Codigo Prevention

Hello, Users!

Welcome to your genetic code!

YOUR DNA PROFILING FOR PREVENTION OF DISEASES

We present to you the results of your DNA profiling for disease prevention according to the genotyping test. The following report contains information about the genetic variants that we found in your genome and have been associated, according to medical and scientific studies, with a potential increased risk of an individual to develop specific diseases throughout his/her life.

In this report we have calculated a parameter for your predisposition or risk to develop some diseases according to your genotype. Our quantitative analysis includes markers for Alzheimer's disease, Parkinson's disease, diabetes, breast cancer, ovarian cancer, prostate cancer, some common metabolic diseases such as: congenital hypothyroidism, cystic fibrosis, tyrosinemia, etc.

You must take into account that the results of this genetic analysis do not contain information for all the genetic variants known in the human genome. This is due to the ongoing discovery of new variants associated with specific diseases in research studies under development.

In this study we analyzed about 600,000 genetic variants distributed among your 23 pairs of chromosomes. Our genetic test mainly analyzes single nucleotide variants (SNVs) and some small deletions and insertions (INDELs) in your genomic DNA. Due to the number and the genomic distribution of the variants analyzed by Código 46, this study is useful to know, in general terms, a large number of diseases associated with these markers. This report includes information of variants classified as pathogenic or high risk in the database of clinical variants (ClinVar) of the National Center for Biotechnology (NCBI) of the United States of America.

It is our responsibility to inform you that even when you have a predisposition or risk to certain diseases associated with some genetic markers, it does not mean that you are going to develop a disease. There are other non-genetic factors such as: nutrition, lifestyle, environment or stress, etc., which may be related to the development of the described health conditions.

Below you will find a section entitled How to read your report. This section will serve you as a guide to read the sections of the document and the information that each one contains. At the end of this report you will find two other sections; the first one specifies the limitations of the test and the second one contains a glossary that you or your doctor can use as a complement to read the information contained in the test.

HOW TO READ YOUR PREVENTION REPORT

The disease prevention report contains several sections, each corresponding to different health conditions for which associations were found with the variants studied in your genome. In turn, each of the sections is divided into additional parts.

There is a brief description of the disease studied at the beginning of each section for each disease. After this brief description, a summary table appears with data associated with your genotype and the general characteristics of the variants detected in your genome; the affected gene, the identifier of the variant (dbSNP), your personal genotype for the variant in the two chromosomes, the number of the chromosome in which the variant is located, the position of the variant within the chromosome and the state of the variant analyzed in your two chromosomes. If they are in a homozygous or heterozygous state.

With the information from you allelic variants, we can approximate the risk grade you have to develop the reported diseases, with two distincts parameters. The first, of these parameters, is known as Lifetime Risk or LTR, while the second is known as Disease Risk Score or DRS.

LTR parameter is based on statistic data at population level and include the multiple factors from the disease can occur. Within these factors, your LTR score indicates the incidence you have, throughout your lifetime, within the number of inhabitants with a risk profile similar to yours.

DRS parameter is based on comprehensive studies that associate the effect level of an observed allelic variant in a particular trait. Some genetic variants would have either a major or minor effect in comparison. With such studies, it is possible to approximate an overall genetic score regarding all the variants found in your genotype. Therefore, your DRS indicates the odd risk because of the overall quantitative effect of all the variants found in your genotype.

To ease the interpretation of these parameters, we have harmonized the parameter units to help you understand the values of risk in your genetic context (DRS) and in the epidemiological context (LTR) respect to the overall population .

LTR interpretation scale is expressed in terms of a given population segmentation. If your LTR score is, for example, 1 out of 15 for a given disease, it means that the risk of developing a disease is observed in 1 person from a subset of 15, with a similar population profile. The smaller the LTR is, the greater the risk of incise in the trait of the profile.

DRS score scale is expressed in terms of a base-2 exponential function. So, whether your risk score is, for example, 1.0 for a given trait, it means that your genetic risk is 2 times more likely to suffer such disease. When your risk score is negative, it implies that your genetic risk score is lesser, in terms of the same value (If it is -1.0, you have 2 times lesser risk). When your risk score si zero, it means that your genetic risk does not increase nor decrease the risk of suffer the given disease. In order to understand this risk score, we help you to visualize it graphically

After the summary table, a qualitative description of the pathogenic variants that we detect in your genotype appears. We consider it important to give you more details about these variants because for these a direct association with the development of the disease has been found and reported. After part, it is displayed a section that contains a list of the molecular and physiological functions of the proteins encoded by the genes affected by your genetic variants. If your genetic test reveals the existence of more than one disease variant associated with the same gene or multiple genes, these will be listed as well in a list. There are times when a particular variant can cause more than one disease. If this is the case, the additional health conditions associated with the variant will be described in the form of a list in a dedicated section. After the sections that describe the variants, genes and other diseases associated with the variants, another section appears with additional details of the risk parameters you have for the disease. That is, you will find a broader description of the LTR for people of your age in the population and the DRS that we calculated given your genetic variants analyzed.

We recommend that you go with a medical professional if you have any risk for any condition reported here, if any member of your family has any of the diseases analyzed or if you have any questions about the results of your genetic test. It is important that you be more careful with your health if any of the risk variants analyzed were detected in your genotype. Keep in mind that not having any of the risk variants analyzed does not mean that the disease under study will not be developed. There are other genetic risk markers that are not included in this analysis and continue in research. Take into account that other non-genetic factors can also affect your risk of developing a disease.

This genetic analysis can not determine with total certainty if you are going to present a health condition in the future. This study offers you a probabilistic estimate of presenting a disease given the population risk of the disease and a set of variants that we analyze in your genotype. This study offers you an overview of your health given certain genetic variants of risk; however, it should not be used to make decisions without medical consultation.

DISEASE PREVENTION REPORT

Methylmalonic aciduria

Methylmalonic academia (MMA) consists of a group of genetic disorders of autosomal recessive inheritance, caused by mutations of the MMUT gene, which lead to a partial or complete deficiency of activity of the methylmalonyl-CoA mutase (MCM). MCM is an enzyme that is involved in the degradation pathway of certain amino acids (building blocks of proteins) and fatty acids. It also plays a role in the metabolism of vitamin B12 and its presence is needed for this vitamin to function properly. The deficiency of MCM causes an accumulation of organic acids and other toxic substances in the blood, increasing its acidity. Children with MMA may not show signs or symptoms at birth and posteriorly deteriorate showing poor feeding, vomiting, abdominal distension, alterations in blood glucose levels, anemia, drowsiness, kidney failure, coma, and even death. Some cases have a late onset, in early adulthood and the symptoms appear after situations of metabolic stress like infections, traumatisms or surgery. Treatment is based fundamentally on a low protein diet and taking vitamin B12 supplements.

Código 46 looks for pathogenic variants in your genome, these variants are directly related to the development of such disease. When the number of pathogenic variants detected is 0, it means that you do not have mutations that have already been clinically reported in patients who develop that disease. On the other hand, for some diseases, DRS is also calculated, which serves to compare a profile of variants with respect to populations, whether healthy or sick. Therefore, the number of pathogenic variants and the DRS are independent measurements.

Abstract **Pathogenic variants** **Tested** 29 **Detected** 2

Gene	Variant	Genotype	Chromosome	Position	Condition
MUT	rs398123276	T/C	6	49415500	Heterozygous
ACSF3	rs387907119	G/A	16	89211720	Heterozygous

Characteristics of the genetic variants detected

rs398123276

The variant rs398123276 was detected in a heterozygous state (genotype T/C) in your genome. This variant is located in the position 49415500 of chromosome 6 in an exonic region of the MUT gene. It is known that the variant causes a change in the coding of the protein and this probably alters its folding and physiological function.

rs387907119

The variant rs387907119 was detected in a heterozygous state (genotype G/A) in your genome. This variant is located in the position 89211720 of chromosome 16 in an exonic region of the ACSF3 gene. It is known that the variant causes a change in the coding of the protein and this probably alters its folding and physiological function. The genetic mutation specifically alters the amino acid at position 471 the protein, causing a change from arginine to glutamine (R471Q).

Characteristics and functions of the affected genes

MUT

Methylmalonyl-CoA mutase

The MUT gene is located on chromosome 6. The protein encoded by this gene is a mitochondrial enzyme called methylmalonyl-CoA mutase (MUT). This enzyme is involved in the degradation of amino acids, specifically; isoleucine, methionine, threonine and valine, certain types of lipids and cholesterol. To carry out the decomposition, the enzyme catalyses the molecules in methylmalonyl-CoA, subsequently and in conjunction with adenosylcobalamin (AdoCbI), catalyzes the isomerization of methylmalonyl-CoA to succinyl-CoA. Once succinyl-CoA is formed, other enzymes break it down into molecules that will then be used as energy. Mutations or deficiencies of the protein can cause mut-methylmalonic aciduria (MMAM), characterized by an often deadly disorder of the metabolism of organic acids. Common clinical features include lethargy, vomiting, growth retardation, hypotonia, neurological deficit and premature death.

ACSF3

Acyl-CoA synthetase family member 3, mitochondrial

combined malonic and methylmalonic aciduria

Other pathologies related to the variants found

Scientific reports have shown that the variant rs398123276 is associated with the occurrence of methylmalonic aciduria due to methylmalonyl-coa mutase deficiency. This disease is characterized by having autosomal recessive inheritance pattern.

Scientific reports have shown that the variant rs387907119 is associated with the

occurrence of null. This disease is characterized by having autosomal recessive inheritance pattern.

Tyrosinemia

Tyrosinemia is an autosomal recessive disorder in which the body cannot break down an amino acid called tyrosine. This condition causes a harmful accumulation of tyrosine, other amino acids and toxins in the body which cause serious health problems. The signs and symptoms of untreated tyrosinemia usually begin during childhood and include diarrhea, vomiting, swollen abdomen, insufficient weight gain, lethargy, irritability, smell similar to cabbage, bleeding problems, respiratory problems and developmental delays. If this disease is not treated, liver and kidney failure may occur, as well as nervous system problems. Babies with tyrosinemia need dietary and medical treatment for life. Early treatment can help prevent liver, kidney and brain problems. Children who receive treatment early in life can often have normal growth and development.

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Abstract	Pathogenic variants		Tested 5		Detected 1
Gene	Variant	Genotype	Chromosome	Position	Condition
FAH	rs80338901		15	80472572	

Characteristics of the genetic variants detected

rs80338901

Characteristics and functions of the affected genes

FAH

Fumarylacetoacetase

The FAH gene is encoded on chromosome 15. The protein encoded by this gene is Fumarylacetoacetase. The FAH gene provides instructions for producing an enzyme

called fumarateacetoacetate hydrolase. This enzyme is abundant in the liver, kidneys and there are smaller amounts that are found in many tissues throughout the body. Fumarylacetoacetate hydrolase is the latest in a series of five enzymes that work to break down the amino acid tyrosine. Specifically, fumarylacetoacetate hydrolase converts a by-product of tyrosine called acetoacetic fumarate into smaller molecules that are excreted by the kidneys or used to produce energy or produce other substances in the body. Several mutations of FAH can cause type I tyrosinemia. This condition is characterized by severe liver and kidney disease, neurological problems, and other signs and symptoms that begin in childhood. The altered FAH gene causing this condition produces an unstable or inactive enzyme, which results in reduced or absent fumarateacetoacetate hydrolase activity. The most common FAH mutation interrupts the way in which the instructions of the gene are used to produce the enzyme.

Other pathologies related to the variants found

Scientific reports have shown that the variant rs80338901 is associated with the occurrence of hypertyrosinemia. This disease is characterized by having autosomal recessive inheritance pattern.

Diabetes

Diabetes mellitus (DM) is a group of metabolic disorders, whose main characteristic is the presence of high concentrations of glucose in the blood persistently or chronically, due to either a defect in the production of insulin, a resistance to the action of it to use glucose, an increase in the production of glucose or a combination of these causes. It is also accompanied by abnormalities in the metabolism of lipids, proteins, mineral salts and electrolytes.

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Abstract Pathogenic variants Tested 20 Detected 0

Risk parameters LTR 1/0 DRS 0.0

Your Diabetes predisposition

Although we did not detect pathogenic variants, due to set of variants that we detect in your genotype, your risk of having Diabetes is equals than a person randomly chosen from open population.

The lifetime risk (LTR) that a common person has of developing Diabetes is 1/0.

Parkinson's disease

Parkinson's disease (PD), also called Parkinson's disease, idiopathic parkinsonism, agitation paralysis or simply Parkinson's disease, is a chronic neurodegenerative disease characterized by slow movement, stiffness, increased muscle tone and tremor. cognitive function, depression, pain and alterations in the function of the autonomic nervous system. Parkinson's disease increases its severity over time, as a consequence of the progressive destruction, due to unknown causes, of the pigmented neurons of the substantia nigra.

Código 46 looks for pathogenic variants in your genome, these variants are directly related to the development of such disease. When the number of pathogenic variants detected is 0, it means that you do not have mutations that have already been clinically reported in patients who develop that disease. On the other hand, for some diseases, DRS is also calculated, which serves to compare a profile of variants with respect to populations, whether healthy or sick. Therefore, the number of pathogenic variants and the DRS are independent measurements.

Abstract Pathogenic variants Tested 8 Detected 0

Risk parameters LTR **1/0** DRS **0.0**

Your Parkinson's disease predisposition

Although we did not detect pathogenic variants, due to set of variants that we detect in your genotype, your risk of having Parkinson's disease is equals than a person randomly chosen from open population.

The lifetime risk (LTR) that a common person has of developing Parkinson's disease is 1/0.

Prostate cancer

Prostate cancer refers to the disease that develops in the prostate, one of the glandular organs of the male reproductive system. Cancer occurs when some prostate cells mutate and begin to multiply uncontrollably. These could also spread from the prostate to other parts of the body, especially the bones and lymph nodes causing a metastasis. This

condition can cause pain, difficult urination, erectile dysfunction, among other symptoms.

Código 46 looks for pathogenic variants in your genome, these variants are directly related to the development of such disease. When the number of pathogenic variants detected is 0, it means that you do not have mutations that have already been clinically reported in patients who develop that disease. On the other hand, for some diseases, DRS is also calculated, which serves to compare a profile of variants with respect to populations, whether healthy or sick. Therefore, the number of pathogenic variants and the DRS are independent measurements.

Abstract Pathogenic variants Tested 4 Detected 0

Risk parameters LTR 1/0 DRS 0.0

Your Prostate cancer predisposition

Although we did not detect pathogenic variants, due to set of variants that we detect in your genotype, your risk of having Prostate cancer is equals than a person randomly chosen from open population.

The lifetime risk (LTR) that a common person has of developing Prostate cancer is 1/0.

HEALTH CONDITIONS FOR WHICH YOU DO NOT HAVE PATHOGENIC VARIANTS IN YOUR GENOME

Your genetic test included variants for which you have not shown pathogenic variants. Here the list of conditions for which you don't have variants:

- Alzheimer Disease
- Sickle cell anemia
- Breast cancer
- Hereditary breast and ovarian cancer
- Ovarian cancer
- Prosthate cancer
- Kidney carcinoma
- Cardiomyopaties
- Biotinidase deficiency
- Glucose 6 phosphate dehydrogenase deficiency
- 3 Methylcrotonyl-CoA carboxylase 1 deficiency
- Maple syrup urine disease
- Cystic fibrosis
- Congenital adrenal hyperplasia
- Congenital hypothyroidism
- Parkinson Disease

Disease	Gen	Variants analyzed
3 methylcrotonyl-coa carboxylase 1 deficiency	MCCC1	rs119103213, rs185741664, rs544349961, rs727504005, rs727504006
Alzheimer's disease	PSEN1	rs63750082, rs63750687
Biotinidase deficiency	BTD	rs104893686, rs104893687, rs104893688, rs138818907, rs146015592, rs181396238, rs397514371, rs397514380, rs397514392, rs587783005, rs80338686
Breast cancer	BRCA2	rs397507384, rs786202461, rs80358721, rs80359175
	BRCA1	rs398122661, rs730881468, rs786203754, rs80356888, rs80356913, rs80356936, rs80356937, rs80357063, rs80357233, rs80357295, rs80357463, rs80357475
	PRLR	rs72478580
Cystic fibrosis	CFTR	rs113993959, rs121908748, rs121908749, rs121908750, rs121908751, rs121908752, rs121908754, rs121908755, rs121908760, rs121908763, rs121908764, rs121908765, rs121908791, rs121908792, rs121908793, rs121908794, rs121908797, rs121908803, rs121908810, rs121909011, rs121909012, rs121909015, rs121909017, rs121909019, rs121909025, rs121909026, rs121909036, rs121909045, rs121909047, rs139304906, rs139573311, rs141158996, rs143570767, rs149790377, rs151048781, rs193922503, rs201124247, rs267606722, rs368505753, rs372227120, rs374705585, rs374946172, rs387906362, rs387906369, rs397508168, rs397508173, rs397508176, rs397508200, rs397508201, rs397508211, rs397508227, rs397508243, rs397508247, rs397508249, rs397508263, rs397508267, rs397508279, rs397508296, rs397508328, rs397508331, rs397508336, rs397508350, rs397508378, rs397508387, rs397508393, rs397508394, rs397508435, rs397508442, rs397508453, rs397508464, rs397508470, rs397508532, rs397508536, rs397508538, rs397508596, rs397508604, rs397508675, rs397508684, rs397508701, rs397508702, rs397508746, rs397508759, rs397508761, rs397508778, rs397508796, rs397508799, rs74467662, rs74551128, rs74597325, rs74767530, rs75039782, rs75096551, rs75115087, rs75389940, rs75527207, rs755416052, rs75549581, rs75961395, rs76554633, rs76649725, rs76713772, rs77010898, rs77101217, rs77188391, rs77284892, rs77409459, rs77646904, rs77902683, rs78194216, rs78440224, rs78655421, rs78756941, rs78802634, rs79031340, rs79633941, rs79660178, rs797045160, rs79850223, rs80034486, rs80055610
Diabetes	WFS1	rs104893879, rs28937891, rs28937892

	AQP2	rs104894331, rs104894338, rs28931580
	PAX4	rs114202595
	SLC19A2	rs121908540, rs74315374
	INSR	rs121913148
	HNF1B	rs121918671, rs121918673
	DLD	rs121964990
	GCGR	rs1801483
	GCK	rs193922335, rs193922338, rs794727236
	ZFP57	rs77625743
	ABCC8	rs797045209
	INS	rs80356669
Glucose 6 phosphate dehydrogenase deficiency	G6PD	rs398123546, rs398123552, rs76723693, rs78365220
Hereditary breast and ovarian cancer	BRCA1	rs398122661, rs786203754, rs80356888, rs80356913, rs80356936, rs80356937, rs80357063, rs80357233, rs80357295, rs80357463, rs80357475
	BRIP1	rs587780875
	BRCA2	rs786202461, rs80358721, rs80359175
Kidney carcinoma	OGG1	rs104893751
Maple syrup urine disease	DLD	rs121964988, rs121964990
	BCKDHB	rs121965004, rs371518124, rs398124561, rs398124574, rs398124577, rs398124582, rs398124589, rs398124592, rs398124593, rs398124602
	BCKDHA	rs137852871, rs137852874, rs137852875, rs182923857, rs375785084, rs398123490, rs398123491, rs398123496, rs398123497, rs398123503, rs398123508, rs398123509, rs398123513
	DBT	rs398123660, rs398123665, rs398123669, rs398123674, rs398123675, rs794727262, rs794727635
Methylmalonic aciduria	MMAA	rs104893851, rs571038432
	SUCLA2	rs121908538
	MMACHC	rs121918240, rs121918241, rs121918242, rs370596113, rs398124295
	MUT	rs121918251, rs121918252, rs121918254, rs121918255, rs121918256, rs121918257, rs398123276, rs398123278, rs564069299, rs727504020, rs727504022, rs760782399

	ACSF3	rs138680796, rs140986055, rs387907119
	ABCD4	rs201777056
	SUCLG1	rs267607097
	MMAB	rs28941784, rs369296618, rs398124434, rs756414548
Parkinson's disease	PLA2G6	rs121908686, rs121908687
	PRKN	rs137853058, rs137853060
	PINK1	rs28940285, rs74315359
	FBXO7	rs71799110
	PARK7	rs74315352
Prostate cancer	MAD1L1	rs121908982
	AR	rs137852571, rs137852593
	POLK	rs148960463
Tyrosinemia	FAH	rs121965073, rs370686447, rs80338894, rs80338895, rs80338901

TEST LIMITATIONS

The interpretation of the results of the tests carried out by Código 46 is limited by the information currently available. A more extensive interpretation may be possible in the future as more data and knowledge about human genetics and the health conditions accumulate.

When the genotyping does not reveal any difference with respect to the reference sequence, or when a variant is in a homozygous state, it cannot be certain that both alleles of an individual could be detected, this is a limitation of any platform of microarray genotyping.

Occasionally, an individual may carry an allele that is not amplified and detected due to a large deletion or insertion in its genome; in these cases, the marker can not be detected by our technology and therefore, this report does not contain information about this kind of alleles. Our tests do not detect copy number variants (CNV).

We evaluate single nucleotide variants (SNVs) in different coding exons for each gene included in our array. Unless specifically indicated, the report do not contain information on other genomic regions that have not been characterized. Unless otherwise indicated, the DNA sequence data is obtained from a specific cell type (from the sample of epithelial tissue collected by our kit). The results of this report do not contain information about the DNA sequence in other types of cells, tissues or organs. Because of this, our ability to detect variants due to somatic mosaicism is limited.

For this test we use the following reference genome (Genome build hg19, version GRCh37), a reinterpretation of your data under any other version of the human genome may differ from the results shown here.

We trust in our ability to track a sample once it has been received by Código 46. However, we are not responsible for any sample labeling error that occurs before the sample reaches Código 46.

These results should be used in the context of available clinical findings, and should not be used as the sole basis for treatment. This test was developed and its performance characteristics were determined by Código 46, which is certified under the ISO 9001: 2015 standard to perform highly complex clinical laboratory tests.

We recommend genetic counseling to help explain the results and analyze options for replication.

GLOSSARY

Allele
We have two copies of the same gene provided by each of our parents. An allele is the reference to each copy of the gene. That is, an individual inherits two alleles for each gene, one from the father and the other from the mother.

Autosomal Inheritance (Dominant or Recessive)
It is a pattern of inheritance in which the transmission of characteristics depends on the presence or absence of certain alleles. 'Autosomal' means that the gene in question is located on one of the non-sexual chromosomes (i.e., chromosome number 1 to 22). 'Dominant' means that a single copy of the mutation related to a disease is sufficient to cause the disease. On the contrary, a 'Recessive' character requires that both copies of the gene in question be altered, or mutated, in order for the disease to occur.

Chromosome
A chromosome is an ordered package of DNA found in the nucleus of the cell. Humans have 23 pairs of chromosomes - 22 autosomal pairs, and one pair of sex chromosomes, X and Y. Each parent contributes one chromosome of their pair so that the children get half of their chromosomes from their mother and half of their chromosomes from his father.

CNV
The copy number variation or CNV is when a section of the genome is repeated or absent. This is because the genome experiences gains and losses of genetic material.

DNA
The Deoxyribonucleic acid is a macromolecule that encodes the genes of cells, bacteria and some viruses. The DNA is the hereditary material of living beings and this information is used to make the proteins necessary for its development and functioning, as well as to regulate other functions of the organism. It is formed by a long sequence of nucleotides. Each nucleotide contains one of the 4 nitrogenous bases of DNA: adenine (A), thymine (T), cytosine (C) and guanine (G).

Exon
An exon is an element of the gene that contains the instructions for encoding amino acids.

Gene
The gene is the basic functional unit of inheritance. Genes are transmitted from parents to children and contain the necessary information to determine their traits. They are arranged in the chromosomes. Humans have approximately 20,000 genes organized into their chromosomes, representing ~ 1% of all the genetic information contained in each cell.

Genome
All the genetic information that is stored in the nucleus of each cell.

Genotype
A genotype is the collection of genes of an individual. The term can also refer to the two alleles inherited for a particular gene.

Genotyping / Genotyping
It is the procedure by which the genetic variants of an individual are determined. Their analysis allows to know the alleles that an individual has inherited from their biological parents.

Homozygous
If the two alleles are identical, i.e., when your genotype for the detected variant is identical in both copies of the gene.

Heterozygous
If the two alleles are different, i.e., your genotype for the detected variant is different in both copies of the gene.

Pathogenic variant
A genetic variant related with producing some type of disease.

Phenotype
The observable traits of an individual by the expression of the genotype, such as height, eye color, blood group and disease.

SNP
Single nucleotide polymorphisms are a type of change that occurs by a variation in a single DNA base pair. SNPs have been linked to health conditions, drug response and other phenotypes, this is what we explore and report in your genetic test.