CGenome

Medical Genome Check up

cBioinformatics

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Introduction

Since the early 2000s, large-scale genetic studies involving thousands to hundreds of thousands of individuals have been conducted worldwide. These studies, known as genome-wide association studies (GWAS), are compiled in the GWAS Catalog. Based on this evidence, diseases that have been validated in Japanese or East Asian populations were selected, and your genetic risk of developing these conditions was estimated by referring to your genotype.

Disease risk is not determined by genetic variation alone. It is also significantly influenced by lifestyle and environmental factors. Therefore, the results of this report should be used as a reference to support your health management and preventive care.

Genetic variants (such as A/T) are described based on the human reference genome GRCh38, which is the current global standard in genomic analysis. As a result, the notation may differ from that used in earlier studies or other genetic testing services.

Risk estimates for each condition are calculated using data from previous genomic studies involving individuals with genetic profiles similar to yours. Even if a higher risk score is indicated, maintaining a healthy lifestyle, paying attention to early symptoms, and undergoing regular medical checkups can help support long-term health.

Please note that some sex-specific conditions, such as prostate cancer, may appear in reports for female individuals. These entries are based on genetic variation analysis only and do not indicate that the condition will actually occur.

Prediction of Disease Susceptibility

This section provides detailed information on conditions for which a higher genetic risk has been identified.

Cancer

Breast cancer 1.21	Ovarian cancer 1.02	Pancreatic cancer	Diffuse gastric adenocarcinoma	Non-small cell lung cancer	Gastrointestinal-type adenocarcinoma	Papillary thyroid carcinoma
Renal cell carcinoma	Acute lymphoblastic leukemia	Differentiated thyroid carcinoma	Hepatocellular carcinoma	Luminal A breast cancer	Gallbladder tumor	Esophageal squamous cell carcinoma
Chronic myeloid leukemia	Intracranial germ cell tumor	Gastric cardia cancer	Esophageal cancer	Bladder cancer	Upper laryngeal cancer	Lung squamous cell carcinoma
Stomach cancer	B-cell non-Hodgkin lymphoma	Colorectal cancer	Pancreatic cancer	Liver cancer	Lung adenocarcinoma	Prostate cancer

Metabolism, Endocrinology, and Urinary System

Chronic kidney disease	Urolithiasis	Nephrolithiasis	Primary aldosteronism	Metabolic syndrome	Pregnancy-induced hypertension syndrome	Membranous nephritis
Gestational diabetes	Type 1 diabetes	Hyperthyroidism	Hypertensive nephropathy	Polycystic ovary syndrome (PCOS)	Thyroid nodule	Hypertriglyceridemia
Hyperuricemia	Type 2 diabetes	IgA nephropathy	Sjögren's syndrome	Nephrotic syndrome	Graves' disease	Dyslipidemia

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Cardiovascular and Respiratory System

Chronic obstructive pulmonary disease (COPD)	Myocardial infarction	Takayasu arteritis	Peripheral artery disease	Cardiac arrhythmia	Brugada syndrome	Resistant hypertension
Large-vessel stroke	Cardioembolic stroke	Stroke	Vasculitis	Nontuberculous mycobacterial lung infection	Small-vessel stroke	Coronary artery aneurysm
Orthostatic hypotension	Childhood-onset asthma	Interstitial lung disease	Intracerebral hemorrhage	Familial long QT syndrome	Cardioembolism	Small-vessel occlusion
Hypertrophic cardiomyopathy	Hemorrhagic stroke	Coronary spasm	Congestive heart failure	Atopic asthma	Aspirin-induced asthma	Moyamoya disease
Coronary artery disease	Idiopathic pulmonary fibrosis	Asthma	Hypertension			

Gastrointestinal System

Inflammatory bowel disease (IBD)	Non-alcoholic fatty liver disease (NAFLD)	Liver cirrhosis	Biliary cirrhosis	Gastritis	Duodenal ulcer	Colonic diverticu-lum
Chronic hepatitis B	Chronic hepatitis C	HCV-related cirrhosis	Crohn's disease	Primary biliary cholangitis (PBC)	Duodenal ulcer	Ulcerative colitis

Prediction of Disease Susceptibility

This section provides detailed information on conditions for which a higher genetic risk has been identified.

Eye and Ear

Astigmatism	Hearing loss	Glaucoma	Diabetic retinopathy	Myopia	Low-tension glaucoma	Pathologic myopia
Age-related hearing loss	Olfactory dysfunction	COVID-19 symptom susceptibility	Taste disorder	Age-related macular degeneration	Open-angle glaucoma	Corneal astigmatism
High myopia	Vitiligo	Normal-tension glaucoma	Angle-closure glaucoma			

Bone and Joint

Gout	Osteoarthritis	Hip osteoarthritis	Knee osteoarthritis	Osteoporosis	Ossification of the posterior longitudinal ligament (OPLL)	Plantar fibromatosis
Temporomandibular disorder	Idiopathic femoral head necrosis	Behçet's disease	Rheumatoid arthritis	Ankylosing spondylitis	Osteoporosis	Systemic lupus erythematosus (SLE)
Plantar fasciitis						

Prediction of Disease Susceptibility

This section provides detailed information on conditions for which a higher genetic risk has been identified.

Other Conditions

Endometriosis	Migraine	Generalized epilepsy	Attention-deficit/hyper-activity disorder (ADHD)	Bipolar disorder
Headache	Androgenetic alopecia	Herpes zoster	Uterine fibroids	Urticaria
Epilepsy	Bullous pemphigoid	Alopecia	Narcolepsy with cataplexy	Hypersomnia
Refractory atopic dermatitis	Sarcoidosis	Dysmenorrhea	Vitiligo	Raynaud's disease
Hyperhidrosis	Major depressive disorder	Psoriasis vulgaris	Psoriasis	Periodontitis
Obstructive sleep apnea syndrome	Atopic dermatitis	Keloid	Hansen's disease	Amyotrophic lateral sclerosis (ALS)
Kawasaki disease	Cerebral aneurysm			

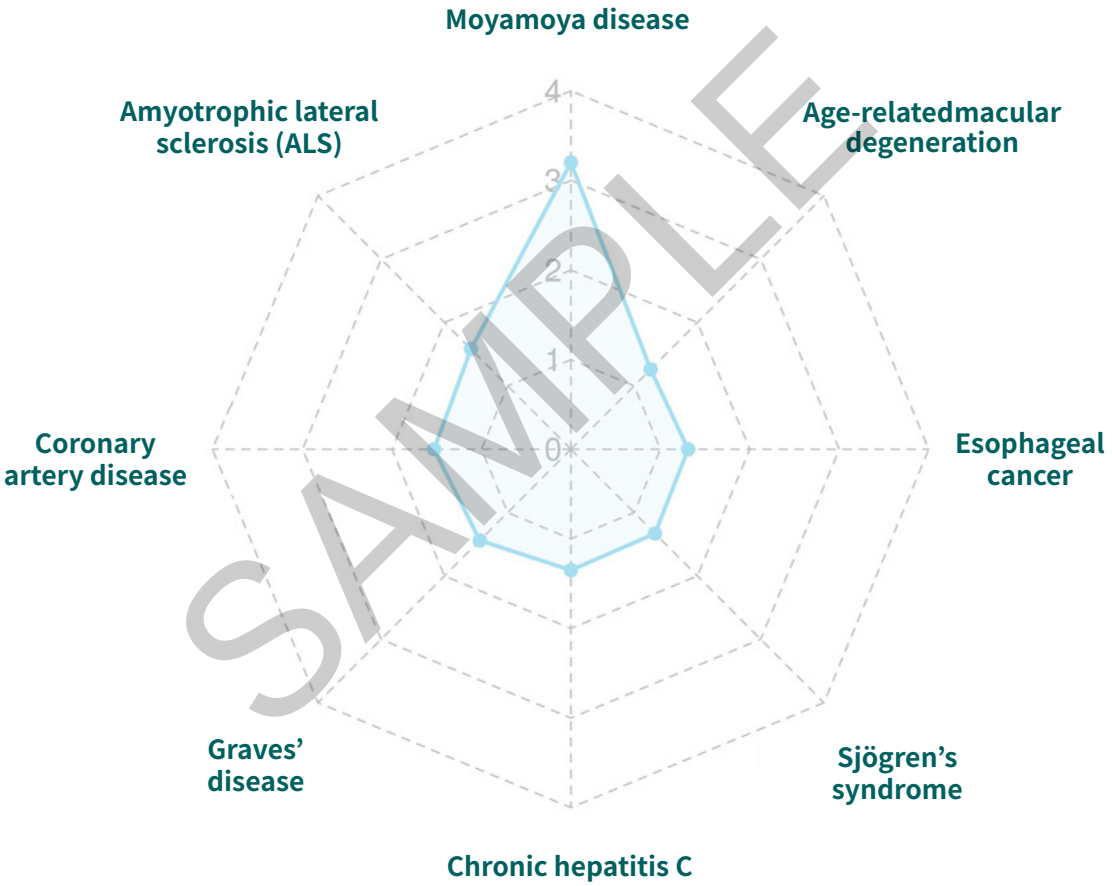
Details of High-Risk Conditions

Risk scores for each condition are presented by category.

Chronic Hepatitis C  Markers: HCV antibody, HCV-RNA	Symptoms: Often asymptomatic in the early stages. Loss of appetite, abdominal pain, fatigue, etc.	Diagnosis: HCV antibody, HCV-RNA	Risk: Exposure to the blood or body fluids of an infected individual
Sjögren's Syndrome  Markers: None	Symptoms: Dry mouth or eyes, joint pain, pallor of the fingers and toes	Diagnosis: Biopsy of salivary or lacrimal glands, salivary secretion test, tear secretion test, autoantibody testing	Risk: Unknown
Graves' Disease  Markers: TSH, fT3, fT4	Symptoms: Palpitations, rapid heartbeat, tremor of the hands, weight loss, nervousness and anxiety, protrusion of the eyes	Diagnosis: Blood thyroid hormone levels, TSH, thyroid ultrasound examination	Risk: Certain medications (amiodarone, interferon-alpha, PD-1 inhibitors) and excessive iodine intake

Details of High-Risk Conditions

This section shows conditions for which an increased risk has been identified.
"High risk" indicates a relative risk of 1.2 times or higher compared with the average risk in the Japanese population.



Medications Requiring Caution

This section lists medications that require caution based on your genetic profile.
Recommended administration guidelines are provided in the original English.
Please consult your physician before taking any medication.

- **Methylene Blue**
  - Indication: Treatment of toxic methemoglobinemia
  - Population: General
  - Phenotype: G6PD: Normal
  - Gene: G6PD Genotype: B (reference) / B (reference)
- **Azathioprine**
  - Indication: Immunosuppressant
  - Population: General
  - Phenotype: NUDT15: Normal Metabolizer | TPMT: Normal Metabolizer
  - Gene: NUDT15 | TPMT
  - Genotype: *1/*1 | *1/*1
- **Tacrolimus**
  - Indication: Immunosuppressant
  - Population: General
  - Phenotype: CYP3A5: Normal Metabolizer
  - Gene: CYP3A5
  - Genotype: *1/*1
- **Dapsone (Diaminodiphenyl Sulfone)**
  - Indication: Synthetic antibacterial agent / Immunosuppressant
  - Population: General
  - Phenotype: G6PD: Normal
  - Gene: G6PD
  - Genotype: B (reference) / B (reference)
- **Desflurane**
  - Indication: Inhalational anesthetic
  - Population: General
  - Phenotype: CACNA1S: Uncertain Susceptibility | RYR1: Uncertain Susceptibility
  - Gene: CACNA1S | RYR1
  - Genotype: Reference / Reference | Reference / Reference

Exercise Type

This section describes whether your genetic profile indicates a higher proportion of fast-twitch or slow-twitch muscle fibers.

Power Type (C/C genotype)

Slow-twitch fibers, also known as “red muscle,” contract slowly and are resistant to fatigue.
They produce energy through aerobic metabolism by using oxygen to burn carbohydrates and fats, making them suitable for endurance activities.

Recommended sports: Long-distance running, triathlon, Pilates, and other aerobic activities.

The proportion of fast-twitch and slow-twitch muscle fibers is associated with the ACTN3 gene (rs1815739, R577X). This gene has three polymorphisms, each with the following characteristics.

Genotype	SNP (rs1815739)	Muscle Fiber Profile	Performance Characteristics
R/R type	C/C (Arginine, Arg)	Fast-twitch dominant	Advantageous for sprinting and power-based activities
R/X type	C/T (Arginine / Stop codon)	Balanced fast- and slow-twitch fibers	Suitable for both power and endurance activities
X/X type	T/T (Stop codon)	Slow-twitch dominant	Advantageous for endurance and long-duration activities

Reference: Other Exercise Types

Stamina Type (T/T genotype)

Slow-twitch muscle fibers, also known as “red muscle,” contract slowly and are resistant to fatigue. They produce energy by using oxygen to metabolize carbohydrates and fats. These fibers support sustained muscle activity and are well suited for aerobic exercise.

Recommended sports:
Long-distance running, triathlon, Pilates, and other aerobic activities.

Balance Type (C/T genotype)

This type has a well-balanced distribution of fast- and slow-twitch muscle fibers. It provides both explosive power and endurance, making it suitable for sports that require a combination of both.

Recommended sports:
Middle-distance running, soccer, tennis, and boxing.

Physical Characteristics

Power Athlete Profile

Overall Rating 

Your Score	rsID	Effect	Gene
☆	rs4363	Involved in blood pressure regulation by converting angiotensin into angiotensin II, which causes vasoconstriction. The performance-enhancing type is reported to be more common among power-oriented athletes.	ACE
☆	rs1815739	Associated with deficiency of α-actinin-3, which is linked to slow-twitch muscle dominance.	ACTN3
★	rs699	Involved in the renin–angiotensin system, similar to ACE, and associated with regulation of blood pressure and body fluid balance.	AGT
★	rs1800795	Associated with post-exercise recovery and maintenance of muscle mass.	IL6-174
★	rs4880	Associated with antioxidant capacity and recovery after exercise.	SOD2
☆	rs1799983	Involved in the production of nitric oxide (NO), which promotes vasodilation and helps regulate blood pressure and circulation. It may also contribute to improved blood flow.	NOS3
★	rs2070744	Involved in the production of nitric oxide (NO), which promotes vasodilation and helps regulate blood pressure and circulation. It may also contribute to improved blood flow.	NOS3

Physical performance is influenced not only by muscle characteristics but also by factors such as blood pressure, cardiopulmonary function, oxygen delivery to muscles via the blood, and vascular function. These functions are affected by both environmental factors and genetically inherited traits.

Understanding your genetic characteristics may help you optimize your daily training and physical activity management.

Endurance Athlete Profile

Overall Rating 

Your Score	rsID	Effect	Gene
☆	rs1799945	A key gene involved in iron metabolism. It is associated with increased oxygen transport capacity and may contribute to improved endurance performance.	HFE
★	rs1815739	Associated with deficiency of α-actinin-3, which is linked to slow-twitch muscle dominance.	ACTN3
☆	rs8192678	Associated with mitochondrial biogenesis and may contribute to enhanced endurance capacity.	PPARGC1A
☆	rs17602729	Involved in the activation of AMP deaminase, which plays an important role in ATP production and energy metabolism, including carbohydrate and lipid utilization.	AMPD1
★	rs1799945	A key gene involved in iron metabolism. It is associated with increased oxygen transport capacity and may contribute to improved endurance performance.	HFE

Physical performance is influenced not only by muscle characteristics but also by factors such as blood pressure, cardiopulmonary function, oxygen delivery to muscles via the bloodstream, and vascular function.
These functions are affected by both environmental factors and genetically inherited traits.

Understanding your genetic characteristics may help you optimize your daily training and physical activity management.

This section describes your alcohol metabolism profile.

High alcohol tolerance

Low alcohol tolerance

EffAlcohol Toleranceect	Characteristics	ADH1B (rs1229984)	ALDH2 (rs671)
Very High Tolerance	Alcohol is metabolized quickly, making hangovers less likely.	TT (Fast metabolizer)	GG (Active type)
Average to Slightly Low Tolerance	You may become intoxicated easily but can consume alcohol.	CC (Slow metabolizer)	GG (Active type)
Low Tolerance	Alcohol can be consumed, but accumulation of acetaldehyde may cause discomfort or nausea.	TT (Fast metabolizer)	GA (Reduced activity type)
Very Low Tolerance	Even small amounts may cause rapid intoxication and increase the risk of hangover.	CC (Slow metabolizer)	GA (Reduced activity type)
Alcohol Intolerant	Alcohol cannot be properly metabolized, and drinking may be unsafe.	–	AA (Inactive type)

Alcohol metabolism is the process by which alcohol is broken down in the body and eliminated as harmless substances. This process occurs primarily in the liver, where enzymes break down alcohol. Metabolic capacity is influenced by genetic variations.

Understanding your genetic characteristics can help you drink responsibly and in a manner suited to your individual tolerance.