genografi

Listen to Your Genes

www.genografi.com | info@genografi.com

genografi

Welcome,

We congratulate you for discovering your DNA and stepping into a healthier and happier life. This report contains information about your genetic characteristics, your inherited disease risks, your carrier status, and your drug sensitivity. This information has been compiled in the light of recent scientific studies and is produced with the help of Next Generation Sequencing technology which enables us to do the most accurate and detailed analysis in the field.

Your sample has been analyzed according to the norms of the ISO9001 quality management system, ISO 15189 laboratory standard and ISO 27001 information security standard, without being shared with any personnel or third parties. The resulting data was analyzed and interpreted according to all recognized and accepted ACMG standards.

Genografi is a once in a lifetime test that you can always use. It is a starting point for personal, protective and preventive health. In the light of the information provided by Genografi analysis, you can take important steps to keep your life healthier, to have knowledge about diseases and other genetic features that your children can inherit, and to stay in your ideal form. If you share the results of this report with physicians in different aress of expertise, they can offer you precautions and proposals to protect your health for a long time. We hope that this report will guide you through your life.

We congratulate you on your desire to discover your DNA and hope that you will be pleased with our service.

*Sue Richards, Nazneen Aziz, Sherri Bale, David Bick, Soma Das, Julie Gastier-Foster, Wayne W. Grody, Madhuri Hegde, Elaine Lyon, Elaine Spector, Karl Voelkerding, and Heidi L. Rehm. "Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology." Genetics in Medicine Genet Med (2015): 405-23.

genografi

Genome and Genes

There are around 25,000 genes in every human, and all of these genes are called genome. The genome consists of a sequence of four types of DNA bases symbolized by the letters A, C, T and G. There are a total of 3 billion base pairs in a genome, and a very large part of the sequence of these bases is the same in every human being, with regions of difference providing genetic diversity.

What is Variation?

Each human's genome has some DNA differences in the millions of regions compared to the genome considered as reference. These differences are called variations. Most of these variations are harmless and allow diversification. Features such as hair color, freckles or baldness are the result of these variations; familial mediterranean fever, cystic fibrosis or lactose intolerance are also the consequences. Variations that can be associated with diseases are called "pathogenic variations". The Genografi report is based on pathogenic and likely pathogenic variations that are genetic risk factors according to ACMG standards.

Genetic and environmental factors play a role in the formation of complex diseases. The risk factors identified in the light of the scientific researches are of great importance in order to be able to take preventive measures before diseases occur. Having a genetic risk does not necessarily mean you will be affected. Only, for a normal person, it means that you have a higher probability of having this disease.

Genografi is intended for informative purposes only and is not intended for diagnosis or treatment. All variants listed should be evaluated for current relevance in relation to the disease being tested. The pathogenicity classes of the reported variants may change over time in the course of scientific studies. The negative test result means a reduced likelihood for the disease being genetic, it shouldn't be interepreted as having no genetic risk. Variations in genomic regions that are not scanned by the test or technical limitations that the test has may result in subsequent changes.

Scientific Reliability

The sample of saliva that we receive from you turns into a result report by following the steps below.

Saliva DNA Sequencing Analysis Genetic Counseling

Saliva

The sample given using the saliva collecting device which comes out of the Genografi kit is sent to our laboratory.

Sequencing

We use special devices designed for DNA sequencing in order to transform the DNA we isolate in the laboratory environment. Unlike other alternative tests, we use Next Generation Sequencing (NGS) technology instead of Microarray technology to ensure that the quality of sequencing is at the highest possible level.

Analysis

The DNA data from the Next Generation Sequencing platform is analyzed with the filter of various databases, population studies and bioinformatics algorithms. As a result of this analysis, the pathogenicity classification of the variations determined according to the ACMG standards is made Limitations: This test can not detect changes in copy numbers of copies that can affect a large part of a gene or the entire population. In genomic regions where the total number of readings is insufficient or too low, analysis may not yield a definitive result.

Genetic Counseling

Your Genografi result is assessed by a specialist geneticist after obtaining the necessary clinical information. Your Genografi report is given by our specialist physician in connection with genetic counseling and the risks related to the diseases and suggestions for solutions.

genografi

Patient Information

Patient ID: GNOG4_S7
Sample Date: 13.10.2016
Report Date: 30.11.2016
Analysis Version: 9-2.8.2-1.2

Content of Genografi Report

Genetic Characteristics

You will find information about your visible genetic characteristics that distinguish you from other people.

Disease Risks

You will finf important data about any hereditary diseases you may have and their risk levels in this section.

Carrier Status

In this section, you will see the diseases and risk levels hidden in the recessive character in your genes that can be passed on to future generations.

Drug Sensitivity

You will get useful information about possible sensitivities and reactions to different medications and treatments.

Pain Sensitivity

Decreased sensitivity to pain.

People's sensitivity to pain varies greatly: some people can tolerate the pain that their peers cannot tolerate. Some syndromes cause people to feel much more pain than others. There are genetic variables that determine the sensitivity of people to pain.

Early Menopause

Typical odds of early menopause.

Menopause is the state in which the production of reproductive hormones in women's lives has ceased and fertility has been lost. The time to start menopause can vary within the 40-60 age range.

Runner Performance

No working copies of alpha-actinin-3 in fast-twitch muscle fiber. Few world-class sprinters have this genotype, but many world-class endurance athletes do.

Why are some of us better in long distance runs, when some of us run 100 meters better? The answer is hidden in the structure of our muscles and therefore in our DNA.

Hair Type

Slightly curlier hair on average.

Having curly hair comes from our genes as well as many other features.

genografi

Reading Ability

Typical nonword reading score.

Nonword reading tests the ability to decode words that do not have any meaning in the sequence of letters. Knowing the genetic factors that influence nonword reading ability can help to understand reading illnesses such as dyslexia.

Bitter Perception

80% chance of not being able to taste certain bitter flavors.

25% of people cannot taste a chemical called propylthiouracil: and there fore cannot percent bitter tastes.This chemical also affects people's eating habits because it is found in many food such as pumpkin, broccolli and coffee.

Ear Wax Type

Wet earwax.

There are two types of ear wex, either dry (flaky, greyish) or wet (sticky and honey colored). This feature is almost completely determined by on ABCC1 gene.

Alcohol Flush Reaction

Two working copies of ALDH2. Little or no flushing reaction to alcohol.

The reason for the redness reaction after alcohol use is due to the non-digestion of a chemical called acetaldehyde. We can understand whether or not this reaction exists by looking at whether the ALDH2 gene is working properly.

Eye Color

56% chance of brown eyes; 37% chance of green eyes; 7% chance of blue eyes.

Eye color is a feature determined by the amount of melanin. The amount of melanin is a complex condition (feature) due to many different genes.

genografi

Disease Risks

What is risk?

Genetic and environmental factors play a role in the formation of complex diseases. Genetic factors are evaluated with the knowledge of the variants you have. These variations are interpreted with clinical data and family history and then your risk profile is created.

For certain diseases, just because you have higher risk profile than average of the society does not mean you will be affected. On the other hand knowing your risk profile and actions you need to take in order to prevent it has huge importance.

For instance, smoking is the most known risk factor for several diseases. Smoking men are 23 times more likely to get lung cancer than the ones who do not smoke. But highness of the risk number doesn't mean every person who is a smoker will get lung cancer. While 18-25% of the smokers have lung cancer, other do not have lung cancer even though they are smokers. So, just being in environmental and genetic risk group doesn't mean you will certainly get the disease.

How can I use the result of this report?

By using Genografi report, if you see a doctor who is a specialist in your identified health risk, you can get information about preventive actions and you can arrange a regular doctor control so that you can create a chance for early diagnosis in case of possibility of catching the disease. Besides Genografi can help diagnose the unknown and undiagnosable symptoms you have.

Preventing the occurrence of a disease could be safer and more successful than trying to cure an uncontrollable disease.

One genetic disease risk is detected. It is:

Hypertrophic Cardiomyopathy

Hypertrophic Cardiomyopathy

1 pathogenic variation is detected for Hypertrophic Cardiomyopathy

Doctor's Recommendation

You are in increased risk group for this disease. Therefore you should get information about the signs and sym ptoms of this disease and the necessary precautions under doctor's control.

Hypertrophic cardiomyopathy (HCM) is a disease in which the heart muscle becomes abnormally thick. The thickened heart muscle can make it harder for the heart to pump blood.Hypertrophic cardiomyopathy (HCM) is very common and can affect people of any age and gender. HCM occurs if heart muscle cells enlarge and cause the walls of the ventricles to thicken. Despite this thickening, the ventricle size often remains normal. However, the thickening may block blood flow out of the ventricle. If this happens, the condition is called obstructive hypertrophic cardiomyopathy. HCM also can affect the heart's mitral valve, causing blood to leak backward through the valve. Sometimes, the thickened heart muscle doesn't block blood flow out of the left ventricle. This is called non-obstructive hypertrophic cardiomyopathy. The entire ventricle may thicken, or the thickening may happen only at the bottom of the heart. The right ventricle also may be affected.In both types of HCM (obstructive and non-obstructive), the thickened muscle makes the inside of the left ventricle smaller, so it holds less blood. The walls of the ventricle also may stiffen. As a result, the ventricle is less able to relax and fill with blood.

genografi

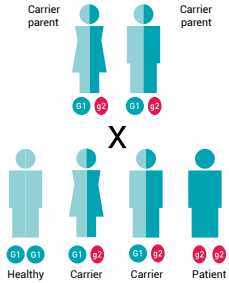
Your Genetic Data

Gene	Variation
MYBPC3	NM_000256.3:c.1210C>T

	dbSNP	Chromosome	Position	Genotype	Exon/Intron	Result of change	Pathogenicity
1.	rs727504329	11	47365056	Heterozygous	Exon 13	Stop Gained	Pathogenic

genografi

Carrier Status



You will see the risk levels of diseases and diseases with recessive characteristics which is disguised and can be transferred through generations. When you are a carrier of a recessive pathogenic variation the syndromes of this variation may not be seen in you but it can be transferred to the next generation.

For the diseases that we scanned, in this section, you are probably not ill because you received them one of your parents (just your mother or father) as a working copy. But if your husband has similar variations in the same gene the possibility of you child to born ill would be minimum 25%.

One carrier status variation is decected. It is;

Autosomal Recessive Polycystic Kidney Disease

Autosomal Recessive Polycystic Kidney Disease

You are a carrier for Autosomal Recessive Polycystic Kidney Disease.

Your Doctor's Recommendation

You have only one copy for this recessive disease, therefore you are a carrier. If you have family history and planning to have children, please see a genetic consultant. We recommend your spouse to be tested as well for your children's risk profiling.

Autosomal Recessive Polycystic Kidney Disease is a very rare condition. In general, polycystic kidney disease is a disorder that affects the kidneys and other organs. Clusters of fluid-filled sacs, called cysts, develop in the kidneys and interfere with their ability to filter waste products from the blood. The growth of cysts causes the kidneys to become enlarged and can lead to kidney failure. Cysts may also develop in other organs, particularly the liver.

genografi

Your Genetic Data

Gene	Variation
PKHD1	NM_138694.3:c.10444C>T

	dbSNP	Chromosome	Position	Genotype	Exon/Intron	Result of change	Pathogenicity
1.	rs148617572	6	51524480	Heterozygous	Exon 68	Missense(R3	Pathogenic

genografi

Drug Sensitivity

According to your Genografi result, atypical drug sensitivity has been detected for 1 out of 6. You do not have to do anything for the typical results. If you have medicines or treatments that have atypical sensitivity, you should keep this situation in mind and share with your doctor if it is necessary. Please, never use this information before consulting to your physician.

Medication/Treatment	Disease Used	Your Sensitivity
Choline ester Metabolism	General Internal Medicine	Typical
Abacavir Hypersensitivity	General Internal Medicine	Typical
Tolbutamide Metabolism	Metabolism	Typical
Statin Sensitivity	**Choloestrol**	**Atypical**
Mefenitoin Metabolism	Metabolism	Typical
Fluourasil Reaction	Cancer	Typical

Statin Sensitivity

When you are using Statin, your chances of having myopathy are relatively high.

Statins are drugs which are used to lower the level of cholesterol in people at high risk for cardiovascular disease. Although they are generally quite safe; they can lead to liver problems and muscle pain and rhabdomyolysis which cause muscle breakdown. In people who use Statin in 10000, myopathy causing muscle pain and weakness can be seen. 1 of the 10000 people who use statin, myopathy can be seen causing muscle pain and weakness.

	dbSNP	Chromosome	Position	Genotype	Exon/Intron	Result of change
1.	rs2395029	6	31431780	Homozigot	Promoter	Upstream Gene

genografi

Your Genetic Data

Gene	Variation
1. HCP5	NA

genografi

Genetic Diseases Screened in Genografi

Genetic Features

Indifference To Pain	Baldness
Pain Sensitivity	Athletic Performance
Bitter Perception	Ear Wax Type
ALDH reaction	Reading Ability
Early Menopause	Hair Type
Eye Color	Smoking Behavior

Pharmacogenetics

Florouacil Response	Tolbutamide Metabolism
Choline Ester Metabolism	Statine Sensivity
Abacavir Hypersensitivity	Mephenytoin Metabolism

Dermatology

Epidermolysis Bullosa	Keratoderma
Cutis Laxa	
Vohwinkel Syndrome	

Endocrinology

Diabetes Mellitus Insulin-Dependent	Hereditary Fructose Intolerance
Protoporphyria	Fabry Disease
Sweat Chloride Elevation Without Cystic Fibrosis	Phosphoribosylpyrophosphate Synthetase Superactivity
Familial Mediterranean Fever	Galactokinase Deficiency
Gangliosidosis	Maturity-Onset Diabetes Of The Young Type
Glycogen Storage Disease	Glutaric Acidemia
Hyper Ige Syndrome	Diabetes Mellitus Transient Neonatal
Permanent Neonatal Diabetes Mellitus	Classic Galactosemia
Congenital Disorder Of Glycosylation	Congenital Hyperinsulinism
Corticosterone Methyl Oxidase Deficiency	Niemann-Pick Disease
Lipoprotein Lipase Deficiency	Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
Methylmalonic Aciduria	Pendred Syndrome
Thylmalonic Aciduria	Porphyria
Mucolipidosis	Sandhoff Disease
Mucopolysaccharidosis	Amyloidosis Hereditary Transthyretin-Related

Gastroenterology

Hereditary Pancreatitis	Cholestasis
Obesity	

Developmental Disorders

Bannayan-Riley-Ruvalcaba Syndrome	LADD syndrome
Beckwith-Wiedemann Syndrome	Mandibuloacral Dysostosis
Ectrodactyly Ectodermal Dysplasia And Cleft Lip/Palate Syndrome	Achondroplasia
Hystrix-like ichthyosis with deafness	Anauxetic Dysplasia
Crouzon Syndrome With Acanthosis Nigricans	Gracile Syndrome
Greig Cephalopolysyndactyly Syndrome	Hutchinson-Gilford Syndrome
IMAGE Syndrome	Keratitis-Ichthyosis-Deafness Syndrome
Cranioectodermal Dysplasia	Craniosynostosis
Leopard Syndrome	Microcephaly
Opitz-Kaveggia Syndrome	Orofacial Cleft
Postaxial Polydactyly	

Chest Diseases

Pneumothorax Primary Spontaneous	Pulmonary Fibrosis And/Or Bone Marrow Failure Telomere-Related
Cystic Fibrosis	

Eye Diseases

Senior-Loken Syndrome	Age-Related Macular Degeneration
Stargardt Disease	Achromatopsia
Marfan Syndrome	Cataract
Progressive External Ophthalmoplegia	Optic Atrophy
Usher Syndrome	

Genetic Diseases Screened in Genografi

Hematology

Sea-Blue histiocyte disease	Thrombotic Hyperhomocysteinemia
Dysprothrombinemia	Homocysteinemia due to MTHFR deficiency
Erythrocytosis	Angiopathy Hereditary With Nephropathy Aneurysms And Muscle Cramps
Hemophilia	Sickle Cell Anemia
Hemolytic Anemia	Prekallikrein Deficiency
Familial Erythrocytosis	Thrombophilia
Alpha-Thalasemia	Thrombocytopenia-Absent Radius Syndrome
Beta Thalassemia	Fanconi Anemia
Factor Xi Deficiency	Heinz Body Hemolytic Anemia
Hemochromatosis	

Cardiology

Familial Hypertrophic Cardiomyopathy	Atrial Fibrillation
Atrial Septal Defect	Dilated Cardiomyopathy
Hyperlipoproteinemia	Long Qt Syndrome
Familial Hypercholesterolemia	Hypercholesterolemia
Brugada Syndrome	Sick Sinus Syndrome
Homocystinuria	Intracardiac Myxoma
Cardiomyopathy	Catecholaminergic Polymorphic Ventricular Tachycardia
Catecholaminergic Polymorphic Ventricular Tachycardia	Cerebrotendinous Xanthomatosis
Wolff-Parkinson-White Syndrome	

Otorhinolaryngology

Deafness

Nephrology

Alport Syndrome	Lipoprotein Glomerulopathy
Nephrotic Syndrome	Polycystic Kidney Disease

Neurological Diseases

Distal Myopathy	Epilepsy
Frontotemporal Dementia With Tdp43 Inclusions Tardbp-Related	Lhermitte-Duclos Disease
Limb-Girdle Muscular Dystrophy	Leukoencephalopathy
Mononeuropathy Of The Median Nerve	Myofibrillar Myopathy
Muscular Dystrophy	Nemaline Myopathy
Neuropathy	Glaucoma
Primary Lateral Sclerosis	Smith-Lemli-Opitz Syndrome
Spastic Paralysis	Familial Dysautonomia
Amyotrophic Lateral Sclerosis	Arts Syndrome
Brain Small Vessel Disease	CAPOS syndrome
Charcot-Marie-Tooth Disease	Cowden Disease
D-Bifunctional Protein Deficiency	Dystonia
Emery-Dreifuss Muscular Dystrophy	Hereditary Sensory And Autonomic Neuropathy
Early Infantile Epileptic Encephalopathy	Phenylketonuria
Hereditary Neuralgic Amyotrophy	Hypokalemic Periodic Paralysis
Holoprosencephaly	Joubert Syndrome
Vldlr-Associated Cerebellar Hypoplasia	Krabbe Disease
Leigh Syndrome	Macrocephaly/Autism Syndrome
Megalencephalic Leukoencephalopathy	Myopathy
Miyoshi Muscular Dystrophy	Nocturnal Frontal Lobe Epilepsy
Neuronal Ceroid-Lipofuscinosis	Parkinson Disease
Peters Anomaly	Pontocerebellar Hypoplasia
Porencephaly	Progressive Myoclonic Epilepsy
CARASIL Syndrome	Anemia Sideroblastic And Spinocerebellar Ataxia
Snyder-Robinson Syndrome	Spastic Ataxia
Spastic Paraplegia	Spinal Muscular Atrophy
Tay-Sachs Disease	Spinocerebellar Ataxia
Wolfram Syndrome	

genografi

Genetic Diseases Screened in Genografi

Audiology

Bjornstad Syndrome
Deafness Congenital

Oncology

5-Fluorouracil Toxicity
Adenomatous Polyposis Coli
Gardner Syndrome
Colorectal Cancer
Prostate Cancer
Pten Hamartomatous Tumour Syndrome
Mismatch Repair Cancer Syndrome
Breast-Ovarian Cancer Familial
Birt-Hogg-Dube Syndrome
Gastric Cancer
Li-Fraumeni Syndrome
Multiple Endocrine Neoplasia
Paragangliomas
Von Hippel-Lindau Syndrome
Hereditary Neoplastic Syndromes
Pheochromocytoma
Leukemia Juvenile Myelomonocytic Somatic
Leukemia
Prostate Cancer Somatic
Thyroid Carcinoma
Familial Adenomatous Polyposis
Pituitary Adenomas
Carney Complex
Choroid Plexus Papilloma
Muir-Torre Syndrome
Pallister-Hall Syndrome
Retinoblastoma
Wilms Tumor

Orthopedics

Carpal Tunnel Syndrome Familial

Paediatrics

Peroxisome Biogenesis Disorder
Alkaptonuria
Bloom Syndrome
Canavan Disease
Alternating Hemiplegia Of Childhood
Dent Disease
Feingold Syndrome
Hereditary Folate Malabsorption
Short Stature
Cartilage-Hair Hypoplasia
Congenital Cataracts Facial Dysmorphism And Neuropathy
Metachromatic Leukodystrophy
Noonan Syndrome
Stiff Skin Syndrome
Wilson Disease
3-Methylglutaconic Aciduria
Bartter Syndrome
Caffey Disease
Child Syndrome
Costello Syndrome
Dubin-Johnson Syndrome
Gaucher Disease
Hermansky-Pudlak Syndrome
Cardiofaciocutaneous syndrome
Cleidocranial Dysplasia
Congenital Secretory Chloride Diarrhea
Epiphyseal Dysplasia
Pyruvate Carboxylase Deficiency
Vitamin D-Dependent Rickets

Perinatology

Dyskeratosis Congenita
Mitochondrial Complex III deficiency

Psychiatry

Lujan-Fryns Syndrome

Paediatrics

Osteogenesis Imperfecta
Parietal Foramina
Bart-Pumphrey syndrome
Cold-Induced Sweating Syndrome
Thanatophoric Dysplasia
Metachondromatosis
Scapuloperoneal Myopathy
Inclusion Body Myopathy
Solitary Median Maxillary Central Incisor

Urology

Spermatogenic Failure

Genetic Diseases Screened in Genografi

Medical Genetics

Hirschsprung Disease	Leri-Weill Dyschondrosteosis
Meckel Syndrome	Mitochondrial DNA Depletion Syndrome
Mitochondrial Complex II Deficiency	Mitochondrial Recessive Ataxia Syndrome
Schizencephaly vs diseases_covered_moi	ADULT Syndrome
Maple Syrup Urine Disease	Benign Familial Neonatal Seizures
CATSHL syndrome	Hyperbilirubinemia
Hyperproinsulinemia	Hypophosphatemic Rickets
Hypochondroplasia	Hmg-Coa Lyase Deficiency
Holocarboxylase Synthetase Deficiency	Juvenile Polyposis Syndrome
Congenital Bilateral Absence Of The Vas Deferens	Lipodystrophy
Medulloblastoma	Metaphyseal Dysplasia
Muenke Syndrome	Orofaciodigital Syndrome
Osteosarcoma	Pigmented Nodular Adrenocortical Disease
Tortuosity Of Retinal Arteries	Sjögren-Larsson Syndrome