TOTAL HEALTH

YOUR NAME

VERELST GENETICS

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Dear ...,

In this report you will discover your genetic predispositions related to health.

In the first pages you will find an iconographic summary of the topics which are explained with more detail in later pages.
Taking into consideration that there are almost 10.000 diseases known with genetic background, everybody in the world has some genetically inherited genes that affect their medical future. This report contains those variants that have been scientifically proven to affect your health or wellbeing, following the guidelines established by the American College of Medical Genetics and Genomics (ACMG).

Some single genetic mutations are enough to cause a disease, although most genetic variants do not cause a disease by themselves alone. The susceptibility to most diseases is generally increased when genetic variations are combined with other genes and/or environmental factors. By keeping a watchful eye and following the outcome and recommendations of this genetic analysis, you can improve your wellbeing and prevent or delay developing the disorders of your genetic inheritance.

The icons next to the topics represent your genetic predisposition for the disease for which you are more susceptible or less susceptible than the general population:

 you are less likely to suffer from the disease

 you have an increased risk for this disease

Diseases for which you have the same susceptibility as the general population are not reported

This analysis report is not a diagnostic tool, but it helps to identify which areas you should pay more attention to. The report is presented in following sections:

COMPLEX DISEASES

This section includes complex diseases that are not caused by a single mutation but are the result of the interaction between several genetic variants and environmental influencers such as food and lifestyle. The data gives your predisposition compared with the rest of the population. It does not mean that you are going to suffer the disease, it only indicates that statistically you have a higher or lower predisposition than the majority (90 %) of the population.

MUTATIONS

This section contains mutations in the most important genes from an oncological point of view. We look for mutations suspected of being pathogenic according to the ClinVar database.

CARRIER STATUS

Some diseases are genetically caused but they only affect persons who carry a mutation in homozygosis, meaning when both copies of the specific gene contain a mutation. People carrying the mutation only in 1 copy of the gene do not present any symptoms at all. The mutated gene however is passed on to the children with a chance of 50%. In case your partner also is a carrier of a mutation in the same gene, this “harmful” gene also has a 50% chance of being passed on, which means that your children have a 25 % chance of ending up with two copies of the mutated gene. In that case they will develop the disease. Since these mutations go unnoticed in carriers and are only noticed when children are born, it is of high value knowing which possible genetic disorders could affect your children. Genetic testing of both parents can provide peace of mind or help make informed decisions regarding family planning.

BIOMARKERS

This section calculates your genetic predisposition to have abnormal levels of certain metabolic parameters. It is also compared to the general population and reported if your values are higher or lower than the majority of 90 % of all people. Due to statistical distribution it is normal that several parameters within this analysis will differ.

We remind you that this report is only valid for research and education. It is not valid for clinical or diagnostic use. Any changes you want to make regarding your health should be guided by your medical doctor.

SUMMARY

Complex diseases


Rheumatoid arthritis


Ovarian cancer in BRCA1 mutation carriers


Upper aerodigestive tract cancers


Endometriosis


Celiac disease


Alzheimer's disease (late onset)


Glioma


Osteosarcoma


Psoriasis

Mutations

We did not detect any mutations that are considered pathogenic in oncological sense.

Carrier status

These diseases do not cause any symptoms for the carrier of the mutation, but can be passed on to the children, who could possibly suffer from the disease if both parents are carriers of a mutation.

Lissencephaly

Biomarkers


Bilirubin levels


C-reactive protein and white blood cell count


Calcium levels


Liver enzyme levels (Gamma-Glutamyl Transferase)


Liver enzyme levels


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COMPLEX DISEASES

Rheumatoid arthritis

Rheumatoid arthritis (RA) is a form of arthritis that causes pain, swelling, stiffness and loss of function in your joints. It can affect any joint but is common in the wrist and fingers.

More women than men get rheumatoid arthritis. It often starts in middle age and is most common in older people. You might have the disease for only a short time, or symptoms might come and go. The severe form can last a lifetime.

Genes analyzed: ACOXL, AFF3, ANKRD55, ARID5B, ATG5, BLK, C1QBP, C4ORF52, C5ORF30, CCL19, CCR6, CD2, CD226, CD28, CD40, CDK6, TYR, CASP8, CLNK, CTLA4, ABDH6, EOMES, ETS1, FADS1.

You are less likely to suffer the disease.

Upper aerodigestive tract cancers

Cancer of the upper aerodigestive tract includes tumors of the oral cavity, pharynx, larynx, nasal cavity and paranasal sinuses, ear and salivary glands. Head and neck carcinoma is the most common among them and has a high mortality rate. Alcohol and tobacco use are the main factors that increase the risk, although human papillomavirus infection also play an important role.

Genes analyzed: ADH1B, ADH7, ADH, ALDH2.

 You are more likely to suffer the disease.

Heavy alcohol intake and tobacco use significantly raise the risks of developing this kind of cancer.
Also, salt-cured foods such as fish and preserved vegetables release harmful chemicals when cooked, which increase the risk even more. By following a healthy lifestyle and a diet based on fresh ingredients your susceptibility for this disease can be reduced.
In general, risks for cancer are knowingly reduced if DNA is well maintained. This is achieved by the methylation cycle, a metabolic pathway in your body that is highly dependent on vitamin intake. The Weight and Fitness management plan contains all information about your genetics and vitamin needs, which will help you to optimize the methylation biochemical reactions and avoid disease.

Ovarian cancer in BRCA1 mutation carriers

Ovarian cancer is the fourth cancer in order of incidence among women and is the leading cause of death from gynecological cancer in Europe. Because there are no tools for early diagnosis or screening for ovarian cancer, early diagnosis has a direct impact on patient survival. Women with a mutation in the BRCA1 gene are at greater risk than the general population of having ovarian cancer. Large-scale association genetic studies have shown that the risk of developing BRCA1-associated pathogenic mutations may vary due to the existence of certain genetic variants.

This section is only valid for women with pathogenic markers in the BRCA1 gene.

Genes analyzed: NR, MDM4, PTHLH.

You are less likely to suffer the disease.

Endometriosis

The uterus, or womb, is the place where a baby grows when a woman is pregnant. Endometriosis is a disease in which the kind of tissue that normally grows inside the uterus grows outside the uterus. It can grow on the ovaries, fallopian tubes, bowels, or bladder. Rarely, it grows in other parts of the body.

Genes analyzed: WNT4, GREB1, NR, ARF, VEZT.

You are less likely to suffer the disease.

Celiac disease

Celiac disease is an immune disease in which people can't eat gluten because it will damage their small intestine. If you have celiac disease and eat food with gluten, your immune system responds by damaging the small intestine. Gluten is a protein found in wheat, rye, and barley. It may also be in other products like vitamins and supplements, hair and skin products, toothpastes, and lip balm. Celiac disease affects each person differently. Symptoms may occur in the digestive system, or in other parts of the body. One person might have diarrhea and abdominal pain, while another person may be irritable or depressed. Irritability is one of the most common symptoms in children. Some people have no symptoms.

Genes analyzed: RGS1, AHSA2, IL18R1, ITGA4, CTLA4, CCR5, IL12A, LPP, IL21, HLA-DQB1, TNFAIP3, SH2B3, PTPN2, TNRFSF14, RUNX3, PLEK, CD80, MAP3K7, THEMIS, ZMIZ1, ETS1, CLEC16A, ICOSLG, CD247, TNFSF18, FRMD4B, ELMO1, YDJC, TLR7.


You are more likely to suffer the disease.

If you have celiac disease, eating gluten triggers an immune system in your small intestine. Following a gluten-free helps to manage the symptoms and promote intestinal healing.

Alzheimer's disease (late onset)

Alzheimer's disease (AD) is the most common form of dementia among older people. Dementia is a brain disorder that seriously affects a person's ability to carry out daily activities. AD begins slowly. It first involves the parts of the brain that control thought, memory and language. People with AD may have trouble remembering things that happened recently or names of people they know. A related problem, mild cognitive impairment (MCI), causes more memory problems than normal for people of the same age. Many, but not all, people with MCI will develop AD. This section analyzes the predisposition to the Late Onset type of Alzheimer.

Genes analyzed: RGS1, AHSA2, IL18R1, ITGA4, ICOS, CCRL2, IL12A, LPP, IL21, HLA-DQB1, TNFAIP3, SH2B3, PTPN2, MMEL1, RUNX3, PLEK, CD80, MAP3K7, THEMIS, ZMIZ1, ETS1, CLEC16A, ICOSLG, CD247, TNFSF18, FRMD4B, ELMO1, YDJC, TLR7.


You are more likely to suffer the disease.

Diet, physical activity and brain stimulation are important factors that modulate the susceptibility to neurological problems. Risk factors that increase the risk for Alzheimer include high levels of cholesterol, diabetes (because they are a potential cause of stroke, which often precedes the onset of Alzheimer) and lower education. By keeping brain and body active you can prevent neurological complications. High level of homocysteine is also an important factor that contributes to the development of the Alzheimer. This can be controlled through adequate food supplementation. The Weight and Fitness management plan contains all information about your genetics and vitamin needs, which will help you to optimize homocysteine levels and avoid disease.

Glioma

Glioma is a type of neoplasm that occurs in the brain or spinal cord. It is called glioma because it arises from glial cells, its most frequent location is the brain.

Genes analyzed: TERT, CCDC26, CDKN2A, PHLDB1, RTEL1.

 You are more likely to suffer the disease.

In general, risks for cancer are knowingly reduced if DNA is well maintained. This is achieved by the methylation cycle, a metabolic pathway in your body that is highly dependent on vitamin intake. The Weight and Fitness management plan contains all information about your genetics and vitamin needs, which will help you to optimize the methylation biochemical reactions and avoid disease.

Osteosarcoma

Osteosarcoma is a very rare type of cancerous bone tumor that usually develops in teenagers. It often occurs when a teen is growing rapidly. Osteosarcoma is the most common bone cancer in children. Average age at diagnosis is 15. Boys and girls are just as likely to develop this tumor until the late teens, when it occurs more often in boys. Osteosarcoma is also common in people over the age of 60.

The cause is not known, but it runs in some families, which indicates a genetic component and some genes have been linked to an increased risk. These genes are also associated with familial retinoblastoma, a type of cancer that occurs in the eye.

Genes analyzed: GRM4, AJ412031, ADAMTS6.


You are more likely to suffer the disease.

This kind of cancer tends to affect children during the phase of rapid growth, and it is very unlikely it will affect you as an adult. Keep a watchful eye for any type of swelling near a bone, especially in arms and legs.

Psoriasis

Psoriasis is a skin disease that causes itchy or sore patches of thick, red skin with silvery scales. You usually get the patches on your elbows, knees, scalp, back, face, palms and feet, but they can show up on other parts of your body. Some people who have psoriasis also get a form of arthritis called psoriatic arthritis. A problem with your immune system causes psoriasis. In a process called cell turnover, skin cells that grow deep in your skin rise to the surface. Normally, this takes a month. In psoriasis, it happens in just days because your cells rise too fast. The disease is not hereditary, but there is a genetic predisposition to it, and a third of those affected have direct relatives with psoriasis.

Genes analyzed: TP63, COG6, LOC144817, RUNX1, CLIC6, OSTN, IL12B, TNIP, IFIH1, LCE, TNFAIP3, REL, PSMA6, NOS2, IL13, DDX58, IL28RA, ILF3, IL23A.

You are less likely to suffer the disease.

CARRIER STATUS

Lissencephaly

LIS1-associated lissencephaly includes Miller-Dieker syndrome (MDS), isolated lissencephaly sequence (ILS), and (rarely) subcortical band heterotopia (SBH). Lissencephaly and SBH are cortical malformations caused by deficient neuronal migration during embryogenesis. Lissencephaly refers to a "smooth brain" with absent gyri (agyria) or abnormally wide gyri (pachygyria). SBH refers to a band of heterotopic gray matter located just beneath the cortex and separated from it by a thin zone of normal white matter. MDS is characterized by lissencephaly, typical facial features, and severe neurologic abnormalities. ILS is characterized by lissencephaly and its direct sequelae: developmental delay, intellectual disability, and seizures.

Genes analyzed: PAFAH1B1.

We have detected at least one mutation that could be pathogenic to your children.

We strongly recommend a genetic test of your partner if you are thinking about family planning. This way you can find out if there is any reason for concern about your children getting any recessive inheritable disease.

BIOMARKERS

Bilirubin levels

Bilirubin is an orange-yellow substance made during the normal breakdown of red blood cells. Bilirubin passes through the liver and is eventually excreted out of the body. Due to lower elimination rate of bilirubin, occasionally this can result in high levels of bilirubin in blood, which can manifest itself as a yellowish tinge of skin or eyes. The condition is harmless and does not cause any health risk.

Genes analyzed: UGT1A8, HIST1H1T, ARHGEF7, SLCO1B1


You are more likely to suffer abnormal levels.

High levels generally become visible in case of illness such as the flu, dehydration, stress, strict low-calorie diet, high-intensity exercise or lack of sleep. Many people with abnormal levels never present any symptoms and are frequently unaware of the condition.

C-reactive protein and white blood cell count

C-reactive protein (CRP) and white blood cell (WBC) have been utilized as critical markers contributing to acute and chronic inflammation.

Genes analyzed: DPF3, FLJ20021, DOCK4, LOC105377910, KCNE4, HNF1A, LOC105374322, PSMD3, LOC100506403

 You are less likely to suffer abnormal levels.

Calcium levels

Calcium is vital to the normal functioning of multiple organ systems and its serum concentration is tightly regulated.

Genes analyzed: CASR, DGKD, GCKR, LOC101928272, LOC105370176, CYP24A1, WDR81.

 You are more likely to suffer abnormal levels.

Calcium is important for optimal bone health throughout your life, but also for heart, muscles and nerves. Although diet is the best way to get calcium, calcium supplements may be an option if your diet falls short.

Liver enzyme levels (gamma-glutamyl transferase)

Concentrations of liver enzymes in plasma are widely used as indicators of liver disease. The liver is essential for digesting food and ridding your body of toxic substances. Genetic heritability, viruses, alcohol use and obesity all influence the health of the liver.

Genes analyzed: PNPLA3, NBPF3, RNU6, LOC105376184, ABO, JMJD1C, FADS2, ST3GAL4, ASGR1, ABHD12, LOC101927479, CEPT1, EFHD1, SLC2A2, HPRT1P2, MLXIPL, DLG5, HNF1A, EXOC3L4, RORA, CD276, LOC102724084, SOX9-AS1, FUT2, MICAL3, GGT1.


You are more likely to suffer abnormal levels.

To protect the liver, it is recommended to drink alcohol in moderation, get vaccination against Hepatitis A and B, and maintain a healthy weight.
Also avoid direct skin contact with toxic substances such as insecticides, fungicides, paint, and make sure not to take wrong medication. Consult the results of your pharmacogenomics test as a guide to avoid drug toxicity.

Liver enzyme levels (other enzymes)

Plasma liver-enzyme tests are widely used in the clinic for the diagnosis of liver diseases and for monitoring the response to drug treatment. There is considerable evidence that human genetic variation influences plasma levels of liver enzymes.

Genes analyzed: JMJD1C, LINC01363, ADMTS13, PNPLA3, NBPF3, GPLD1, GGT1.

 You are less likely to suffer abnormal levels.

Phospholipid levels (plasma)

Long-chain n-3 polyunsaturated fatty acids (PUFAs) can derive from diet or from α-linolenic acid (ALA) by elongation and desaturation

Genes analyzed: TMEM258, MYRF, RPLPOP2, FADS1, FADS2, FEN1, UBXN4, TMEM258, MYRF, ELOVL2, BEST1, LOC101926964, SYCP2L, RAB3IL1, DAGLA, GCKR, LOC105370339, RPS2P37, STIM2.

You are less likely to suffer abnormal levels.

Plasma omega-3-6 polyunsaturated fatty acid levels

Omega 3 and Omega 6 polyunsaturated fatty acids (PUFAs) and their metabolites are involved in cell signaling, inflammation, clot formation, and other crucial biological processes. Genetic components determine the composition and availability of PUFAs for your body.

Genes analyzed: PDXDC1, TMEM258, IL23R, C10ORF128, FADS1, FADS2, NTAN1, FTLP19, TMEM39A, ELOVL2.

 You are more likely to suffer abnormal levels.

The contemporary diet is rich in Omega 6 but short in Omega 3. Both types of fatty acids should be consumed in equal quantities, but the required level of Omega 3 is often not obtained. Supplement your diet with an extra source of Omega 3 EPA and DHA (eicosapentaenoic acid and docosahexaenoic acid) to keep the essential lipids at optimum levels.

Red blood cell count

Hemoglobin is a protein present in red blood cells that carries oxygen to your body's organs and tissues and transports carbon dioxide from organs and tissues back to the lungs. If the level of hemoglobin is lower than normal, it means that we have a low red blood cell count (anemia).

Genes analyzed: PRKCE, ABO, LOC105370987, ALPL, GPLD1, PNPLA3, BRAP, MRC1, LOC105374266, SLC14A2, LOC105377143, CD163, GGT1, ALDH2, TMPRSS6, LIPC, LOC101929011, CETP, LPL, LOC107986647, WDR72, TNFRSF13B, RPS11, HBA2, RCL1, LINC00885, LOC645434, DENND4A, TYMP, LOC105377656, THRB, ATP6V1G3, TIMM23, MARCH8, CCDC162P, UBE2L3.

You are less likely to suffer abnormal levels.

Sex hormone levels

Genetic factors contribute strongly to sex hormone levels, yet knowledge of the regulatory mechanisms remains incomplete.

Genes analyzed: ZNF789, LOC146253, LOC105376607, ANO2, ZKSCAN5, SLC22A24, SULT2A1, LOC102723403.

You are less likely to suffer abnormal levels.

White blood cell count

White blood cells are a type of blood cell that is produced in the bone marrow and found in blood and lymphatic tissues. White blood cells are part of the body's immune system. These help the body fight infections and other diseases. The types of white blood cells are granulocytes (neutrophils, eosinophils, and basophils), monocytes, and lymphocytes (T cells and B cells).

The white blood cell count is a common clinical measure from the whole blood count assays and varies widely among healthy individuals.

Genes analyzed: LINC01565, EPS15L1, LOC101927156, CCDC26, LOC105376219, PSMD3, HCG22, PSMD3.

You are less likely to suffer abnormal levels.

Aortic root size

Echocardiographic measures of left ventricular (LV) structure and function are heritable phenotypes of cardiovascular disease.

Genes analyzed: SLC35F1, TMEM232, SMG6, PRDM6, HMGA2, LOC100506393, LOXL1.

You are less likely to suffer abnormal levels.

RECOMMENDATIONS

- Avoid heavy alcohol intake and smoking.
- Do not consume salt-cured foods such as fish and preserved vegetables, and especially avoid heating them before consumption.
- If you present any symptoms related with celiac disease (such as fatigue, bloating, nausea, constipation, itchy skin, mouth ulcers, headaches, joint pain) try following a gluten-free diet.
- Follow a diet rich in calcium or take supplements.
- Supplement your diet with an extra source of Omega 3 fatty acids.
- Consult the results of your Weight and Fitness management plan and follow the recommendations regarding food supplementation. The right diet with the vitamins and minerals that are suited for your unique metabolism helps you to prevent disease.
- Get vaccination against Hepatitis A and B.
- Consult your Pharmacogenomics genetic results to avoid taking wrong medication that could cause toxicity.

Any changes you want to make to your diet or health treatment should be discussed with a health professional.