to General care

Risk of Disease Moderate

Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
34	10	

As a result of the examination, 10 out of 34 risk alleles that may lead to the development of Type 2 Diabetes Mellitus were detected. This corresponds to less than 19 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Type 2 Diabetes Mellitus occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph** You have 10 risk alleles of Type 2 Diabetes Mellitus, while Asian people have 14 risk alleles on average. Type 2 Diabetes Mellitus risk allele distribution of Asian people Population frequency (%) Top 5% of risk alleles 0 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 23 24 25 26 27 28 29 30 31 32 33 34 Your results Average of Asian people

## **Genotype Analysis Results**

10 out of 34 risk alleles that may lead to the development of Type 2 Diabetes Mellitus were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information)	Your genotype	No. of risk alleles detected
ADCY5 123065778	TT		LTK 42201410	GG	
CDC123-RN7SL198P 12307894	CC		MTNR1B 92708710	CG	
CDKAL1 20661250	GG		PAX4 127253550	GG	
DGKB 14898282	GG		RMST 97562756	GG	
FTO 53800954	TC		SLC30A8	CC	
HNF1B 36098040	TT		TCERG1L 132947962	CC	
IGF2BP2 185529080	TT		TCF7L2 114758349	CC	
KCNQ1 2839751	CC		TLE1 84308948	TT	
KCNQ1 2857194	AA				

Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)

[75/107]





## Type 2 Diabetes Mellitus

#### **Description**

Type 2 diabetes mellitus is characterized by higher blood sugar than healthy people. If glucose in the blood fails to enter the cells and turns into an energy source, blood sugar levels rise. Type 2 diabetes mellitus occurs when there is a lack of insulin secreted from the pancreas or insulin does not function properly, which makes up about 90% of diabetes cases. Common symptoms include excessive drinking, increased hunger, and frequent urination, which are sometimes accompanied by unexplained weight loss, blurred vision, fatigue, and lethargy. Diabetes generally occurs slowly. Many patients do not experience any symptoms, so it is important to get regular check rups.

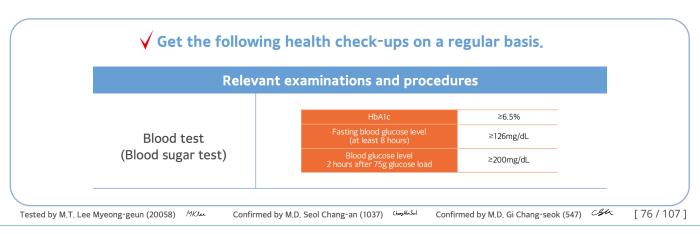
#### **Risk Factors**

Type 2 diabetes mellitus is known to occur due to a combination of genetic and environmental factors. Typically, it begins in mid to old age. The older you become, the higher the risk. Obesity interrupts insulin function and impairs the insulin secretion of the pancreas, which causes diabetes.



#### Recommendation

Smoking appears to increase the risk of diabetes. Some people gain weight after quitting smoking. In this case, the risk of diabetes may increase. Some hypoglycemic agents and antiobestic drugs were found to be effective for reducing the risk of diabetes. However, the most important thing is to correct your lifestyle. A healthy lifestyle including maintaining a healthy weight, exercise, and healthy dietary habits delays the onset of diabetes in the high-risk group.







# Hypercholesterolemia



## Patient Asian-male is subject to General care

Risk of Disease Moderate

Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
5	0	

As a result of the examination, 0 out of 5 risk alleles that may lead to the development of Hypercholesterolemia were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Hypercholesterolemia occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have **0** risk alleles of Hypercholesterolemia, while Asian people have 0 risk alleles on average.

Hypercholesterolemia risk allele distribution of Asian people Population frequency (%) 100 90 80 Top 5% of risk alleles 40 30

## **Genotype Analysis Results**

0 out of 5 risk alleles that may lead to the development of Hypercholesterolemia were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[77/107]





## Hypercholesterolemia

#### **Description**

Hypercholesterolemia is a medical condition in which abnormal lipid metabolism of blood increases total cholesterol and LDL cholesterol levels. Patients with hypercholesterolemia have high cholesterol levels but their triglyceride levels are normal. There are a variety of causes of hypercholesterolemia. It is a common medical condition that can be found in approximately 18% of adults 30 years and older. Hypercholesterolemia is a major cause of cardiovascular diseases such as atherosclerosis and cerebrovascular disease such as stroke. Patients with familial hypercholesterolemia, associated with mutations in the PCSK9 and LDLR genes, may experience xanthomata.

#### **Risk Factors**

General risk factors include a high-fat diet and lack of exercise. Heavy alcohol use and smoking may increase blood lipid levels. Hypercholesterolemia may be caused by underlying medical conditions such as diabetes, hypothyroidism, and chronic liver disease.

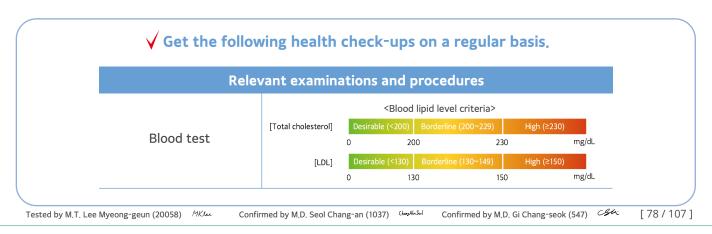


#### Recommendation

It is important to maintain a healthy weight and work out regularly combining both aerobic exercise and strength training exercise. Control fat intake and avoid excess intake of carbohydrates and sugars. It is necessary to have healthy dietary habits such as consuming fiber-rich foods.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of APOE, PCSK9 and other hypercholesterolemia related genes.







# Hypertriglyceridemia



## Patient Asian-male is subject to General care

Risk of Disease Moderate

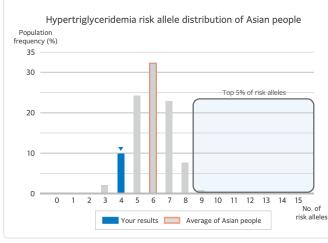
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
15	4	

As a result of the examination, 4 out of 15 risk alleles that may lead to the development of Hypertriglyceridemia were detected. This corresponds to less than 9 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Hypertriglyceridemia occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 4 risk alleles of Hypertriglyceridemia, while Asian people have 6 risk alleles on average.



### **Genotype Analysis Results**

4 out of 15 risk alleles that may lead to the development of Hypertriglyceridemia were detected.

Gene (Variant information)	Your genotype	No. of risk alleles detected	Gene (Variant information	Your genotype	No. of risk alleles detected
APOA5 116652207	GG				
APOA5 116661392	GG				
APOA5 116662407	CC				
APOE 45411941, 45412079	e3/e4				
GCKR 27730940	CC				
GCKR 27741237	CC				
MLXIPL 72982874	GG				
MLXIPL 73020337	GG				
High risk geno	otype	Risk alleles det	ected	Risk alleles not de	etected

Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[79/107]





## Hypertriglyceridemia

#### **Description**

Hypertriglyceridemia is a medical condition in which abnormal lipid metabolism of blood increases triglyceride levels. Patients with hypertriglyceridemia have increased blood triglyceride levels, around 200~500mg/dL, but their LDL cholesterol levels are normal. There are various causes of hypertriglyceridemia. Its accurate mechanism is not known. Hypertriglyceridemia may be caused by some medical conditions such as obesity, diabetes, and hypothyroidism. The risk of pancreatitis increases in people whose triglyceride levels are above 1000mg/dL, which is a major cause of cardiovascular disease such as atherosclerosis and cerebrovascular diseases such as stroke. Hypertriglyceridemia is more common in males than in females. Hypertriglyceridemia is found in approximately 17% of adults 30 years and older.

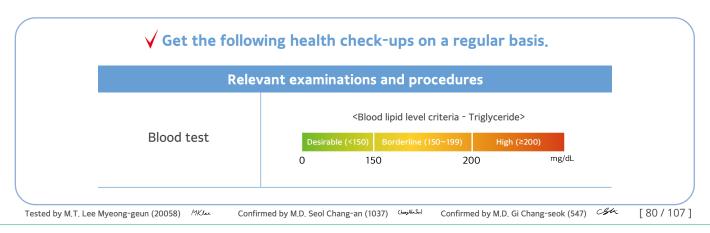
#### **Risk Factors**

General risk factors include a high-fat diet and lack of exercise. Heavy alcohol use and smoking may increase blood lipid levels. Hypertriglyceridemia may be caused by underlying medical conditions such as diabetes, hypothyroidism, and chronic liver disease.



#### Recommendation

It is important to maintain a healthy weight and work out regularly basis combining both aerobic exercise and strength training exercise. Control fat intake and avoid excess intake of carbohydrates and sugars. It is necessary to have healthy dietary habits such as consuming fiber-rich foods.









## Vitamin B12 Concentration



## Patient Asian-male is subject to General care

Risk of Disease Moderate

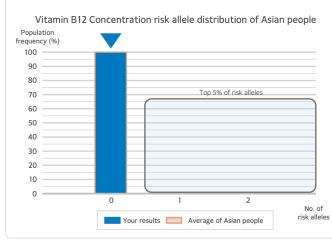
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
2	0	

As a result of the examination, 0 out of 2 risk alleles that may lead to the development of Vitamin B12 Concentration were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Vitamin B12 Concentration occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 0 risk alleles of Vitamin B12 Concentration, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

0 out of 2 risk alleles that may lead to the development of Vitamin B12 Concentration were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[81/107]





## **Solution** Vitamin B12 Concentration

#### **Description**

Vitamin B12, one of the B vitamins, is involved in erythropoiesis, DNA synthesis, and tissue and cell regeneration, and is important in nervous tissues. Vitamin B12 concentration is known to be associated with various diseases such as malignant anemia, cardiovascular disease, cancer, and degenerative neurological disorder. Vitamin B12 deficiency may cause anemia, accompanied by dizziness, weakness, and fatigue. The FUT2 gene is known to affect vitamin B12 levels.

#### **Risk Factors**

Vitamin B12 deficiency may occur when you do not take enough vitamin B12 or the vitamin is not absorbed well in the gut, Medical conditions that interfere with vitamin B12 absorption include intestinal disease and bacterial overgrowth within the stomach and intestine. Decreased gastric acid secretion interferes with vitamin B12 separation, which consequently interrupts absorption. In addition, vitamin B12 deficiency may occur in patients with malignant anemia.







Low intake of vitamin B12

Intestinal disease

Malignant anemia

#### Recommendation

The most important thing is to eat plenty of foods rich in vitamin B12. Vitamin B12 is contained in a variety of foods, especially rich in animal products such as beef liver, fish, shellfish, meat, milk, and dairy products. Decreased vitamin B12 may easily occur in vegetarians. They should check if their dietary supplements contain vitamin B12.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Blood test (Blood vitamin B12 level test)

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[82/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

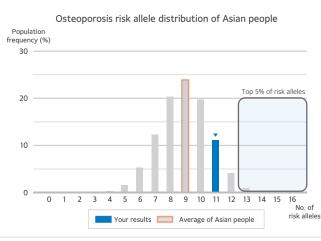
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
16	11	

As a result of the examination, 11 out of 16 risk alleles that may lead to the development of Osteoporosis were detected. This corresponds to less than 13 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Osteoporosis occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

### **Risk Allele Distribution Graph**

You have 11 risk alleles of Osteoporosis, while Asian people have 9 risk alleles on average.



## **Genotype Analysis Results**

11 out of 16 risk alleles that may lead to the development of Osteoporosis were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[83/107]







#### **Description**

Osteoporosis is a skeletal disorder characterized by low bone density and strength leading to increased fracture risk. Bone density increases in your 20s and 30s, and then gradually decreases afterward. In women, bone density decreases fast after menopause. Symptoms barely appear in the initial phase. When the vertebrae weaken, the spine may curve or other symptoms such as pain may manifest. The most common symptom is a fracture of a weakened bone.

#### **Risk Factors**

Generally, osteoporosis is known to be associated with aging, but other risk factors may affect the disease as well. Calcium absorption disorder and vitamin D deficiency affect bone formation and degradation, which may cause osteoporosis. Heavy drinking and smoking also interfere with bone formation. In women, menopause may affect the development of osteoporosis since estrogen maintains bone density levels.









Aging

Menopause

Changes in hormones

Drugs

#### Recommendation

It is easier to prevent a decrease in bone density than to recover from it. Prevent osteoporosis before it develops. Avoid smoking, heavy alcohol use, and excess caffeine intake. It is recommended to take in calcium and vitamin D, which are important nutrients for bone health. Vitamin D is generated through exposure to UV light. Get plenty of sunlight to prevent osteoporosis.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Bone density test, Vitamin D test, Blood test

Those who need a bone density test: Women 65 years of age or older / Men 70 years of age or older Those who have a disease or are taking any medication that may cause osteoporosis

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[84/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

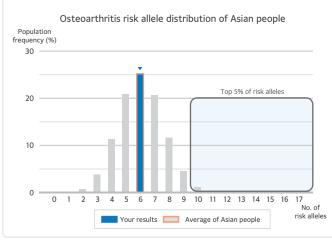
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
17	6	

As a result of the examination, 6 out of 17 risk alleles that may lead to the development of Osteoarthritis were detected. This corresponds to less than 10 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Osteoarthritis occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

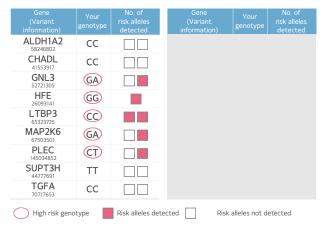
### **Risk Allele Distribution Graph**

You have 6 risk alleles of Osteoarthritis, while Asian people have 6 risk alleles on average.



## **Genotype Analysis Results**

6 out of 17 risk alleles that may lead to the development of Osteoarthritis were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[85/107]





## Osteoarthritis

#### **Description**

Osteoarthritis is a medical condition in which the cartilage surrounding the joint wears away, the bone becomes exposed, and consequently inflammation and pain develop due to damage to the cartilage and the surrounding tissues. The risk of osteoarthritis increases with age, so it is also called degenerative arthritis. The most common symptom is a pain in the lesion. The degree of pain varies from person to person. In the initial phase, pain appears or gets better depending on the physical activity or weather conditions. As osteoarthritis progresses over time, patients experience consistent pain. Other symptoms include stiffness, swelling, and bone outgrowths.

#### Risk Factors

Osteoarthritis occurs due to cartilage damage resulting from various causes. Obesity, vigorous physical activities, and joint trauma may put a strain on the joints and consequently may cause osteoarthritis. Aging and genetic factors are also known to be contributors to osteoarthritis. Osteoarthritis is more common in females than in males.









Aging

Overweight

Bad posture

Vigorous physical activities

#### Recommendation

Maintain a healthy weight not to put stress on the joints. Get moderate exercise regularly to strengthen your muscles. Avoid bad posture such as kneeling or sitting in a squatting position. Antioxidants such as vitamin C, vitamin E, and beta carotene may prevent cartilage damage. Eat plenty of vegetables and fruits rich in antioxidants.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

X-ray, MRI

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[86/107]







# Rheumatoid Arthritis



## Patient Asian-male is subject to General care

Risk of Disease Moderate

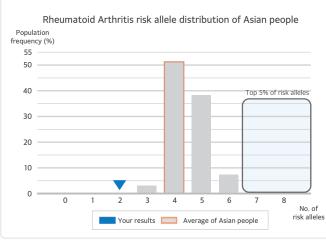
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
8	2	

As a result of the examination, 2 out of 8 risk alleles that may lead to the development of Rheumatoid Arthritis were detected. This corresponds to less than 7 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Rheumatoid Arthritis occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 2 risk alleles of Rheumatoid Arthritis, while Asian people have 4 risk alleles on average.



## **Genotype Analysis Results**

2 out of 8 risk alleles that may lead to the development of Rheumatoid Arthritis were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[87/107]





## Rheumatoid Arthritis

#### **Description**

Rheumatoid arthritis is a chronic inflammatory disease in which consistent inflammation occurs in the synovium of the joint. Unlike osteoarthritis in which inflammation manifests due to wear of cartilage, rheumatoid arthritis is known to be caused by abnormalities of the immune system. Rheumatoid arthritis may develop in all ages, but most commonly occurs between 35 and 50 years of age. It is the second most common osteoarthritis following osteoarthritis. In the initial phase, stiffness appears in the hands. The symptoms persist for over one hour after waking up. Other symptoms include hand/foot joint swelling, pain, and a burning sensation.

#### **Risk Factors**

Risk factors are not clearly identified, but it is known that rheumatoid arthritis develops when those with genetic factors are exposed to environmental factors such as smoking. It also occurs as an immune response to infectious agents such as bacteria and viruses.



#### Recommendation

Early diagnosis and treatment prevent joint deterioration. Maintain a healthy weight and work out for at least 30 minutes every day in a way not to put a strain on the joints. Avoid maintaining the same posture for a long time or lifting heavy objects which may put stress on the joints.



#### Relevant examinations and procedures

Blood test (rheumatoid factor), X-ray, Clinical inquiry and examination

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[88/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

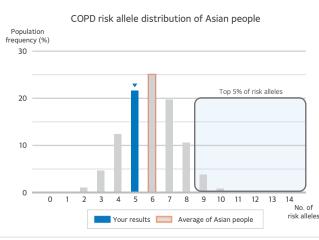
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
14	5	

As a result of the examination, 5 out of 14 risk alleles that may lead to the development of Chronic Obstructive Pulmonary Disease were detected. This corresponds to less than 9 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Chronic Obstructive Pulmonary Disease occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

### **Risk Allele Distribution Graph**

You have 5 risk alleles of COPD, while Asian people have 6 risk alleles on average.



## **Genotype Analysis Results**

5 out of 14 risk alleles that may lead to the development of COPD were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[89/107]







#### **Description**

Chronic obstructive pulmonary disease is a medical condition characterized by a decline in lung function due to inflammation in the bronchus or lung resulting from exposure to harmful particles or gases. Chronic obstructive pulmonary disease includes chronic bronchitis and emphysema. Chronic inflammation changes the structure of the small airways or damages alveoli, which consequently causes shortness of breath. Symptoms may not emerge at all in the early stages of the disease. Patients with advanced chronic obstructive pulmonary disease may experience chronic coughing, sputum, shortness of breath, and decreased vital capacity. Drug treatment is not effective for chronic obstructive pulmonary disease. Prevention is more important than anything else.

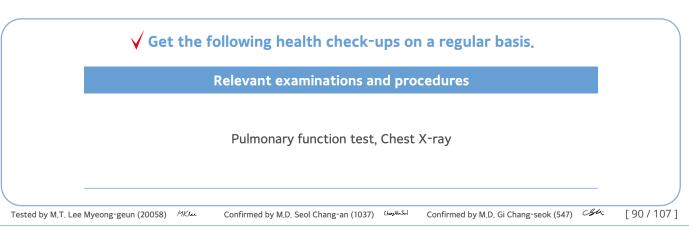
#### **Risk Factors**

Long-term exposure to toxic gases or particles causes chronic obstructive pulmonary disease. The most important cause of chronic obstructive pulmonary disease is smoking. Of those who smoke, about 15~20% are assumed to have chronic obstructive pulmonary disease. A working environment exposed to dust, and indoor and outdoor air pollution may cause chronic obstructive pulmonary disease as well.



#### Recommendation

The most important way to prevent chronic obstructive pulmonary disease is to avoid risk factors. Patients with chronic obstructive pulmonary disease must quit smoking. By doing so, you can prevent your lung functions from declining. It is also recommended to avoid going outside when the air quality is bad, and to manage the indoor air quality through ventilation and using an air purifier.











## Patient Asian-male is subject to General care

Risk of Disease **Moderate** 

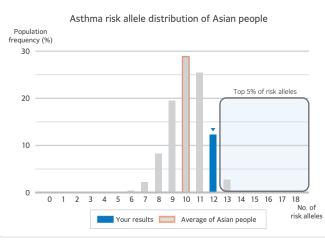
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
18	12	

As a result of the examination, 12 out of 18 risk alleles that may lead to the development of Asthma were detected. This corresponds to less than 13 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Asthma occurrence among Asian people. Therefore, the disease risk assessment result is moderate.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## Risk Allele Distribution Graph

You have 12 risk alleles of Asthma, while Asian people have 10 risk alleles on average.



## **Genotype Analysis Results**

12 out of 18 risk alleles that may lead to the development of Asthma were detected.

Gene (Variant information)		No. of risk alleles detected	Gene (Variant information	Your genotype	No. of risk alleles detected
D2HGDH 242698640	GG				
FLG 152285861	CC				
GSDMA 38121993	GA				
LINC00536					
RAD50 131901225	AG				
RORA 61069988	GG				
SEMA3E 83157804	T				
SERPINB7 61442619	AA				
SLC7A10-CEBPA 33726578	GG				

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[91/107]







#### **Description**

Asthma is a chronic disease in which the bronchi narrow due to allergic inflammation. The inflammation of the bronchus makes the bronchial mucosa swell and narrow. Common symptoms include shortness of breath, coughing, and wheezing and may be accompanied by chest tightness and sputum. Symptoms, severity, and recovery periods vary from person to person and are repeated. This chronic disease generally begins in childhood, but sometimes develops in adulthood or old age. It is known that approximately 5% of Korean people have experienced asthma. The prevalence of asthma is increasing every year.

#### Risk Factors

Asthma is caused by a combination of genetic and environmental factors. Asthma may develop when those with an allergy are exposed to allergens such as pollen, house dust mites, fungus, and animal hair. Tobacco smoke, air pollution, and stress may aggravate symptoms.



#### Recommendation

The most important thing is to avoid the causes of asthma. Identify and avoid allergens. Properly ventilate and clean the surroundings to reduce exposure to the common causes of asthma such as house dust mites and dust. Stopping smoking and stress management may help in the prevention of asthma. It is important to take a pulmonary function test and get treatment when symptoms emerge.



Pulmonary function test, Bronchial challenge test, Skin prick test

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[92/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

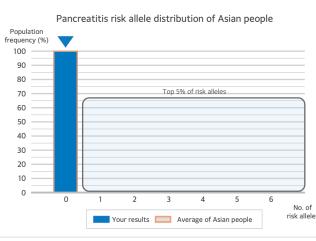
Risk allele analysis results			
No. of risk alleles tested	No. of risk alleles detected		
6	0		

As a result of the examination, 0 out of 6 risk alleles that may lead to the development of Pancreatitis were detected. This corresponds to less than 1 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Pancreatitis occurrence among Asian people. Therefore, the disease risk assessment result is moderate.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

## **Risk Allele Distribution Graph**

You have 0 risk alleles of Pancreatitis, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

0 out of 6 risk alleles that may lead to the development of Pancreatitis were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[93/107]





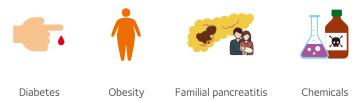


#### Description

Pancreatitis is a medical condition in which the pancreas gland is damaged by pancreatic enzymes (mainly trypsin) or part or the whole of the pancreas becomes inflamed. Pancreatitis is divided into acute pancreatitis and chronic pancreatitis. In acute pancreatitis in which acute inflammation occurs in the pancreas, the function of the pancreas returns to normal after recovery. In chronic pancreatitis, inflammation is repeated in the pancreas and an irreversible disorder occurs in exocrine and endocrine pancreatic functions. One of the most common symptoms of pancreatitis is pain. Patients with pancreatitis experience pain generally in the epigastrium or the left side of the body. Pain is sometimes reflected in the back, chest, or flank. Severe pancreatitis may cause vomiting.

#### **Risk Factors**

Common causes of pancreatitis include gallstones and alcohol. The end of the bile duct (ampulla of Vater) is blocked with the pancreatic duct due to a gallstone in the gallbladder, common bile duct, or liver, which interferes with the flow of bile and pancreatic juice. When bile and pancreatic juice flow backward, inflammation occurs in the pancreas. The mechanism of alcohol causing pancreatitis is not clearly identified yet.



#### Recommendation

Both acute pancreatitis and chronic pancreatitis are caused by alcohol. The best way to prevent pancreatitis is to avoid alcohol use. Even after you recover from acute pancreatitis completely, pancreatitis is likely to recur if you consume alcohol. Patients with chronic pancreatitis must avoid alcohol use for treatment and prevention of recurrence. There is a chance that pancreatitis will recur in patients along with pain even though they do not consume alcohol. However, alcohol greatly increases the risk of recurrence. Gallstone pancreatitis is more likely to occur when you have a small gallstone rather than a large gallstone. If you have any symptoms of gallstones, you must receive treatment.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of PRSS1, SPINK1 and other pancreatitis related genes.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Blood test (amylase and lipase concentration), Abdominal CT, Abdominal MRI

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[94/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030

# Corneal Dystrophy



## Patient Asian-male is subject to Intensive care

Risk of Disease High

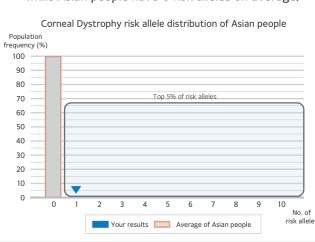
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
10	1	

As a result of the examination, 1 out of 10 risk alleles that may lead to the development of Corneal Dystrophy disease was(were) detected. It contains particularly high risk alleles relevant to the diseases. Therefore, he disease risk assessment result is high. Consultation with a medical professional is recommended

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

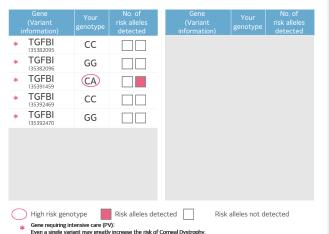
## **Risk Allele Distribution Graph**

You have 1 risk alleles of Corneal Dystrophy, while Asian people have 0 risk alleles on average.



## **Genotype Analysis Results**

1 out of 10 risk alleles that may lead to the development of Corneal Dystrophy were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[95/107]





## Corneal Dystrophy

#### **Description**

Corneal dystrophy is a medical condition characterized by non-inflammatory clouding in the transparent central front part of the eye called the cornea. This hereditary disorder develops in both eyes at birth and advances slowly. Corneal dystrophy is divided into several types depending on the histological part of the lesion. Different types of corneal dystrophy have different names depending on their shapes and properties. The most common type is Avellino corneal dystrophy. In Avellino corneal dystrophy, whitish granular lesions occur in childhood, followed by blurred vision. Avellino corneal dystrophy is relatively common, with a reported prevalence of 1 in 870 people in Korea, If patients with Avellino corneal dystrophy undergo laser-assisted keratectomy (LASIK, LASEK, etc.), blurred vision may be accelerated.

#### **Risk Factors**

Corneal dystrophy is generally caused by genetic factors. Nongenetic factors are not known. The disease is generally caused by mutations in the TGFBI gene and may worsen due to laser-assisted keratectomies such as LASIK and LASEK.





Inheritance

LASIK and LASEK

#### Recommendation

Corneal dystrophy is a hereditary disease. If one of the parents has been diagnosed with corneal dystrophy, the child must check and manage his/her medical condition. If a patient with corneal dystrophy undergoes keratectomy such as LASIK and LASEK, the disease may swiftly progress and he/she may lose his/her sight in several years. You must undergo an examination if you consider such surgery. If you have corneal dystrophy, it is recommended that you avoid stimuli such as UV light to slow down the progress of the disease.

#### **Cautions**

Even though any risk alleles are not found in this examination, risk alleles may be detected in other parts of TGFBI and other corneal dystrophy related genes.

✓ Get the following health check-ups on a regular basis.

Relevant examinations and procedures

Slit-lamp biomicroscopy

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[96/107]









## Patient Asian-male is subject to General care

Risk of Disease Moderate

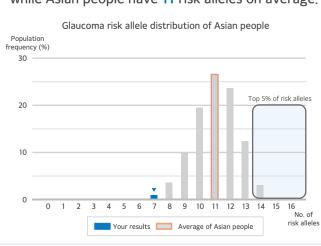
Risk allele analysis results			
No. of risk alleles tested	No. of risk alleles detected		
16	7		

As a result of the examination, 7 out of 16 risk alleles that may lead to the development of Glaucoma were detected. This corresponds to less than 14 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Glaucoma occurrence among Asian people. Therefore, the disease risk assessment result is moderate.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

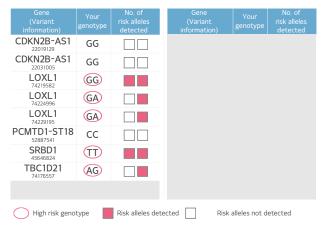
### **Risk Allele Distribution Graph**

You have 7 risk alleles of Glaucoma, while Asian people have 11 risk alleles on average.



## **Genotype Analysis Results**

7 out of 16 risk alleles that may lead to the development of Glaucoma were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[97/107]





## Glaucoma

#### Description

Glaucoma causes visual field defects resulting from damage to the optic nerve that carries light from the eye to the brain. There are no symptoms early in the disease process. In later stages, patients experience visual field defects, narrowing of the field of view, and blindness. The aqueous humor, secreted from the trabecular meshwork and circulating through the entire eye, provides nutrition for the cornea and the lens. Open-angle glaucoma is caused by high eye pressure resulting from the aqueous humor blocked due to a problem in the pathway after the trabecular meshwork.

#### **Risk Factors**

The most important risk factor is ocular hypertension. Other risk factors include asymmetry of the cup-to-disc ratio, being 40 or older, a family history of glaucoma, diabetes, thyroid insufficiency, myopia, eye injuries, surgery, chronic inflammation, use of steroid drops, and long-term use of steroid medication. If you have any risk factors, it is recommended that you get regular check-ups for early diagnosis.





Ocular hypertension Age of 40 or older

#### Recommendation

High blood pressure during periods of excitement and high abdominal pressure resulting from such activities as a headstand or sit-ups may raise eye pressure. Be careful in such situations. It is recommended that you avoid smoking and alcohol use. An attack may develop in the cold winter and the hot summer. Pay attention to temperature changes. This medical condition is associated with a family history. If any member of your family has glaucoma, get regular check-ups for early diagnosis and treatment.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Eye pressure test, Anterior segment eye examination, Optic nerve examination, Visual field test

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[98/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030

## Sjogren's Syndrome



## Patient Asian-male is subject to General care

Risk of Disease Moderate

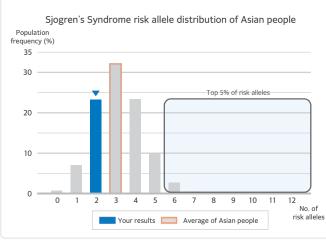
Risk allele analysis results			
No. of risk alleles tested	No. of risk alleles detected		
12	2		

As a result of the examination, 2 out of 12 risk alleles that may lead to the development of Sjogren's Syndrome were detected. This corresponds to less than 6 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Sjogren's Syndrome occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

### **Risk Allele Distribution Graph**

You have 2 risk alleles of Sjogren's Syndrome, while Asian people have 3 risk alleles on average.



### **Genotype Analysis Results**

2 out of 12 risk alleles that may lead to the development of Sjogren's Syndrome were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Sool

Confirmed by M.D. Gi Chang-seok (547)



[99/107]





## Sjogren's Syndrome

#### **Description**

Sjogren's syndrome is a systemic autoimmune disease. A problem arises in the immune system protecting the body from external pathogens, and the immune system attacks the body. Lymphocytes infiltrate into the exocrine gland secreting liquid such as salivary glands and lacrimal glands and cause chronic inflammation and secretion disorder. Sjogren's syndrome is characterized by a dry mouth and dry eyes. Other symptoms include nasal dryness, dry throat, dry skin, and dental caries. Females are affected about 10 times as often as are males. Sjogren's syndrome is especially common in middle-aged women in their 30s to 50s.

#### Risk Factors

Sjogren's syndrome is an autoimmune disease in which a problem arises in the immune system that protects the body from external threats. It is assumed to be associated with genetic factors, viral infection, hormones, etc.





Inheritance

Immune system disorder

#### Recommendation

If you find your oral cavity and eyes feel dry, get an examination for early diagnosis. If you have dry eyes, maintain appropriate indoor humidity levels using a humidifier, eye drops, and artificial tears. You may experience symptoms similar to Sjogren's syndrome when you take any medications for hypertension, depressive disorder, Parkinson's disease, etc. If you have such a medical condition, consult your physician.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Schirmer's test (dry eye test), Salivary gland biopsy, Salivary gland test, Serum autoantibody test, Sialography

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[100/107]









## Patient Asian-male is subject to General care

Risk of Disease **Moderate** 

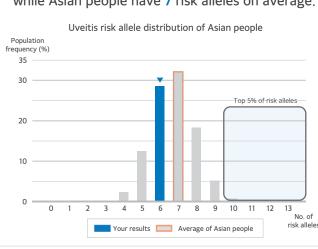
Risk allele analysis results			
No. of risk alleles tested	No. of risk alleles detected		
13	6		

As a result of the examination, 6 out of 13 risk alleles that may lead to the development of Uveitis were detected. This corresponds to less than 10 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Uveitis occurrence among Asian people. Therefore, the disease risk assessment result is moderate.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

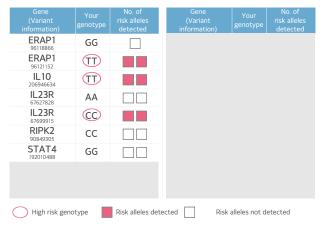
### **Risk Allele Distribution Graph**

You have 6 risk alleles of Uveitis, while Asian people have 7 risk alleles on average.



## **Genotype Analysis Results**

6 out of 13 risk alleles that may lead to the development of Uveitis were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[101/107]







#### **Description**

Uveitis is the inflammation of the uvea forming the middle layer of the eye. The uvea is vulnerable to inflammation since it has many connective tissues and blood vessels providing nutrition for the eyes. Inflammation may be caused by infection, damage, systemic autoimmune diseases, or other unknown reasons. Acute uveitis is accompanied by severe pain, photophobia, and blurred vision. Chronic inflammation may be accompanied by pain in the eye, blurred vision, and redness of the eye. Complications include cataracts, vitreous clouding, abnormalities of the retina, and glaucoma.

#### **Risk Factors**

Causes of uveitis can be divided into infectious causes and noninfectious causes. Infectious causes include infection with mycobacterium tuberculosis, treponema pallidum, etc. Noninfectious causes are not well known but may be associated with autoimmune diseases such as rheumatoid arthritis and lupus erythematosus. Uveitis may develop when you have an eye injury.



#### Recommendation

Since uveitis is associated with the immune system, it may recur due to overwork, common cold, drinking, smoking, etc. If you experience any recurrence symptoms, it is recommended that you get treatment immediately to minimize eye damage. If you have had uveitis, it is advised to get an ophthalmic exam once or twice a year regardless of the recurrence of the disease.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Slit-lamp biomicroscopy, Funduscopy, Fluorescein angiography, Optical coherence tomography, Indocyanine green angiography

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[102/107]





Sample ID Examinee Asian-male 14443 Reception No. 20230221-171-0030



## Macular Degeneration



## Patient Asian-male is subject to General care

Risk of Disease Moderate

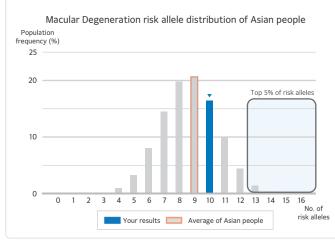
Risk allele analysis results		
No. of risk alleles tested	No. of risk alleles detected	
16	10	

As a result of the examination, 10 out of 16 risk alleles that may lead to the development of Macular Degeneration were detected. This corresponds to less than 13 risk alleles, which is equivalent to the bottom 95% in risk allele distribution for Macular Degeneration occurrence among Asian people. Therefore, the disease risk assessment result is **moderate**.

The more risk alleles there are, the more environmental factors need to be managed for disease prevention.

### **Risk Allele Distribution Graph**

You have 10 risk alleles of Macular Degeneration, while Asian people have 9 risk alleles on average.



## **Genotype Analysis Results**

10 out of 16 risk alleles that may lead to the development of Macular Degeneration were detected.



Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)

[103/107]





## **Macular Degeneration**

#### **Description**

The retina is nervous tissue that serves a function analogous to that of the film in a camera. You can see objects clearly through the macula, the center of the retina. Macular degeneration develops as the retinal cell function is degraded with aging. Major symptoms include dark vision and distorted vision. Other symptoms include visual distortions such as straight lines seeming bent, the center of an object seeming dark, and a decreased ability to distinguish light and shade. Macular degeneration is a major cause of blurred vision of elderly persons. The disease generally develops between the age in 50 and 60.

#### Risk Factors

Causes of macular degeneration are not clearly identified yet. The risk of macular degeneration increases with age and sharply rises after the age of 75. In addition to genetic factors, cardiovascular disease, smoking, and hypercholesterolemia are known as risk factors. Excess exposure to UV light may also be a cause of macular degeneration.



#### Recommendation

Early symptoms are similar to presbyopia. You need to get a retinal exam once a year after the age of 40 at the onset of presbyopia. It is recommended that you take commercially-available vitamins, vegetables, and fruits containing macular pigment reducing damage caused by aging. Eat a healthy diet, work out regularly, and manage your lifestyle factors to reduce risk factors such as obesity, smoking, hypertension, and UV light.

✓ Get the following health check-ups on a regular basis.

#### Relevant examinations and procedures

Slit-lamp biomicroscopy, Eye pressure test, Fundus examination, Fluorescein angiography, Indocyanine green angiography

Tested by M.T. Lee Myeong-geun (20058) / MKLLL

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Su-l

Confirmed by M.D. Gi Chang-seok (547)



[104/107]





# 03. Appendix

- Testing Method & Limitations - References

Tested by M.T. Lee Myeong-geun (20058)

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



[105/107]





## **Testing Method & Limitations**

## **Testing Method**

#### Sequencing

\* This examination uses Next Generation Sequencing among the sequencing techniques.

#### Limitations

We examine your genetic factors that may increase the risk of the diseases and provide customized health care information after analyzing research papers. Be aware that this examination is not relevant to disease diagnosis. To diagnose and treat a disease, you must consult your physician.

## Reference Transcriptome of Genes Requiring Intensive Care

Reference Transcriptome of Genes Requiring Intensive Care				
Gene	Reference transcriptome	Gene	Reference transcriptome	
BRCA1	NM_007294.4	PCSK9	NM_174936.3	
BRCA2	NM_000059.3	PKP2	NM_004572.3	
CDH1	NM_004360.5	PRSS1	NM_002769.4	
GRN	NM_002087.4	PTPN11	NM_002834.5	
KCNH2	NM_000238.3	RNF213	NM_001256071.3	
LMNA	NM_170707.4	SPINK1	NM_003122.3	
MAPT	NM_001377265.1	TGFBI	NM_000358.2	
NOTCH3	NM_000435.3	TNNI3	NM_000363.4	

\* This test was developed and its performance characteristics were determined by GC Genome. It has not been cleared or approved by the Korean Ministry of Food and Drug Safety (MFDS).

Tested by M.T. Lee Myeong-geun (20058) / MKJu.

Confirmed by M.D. Seol Chang-an (1037) Chang-Ala-Seol

Confirmed by M.D. Gi Chang-seok (547)





## References

- Cancer, 2017 Oct 15:123(20):3966-3976.
- Nat Genet. 2010 Sep;42(9):755-8.
- PLoS One. 2018 Jan 31;13(1):e0185490.
- Genet Mol Res. 2014 Oct 7:13(4):8066-79.
- BMC Cancer. 2015 Apr 25;15:314.
- Nat Commun. 2020 Sep 4;11(1):4423.
- Nat Commun. 2017 Feb 14;8:14517.
- Nat Commun. 2017 Jul 13;8:15966.
- Nat Genet. 2009 Apr;41(4):460-4.
- 10 Tumour Biol. 2014 Aug:35(8):7707-12.
- 11 Fur Rev Med Pharmacol Sci. 2014:18(15):2097-101.
- 12 Lancet Oncol. 2017 Apr:18(4):535-544.
- 13 J Hum Genet. 2012 Apr;57(4):235-7.
- 14 Gut. 2018 Aug;67(8):1517-1524.
- 15 PLoS One. 2018 Apr 26;13(4):e0196245.
- 16 Nat Genet, 2019 Jan:51(1):76-87.
- 17 PLoS Genet. 2014 Apr 17;10(4):e1004228.
- 18 Int J Cancer. 2019 Nov 1;145(9):2315-2329.
- 19 Nat Genet. 2016 Dec;48(12):1544-1550.
- 20 Tumour Biol. 2015 Feb;36(2):1023-7.
- 21 Sci Rep. 2017 Aug 29;7(1):9701.
- 22 Hum Mol Genet. 2014 Oct 15;23(20):5545-57.
- 23 Hum Mol Genet, 2015 Feb 15:24(4):1177-84.
- 24 Cancer Res. 2016 Jun 1:76(11):3277-84.
- 25 Front Oncol. 2020 Feb 11;10:134.
- 26 Hum Mol Genet, 2014 Mar 1:23(5):1387-98.
- 27 Nat Genet. 2011 Jun 5;43(7):679-84.
- 28 World J Gastroenterol. 2010 Sep 7;16(33):4210-20.
- 29 PLoS One. 2010 Oct 27;5(10):e13679
- PLoS One. 2015 Feb 13;10(2):e0117173.
- Medicine (Baltimore). 2017 Apr;96(16):e6111.
- 32 Eur J Hum Genet. 2019 Oct;27(10):1589-1598.
- 33 Nat Commun. 2017 Jun 9;8:15724.
- 34 PLoS One. 2015 Mar 31:10(3):e0122589.
- 35 Gut. 2020 Apr:69(4):641-651.
- 36 Nat Genet. 2010 Sep;42(9):764-7.
- 37 World J Gastroenterol, 2014 Aug 28:20(32):11429-38.
- 38 Tumour Biol. 2014 Mar:35(3):2123-32.
- 39 Nat Commun. 2018 Sep 11;9(1):3688.
- 40 PLoS One. 2010 Jul 29;5(7):e11824.
- 41 Nat Genet. 2014 Jul;46(7):736-41.
- 42 Carcinogenesis. 2018 Sep 21;39(9):1135-1140.
- 43 Transl Lung Cancer Res. 2015 Feb;4(1):104-8.
- 44 PLoS One. 2012;7(6):e37970.
- 45 Nat Genet. 2017 Jul;49(7):1133-1140.
- 46 Nat Genet. 2009 Jul;41(7):811-5.
- 47 Nat Genet. 2017 Jul;49(7):1141-1147.
- 48 Hum Mol Genet. 2011 Aug 1;20(15):3109-17.
- 49 Cancer Discov. 2015 Aug;5(8):878-91.

- 50 Hum Genet. 2016 Aug;135(8):923-38.
- 51 Nat Genet. 2021 Jan;53(1):65-75.
- 52 Am J Epidemiol. 2008 Jan 1; 167(1):7-14
- 53 Asian Pac J Cancer Prev. 2014: 15(8):3705-13
- 54 Nat Genet. 2016 Oct;48(10):1151-1161.
- 55 Int J Mol Med. 2018 Nov;42(5):2415-2436
- 56 Nat Genet. 2016 Oct;48(10):1162-70.
- 57 Biomed Rep. 2018 Nov;9(5):383-404.
- 58 Nat Genet. 2017 Sep;49(9):1392-1397.
- 59 Nat Commun. 2020 Jan 9:11(1):163.
- 60 Biomed Rep. 2018 Jul;9(1):8-20.
- 61 Sci Rep. 2017 Dec 20:7(1):17921
- 62 J Cardiol. 2017 Jul;70(1):74-79.
- 63 Nat Genet. 1997 Aug;16(4):379-82.
- 64 Nat Genet. 2010 Mar;42(3):240-4.
- 65 Nat Genet, 2018 Jun 11:50(9):1225-1233.
- 66 Eur Heart J. 2017 Sep 7;38(34):2586-2594.
- 67 Nat Genet. 2017 Jun;49(6):953-958.
- 68 Circ Cardiovasc Genet. 2010 Jun;3(3):248-55.
- 69 Alzheimers Dement. 2019 Oct;15(10):1333-1347
- 70 World J Biol Psychiatry. 2013 Apr;14(3):220-6.
- 71 Exp Ther Med. 2013 Mar:5(3):853-859.
- 72 J Stroke Cerebrovasc Dis. 2017 Nov:26(11):2482-2493.
- 73 Lancet. 2016 Aug 20;388(10046):761-75.
- 74 Development. 2014 Jan;141(2):307-17.
- 75 Eur Neurol. 2014;71(5-6):217-22.
- 76 J Stroke Cerebrovasc Dis. 2013 Jul;22(5):608-14.
- 77 J Stroke Cerebrovasc Dis. 2018 Aug;27(8):2259-2270.
- 78 Stroke, 2014 Nov:45(11):3200-7.
- 79 J Clin Med. 2019 Feb 25;8(2):275
- 80 Neuron. 2007 Jun 7;54(5):713-20.
- 81 Nat Genet. 2019 Mar;51(3):404-413.
- 82 Alzheimers Res Ther. 2019 Jan 12;11(1):5.
- 83 Arch Neurol, 2011 Dec:68(12):1569-79.
- 84 Neurobiol Aging. 2015 Oct;36(10):2904.e13-26.
- 85 Lancet Neurol. 2018 Jun;17(6):548-558.
- 86 Hum Mol Genet. 2016 Sep 1;25(17):3849-3862.
- 87 Hum Mol Genet. 2017 Jan 1;26(1):226-232.
- 88 Hum Genet. 2009 Jan;124(6):593-605.
- 89 BMC Genomics, 2014 Feb 10:15:118,
- 90 Nat Genet, 2009 Dec:41(12):1308-12.
- 91 Nat Genet. 2013 Aug; 45(8): 912-917.
- 92 Cephalalgia. 2018 Mar;38(3):466-475. 93 Mol Psychiatry. 2011 Feb;16(2):193-201.
- 94 Am J Psychiatry. 2013 Feb;170(2):207-17.
- 95 PLoS Genet. 2011 Apr;7(4):e1001363.
- 96 Nat Genet. 2018 Nov;50(11):1505-1513.
- 97 Nat Commun. 2018 Jul 27;9(1):2941.
- 98 PeerJ. 2017 Aug 17;5:e3618

- 99 Am J Hum Genet. 2007 Aug;81(2):338-45.
- 100 Atherosclerosis. 2014 Sep;236(1):54-61.
- 101 Nat Genet. 2008 Feb;40(2):161-9.
- 102 Nature. 2010 Aug 5;466(7307):707-13.
- 103 Nat Genet. 2008 Feb;40(2):149-51.
- 104 Am J Hum Genet. 1996 Aug;59(2):343-50.
- 105 PLoS Genet. 2010 Jan 8;6(1):e1000806.
- 106 PLoS Genet. 2010 Jun 10;6(6):e1000977
- 107 Nat Genet. 2018 Dec;50(12):1681-1687.
- 108 Nat Genet. 2014 May;46(5):498-502. 109 Nat Genet, 2019 Feb:51(2):230-236.
- 110 J Rheumatol, 2017 Nov:44(11):1652-1658
- 111 Mol Med. 2014 Aug 14;20(1):341-9. 112 Arthritis Rheumatol. 2014 May;66(5):1121-32.
- 113 Nat Genet. 2010 Jun;42(6):508-14.
- 114 Ann Rheum Dis. 2015 Mar:74(3):e13.
- 115 Respir Res. 2014 Sep 21;15(1):113.
- 116 Lancet Respir Med. 2014 Mar; 2(3):214-25.
- 117 PLoS Genet. 2009 Mar;5(3):e1000421. 118 J Allergy Clin Immunol. 2010 Feb;125(2):328-335.e11.
- 119 Nat Genet. 2018 Jan;50(1):42-53. 120 Nat Commun. 2019 Feb 20:10(1):880.
- 121 Immunogenetics, 2016 Jul;68(6-7);391-400.
- 122 Nat Genet, 2014 Jan:46(1):51-5.
- 123 Lancet Respir Med. 2019 Jun;7(6):509-522.
- 124 Nat Genet. 2000 Jun;25(2):213-6.
- 125 Hum Mutat. 2006 Aug;27(8):721-30.
- 126 Am J Ophthalmol. 2004 Apr;137(4):765-7
- 127 Nat Genet, 1997 Mar:15(3):247-51.
- 128 Eye (Lond). 2019 Jun;33(6):874-881.
- 129 Nat Genet. 2012 Oct;44(10):1142-1146.
- 130 Sci Rep. 2014 Jun 18;4:5340.
- 131 Ophthalmology. 2010 Jul;117(7):1331-8.e5
- 132 Science. 2007 Sep 7;317(5843):1397-400.
- 133 PLoS One, 2012:7(7):e40107.
- 134 PLoS One. 2012:7(3):e33389.
- 135 Nat Genet, 2015 Apr:47(4):387-92.
- 136 Hum Mol Genet, 2014 Nov 15:23(22):6081-7
- 137 Nat Genet. 2013 Nov;45(11):1361-5.
- 138 Nat Genet, 2013 Nov:45(11):1284-92.
- 139 Arthritis Rheumatol, 2015 Jan:67(1):140-51
- 140 Nat Genet. 2013 Feb;45(2):202-7.
- 141 PLoS One. 2014; 9(7): e102100. 142 Ann Hum Genet. 2013 May;77(3):215-31.
- 143 Ophthalmology. 2012 Sep;119(9):1874-85. 144 Proc Natl Acad Sci U S A. 2010 Apr 20;107(16):7401-6.
- 145 Sci Rep. 2017 Aug 3;7(1):7173.
- 146 Nat Genet. 2016 Feb;48(2):134-43.
- 147 Genetics. 2017 Feb; 205(2): 919-924.

Tested by M.T. Lee Myeong-geun (20058) / MKJue

Confirmed by M.D. Seol Chang-an (1037)

Confirmed by M.D. Gi Chang-seok (547)



[107/107]



