Cancer GENOME screen Report

Cancer Genome Screen

Institution		**Sample ID**	20211231-9710000
Name	Jason Doe	**Medical record No.**	
Age / Sex	57 / M	**Accepted**	2021-12-31
Sample type	WB	**Reported**	2022-01-10

The test result of Jason Doe

Test Description

This is a brief description of cancer genome screening.

This analysis, being the latest gene analysis technique, analyses genes that can cause cancer and reviews the results of existing research papers to provide personalized information to help people manage their health. Even if any pathogenic variant(PV) associated with hereditary cancer is found, individuals may have no symptoms(reduced penetrance). However, such individual may have higher risk of cancer compared to the general population, measures to reduce the cancer risk and regular thorough examinations for early detection are recommended.

Summary of Test Results

A Likely pathogenic variant(LPV)
related to Breast Cancer Susceptibility
was detected.

Gene	DNA Change	Predicted AA Chanage	Zygosity	Class
CHEK2	c.470T>C	p.Ile157Thr	Het	LPV

Medical Technologist : M-K Lee M.T(20058) MKlee

Lab Director(medical doctor) : Ju-Seon Song M.D(997) Song Ju Seon

Lab Director(medical doctor) : Young-gon Kim M.D(1139) YGkim

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Interpretation

This variant has been reported in the literature in large meta-analyses involving several thousand cases and controls. Individuals who carried the Ile157Thr variant had a slightly increased risk of breast cancer (OR=1.48-1.58) (PMID: 22799331, 23713947), and colorectal cancer (OR=1.48-1.67) (PMID: 22901170, 23713947). The risk was found to be more pronounced for lobular type breast tumors (OR=4.17) (PMID: 22799331). ClinVar contains an entry for this variant as 'Conflicting Interpretations' (Likely pathogenic(9);Pathogenic(5);Uncertain significance(7), ID: 5591).

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Relevant Disease Information

Disease	Test guidebook
Breast Cancer, Susceptibility	**03.Prevention and Treatment page14, 16~17**

About 5-20% of breast cancers are caused by genetic abnormalities that have been inherited from Parents. Most of hereditary breast cancers are caused by pathogenic mutations in the BRCA1 and BRCA2 genes, but the incidence rate of breast cancer can increase due to mutations in other genes as well. This is called breast cancer susceptibility gene.

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The result details and related diseases

* Hereditary Breast and Ovarian Cancer

Hereditary diseases that cause breast and ovarian cancer due to the abnormalities in the BRCA1 and BRCA2 gene.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Breast cancer, Ovarian cancer	BRCA1	☐	☑
	BRCA2	☐	☑

* Breast Cancer, Susceptibility

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Breast cancer	ATM	☐	☑
	CDH1	☐	☑
	CHEK2	☑	☐
	NBN	☐	☑
	NF1	☐	☑
	PALB2	☐	☑

* Ovarian Cancer, Susceptibility

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Ovarian cancer	BRIP1	☐	☑
	RAD51C	☐	☑
	RAD51D	☐	☑

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The result details and related diseases

* Li-fraumeni syndrome

Familial cancer diseases that are inherited as autosomal dominant due to the abnormalities in TP53.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Breast cancer, Brain tumor, leukemia, Adrenocortical carcinoma etc.	TP53	☐	☑

* Peutz Jeghers syndrome

Hereditary diseases that show multiple hamartomatous polyposis in the digestive tract and melanin pigmentation on the skin mucosa.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Colorectal cancer, Gastric cancer	STK11	☐	☑

* Lynch syndrome

Diseases that show a risk to several cancers due to genetic abnormalities of the DNA repairing system.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Colorectal cancer, Endometrial cancer, Gastric cancer, Ovarian cancer etc.	EPCAM	☐	☑
	MLH1	☐	☑
	MSH2	☐	☑
	MSH6	☐	☑
	PMS2	☐	☑

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The result details and related diseases

* Polyposis syndrome

Hereditary diseases associated with developing multiple polyps in the stomach, colon, and rectum.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Familial adenomatous polyposis	APC	☐	☑
MUTYH-associated polyposis	MUTYH	☐	☑
Juvenile polyposis syndrome	BMPR1A	☐	☑
	SMAD4	☐	☑

* Von hippel-lindau syndrome

Hereditary diseases that cause malignant and benign tumors, especially in the central nervous system and kidneys.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
CNS hemangioblastoma, Retinal hemangioblastoma, Pheochromocytoma	VHL	☐	☑

* Multiple endocrine neoplasia

Hereditary diseases of in the endocrine system, such as thyroid glands, parathyroid glands, intestinal and pancreatic neuroendocrine system, anterior pituitary, and skin.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Multiple Endocrine Neoplasia Type 1	MEN1	☐	☑
Multiple Endocrine Neoplasia Type 2	RET	☐	☑

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The result details and related diseases

* PTEN hamartoma tumor syndrome

Hamartoma of various organ associated with PTEN gene abnormalities.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Breast cancer, Thyroid cancer	PTEN	☐	☑

* Retinoblastoma

Diseases in which primary malignant tumors occur in the optic nerve cells of the retina, mostly in infants and babies.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Retinoblastoma	RB1	☐	☑

* Hereditary paraganglioma pheochromocytoma syndrome

Endocrine diseases that indicate pheochromocytoma in the adrenal gland, paraganglioma and neuroendocrine tumor.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Paraganglioma, Pheochromocytoma	SDHD	☐	☑
	SDHAF2	☐	☑
	SDHC	☐	☑
	SDHB	☐	☑

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The result details and related diseases

* Tuberous sclerosis complex

Hereditary diseases in which tumors in the central nervous system and various body parts, associated with mental retardation, epilepsy, and skin lesions, appear.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Retinal tumor, Brain tumor, Lung lymphoma etc	TSC1	☐	☑
	TSC2	☐	☑

* WT1-related wilms tumor

Diseases that can accompany congenital abnormality along with malignant tumors in the kidneys.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Renal cell carcinoma	WT1	☐	☑

* Neurofibromatosis type 2

Diseases in which a benign tumor occurs in the acoustic nerve, a type of brain nerve.

Disease	Gene	Pathogenic Variant	
		Detected	Not detected
Acoustic Neuroma	NF2	☐	☑

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What is Cancer Genome Screen?

Hereditary cancer refers to a cancer caused by an abnormality in the gene associated with a tumor occurrence, the oncogene or the tumor suppressor gene. About 5~10 % of all cancers are known to be hereditary cancers.
Early diagnosis through genetic testing is important in hereditary cancers because they can occur earlier than non-hereditary cancers and can lead to cancer in many organs.
Cancer Genome Screen is a test that can be expected to prevent, diagnose early, and improve the treatment effects regarding hereditary cancer by examining 35 genes known to increase the risk of developing various cancers, including breast cancer, ovarian cancer, colon cancer, prostate cancer, pancreatic cancer, and thyroid cancer, with NGS test.

Hereditary cancer diseases have these characteristics.

- Hereditary cancer is caused by a pathogenic variant (PV) of a gene known to cause certain cancers. Hereditary cancers may have different genes for different types of cancers, and abnormalities in more than one gene can cause various cancers.
- Even if pathogenic variant(PV) is found in genes related to hereditary cancer, cancer does not occur 100% (Reduced Penetrance). However, it is important to be aware of and prevent it because the risk of cancer is very high compared to the general population. In particular, regular thorough examinations for early detection are recommended by identifying types and risks of cancer with high incidence.
- Hereditary cancers account for 5-10% of all cancers, and if more than one family member is diagnosed with cancer, the risk of developing cancer at a young age or simultaneously in multiple organs increases.
- Even if genetic testing related to hereditary cancer does not identify any disease-related PV, there is still a possibility of cancer occurrence due to non-hereditary causes such as environmental effects and lifestyle.

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Disease-associated Genes

Cancer genome screen tests following diseases and genes according to the recommendation of the American College of Medical Genetics and Genomics (ACMG).

Diseases		Gene
Hereditary Breast and Ovarian Cancer	Breast cancer, Ovarian cancer	BRCA1
		BRCA2
Breast Cancer, Susceptibility	Breast cancer	ATM
		CDH1
		CHEK2
		NBN
		NF1
		PALB2
Ovarian Cancer, Susceptibility	Ovarian cancer	BRIP1
		RAD51C
		RAD51D
Li-fraumeni syndrome	Breast cancer, Brain tumor, leukemia, Adrenocortical carcinoma etc.	TP53
Peutz Jeghers syndrome	Colorectal cancer, Gastric cancer	STK11
Lynch syndrome	Colorectal cancer, Endometrial cancer, Gastric cancer, Ovarian cancer etc.	EPCAM
		MLH1
		MSH2
		MSH6
		PMS2
Polyposis syndrome	Familial adenomatous polyposis	APC
	MUTYH-associated polyposis	MUTYH
	Juvenile polyposis syndrome	BMPR1A
		SMAD4
Von hippel-lindau syndrome	CNS hemangioblastoma, Retinal hemangioblastoma, Pheochromocytoma	VHL
Multiple endocrine neoplasia	Multiple Endocrine Neoplasia Type 1	MEN1
	Multiple Endocrine Neoplasia Type 2	RET
PTEN hamartoma tumor syndrome	Breast cancer, Thyroid cancer	PTEN
Retinoblastoma	Retinoblastoma	RB1
Hereditary paraganglioma pheochromocytoma syndrome	Paraganglioma, Pheochromocytoma	SDHD
		SDHAF2
		SDHC
		SDHB
Tuberous sclerosis complex	Retinal tumor, Brain tumor, Lung lymphoma etc	TSC1
		TSC2
WT1-related wilms tumor	Renal cell carcinoma	WT1
Neurofibromatosis type 2	Acoustic Neuroma	NF2

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Test Information and Limitations

- Genetic variation is divided into five categories, pathogenic variant (PV), likely pathogenic variant (LPV), variant of unknown significance (VUS), likely benign variant (LBV), and benign variant (BV), according to 2015 ACMG/AMP.

- The disease relevance of sequence variation according to 2015 ACMG/AMP guidelines is analyzed by a specialist in the department of laboratory medicine, combining various evidence such as allele frequency in population databases, frequency, function analysis and computer prediction, and papers and mutation databases. However, the interpretation of the variation could be changed as additional evidence builds up after the results are reported.
- In this test, it is a rule to report mainly pathogenic variant and likely pathogenic variant, which have high or very high disease relevance, and not to report variant of unknown significance, likely benign variant, and benign variant. But in some genes (RET, SDHAF2), it is a rule to report only well-known PVs.

- Among the genes included in the test, the MUTYH gene is inherited as an autosomal recessive gene. For autosomal recessive genes, two PVs must exist to increase the possibility of cancer. Therefore, for MUTYH, it is a rule to report only when two PVs are present.

- The genes included in the test include the entire exon, but in some areas sequencing may not be sufficiently covered. In addition, if a highly homologous sequence exists, the sequencing of the base may not be accurate, and variations in large deletions or duplications or non-protein-coding sequence areas may be difficult to detect.

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Test Information

Specimen	**Peripheral Blood**
Method	**Next-Generation Sequencing; NGS**

Next-Generations Sequencing Test

This technique breaks down the genome into many pieces, reads each piece at the same time, and combines the data obtained with bioinformatic techniques to quickly decode vast amounts of genome information.

Reference Transcript			
APC	NM_000038.5	PMS2	NM_000535.5
ATM	NM_000051.3	PTEN	NM_000314.4
BMPR1A	NM_004329.2	RAD51C	NM_058216.2
BRCA1	NM_007294.2	RAD51D	NM_002878.3
BRCA2	NM_000059.3	RB1	NM_000321.2
BRIP1	NM_032043.2	RET	NM_020975.4
CDH1	NM_04360.3	SDHAF2	NM_017841.2
CHEK2	NM_007194.3	SDHB	NM_003000.2
EPCAM	NM_002354.2	SDHC	NM_003001.3
MEN1	NM_130799.2	SDHD	NM_003002.2
MLH1	NM_000249.3	SMAD4	NM_005359.5
MSH2	NM_000251.2	STK11	NM_000455.4
MSH6	NM_000179.2	TP53	NM_000546.5
MUTYH	NM_001128425.1	TSC1	NM_000368.4
NBN	NM_002485.4	TSC2	NM_000548.3
NF1	NM_000267.3	VHL	NM_000551.3
NF2	NM_000268.3	WT1	NM_024426.4
PALB2	NM_024675.3		

Reference

1. Breast Cancer Information Core (http://research.nhgri.nih.gov/bic)
2. The Human Gene Mutation Database (http:www.hgmd.cf.ac.uk)
3. Gene Reviews (http://geneclinics.org)

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Clinical significance of each tested gene

Even in people with positive result in the test, the onset and symptoms of the disease vary and it doesn't mean that necessarily the disease necessarily occur. In addition, even in people with negative result, the disease can be caused by other genes that have not been tested or other factors.

Genes with high clinical significance

APC	BRCA1	BRCA2	CDH1	MEN1	MLH1	MSH2	NF1	NF2
PTEN	RB1	RET	SDHB	SDHD	SMAD4	STK11	TP53	TSC1
TSC2	VHL							

Genes with clinical significance partially proven

BMPR1A	*MedlinePlus*
	More than 60 pathogenic variants in the BMPR1A gene have been found to cause juvenile polyposis syndrome. Most BMPR1A pathogenic variants result in the production of an abnormally short, nonfunctional protein. As a result, the BMPR1A protein cannot bind to ligands in the TGF-β pathway. This disruption in binding interferes with the activation of the SMAD protein complex. This inactive complex is not transported to the nucleus, where it is needed to regulate cell growth and the activity of certain genes. Unregulated cell growth can lead to polyp formation in people with juvenile polyposis syndrome.
MSH6	*MedlinePlus*
	Pathogenic variants in the MSH6 gene have been reported in about 13 percent of families with Lynch syndrome that have an identified pathogenic variant. Lynch syndrome increases the risk of many types of cancer, particularly colorectal cancer. People with Lynch syndrome also have an increased risk of cancers of the endometrium (lining of the uterus), ovaries, stomach, small intestine, gallbladder ducts, upper urinary tract, and brain. By age 75, the risk of developing one of these cancers is 60 percent for women and 40 percent for men with an MSH6 pathogenic variant. Endometrial cancer is especially common in women with Lynch syndrome caused by MSH6 pathogenic variants.
MUTYH	*MedlinePlus*
	Pathogenic variants in the MUTYH gene cause an autosomal recessive form of familial adenomatous polyposis (also called MYH-associated polyposis). Pathogenic variants in this gene affect the ability of cells to correct errors made during DNA replication. In individuals who have autosomal recessive familial adenomatous polyposis, both copies of the MUTYH gene in each cell are mutated. Most pathogenic variants in this gene result in the production of a nonfunctional or low-functioning MYH glycosylase. When base excision repair in the cell is impaired, pathogenic variants in other genes build up, leading to cell overgrowth and possibly tumor formation.
PALB2	*Clinical Cancer Research. 2008;14(18):5931–7*
	Pathogenic variants in the PALB2 gene are associated with an increased risk of developing breast cancer of magnitude similar to that associated with BRCA2 pathogenic variants and PALB2-deficient cells are sensitive to PARP inhibitors.
PMS2	*MedlinePlus*
	Pathogenic variants in the PMS2 gene have been reported in about 6 percent of families with Lynch syndrome that have an identified pathogenic variant. Lynch syndrome increases the risk of many types of cancer, particularly colorectal cancer. People with Lynch syndrome also have an increased risk of cancers of the endometrium (lining of the uterus), ovaries, stomach, small intestine, liver, gallbladder ducts, upper urinary tract, and brain. By age 75, the risk of developing one of these cancers is 30 percent for women and 25 percent for men with a PMS2 pathogenic variant. These athogenic variants lead to a form of Lynch syndrome with a lower risk of cancer development compared to other causes of this condition. Additionally, in people with a PMS2 pathogenic variant, cancer tends to occur at a later age compared to others with Lynch syndrome.

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SDHAF2	*UniProt*
	Pathogenic variants in the SDHAF2 gene cause hereditary paraganglioma, a neuroendocrine tumor. Paraganglioma is a neural crest tumor usually derived from the chemoreceptor tissue of a paraganglion, and may develop at various body sites, including the head, neck, thorax and abdomen.
SDHC	*MedlinePlus*
	More than 30 pathogenic variants in the SDHC gene have been found to increase the risk of hereditary paraganglioma-pheochromocytoma type 3. People with this condition have paragangliomas, pheochromocytomas, or both. An inherited SDHC pathogenic variant predisposes an individual to the condition, and a somatic pathogenic variant that deletes the normal copy of the SDHC gene is needed to cause hereditary paraganglioma-pheochromocytoma type 3.
WT1	*MedlinePlus*
	At least 80 pathogenic variants in the WT1 gene have been found to cause Denys-Drash syndrome, a condition that affects development of the kidneys and genitalia and most often affects males. These pathogenic variants are germline, which means they are present in cells throughout the body. The pathogenic variants that cause Denys-Drash syndrome almost always occur in areas of the gene known as exon 8 and exon 9. Most of these pathogenic variants result in changes in single protein building blocks (amino acids) in the WT1 protein. The most common pathogenic variant that causes Denys-Drash syndrome (found in about 40 percent of cases) replaces the amino acid arginine with the amino acid tryptophan at protein position 394 (written Arg394Trp or R394W).

▶ Genes with low clinical significance

*** The following genes are lacking objective validity for action related to health.**

ATM BRIP1 CHEK2 EPCAM NBN RAD51C RAD51D

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