



# Health Report

OPTIMAL HEALTH FOR LIFE



**Your Lab**

Your Logo

**GENOTYPE  
REPORT**

**NAME**

Sam Slim

**DATE OF BIRTH**

9/9/1990

**REFERRING PRACTITIONER**

**DATE REPORTED**

11/1/2017 4:27:03 PM

**ACCESSION NUMBER**

AJ-HealthSP

# WELCOME TO YOUR DNA HEALTH REPORT

From your buccal swab sample, we use a process called the Polymerase Chain Reaction (PCR), which copies the DNA of your genes many times over so that we can generate sufficient quantities to analyze your genetic material. Then, we identify unique DNA sequences in some of your genes. Certain changes (polymorphisms) in these genes have been studied in detail, with evidence that correlates these polymorphisms with an individual's risk of developing certain chronic disease conditions or altered metabolic processes. Having identified the presence or absence of these polymorphisms, we are able to qualitatively assess particular areas of health risk related to the specific genes.

To make a holistic assessment of health risks, environmental factors (diet and lifestyle) need to be considered in conjunction with the accompanying genetic profile.



## HOW TO READ YOUR RESULTS

You will find your genetic results in the following pages. On the left side, you will see the gene name and description. On the right side, you will find your specific result and an explanation of the results, associated risks, and diet and lifestyle recommendations. Please see the key above to identify each impact level.

### NO IMPACT

Genotype has no effects on the biological area in question.

### LOW IMPACT

Genotype has mild effects on the biological area in question with a small change in responsiveness to environmental influences.

### MODERATE IMPACT

Genotype has moderate effects on the biological area in question. Attention should be paid, and some dietary and lifestyle changes are recommended.

### HIGH IMPACT





Genotype has significant impact on the biological area in question. Cohesive and intensive diet and lifestyle action should be taken.

### BENEFICIAL IMPACT

Genotype result is advantageous to health.

## BONE HEALTH

Our bones are not a fixed structure. Our cells work continuously to dissolve old bone and create new bone tissue. After the age of 30, both men and women start losing bone mass; the loss is particularly marked in women after menopause. According to the latest research, both nutrition and genetic factors play an important role in determining bone health.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Bone Health	COL1A1	1546G>T	C/C	
	VDR	Fok1 T>C	G/A	
		Taq1 C>T	T/C	
		Bsm1 G>A	G/A	

## COL1A1 | 1546G>T



Type 1 Collagen is the major protein of bone and is formed from two collagen alpha 1- and one collagen alpha 2 chains.

### YOUR RESULT: C/C

No genetic variation was detected at the 1546 G>T (C>A) locus.

## VDR | Fok1 T>C



Peak bone mass is to a great extent genetically determined. The vitamin D receptor (VDR) gene accounts for around 70% of the entire genetic influence on bone density, playing an important role in calcium homeostasis, bone cell growth and differentiation, and intestinal calcium absorption.

### YOUR RESULT: G/A

The T (A) allele has poorer calcium absorption compared to the C (G) allele.

## VDR | Taq1 C>T



Peak bone mass is, to a great extent, genetically determined. The vitamin D receptor (VDR) gene accounts for around 70% of the entire genetic influence on bone density, playing an important role in calcium homeostasis, bone cell growth and differentiation, and intestinal calcium absorption.

### YOUR RESULT: T/C

The variation does not lead to an increased risk for osteoporosis.

## VDR | Bsm1 G>A



Peak bone mass is, to a great extent, genetically determined. The vitamin D receptor (VDR) gene accounts for around 70% of the entire genetic influence on bone density, playing an important role in calcium homeostasis, bone cell growth and differentiation, and intestinal calcium absorption.

### YOUR RESULT: G/A

The T (A) allele is associated with reduced bone mineral density in a dose-dependent manner and predisposes carriers to osteoporosis, especially when calcium intake is low.

## DETOXIFICATION

The detoxification process in the body is governed primarily by the GST family of enzymes. Glutathione S-transferases are responsible for catalyzing reactions in which the products of Phase I metabolism are conjugated with glutathione, thus making them more water soluble and more easily excreted from the body through sweat and urine. Cruciferous and allium vegetables help increase the activity of your detoxification system, which aids the removal of harmful substances from your body.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Detoxification	CYP1A1	Msp1 T>C	T/C	
		2454A>G (Ile462Val)	T/T	
	GSTM1	519G>C	G/G	
	GSTP1	313A>G	A/A	
	GSTT1	15G>C	G/G	
	NQO1	609C>T	C/C	

## Phase I Detoxification

### CYP1A1 | Msp1 T>C



The CYP1A1 gene encodes a phase I cytochrome P450 enzyme that converts environmental procarcinogens such as polycyclic aromatic hydrocarbons (PAHs) and aromatic amines to reactive intermediates having carcinogenic effects. In addition, CYP1A1 is involved in the oxidative metabolism of estrogens, which may play a critical role in the etiology of breast and prostate cancers.

#### YOUR RESULT: T/C

The variant allele C is associated with increased enzyme activity resulting in elevated levels of activated metabolites and subsequent DNA damage. Many association studies have reported that this SNP increases the development of several types of cancers, especially amongst smokers. In the presence of the C allele, it is important to reduce exposure to all diet and environmental procarcinogens such as PAHs, aromatic amines, nitrates, and smoking of any kind. In addition, attention should be paid to optimizing phase 2 detoxification.

### CYP1A1 | 2454A>G (Ile462Val)



The CYP1A1 gene encodes a phase I cytochrome P450 enzyme that converts environmental procarcinogens such as polycyclic aromatic hydrocarbons (PAHs) and aromatic amines to reactive intermediates having carcinogenic effects. In addition, CYP1A1 is involved in the oxidative metabolism of estrogens, which may play a critical role in the etiology of breast and prostate cancers.

#### YOUR RESULT: T/T

No variant was detected.

### NQO1 | 609C>T



NADP(H:) quinone oxidoreductase 1 (NQO1) often referred to as Quinone Reductase is primarily involved in the detoxification of potentially mutagenic and carcinogenic quinones derived from tobacco smoke, diet and estrogen metabolism. NQO1 also protects cells from oxidative stress by maintaining the antioxidant forms of ubiquinone and vitamin E.

#### YOUR RESULT: C/C

The analysis identified no genetic variation at the 209 C>T locus.

## Phase II Detoxification

### GSTP1 | 313A>G



Glutathione S-transferase P1 is a member of the GST super-family and is involved in phase II detoxification by conjugating xenobiotics to glutathione and thereby detoxifying cellular environments. Oxidative stress is a risk factor shared by most disorders implicating GST, and it appears that the efficiency of the GSTP1 enzyme may have an impact on the development and prognosis of diseases influenced by oxidative stress. GSTP1 is the most abundant GST subtype in the lungs and is known to metabolize many carcinogenic compounds.

#### YOUR RESULT: **A/A**

No variant was detected.

### GSTM1 | 519G>C



Glutathione S-transferase M1 is the most biologically active member of the GST super-family and is involved in Phase II detoxification in the liver. It is responsible for the removal of xenobiotics, carcinogens, and products of oxidative stress.

#### YOUR RESULT: **G/G**

The absence of a genotype at the GSTM1 519G>C indicates that the gene is deleted.

### GSTT1 | 15G>C



Glutathione S-transferase theta-1 (GSTT1) is a member of a super family of proteins that catalyzes the conjugation of reduced glutathione to a variety of electrophilic and hydrophobic compounds.







#### YOUR RESULT: **G/G**

This genotype indicates the presence of the GSTT1 gene.



## FOOD RESPONSIVENESS

Particular nutrients and certain food components in different foodstuffs can affect individuals in different ways. With new research coming to light in this area, specific genes can be tested to give more insight into how an individual might respond to a particular food component. The areas of food responsiveness covered in this panel include: lactose intolerance, Polyunsaturated fat (PUFA) metabolism, caffeine sensitivity, salt sensitivity and iron overload.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Food Responsiveness	ACE	I/D	G/C	
	AGT	Met235Thr	A/G	
	CYP1A2	-163C>A	A/A	
	HFE	H63D; C282Y	282CC/63HD	
	MCM6	-13910C>T	C/T	
	MYRF/FADS1	-592G>T	G/G	

## Iron overload

**HFE** | H63D; C282Y



Hereditary hemochromatosis is a genetic disorder in which there is excessive accumulation of iron in the body, leading to iron overload. For individuals with this disorder, the daily absorption of iron from the intestines is greater than the amount needed to replace losses. Since the normal body cannot increase iron excretion, the absorbed iron accumulates in the body. Individuals who carry the genes for hereditary hemochromatosis may have no symptoms or signs and the disease is treatable, if detected early. Severe symptoms and signs of iron overload include sexual dysfunction, heart failure, joint pains, liver cirrhosis, diabetes mellitus, fatigue, and hypermelanotic pigmentation.

### YOUR RESULT: 282CC/63HD

The analysis detected no genetic variation increasing risk for the disorder.

## Caffeine sensitivity

**CYP1A2** | -163C>A



Coffee is a major source of caffeine, which is metabolized by the cytochrome P450 1A2 enzyme (CYP1A2).

### YOUR RESULT: A/A

Individuals with the AA genotype are fast metabolizers of caffeine.

## Salt Sensitivity

**ACE** | I/D



ACE codes for the angiotensin-converting enzyme and is part of the renin-angiotensin system, which controls blood pressure by regulating the volume of fluids in the body.

### YOUR RESULT: G/C

Studies show that patients with essential hypertension homozygous for the insertion allele of the ACE gene have a significantly higher blood pressure increase with high salt intake compared to DD (GG) individuals.

**AGT** | Met235Thr



Angiotensinogen is expressed in tissues involved in blood pressure regulation such as the kidneys, adrenals and brain. Increased angiotensinogen levels correlate with increased blood pressure. The gene also influences salt sensitivity of blood pressure.

### YOUR RESULT: A/G

Individuals who carry the C (G) allele are associated with increased risk for hypertension. However, incidence of hypertension was found to be significantly lower among these individuals who reduced sodium intake.



## Lactose intolerance

**MCM6** | -13910C>T



A specific DNA sequence within the MCM6 gene called a regulatory element helps control the activity of a nearby gene called LCT gene which encodes an enzyme called lactase. This enzyme helps to digest lactose, a sugar found in milk and other dairy products. In most individuals, the expression of LCT gene decreases after infancy leading to lactase deficiency. After ingestion of milk or other dairy products, these individuals may experience abdominal cramps, bloating, distension, flatulence and diarrhea.

### YOUR RESULT: C/T

The CT genotype is associated with lactase persistence in the Caucasian population.

## Polyunsaturated fatty acids (PUFA) metabolism

**MYRF/FADS1** | -592G>T





The delta 5 and delta 6 desaturases, encoded by FADS1 and FADS2 genes, are key enzymes in polyunsaturated fatty acid (PUFA) metabolism that catalyze the conversion of linoleic acid (LA) into arachidonic acid (AA) and that of alpha-linolenic acid (ALA) into eicosapentaenoic acid (EPA). SNPs in the FADS locus have been associated with blood concentrations of long-chain PUFAs as well as with cholesterol concentrations. Based on genetic variation, individuals may require different amounts of dietary PUFAs or LC-PUFAs to achieve comparable biological effects.

### YOUR RESULT: G/G

The G allele is associated with enhanced conversion of DGLA to AA due to increased enzymatic efficiency, and thus appears to be associated with higher levels of AA, systemic inflammation and inflammatory disorders.

## INFLAMMATION HEALTH

Inflammation is a normal immune response and an essential step in tissue healing. The release of these inflammatory substances is controlled by genes that govern inflammation. However, when these genes are not switched off, the inflammatory response continues. An increasing number of common disorders, such as obesity, heart disease, arthritis and inflammatory bowel disease have been associated with chronic low-grade inflammation.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Inflammation Health	IL6	-174G>C	G/G	
	TNF	-308G>A	G/G	

### IL6 | -174G>C



Interleukin 6 is a pro-inflammatory cytokine that plays a crucial role in inflammation and regulates expression of C-reactive protein (CRP). Low-grade chronic inflammation is associated with obesity and visceral fat deposition, insulin resistance, dyslipidaemia and increased risk of cardiovascular disease.

#### YOUR RESULT: **G/G**

No variant was detected at the 174 G>C locus.

### TNF | -308G>A







Tumor necrosis factor- $\alpha$  (TNF $\alpha$ ), a proinflammatory cytokine secreted by both macrophages and adipocytes, has been shown to alter whole body glucose homeostasis, and has been implicated in the development of obesity, obesity-related insulin resistance and dyslipidaemia.

#### YOUR RESULT: **G/G**

No variant was detected at the 308 G>A locus.

## INSULIN SENSITIVITY

Insulin is a hormone that stimulates the uptake of glucose from the diet into the blood. Those with lower sensitivity to insulin have a limited ability to respond to the hormone's action. The scientific literature suggests that insulin insensitivity or resistance may play an important role in some of the most common disorders, including obesity, type 2 diabetes, high blood pressure, heart disease and disrupted fat metabolism. role in determining bone health.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Insulin Sensitivity	FTO	rs9939609 T>A	T/T	
	PPARG	Pro12Ala	C/G	
	SLC2A2	Thr110Ile	C/C	
	TCF7L2	C>T	C/C	

## FTO | rs9939609 T>A



Fat-mass-and-obesity-associated (FTO) gene is present at high levels in several metabolically active tissues, including heart, kidney, and adipose tissue, and is most highly expressed in the brain, particularly in the hypothalamus which is concerned with the regulation of arousal, appetite, temperature, autonomic function, and endocrine systems. It has been suggested that the FTO gene plays a role in appetite regulation and that it is associated with energy expenditure, energy intake, and diminished satiety.

### YOUR RESULT: T/T

No variant was detected.

## PPARG | Pro12Ala



Peroxisome proliferator-activated receptor gamma (PPARG) is believed to be involved in adipocyte differentiation. It is a transcription factor activated by fatty acids, which has a major role in adipogenesis and expression of adipocyte-specific genes. It is also involved in the regulation of glucose and lipid metabolism. This receptor is the target for the thiazolidinedione antidiabetic drugs.

### YOUR RESULT: C/G

The G allele is associated with reduced promoter activation, reduced transcriptional activity and reduced adipocyte differentiation. As a result, the G allele has been associated with lower BMI and fasting insulin, improved insulin sensitivity and reduced risk of insulin resistance and diabetes.

## SLC2A2 | Thr110Ile



GLUT2, coded by the SLC2A2 gene, is a member of the facilitative glucose transport protein (GLUT) family and is expressed in the pancreas, liver, small intestine, kidney, and brain. GLUT2 facilitates the first step in glucose induced insulin secretion, with the entry of glucose into the pancreatic  $\beta$ cell. Due to its low affinity for glucose, it has been suggested as a glucose sensor and is considered to be important in the postprandial state. It is also involved in food intake and regulation.

### YOUR RESULT: C/C

The analysis detected no variant.

## TCF7L2 | C>T



Transcription factor 7-like 2 (TCF7L2) gene encodes a transcription factor that regulates blood glucose homeostasis. This SNP influences both insulin secretion and resistance and has been associated with an increased risk of insulin resistance and type 2 diabetes mellitus.

### YOUR RESULT: C/C

No variant was detected.



## LIPID METABOLISM

Heart health depends on a complex balance of environmental, dietary and genetic factors. Certain genes influence LDL and HDL cholesterol levels; higher levels of LDL, or 'bad' cholesterol, and lower levels of HDL or 'good' cholesterol, are associated with a higher risk of heart disease.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Lipid metabolism	APOC3	3175C > G	G/G	
	APOE	388T>C; 526C>T	ε3/ε2	
	CETP	279G>A	G/A	
	LPL	1595C>G	C/C	

## APOE | 388T>C; 526C>T



Apolipoprotein E has a multi-functional role in lipoprotein metabolism and is essential for the normal catabolism of triglyceride-rich lipoprotein constituents. Two SNPs result in three allelic isoforms, affecting the protein conformation and thus, the receptor binding activity and lipoprotein preference of the APOE protein.

### YOUR RESULT: $\epsilon 3/\epsilon 2$

In general, E2 carriers have lower total cholesterol levels. Studies suggest that the APOE E2 allele may have a slight protective effect against CVD. However, despite lower cholesterol levels, E2 carriers are not immune to dyslipidemia and raised triglycerides. E2 carriers appear to respond less to dietary intervention and are more responsive to statin therapy.

## APOC3 | 3175C > G



Apolipoprotein C3 plays an important role in cholesterol metabolism. It inhibits lipoprotein lipase and hepatic lipase, delaying catabolism of triglyceride-rich particles.

### YOUR RESULT: G/G

The G allele is associated with elevated plasma triacylglycerol, cholesterol, and APOC3 concentrations. Carriers of the G variant have an approximate 4-fold increased risk of hypertriglyceridemia. However, they are responsive to dietary intervention. Decrease saturated fat and increase MUFA. If triglycerides are raised, modify carbohydrates intake. Carriers of the G variant may also show enhanced benefit to statin therapy.

## CETP | 279G>A



Cholesterol ester transfer protein plays a key role in the metabolism of HDL and mediates the exchange of lipids between lipoproteins, resulting in the eventual uptake of cholesterol by hepatocytes (reverse cholesterol transport). High plasma CETP concentration is associated with reduced HDL-C concentrations. CETP is a strong and independent risk factor for CAD.

### YOUR RESULT: G/A

The A allele is associated with reduced plasma CETP levels, increased HDL-C levels and reduced risk of cardiovascular disease. An alpha-linoleic acid (ALA)-enriched, low cholesterol diet is effective in decreasing VLDL-C and LDL-C levels in GA and AA individuals.

## LPL | 1595C>G









Lipoprotein lipase is anchored to the vascular endothelium and removes lipids from the circulation by hydrolyzing triglycerides present in VLDL into free fatty acids. The 1595 C>G variant is a strong indicator of body fat, fat distribution, plasma lipids and insulin concentrations.

### YOUR RESULT: C/C

The analysis identified no genetic variation at the 1595 C>G locus.

## METHYLATION

B vitamins provide building blocks for growing cells, which are constantly being renewed, and play an important role in many physiological processes. B vitamins also supply necessary for protecting our genes, so that our DNA doesn't accumulate damage from the wear and tear in the daily lives of our cells. These vitamins – including folate, vitamins B6 and B12 – help make new DNA for cells that are constantly growing and renewing themselves. Folate is also involved in turning many genes on and off, and also helps repair DNA. The process of DNA repair is called methylation. Although B vitamins are only required in small amounts, they are crucial for methylation and in producing new DNA.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Methylation	CBS	699C>T	G/A	
	COMT	472G>A (Val158Met)	A/G	
	MTHFR	1298A>C	GT	
		677C>T	GA	
	MTR	2576A>G	A/A	
	MTRR	66A>G	A/G	

## MTHFR | 1298A>C



Methylenetetrahydrofolate Reductase (MTHFR) is a key enzyme in the folate metabolism pathway, directing folate from the diet either to DNA synthesis or homocysteine remethylation.

### YOUR RESULT: GT

The C allele is associated with decreased enzyme function. Folate requirements are increased and supplementation of folate, B2, B6 and B12 may be desirable.

## MTHFR | 677C>T



Methylenetetrahydrofolate Reductase is a key enzyme in the folate metabolism pathway, directing folate from the diet either to DNA synthesis or homocysteine remethylation.

### YOUR RESULT: GA

The T allele lowers MTHFR enzyme activity, which results in an increase in homocysteine levels, a decrease in DNA methylation, and thus an increase in DNA adducts. T allele carriers have increased folate, vitamin B2, B6 and B12 requirements. In addition to folate-rich foods, a supplement may be recommended.

## COMT | 472G>A (Val158Met)



Soluble catechol-O-methyltransferase (S-COMT) helps control the levels of certain hormones and is involved in the inactivation of the catecholamine neurotransmitters (e.g. dopamine, epinephrine, and norepinephrine). The enzyme introduces a methyl group to the catecholamine, which is donated by S-adenosyl methionine (SAM). Any compound having a catechol structure, like catechol estrogens and catechol-containing flavonoids, are substrates of COMT.

### YOUR RESULT: A/G

The A allele is associated with a 3-4 fold reduction in the methylation activity of the COMT enzyme and is associated with increased risk for breast cancer. Key interventions for beneficial modulation of estrogen metabolism can be accomplished by increasing insoluble fiber, managing the quality of dietary fat intake, losing weight, and increasing exercise. In addition, ensure sufficient antioxidant and magnesium intake. Dietary components that inhibit COMT activity include quercetin and tea catechins.

## CBS | 699C>T



Cystathionine beta synthase catalyzes the conversion of homocysteine to cystathione and is directly involved in the removal of homocysteine from the methionine cycle. Any alterations in its activity could affect homocysteine levels.

### YOUR RESULT: G/A

The T (A) allele is associated with decreased risk of CAD and an increased responsiveness to the homocysteine-lowering effects of folic acid. Check dietary folate intake and homocysteine levels and supplement if necessary.

## MTR | 2576A>G



MTR gene encodes the Methionine Synthase, an enzyme that catalyzes the remethylation of homocysteine to methionine.

### YOUR RESULT: A/A

No variation was detected at the 25776 A>G locus.







Methionine Synthase Reductase (MTRR) catalyzes methylcobalamin, an essential cofactor of methionine synthase (MTR), which is necessary for maintaining adequate intracellular pools of methionine and maintaining homocysteine concentrations at non-toxic levels.

**YOUR RESULT: A/G**

The G allele is associated with increased risk for premature CAD when cobalamin (vitamin B12) status is low. Ensure adequate intake of folate, vitamin B12 and vitamin B6.

## OXIDATIVE STRESS HEALTH

Free radicals are a normal by-product of the body's energy-generating biochemical processes. They are highly reactive with other molecules, and can damage DNA, proteins and cellular membranes. The balance between oxidation and antioxidation is believed to be critical in maintaining healthy biological systems. Dietary antioxidants such as vitamin C, vitamin E, carotenoids and polyphenols are free radical scavengers that interact with the free radical to ensure it is no longer a reactive molecule, and play an essential role in many antioxidant mechanisms in living organisms. However, the major role in antioxidant defense is fulfilled by the body's own antioxidant enzymes.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Oxidative Stress Health	NOS3	894G>T	G/G	
	SOD2	-28C>T (Ala16Val)	C/T	

## NOS3 | 894G>T



NOS3 gene encodes the endothelial nitric oxide synthase 3 (eNOS), an essential enzyme for a healthy cardiovascular system. eNOS produces nitric oxide (NO), a small gaseous molecule involved in several biological processes. In the vascular endothelium NO plays crucial roles in the regulation of vascular tone and peripheral resistance. It has vasoprotective effects by suppressing platelet aggregation, leukocyte adhesion and smooth muscle cell proliferation.

### YOUR RESULT: G/G

No variant was detected at the 894 G>T locus.

## SOD2 | -28C>T (Ala16Val)



The SOD2 gene encodes the superoxide dismutase 2, a mitochondrial enzyme involved in the elimination of free radicals which are normally produced within cells and which are damaging to biological systems. The enzyme thus has important antioxidant activity within the cell, especially within the mitochondria.

### YOUR RESULT: C/T

Individuals with the C allele and with a lower consumption of fruits and vegetables may have an increased risk of experiencing the damaging effects of oxidative stress resulting from free radicals. These effects are also enhanced by the presence of additional risk factors such as smoking. Therefore, it is important for individuals with the C allele to ensure adequate fruit and vegetable intake. Supplementation with antioxidant nutrients can also be considered.

Approved  
By:

Houda Hachad, Pharm.D.

Laboratory Director

Translational Software

CLIA: 12345678





# Diet Report

OPTIMAL HEALTH FOR LIFE



**Your Lab**

Your Logo

**GENOTYPE  
REPORT**

<b>NAME</b>	Deborah Osmond
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<b>DATE OF BIRTH</b>	5/6/1965
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<b>REFERRING PRACTITIONER</b>	
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<b>DATE REPORTED</b>	01 November 2017
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<b>ACCESSION NUMBER</b>	Oest-DietSP
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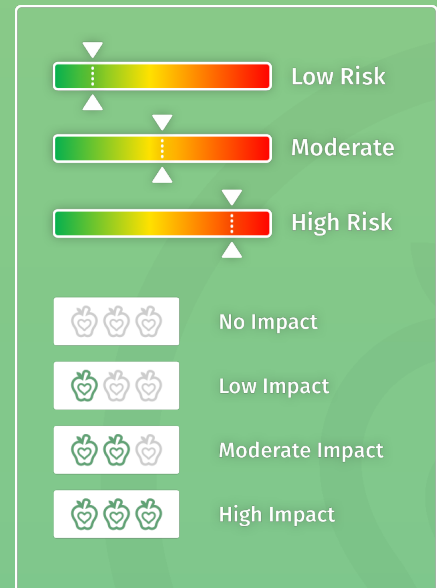
## HOW TO READ THIS REPORT

This genetic report contains two primary pieces of information:

- Based on our analysis of your genes we have calculated your score to determine which of the three possible diet plans (low fat, low carb, and Mediterranean) is likely to be the most effective for you.
- Once you have established the optimal diet type, there is scope for further personalization by considering the genetic contribution of relevant diet and lifestyle factors.

We consider four primary diet and lifestyle factors: exercise, obesity risk, sensitivity to carbohydrates and sensitivity to saturated fats.

In crafting the ideal diet type, take particular note of the lifestyle categories showing medium or high risk.



When devising a diet plan, pay special attention to the medium and high risk lifestyle factors. A brief explanation of these lifestyle factors is given below:

### OBESITY RISK

This gives some indication of the likelihood that you may gain weight easily, but find it more difficult to lose weight than those around you.

### SATURATED FAT

Individuals differ in their response to the quantity and quality of fat in their diet. Your genes may influence how you absorb fat, as well as your ability to burn up fat.

### CARBOHYDRATE

Research has clearly shown that individuals respond differently to carbohydrates in the diet. For some, reducing carbohydrate intake improves weight loss and prevents weight gain.

### EXERCISE

Exercise is an important part of weight loss, but some individuals require higher exercise intensities and greater time spent exercising to mobilize their fat stores. It is important to understand the contribution of exercise in your weight management plan.

## SUMMARY OF YOUR PERSONALIZED WEIGHT MANAGEMENT PLAN

### YOUR DIET PLAN

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A MEDITERRANEAN DIET is the best possible plan for you to manage your weight.

### YOUR EXERCISE PLAN

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A MODERATE INTENSITY exercise program that includes 15 MET HOURS a week.

## BACKGROUND TO THE ANALYSIS

Translational Software received your swab sample and used special molecular techniques to amplify your DNA for further analysis.

This process, called the Polymerase Chain Reaction (PCR), copies the DNA of your genes many times over, so that we can generate sufficient quantities to analyze your genetic material. We then identify unique DNA sequences in some of your genes. Certain changes (polymorphisms) in these genes have been studied in detail, and evidence has emerged that correlates these polymorphisms with an individual's weight management and response to diet and exercise intervention. Having identified the presence or absence of these polymorphisms, we are able to qualitatively assess particular areas of intervention for improved weight management related to the specific genes. To make a holistic assessment of weight management, environmental factors (diet and lifestyle) and previous medical and weight history need to be considered in conjunction with the accompanying genetic profile.

We therefore strongly recommend that these results be discussed with an accredited health professional.

In the following pages you will find a table of your genetic results, and an explanation of these results and associated impacts including diet and lifestyle recommendations.

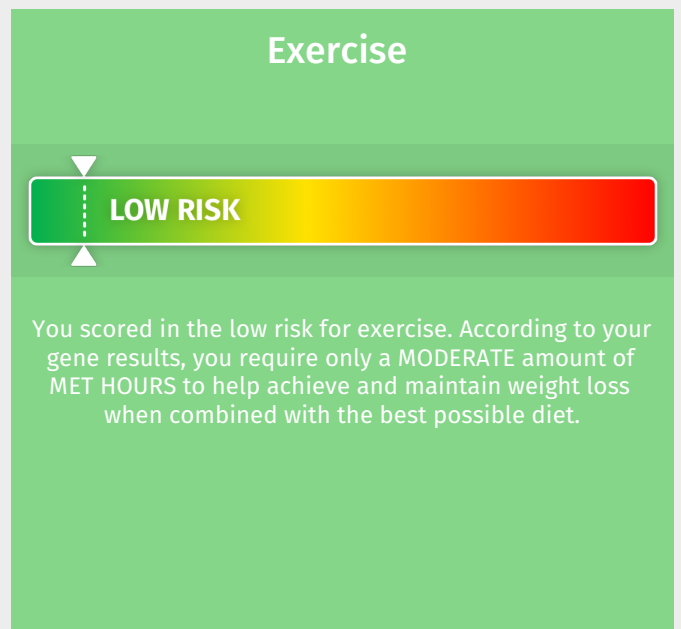
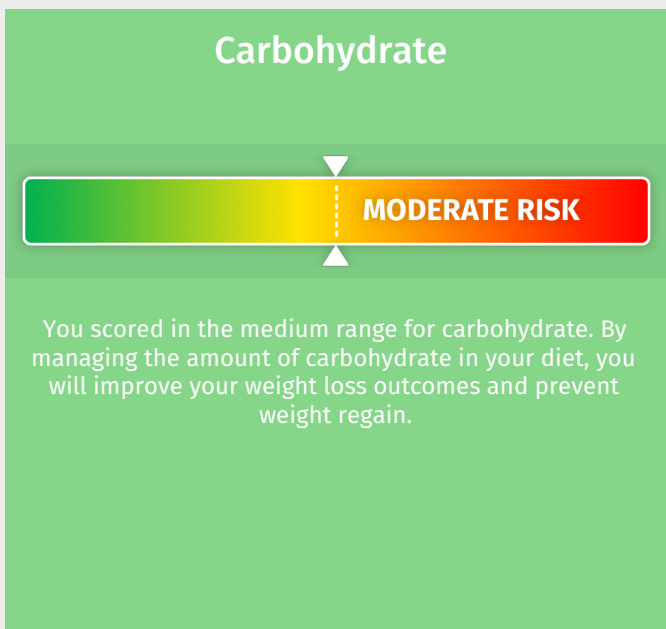
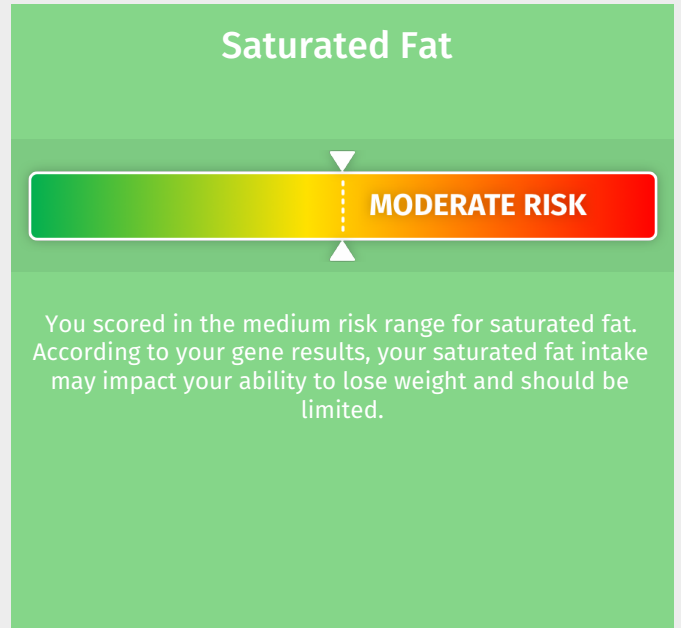
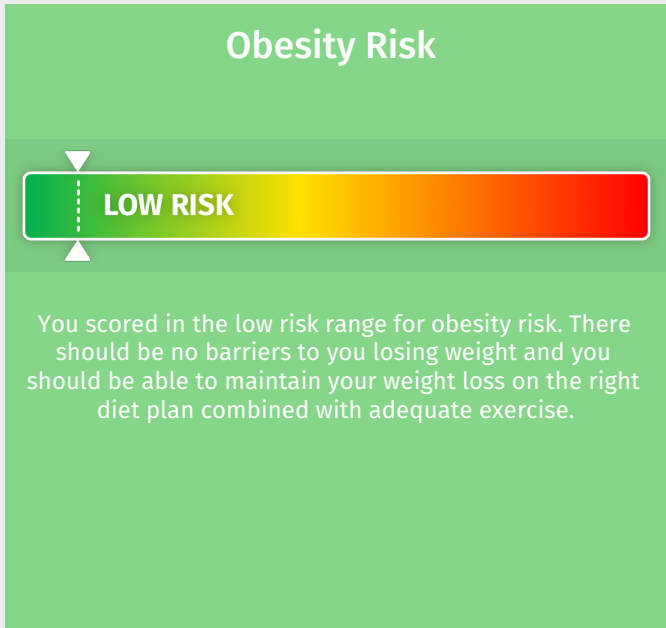
## GENETIC RESULTS

Area of Activity	Gene Name	Genetic Variation	Result	Gene Impact
Absorption and metabolism	ADRB2	46A>G (Arg16Gly)	G/A	
	APOA5	-1131T>C	A/A	
	FABP2	Ala54Thr	C/C	
	PPARG	Pro12Ala	C/C	
Carbohydrate responsiveness	ADRB2	79C>G (Gln27Glu)	C/G	
	ANKK1	Taq1A A1/A2	G/G	
	SLC2A2	Thr110Ile	C/C	
	TAS1R2	Ile191Val	A/A	
Circadian rhythms	CLOCK	3111T>C	A/A	
Exercise responsiveness	ADRB3	Trp64Arg	T/T	
Fat metabolism, obesity and satiety	APOA2	-265T>C	A/G	
Fat storage	PLIN1	11482G>A	G/G	
Inflammation Diet	TNF	-308G>A	G/G	
Regulation of energy intake	FTO	rs9939609 T>A	T/T	
	TCF7L2	C>T	C/T	
Regulation of metabolism and feeding behaviour	MC4R	V103I	T/T	

# WEIGHT MANAGEMENT PRIORITIES

Four diet and lifestyle variables have been analyzed for the role they play in your weight management. Based on your Best Possible Diet plan and the contribution of the weight management variables below, you will be able to customize a weight loss program best suited to your needs.

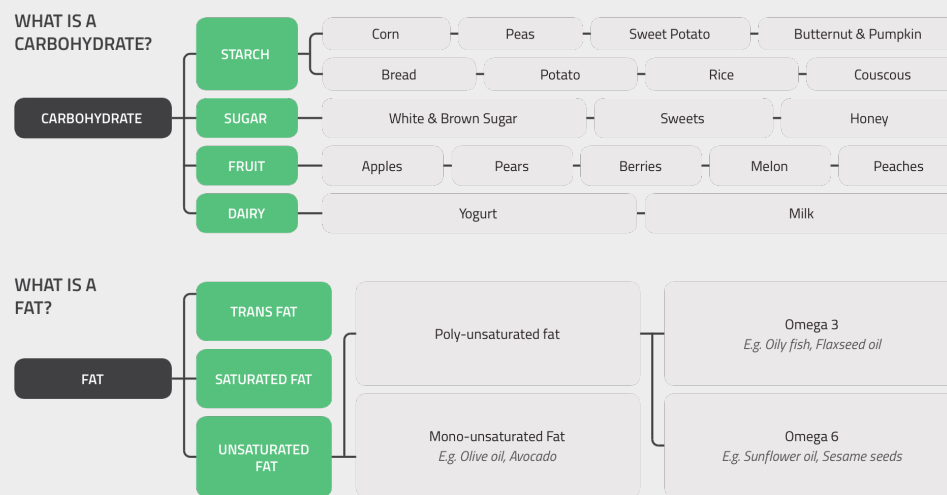
The graphs below give an indication of the significance of each diet and lifestyle variable. From this, you will be able to see which factors need the most attention.



# DIET PLAN PRINCIPLES

## MEDITERRANEAN DIET

The Mediterranean food patterns are typical of Crete, Greece and Southern Italy in the early 1960's. The term is closely tied to traditional areas of olive cultivation in the Mediterranean region more than 30 years ago and not to the urbanized diet eaten in those countries today. Several studies have established the health benefits of the Mediterranean diet in reducing the risk of metabolic syndrome, type 2 diabetes, cardiovascular diseases, and some neuro-degenerative diseases and cancers. In addition, it has been shown to be an extremely effective eating plan for weight loss.



### THE DIET IS BEST DESCRIBED AS:

- Rich in plant foods (whole-grain cereals, fruits, vegetables, legumes, tree nuts, seeds and olives)
- Extra virgin olive oil is the principle source of added fat
- High to moderate intakes of fish and seafood
- Moderate consumption of eggs, poultry and low fat dairy products (mainly cheese and yogurt)
- Low consumption of red meat
- Moderate intake of alcohol (mainly wine during meals)
- In addition all foods in this plan should be as fresh as possible, minimally processed, local and seasonal whenever possible.

## YOUR EXERCISE PLAN

By now, you know the recommended amount of weekly exercise to maximize your chances of weight loss. This recommendation would have been given as MET HOURS. Below you will find a detailed explanation of exactly what MET HOURS are, and a guide to plan your exercise week to meet your recommended MET HOURS. Remember to consult your physician before embarking on a new exercise program, and to stop exercising if you feel nauseous or short of breath.

### WHAT IS A MET?

MET stands for Metabolic Equivalent Task. METs are a way to measure how much energy you burn up during any chosen physical activity. Every activity, from watching TV to going for a run, has a MET value. The more vigorous the activity, the higher the MET value.

### WHAT ARE MET HOURS?

Whereas METs are a way to measure the intensity of a particular activity, MET HOURS allow you to calculate how many hours of your chosen activities you need to do in a week.

### 3 EASY STEPS TO CALCULATING YOUR WEEKLY MET HOUR SCORE

1. Below is a list of activities divided into light, moderate, and vigorous intensity. Find the activity closest to yours.
2. Use this equation to calculate the MET HOURS for each activity.

$$\text{MET Value} \times \text{Duration} = \text{MET HOURS}$$

For example: if you play singles tennis for 1 hour and 40 minutes (1.60 hours) – 8 METS X 1.60 = 13 MET HOURS.

3. To calculate your weekly MET HOURS score, add the MET HOURS score of each workout for that week.

For example, if you played singles tennis for 1 hour and 40 minutes ( $8 \times 1.6 = 13$ ), ran for 30 minutes at a pace of 8 km/hour ( $8 \times .5 = 4$ ) and played 2 hours of golf ( $4.5 \times 2 = 9$ ), then your weekly MET HOURS score would be 26 ( $13 + 4 + 9$ ). See how this compares to the MET HOURS recommendations in your report.

Below is a list of MET VALUES, divided into light, moderate and vigorous intensity activities. Talking during exercise is a reliable way to measure your exercise intensity. If you can talk without puffing at all, you're not pushing too hard and it's very likely a light intensity activity. If you can talk but not sing, you're exercising at a moderate intensity. If you can't talk without gasping, then you are exercising at a high intensity.

### LIGHT INTENSITY

### LESS THAN 5 METS

Stretching, Hatha Yoga	2.5
Horse Riding	2.5
Walking, less than 3.2km/hr, flat ground	2
Walking, 3.2km/hr, firm, flat ground	2.5
Walking, 4km/hr, downhill	2.8
Cycling, less than 16km/hr for leisure	3.4
Rowing, stationary, 50 watts, light effort	4
Tai chi	4
Walking, 5.6km/hr, brisk pace, firm pace	3.8
Water aerobics	4
Golf	4.5
Badminton	4.5

### MODERATE INTENSITY

### 5 - 9 METS

Cycling, stationary, 100 watts, light effort	5.5
Weigh lifting, virgrous effort	6
Jogging/walking combination, 10 minutes	6
Boxing, punching bag	6
Hiking, cross country	6
Walking, 5.6km/hr uphill	6
Mountain biking	8.5
Cycling, general	8
Cycling, stationary, 150 watts	7
Circuit training	8
Stationary rowing, 150 watts	8.5
Aerobics, high impact	7
Running, 8km/hr	8
Cross country running	8
Hockey	8
Tennis, singles	8
Mountain climbing	8
Swimming, freestyle, moderate	7
Walking, 8km/hr	8

### HIGH INTENSITY

### 9 AND ABOVE METS

Cycling, 22-26km/hr, vigrous	10
Running, 9.6km/hr	10
Running, 12.8km/hr	13.5
Kickboxing, judo, etc	10
Rollerblading	12
Cycling, >32km/hr	16
Stairmaster	9
Stationary rowing, 200 watts, very virgrous	12
Boxing, sparring	9
Soccer, competitive	9
Orienteering	9
Rope jumping, fast	12
Squash	12
Swimming, butterfly	11
Swimming, treading water, fast	10

## GENE EXPLANATIONS

Below follows an explanation of all the genes analyzed in this test. Pay particular attention to those genes where you received moderate or high impact scores within the Genetic Results section.

### ANKK1 | Taq1A A1/A2



Midbrain dopamine circuits, particularly dopaminergic signaling via dopamine receptor 2 (DRD2) may play an important role in both addiction and normal eating behavior as they are involved in reward processing. Low DRD2 density is associated with the T (A) allele, putatively making individuals less sensitive to the activation of dopamine-based reward circuitry and rendering them more likely to overeat, especially on energy dense foods. T (A) allele carriers should avoid all high-sugar foods and reduce total carbohydrate intake.

**YOUR RESULT: G/G**

No variant was detected.

### ADRB2 | 79C>G (Gln27Glu)



This ADRB2 receptor protein is involved in energy expenditure regulation through stimulating thermogenesis and lipid metabolism in adipose tissue. The G allele has been associated with increased BMI and fat mass. Subjects with the CG and GG genotypes are less able to mobilize fat stores for energy and have been shown to have a greater risk of obesity and elevated insulin levels when carbohydrate (CHO) intake is greater than 49%. Decreasing intake of CHO has been shown to reduce insulin levels and is beneficial in weight management.

**YOUR RESULT: C/G**

Variant was detected.

### SLC2A2 | Thr110Ile



GLUT2, coded by the SLC2A2 gene, facilitates the first step in glucose induced insulin secretion, with the entry of glucose into the pancreatic  $\beta$ cell. Due to its low affinity for glucose, it has been suggested as a glucose sensor, is considered to be important in the postprandial state, and is involved in food intake and regulation. Individuals with the TT genotype have a higher daily intake of sugars, more so from sweets, such as baked goods, chocolate and sweetened beverages. In T allele carriers, it is important to avoid all high-sugar foods in the diet, especially foods that contain refined sugars.

**YOUR RESULT: C/C**

No variant was detected.

## TAS1R2 | Ile191Val



TAS1R2 encodes the taste receptor type 1 member 2, a key protein involved in the sweet taste recognition. The diverse tissue distribution of TAS1R2 gene affects food intake beyond the detection of sweet taste on the tongue and palate. These tissues include the gastrointestinal tract, pancreas, and hypothalamus -- tissues known for regulating metabolic and energy homeostasis. It is important to decrease intake of high-carbohydrate containing foods, especially high sugar foods.

**YOUR RESULT: A/A**

Variant was detected.

## TNF | -308G>A



Tumour necrosis factor- $\alpha$  (TNF $\alpha$ ), a proinflammatory cytokine secreted by immune cells and fat cells, has been implicated in the development of obesity and insulin resistance. The A allele increases TNF $\alpha$  production and is associated with increased obesity risk, especially when dietary saturated fat and omega-6 fatty acids intake is high. Manage inflammation and emphasize dietary intake of omega 3 fatty acids.

**YOUR RESULT: G/G**

No variant was detected.

## ADRB2 | 46A>G (Arg16Gly)



This ADRB2 receptor protein is involved in the mobilization of fat from fat cells for energy in response to catecholamines and modulates lipolysis during exercise. The G allele has been associated with obesity, and G allele carriers are more likely to gain and regain weight as well as lose weight more slowly. These carriers are less able to mobilize fat stores in response to exercise. For these individuals, it is important to emphasize the use of diet for weight management and to adjust weight management goals.

**YOUR RESULT: G/A**

Variant was detected.

## APOA5 | -1131T>C



Apolipoprotein A5 encodes a protein that plays an important role in regulating plasma triglyceride levels. The T (A) allele has been associated with greater weight and less weight loss, especially when on a high fat, high saturated fat diet. For these individuals, it is important to monitor saturated fat intake.

**YOUR RESULT: A/A**

Variant was detected.

## FABP2 | Ala54Thr



Fatty acid binding protein 2 (FABP2) protein is found in the small intestine epithelial cells where it strongly influences fat absorption and metabolism. The A (T) allele is associated with obesity, elevated BMI, increased abdominal fat, insulin resistance, higher insulin levels, and hypertriglyceridemia. A (T) allele carriers have greater fat absorption and tend to have a slower metabolism, leading to a tendency for weight gain, slower weight loss and difficulty in losing abdominal fat.

**YOUR RESULT: C/C**

No variant was detected.

## PPARG | Pro12Ala



The Ala12 (G) allele is associated with impaired transcriptional activity in adipose tissue and with increased obesity risk in the presence of an obesogenic environment. Focusing on a calorie-restricted diet and increasing physical activity levels in G allele carriers has been associated with improved weight management outcomes.

**YOUR RESULT: C/C**

No variant was detected.

## ADRB3 | Trp64Arg



The beta-3 adrenergic receptor (ADRB3) protein is expressed primarily in visceral adipose tissue. The C allele is associated with increased BMI and weight loss resistance. These individuals break down abdominal fat less easily in response to exercise. As a result, they may have a slower energy metabolism and are less responsive to the beneficial effects of exercise for weight management. The higher risk of obesity among carriers of the C allele may be diminished by higher levels of vigorous physical activity.

**YOUR RESULT: T/T**

No variant was detected.

## APOA2 | -265T>C



Apolipoprotein A2 (APOA2), the second most abundant apolipoprotein in HDL, plays a complex and relatively undefined role in lipoprotein metabolism, insulin resistance, obesity and atherosclerosis susceptibility. The CC (GG) genotype is associated with obesity and increased food consumption, especially total fat and saturated fat intake. When saturated fat intake is high, the CC (GG) genotype is strongly associated with increased BMI and obesity. This diet-gene interaction may also play a role in insulin resistance (IR).

**YOUR RESULT: A/G**

Variant was detected.

## CLOCK | 3111T>C



Circadian Locomotor Output Cycles Kaput (CLOCK), an essential element of the human biological clock, is involved in metabolic regulation. Carriers of the C (G) allele are less successful at losing weight compared to the TT (AA) genotype. In addition, those with the C (G) allele have reduced sleep, report morning fatigue and show an evening preference for activities. They also have higher ghrelin levels which regulates appetite, potentially altering eating behavior and weight loss.

**YOUR RESULT: A/A**

No variant was detected.



Fat-mass-and-obesity-associated (FTO) gene is present at high levels in several metabolically active tissues, including, heart, kidney, and adipose tissue, and is most highly expressed in the brain, particularly in the hypothalamus which is concerned with the regulation of arousal, appetite, temperature, autonomic function, and endocrine systems. It has been suggested that the FTO gene plays a role in appetite regulation and that it is associated with energy expenditure, energy intake, and diminished satiety. The A allele has been associated with higher body mass index (BMI), body fat percentage and waist circumference, especially in individuals with a sedentary lifestyle. Overweight individuals with the A allele are at increased risk for insulin resistance and diabetes, especially when there is a high fat intake. Modify the diet to include a moderate amount of carbohydrate, increase monounsaturated fatty acids (MUFA) intake and decrease saturated fats (SAT FAT); manage the overall fat intake. Regular physical activity is recommended.

**YOUR RESULT: T/T**

No variant was detected.



Transcription factor 7-like 2 (TCF7L2) gene encodes a transcription factor that regulates blood glucose homeostasis and may operate via impaired glucagon-like peptide 1 secretion, which is stimulated more by fat than by carbohydrate ingestion. Individuals with the T allele, and more so the TT genotype, experience less weight loss than CC genotype. Diet and exercise intervention is very important for T allele carriers to prevent weight regain and development of Insulin resistance and diabetes. T allele carriers lose more weight on a low fat hypo-energetic diet than on a high fat diet. A low glycemic load diet and all interventions to manage insulin is also recommended.

**YOUR RESULT: C/T**

Variant was detected.



Melanocortin 4 Receptor (MC4R) is a strong obesity candidate gene, significantly associated with energy intake and expenditure. The C allele is associated with higher intakes of total energy and dietary fat, as well as increased snacking in children and adults, increased hunger and a higher prevalence of eating large amounts of food. Provide a personalized, calorie-restricted eating plan that includes mindful eating strategies and methods to promote satiety.

**YOUR RESULT: T/T**

No variant was detected.



PLIN1 gene encodes a protein called perilipin-1. This protein coats lipid storage droplets in adipocytes, thereby protecting them until they can be broken down by hormone-sensitive lipase. The A allele is associated with greater obesity risk. A allele carriers are more weight loss resistant and show greater decrease in lipid oxidation rate compared to the GG genotype. When there is a higher intake of complex carbohydrates, and lower saturated fat intake, the allele is protective against obesity. Avoid all refined carbohydrates.

**YOUR RESULT: G/G**

No variant was detected.

Approved  
By:

Houda Hachad, Pharm.D.

Laboratory Director

Translational Software

CLIA: 12345678





# Sport Report

OPTIMAL HEALTH FOR LIFE



**Your Lab**

Your Logo

**GENOTYPE  
REPORT**

**NAME**

Mike Muscles

**DATE OF BIRTH**

10/11/1984

**REFERRING PRACTITIONER**

**DATE REPORTED**

11/1/2017 4:26:05 PM

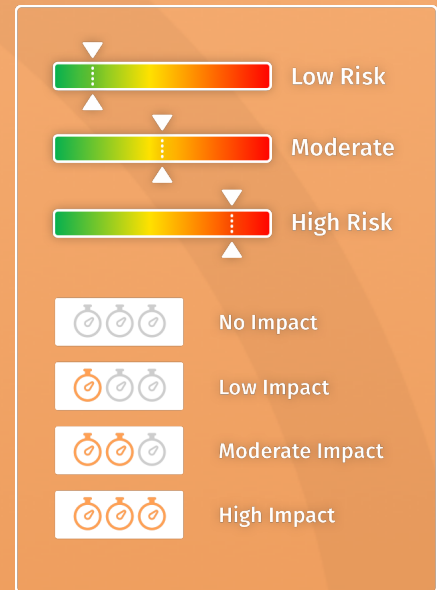
**ACCESSION NUMBER**

CL-SPortSP

## WELCOME TO YOUR DNA SPORT REPORT

From your buccal swab sample we have used a process called the Polymerase Chain Reaction (PCR), which copies the DNA of your genes many times over so that we can generate sufficient quantities to analyse your genetic material. We then identify unique DNA sequences in some of your genes. Certain changes (polymorphisms) in these genes have been studied in detail, with evidence that correlates these polymorphisms with an individual's risk of developing certain chronic disease conditions or altered metabolic processes. Having identified the presence or absence of these polymorphisms, we are able to qualitatively assess particular areas of health risk related to the specific genes.

To make a holistic assessment of health risks, environmental factors (diet and lifestyle) need to be considered in conjunction with the accompanying genetic profile.



## HOW TO READ YOUR RESULTS

The results will give the gene name and variation we tested, as well as a brief description of this gene. You will find your specific result and an explanation of how this may impact your training and/or nutritional requirements. Certain genetic variants are advantageous for athletic performance, while some variants may contribute to an increased risk for injury or a delayed recovery time. Training and nutritional recommendations that may benefit you will also be made, with any additional health recommendations indicated as well. The impact of your specific result can be identified by the DNA Sport symbols (please see the above key).

### **NO IMPACT**

Genotype has no effects on the biological area in question.

### **LOW IMPACT**

Genotype has mild effects on the biological area in question with a small change in responsiveness to environmental influences.

### **MODERATE IMPACT**

Genotype has moderate effects on the biological area in question. Attention should be paid, and some dietary and lifestyle changes are recommended.

### **HIGH IMPACT**

Genotype has significant impact on the biological area in question. Cohesive and intensive diet and lifestyle action should be taken.



# YOUR GENETIC RESULTS PART 1

## INJURY AND RECOVERY

We only need to look around at other individuals whom we exercise with to realize that some individuals seem to be 'injury prone', while others are never forced to skip a day of training. Additionally, some individuals are able to recover quickly from exercise and are ready to train hard again after just a day's rest whereas some individuals don't seem to 'bounce back' from hard sessions quite so quickly and need a longer break between intense training sessions. Research has revealed that certain genetic variations infer a delayed recovery from hard exercise training, while other variants put some individuals at a significantly increased risk of certain injuries. Injury and recovery are very much intertwined because being slow at recovering from heavy exercise is likely to place you at a greater risk of injury, and this increased injury risk means that you will need to incorporate appropriate recovery strategies into your training program. Delayed recovery or increased susceptibility to injury means that a balanced, well managed training program is required, with strong emphasis on recovery strategies, conditioning exercises and nutrition.

With regards to injury and recovery, three important biological systems have been well researched and are examined in the DNA Sport test: injury susceptibility (connective tissue remodeling), inflammation and oxidative stress. The table below gives your genetic results for these three categories, with gene explanations following thereafter.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Inflammation Sport	CRP	219G>A	C/C	
	IL6	-174G>C	G/G	
	IL6R	481A>C	C/C	
	TNF	-308G>A	G/G	
Injury Susceptibility	COL1A1	1546G>T	C/C	
	COL5A1	267C>T	C/T	
	GDF5	-275C>T	C/C	
Oxidative Stress Sport	NOS3	894G>T	G/T	
	SOD2	-28C>T (Ala16Val)	C/C	

## INFLAMMATION SPORT

Inflammation is a normal immune response and an essential part of tissue healing following exercise. The release of inflammatory cytokines is controlled by various genes, however when there is a greater than normal increase in inflammatory cytokines following exercise, or a prolonged increase in these cytokines, increased recovery time is required between hard sessions in order to avoid tissue damage.

### IL6 | -174G>C



Interleukin 6 is an inflammatory cytokine that stimulates an immune response to strenuous exercise. Excess release of this cytokine can lead to a chronic inflammatory state.

#### YOUR RESULT: G/G

The G allele does not influence post-exercise inflammation. Since you do not release excess amounts of IL6, you may have improved muscle repair after eccentric exercise. This gives you an advantage for strength and power training. Adding eccentric loading to your training regimen may be beneficial to power development.

### TNF | -308G>A



Tumour necrosis factor- $\alpha$  (TNFA), like IL6, is a pro-inflammatory cytokine that stimulates the acute phase reaction of inflammation. Levels of TNFA increase after intensive exercise.

#### YOUR RESULT: G/G

Your genotype does not influence the inflammation you experience with exercise.

### CRP | 219G>A



CRP increases in response to inflammation and plays a role in activating parts of the innate immune system. Regular moderate intensity exercise and favorable nutritional choices can help reduce baseline inflammatory markers such as CRP.

#### YOUR RESULT: C/C

The G (C) allele is linked to moderately higher levels of CRP which is associated with higher levels of inflammation. This will impact your required recovery time between training sessions. Regular moderate intensity exercise, as well as a diet low in saturated fats, can lead to improvements in CRP levels.



IL6R gene encodes the Interleukine 6 receptor that mediates the action of Interleukin 6 (IL6). This gene influences the fatigue experienced following a physical exercise and the ability to recover.

#### YOUR RESULT: C/C

The C allele leads to higher levels of Interleukin 6 receptor (IL6R) as well as Interleukin 6 (IL6), and increases the acute inflammatory effects of exercise. It may be necessary to increase your recovery time between training bouts and sessions. It is also important to increase your anti-inflammatory nutritional support. Post-exercise IL6 levels can be reduced by ingesting a good carbohydrate drink during exercise.

## INJURY SUSCEPTIBILITY

Multiple stimuli, including exercise and mechanical load, can lead to connective tissue remodeling. Although remodeling may lead to physical gains, alterations in the structural properties of tissues may also lead to increased injury susceptibility. The variations examined in the DNA Sport test are linked to the ability of soft tissues to repair and remodel following tissue degradation, thus being implicated in injury risk.

### COL1A1 | 1546G>T



COL1A1 is one of the major collagens in connective tissues. Altered expression of this gene may lead to injury risk due to a structural change in the properties of the tissue.

#### YOUR RESULT: C/C

The G (C) allele leads to a decreased expression of COL1A1 and puts you at a moderately increased risk for tendon and ligament injuries. Base strength, conditioning and flexibility training, and nutritional intervention are important parts of injury prevention.

### COL5A1 | 267C>T



COL5A1 is one of the minor collagens that regulates the formation of new soft tissue fibers. Altered expression of this gene can lead to injury risk.

#### YOUR RESULT: C/T

The T allele is associated with a moderately increased risk of injury. Make sure you are including conditioning and flexibility training in your exercise program to try and decrease your risk of injury. Nutrition is also important in injury prevention.

### GDF5 | -275C>T



The growth differentiation factor 5 (GDF5) plays a role in the development and healing of skeletal, joint, and soft tissues. This gene influences the ability to recover from tissue damage.

#### YOUR RESULT: C/C

The C allele does not impact injury risk.

## OXIDATIVE STRESS SPORT

Free radicals are a normal by-product of the biological processes that generate energy, such as those that occur during exercise. They are highly reactive with other molecules, and can damage DNA, proteins and cellular membranes. Antioxidants are free radical scavengers that interact with the free radical to ensure that it is no longer a reactive molecule. Long term regular light and moderate intensity exercise leads to an increase in antioxidant enzymes, as well as a decrease in baseline inflammatory cytokines: beneficial to exercise training, performance and optimal health.

### NOS3 | 894G>T



NOS3 gene encodes the endothelial nitric oxide synthase 3 (eNOS), an essential enzyme for a healthy cardiovascular system. eNOS produces nitric oxide (NO), a small gaseous molecule involved in the regulation of vascular tone and peripheral resistance. Decreased activity of eNOS has been associated with an increase in free radicals.

#### YOUR RESULT: G/T

The T allele leads to a decreased activity of eNOS. This negatively affects oxidative stress and may also promote atherosclerosis. Ensure adequate antioxidant intake, as well as omega 3 fatty acids.

### SOD2 | -28C>T (Ala16Val)



The SOD2 gene encodes the superoxide dismutase 2, a mitochondrial enzyme involved in the elimination of free radicals which are normally produced within cells and which are damaging to biological systems. Oxidative stress during intensive training can lead to muscular fatigue and antioxidant enzymes are thus very important for physical performance.

#### YOUR RESULT: C/C

The C allele is linked to higher levels of oxidative stress. This could be a risk for developing long term disease if fruit, vegetable and other antioxidant intake is not adequate. Low and moderate intensity exercise training can also help to increase baseline levels of antioxidant enzymes.

## YOUR INJURY RISK

Based on our analysis and interpretation of your genetics, your DNA Sport tests indicate that it is likely that you have a moderate risk of a soft tissue injury. This means that you will need to be careful, ensuring that your training volumes and intensities are appropriate to your fitness level and that you engage in some degree of injury-preventing conditioning exercises. Nutrition also plays a role in injury prevention.



## RECOMMENDATIONS

Your genetic results reveal that you may need to be taking some preventative steps to try and anticipate the strains that will occur on your musculoskeletal system with training. We call this conditioning work 'prehabilitative training' – i.e. injury prevention. Resistance and flexibility training are the cornerstones for prehabilitation, and rehabilitation if an injury occurs. They include classical weight training, plyometrics, Pilates, yoga and specific exercises that have been designed to target particular injury risks. If you are training regularly it would be worthwhile doing at least one or two sessions per week which are focused on general conditioning, helping to reduce your injury risk. If you are an elite athlete or focused on a specific sport, consider setting up a practice of sport-specific conditioning exercises 3-4 times per week. It is important to consider the most common musculoskeletal injuries that occur in your particular sport and take specific advice from a coach or exercise professional who specializes in your event. For example: runners are prone to Achilles tendonitis, calf strains, hamstring strains, patellar tendonitis, IT band syndrome; cyclists are prone to knee, back and neck pain (although a good bike set up can make a large difference); swimmers are prone to swimmers shoulder and breast-stroke knee. With regards to nutrition, ensure adequate intake of vitamin C and iron as these are important for collagen turnover. A good protein source is important for amino acid building and should be taken in after intense training sessions.

## YOUR RECOVERY

Based on our analysis and interpretation of your genetics, your DNA Sport tests indicate that it is likely that you are able to recover quickly from hard exercise. This means that you have the capacity to undertake a heavy training load with frequent inputs of exercise. However, it should be noted that hard training comes from a mixture of good genetics and slowly building a training foundation over the course of many years. If you have already been training at a high level for some years, take this as an indication that your body can theoretically handle high loads of exercise under ideal conditions. Otherwise, it is suggested that you build up to this level slowly, taking into account your risk for injury.



## RECOMMENDATIONS

Due to your likely moderate rate of recovery from hard exercise sessions, if you would like to gain the best returns from your training and optimize performance in your chosen sport, we recommend that you follow some planned recovery strategies. Recovery is classically considered as the time between sessions. According to training theories, we require two to three days between hard training sessions. Since you have a moderate recovery rate, once a training base is established, you may expect to hit two to three hard sport-specific sessions per week. Other steady recovery and conditioning sessions can be built around these big two to three sessions. If you are a seasoned athlete, you could potentially progress to a once per day routine with an additional session on one or two days of the week. Always allow one full day off per week. Recreational athletes with other commitments might wish to max out at a total of five sessions per week. In addition to carefully planning your recovery times between sessions, you should also consider these following factors. Sleep is vitally important and you should look to obtain enough sleep so that you feel refreshed upon rising in the morning. This might be +/- eight hours at night and a nap in the day is especially useful for optimizing recovery. Managing your nutrition is also important for optimal recovery and keeping the inflammatory process under control. Since inflammation influences recovery rates, you should look to consume mostly anti-inflammatory and antioxidant foods in your diet and avoid those that are pro-inflammatory. A diet low in carbohydrates can help to reduce post exercise inflammation, however, consuming carbohydrate based beverages during exhaustive exercise can help to reduce levels of inflammatory cytokines such as IL6 and CRP. Consuming a good protein source after exercise is also known to decrease inflammation and assist recovery. Antioxidants can also be consumed in a wide variety of foods, especially fruits and vegetables, as well as green tea. Focus on fruits and vegetables of many different colors; green leafy vegetables and cruciferous vegetables have particularly good antioxidant properties. Avoid smoking of any kind.

# YOUR GENETIC RESULTS PART 2

## PERFORMANCE

It is well established that a high percentage of the variance observed in athletic status can be explained by genetic factors. These genetic factors, as examined in your DNA Sport test, can determine how well you will respond to certain types of exercise training. Although both aerobic training as well as strength and weight training are important for overall health and fitness, the ratio of these types of training should vary between individuals, even between those working towards the same goals. The overall results of this genetic test will enable you to focus your training towards the type of exercise that is going to give you the best outcomes for your hard work, whether that be aerobic or power type training.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Blood Flow and Respiration	ACE	I/D	C/C	
	AGT	Met235Thr	A/A	
	BDKRB2	-275C>T	C/T	
	VEGFA	94C>G	G/G	
Energy Mobilisation	GABPB1	A>G	A/A	
	PPARA	1160-396G>C	G/C	
	PPARGC1A	1444G>A (Gly482Ser)	G/A	
Fuel Metabolism	ADRB2	79C>G (Gln27Glu)	C/C	
	ADRB2	46A>G (Arg16Gly)	G/A	
	CYP1A2	-163C>A	A/A	
	TRHR	rs7832552	C/T	
Musculoskeletal Properties	ACTN3	R577X	T/T	
	VDR	Taq1 C>T	T/C	

## BLOOD FLOW AND RESPIRATION

Sporting performance is largely dependent on oxygen diffusion, and thus the vascular and pulmonary systems. Oxygen transport to the musculature is the key determinant of aerobic capacity and resistance to fatigue.

### ACE | I/D



ACE is a potent vasoconstrictor in the renin-angiotensin system. This enzyme is key in blood pressure regulation. ACE impacts aerobic capacity, muscular strength and lean body mass.

#### YOUR RESULT: C/C

The II (CC) genotype is associated with lower ACE activity and greater aerobic capacity due to increased vasodilation. Aerobic training and low level repetitive weight training will lead to increased muscle efficiency and aerobic capacity. II (CC) also contributes to salt sensitivity, and thus salt intake should be monitored in order to avoid potential blood pressure spikes.

### AGT | Met235Thr



This gene is important in the regulation of electrolyte and body fluid balance, as well as blood pressure. Upregulation of AGT potentially leads to vasoconstriction and increased blood pressure. This gene contributes to the development of power.

#### YOUR RESULT: A/A

The TT (AA) genotype does not impact sporting performance.

### BDKRB2 | -275C>T



Bradykinin is a vasodilator that acts via the bradykinin B2 receptor. BDKRB2 is involved in blood pressure regulation, having the opposite effect to ACE.

#### YOUR RESULT: C/T

The T allele is associated with a moderate advantage for aerobic exercise. It increases expression of this gene, increasing vasodilation which is believed to increase muscle contraction efficiency.

### VEGFA | 94C>G



Vascular endothelial growth factor (VEGF) is a protein involved in the formation and growth of new blood vessels. Therefore, the levels of VEGF impact blood flow and oxygenation - these factors influence muscle efficiency and aerobic capacity.

#### YOUR RESULT: G/G

This genotype does not impact sporting performance.

## ENERGY MOBILISATION

In order to avoid fatigue during exercise the rate of energy production needs to match the rate of energy consumption. The mitochondria are the key sites of energy production (in the form of ATP) for muscle fibers, and the oxidative capacity of muscle fibers is directly linked to the formation of new mitochondria.

### GABPB1 | A>G



Nuclear factor (erythroid-derived 2)-like 2 (NRF2) is a protein that serves as a regulator of the body's antioxidant response against oxidative damage triggered by injury and inflammation. This protein is also important in the formation of new mitochondria: the 'power house' of the cell where energy is produced. NRF2 improves respiratory capacity and the rate of energy production during exercise.

#### YOUR RESULT: A/A

The A allele does not impact sporting performance. However, it must be noted that the AA genotype is present in 98% of the population, and only 2% carry a G allele.

### PPARA | 1160-396G>C



Peroxisome proliferator-activated receptor alpha (PPARA) is involved in the uptake, utilisation and break down of fatty acids to ATP - the main source of energy during prolonged exercise.

#### YOUR RESULT: G/C

GC is advantageous for both aerobic capacity and strength training. You are also likely to have a greater percentage of Type I (slow twitch) muscle fibers.

### PPARGC1A | 1444G>A (Gly482Ser)



Peroxisome Proliferator-Activated Receptor Gamma, Coactivator 1 Alpha (PPARGC1A) plays an essential role in energy regulation. It is expressed in tissues that have high energy demands and is therefore abundant in mitochondria and associated with aerobic capacity. PPARGC1A is also involved in the exercise-induced increase in mitochondria.

#### YOUR RESULT: G/A

This does not impact sporting performance. However, mitochondrial biogenesis and aerobic performance can be heightened by training and caloric restriction.

## FUEL METABOLISM

Carbohydrates and fats are the main contributors to the fuel supply that is necessary to perform exercise. These sources are converted to energy, in the form of ATP, when required. Protein is generally involved in the maintenance and remodeling of tissues rather than an energy source to fuel muscles.

### CYP1A2 | -163C>A



Caffeine is a central nervous system and metabolic stimulant used to reduce physical fatigue. In athletics, moderate doses of caffeine have been known to improve both sprint and endurance performance. CYP1A2 is one of the main enzymes that catalyzes the oxidation of caffeine in humans.

#### YOUR RESULT: A/A

This indicates that you are able to metabolize caffeine at a fast rate. You may want to take caffeine 30 minutes to an hour before a race in order to benefit from the effects. Depending on how long your race is, you could take caffeine during the race as well. Due to health implications, it is not recommended to have more than three cups of coffee on an average day.

### ADRB2 | 46A>G (Arg16Gly)



ADRB2 regulates cardiac, pulmonary, vascular, endocrine and central nervous system functions. Adrenaline acts via ADRB2 to maintain blood glucose levels during prolonged exercise by promoting glycogenolysis. Arg16Gly is involved in the modulation of cardiac output during exercise through vasodilation.

#### YOUR RESULT: G/A

The A allele is associated with the ability to achieve a moderately higher aerobic capacity with endurance training. Focus on aerobic training that stimulates VO2max and aerobic capacity.

### ADRB2 | 79C>G (Gln27Glu)



Gln27Glu within ABRB2 is associated with aerobic capacity and the ability to lose weight as a result of exercise.

#### YOUR RESULT: C/C

The C allele is linked to greater aerobic capacity with training. Individuals with this genotype also have a greater ability to mobilize fats for fuel during exercise, which would suggest the ability to lose weight through exercise. Sub-maximal and maximal endurance training is likely to lead to an increase in aerobic capacity. If you wish to gain a more comprehensive understanding of your weight loss responsiveness to exercise, we recommend performing the DNA Diet test.



Thyrotropin releasing hormone receptor (TRHR) stimulates the release of thyroid hormones T3 and T4 leading to an increased metabolic rate which is required to mobilize fuels during exercise. The TRHR gene has been linked to lean body mass.

**YOUR RESULT: C/T**

This genotype does not impact sporting performance.

## MUSCULOSKELETAL PROPERTIES

The properties of the musculoskeletal system, including bones, muscles, cartilage, tendons, ligaments and joints, greatly affect our ability to perform. Although these tissues can be potentially altered with training, our genetics form the basis of the structural properties of these tissues.

### ACTN3 | R577X



ACTN3 is only present in Type II (fast twitch) muscle fibers and greatly influences power development. ACTN3 also plays a role in muscle fiber type specialization, diameter and metabolism.

#### YOUR RESULT: T/T

The XX (TT) genotype indicates that you do not have a coding copy of the ACTN3 gene, and thus should not have overly high expectations when it comes to strength training. However, this gives you an advantage with regards to aerobic training, possibly due to a greater percentage of slow twitch muscle fibers.

### VDR | Taq1 C>T



Activation of vitamin D receptor (VDR) leads to the maintenance of calcium and phosphorus levels in the blood and bones, which is necessary for bone formation and replacement, and the preservation of bone mineral density. VDR has been linked to muscle strength.

#### YOUR RESULT: T/C

The TC genotype does not impact sporting performance and is unlikely to influence bone mineral density.

## YOUR ATHLETIC POTENTIAL

The performance potential graph gives you an indication of your genetic “score” as a potential of the total “aerobic” and “power” points available.



Based on our analysis and interpretation of your genetics, your DNA Sport tests indicate that in order to gain the best results from your training, you should primarily stimulate your aerobic capacity with endurance-style training. This means that you are likely to benefit from including long-duration, moderate intensity activities in your training regimen. This type of training relies on the “aerobic” energy system. Generally, light to moderate intensity activities can be sufficiently supported by this system. You should also include some shorter duration, higher intensity type exercises in your training, but your greatest potential lies in stimulating your aerobic system. Remember that many variables influence your success with regards to training and performance; genetics is one of these variables that should be used to understand the total outcome.

## KEY TRAINING PRINCIPLES

Your genetic results indicate that you will likely see improvements and gain the greatest returns with aerobic based exercise. Your training should ideally include mostly moderate intensity and moderate to long duration cardiovascular training. In addition, include high-intensity training and incorporate strength and conditioning in order to stimulate a range of energy systems for a well-balanced fitness. The types of training to include are long duration running, cycling, swimming, or similar types of cardio exercise. You could vary your sessions from 30-60 minutes at a steady pace, to 3 x 10-minute repeats at a harder pace. Remember to include an easy pace warm up before sessions, as well as cooling down sufficiently afterwards. Your training will need to be tailored depending on your goal, but always bear in mind that your success is likely to come from longer duration moderate intensity sessions. The aerobic fitness that you gain from this type of training will be of great benefit in endurance style events, but if you are involved in a particular sport, you need to be aware of the importance of combining this training with sport-specific training. It is also important to include strength and conditioning training into your program. Even if you are training for endurance performance, strength and speed training as well as stability training and conditioning will greatly benefit you. Strength can be increased with weight training and speed can be increased with structured interval training. With regards to weight training, your efforts may be best focused on doing multiple repeats with relatively light weights rather than only a low number of repeats with heavy weights. This will help develop good muscle contraction efficiency; circuit training fits this profile well. Plyometrics can also be used to train for greater power output. Resistance training, core-stability, flexibility, general conditioning and sport-specific conditioning are all important parts of general fitness, optimal performance and injury prevention. Remember to efficiently manage your recovery time between sessions in order to avoid superfluous fatigue and injury. What follows is a table which refers to the various levels of cardio training. As someone who has good endurance potential, we recommend that you focus your efforts on the low to medium intensity sessions such as Levels 1 to 4. Your core sessions should be moderate intensity, long duration at levels 2 and 3.

## SUPPLEMENTARY INFORMATION

### MEASURING YOUR THRESHOLD HEART RATE AND SETTING YOUR TRAINING ZONES

Perform a solid warm-up, and then do a 30 minute time trial (all out) on a relatively flat course. Record your average heart rate for the final 20 minutes of the time trial. This is your LTHR. To set your zones, your LTHR is the figure that should go between Level 4 and 5 (100%) in the cardio table above. To work out the other zone heart rates, simply multiply the LTHR by the percentages given.

### CARDIO ZONES TRAINING TABLE

The levels referred to in the cardio training table below represent zone training that can be done either with a Heart Rate (HR) Monitor or simply by your Rate of Perceived Exertion (RPE). You will need to test yourself for your Threshold Heart Rate if you wish determine your training levels with a heart rate monitor (see below). RPE is simply a 0-10 scale of how you perceive a training session to be - 0 being nothing and 10 being maximal output. Levels 1 to 4 are considered endurance style training, whereas above level 4 and above is used in short duration speed and interval training exercises.

LEVEL	INTENSITY	% OF THRESHOLD HR	RPE
1	RECOVERY	<81%	<2
2	AEROBIC	81-89%	2-3
3	TEMPO	90-93%	3-4
4	SUB-THRESHOLD	94-99%	4-5
5	SUPRA-THRESHOLD	100-102%	6-7
6	AEROBIC CAPCTIY	103-106%	>7
7	ANAEROBIC CAPACITY	>106%	MAXIMAL

Approved  
By:

Houda Hachad, Pharm.D.

Laboratory Director

Translational Software

CLIA: 12345678





# Estrogen Report

OPTIMAL HEALTH FOR LIFE



**Your Lab**

Your Logo

**GENOTYPE  
REPORT**

<b>NAME</b>	Deborah Osmond
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<b>DATE OF BIRTH</b>	5/6/1965
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<b>REFERRING PRACTITIONER</b>	
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<b>DATE REPORTED</b>	10/25/2017
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<b>ACCESSION NUMBER</b>	Oest-DietSP
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# WELCOME TO YOUR DNA ESTROGEN REPORT

From your buccal swab sample we have used a process called the Polymerase Chain Reaction (PCR), which copies the DNA of your genes many times over so that we can generate sufficient quantities to analyze your genetic material. Then, we identify unique DNA sequences in some of your genes.

Considerable inter-individual variability has been observed in biological areas that are involved in carcinogen metabolism, metabolism of steroid hormones, and phase I and phase II detoxification. Variations in genes involved in these biological processes help identify a sub-population of women and men with higher lifetime exposure to estrogens, estrogen metabolites and other carcinogens. Understanding an individual's genetic variability will allow for targeted diet, lifestyle and hormone intervention.

	No Impact
	Low Impact
	Moderate Impact
	High Impact

## HOW TO READ YOUR RESULTS

You will find your genetic results in the following pages. On the left side, you will see the gene name and description. On the right side, you will find your specific result and an explanation of the results, associated risks, and diet and lifestyle recommendations. Please see the key above to identify each impact level.

### NO IMPACT

Genotype has no effects on the biological area in question.

### LOW IMPACT

Genotype has mild effects on the biological area in question with a small change in responsiveness to environmental influences.

### MODERATE IMPACT













Genotype has moderate effects on the biological area in question. Attention should be paid, and some dietary and lifestyle changes are recommended.

### HIGH IMPACT

Genotype has significant impact on the biological area in question. Cohesive and intensive diet and lifestyle action should be taken.

## SUMMARY OF YOUR RESULTS

The combination of gene variants identified in this analysis indicates possible inefficiencies in estrogen detoxification, and additional support would be recommended.

AREA OF ACTIVITY	GENE	GENETIC VARIATION	RESULT	GENE IMPACT
Detoxification Oestrogen	CYP17A1	-34T>C	G/G	
	CYP1A1	Msp1 T>C	T/T	
		2454A>G (Ile462Val)	T/T	
	CYP1B1	4326C>G (Val432Leu)	C/G	
	GSTM1	519G>C	indeterminate	
	GSTT1	15G>C	indeterminate	
	NQO1	609C>T	C/C	
	SULT1A1	638G>A	A/A	
Methylation Oestrogen	COMT	472G>A (Val158Met)	A/G	
	MTHFR	677C>T	GG	
Oxidative Stress Oestrogen	SOD2	-28C>T (Ala16Val)	C/T	
Thrombosis	F5	G1691A	CC	

## TEST RESULTS

### F5 | G1691A



Factor V functions as a cofactor to allow factor Xa to activate the enzyme thrombin, and in turn cleaves fibrinogen to form fibrin, which polymerizes to form the dense meshwork that makes up the majority of a clot. Activated protein C (APC) is a natural anticoagulant that acts to limit the extent of clotting by cleaving and degrading factor V. Factor V Leiden gene mutation is characterized by a poor anticoagulant response to APC and an increased risk for venous thromboembolism (VTE). Deep venous thrombosis (DVT) is the most common VTE, with the legs being the most common site. However, VTE can also occur in other parts of the body including the brain, eyes, liver, and kidneys.

#### YOUR RESULT: CC

No variant was detected at the 1691 G>A (C>T) locus.

### MTHFR | 677C>T



Methylenetetrahydrofolate Reductase (MTHFR) is a key enzyme in the folate metabolic pathway and the multistep process that converts the amino acid homocysteine to methionine. Methionine is used to in the synthesis of proteins and other important antioxidant compounds involved in detoxification of estrogens. Reduced activity influences the balance between DNA synthesis, repair and methylation processes.

#### YOUR RESULT: GG

No variant was detected at the 677 C>T locus.

## COMT | 472G>A (Val158Met)



Soluble catechol-O-methyltransferase (S-COMT) helps control the levels of certain hormones and is involved in methylation and inactivation of catechol estrogens. Accumulation of some estrogen metabolites, which leads to oxidative DNA damage, is a recognized risk factor for breast cancer.

### YOUR RESULT: A/G

The A allele is associated with a 3-4 fold reduction in the methylation activity of the COMT enzyme. For A allele carriers, beneficial modulation of estrogen metabolism can be accomplished through dietary and lifestyle modifications. Key interventions include increasing insoluble fiber, managing the quality of dietary fat intake, increasing phytoestrogen intake, losing weight, and increasing exercise. In addition, select nutrients and micronutrients effectively reduce estrogen load by supporting preferred estrogen pathways. These are included at the end of the report.

## CYP1A1 | Msp1 T>C



The CYP1A1 gene encodes a phase I cytochrome P450 enzyme that converts environmental procarcinogens such as polycyclic aromatic hydrocarbons (PAHs) and aromatic amines to reactive intermediates having carcinogenic effects. In addition, CYP1A1 is involved in the oxidative metabolism of estrogens, which may play a critical role in the etiology of breast and prostate cancers. CYP1A1 enzyme catalyzes the 2-hydroxylation of estradiol (E1 and E2) in several extra hepatic tissues including breast tissue. It is also involved in activating cigarette smoke, diet and environmental pollutants, as well as producing carcinogens.

### YOUR RESULT: T/T

No variant was detected at the Msp1 T>C locus.

## CYP1A1 | 2454A>G (Ile462Val)



The CYP1A1 gene encodes a phase I cytochrome P450 enzyme that converts environmental procarcinogens such as polycyclic aromatic hydrocarbons (PAHs) and aromatic amines to reactive intermediates having carcinogenic effects. In addition, CYP1A1 is involved in the oxidative metabolism of estrogens, which may play a critical role in the etiology of breast and prostate cancers.

### YOUR RESULT: T/T

No variant was detected.

## CYP17A1 | -34T>C



CYP17 mediates both steroid 17 $\alpha$ -hydroxylase and 17, 20-lyase activities, and catalyzes a rate-limiting step in ovarian and adrenal biosynthesis leading to the precursor, dehydroepiandrosterone. The C allele increases enzyme activity, thereby increasing the amount of bioavailable estrogen.

### YOUR RESULT: **G/G**

For individuals with the C (G) allele, beneficial modulation of estrogen metabolism can be accomplished through dietary and lifestyle modifications. Key interventions include increasing insoluble fiber, avoiding refined carbohydrates, increasing phytoestrogen intake, losing weight, and increasing exercise. In addition, select nutrients and micronutrients effectively reduce estrogen load by supporting preferred estrogen pathways. These are included at the end of the report.

## CYP1B1 | 4326C>G (Val432Leu)



CYP1B1 enzyme catalyzes the 4-hydroxylation of estradiol; it also oxidizes a variety of procarcinogenic compounds to their activated forms, including polycyclic aromatic hydrocarbons (PAHs) and arylamines.

### YOUR RESULT: **C/G**

This SNP has been found to have the most profound impact on the catalytic properties of CYP1B1, with the 4-hydroxylase activity of the G allele displaying three-fold higher activity compared to the C allele. In the presence of the G allele, it is important to reduce exposure to all diet and environmental procarcinogens such as PAH, aromatic amines, nitrates, and smoking of any kind. In addition, attention should be paid to optimizing phase 2 detoxification.

## GSTM1 | 519G>C



Glutathione S-transferase M1 is the most biologically active member of the GST super-family and is involved in Phase II detoxification in the liver. It is responsible for the removal of xenobiotics, carcinogens, and products of oxidative stress. These enzymes are involved in the conjugation of estrogen quinones to glutathione.

### YOUR RESULT: **indeterminate**

The absence of a genotype at the GSTM1 519G>C indicates that the gene is deleted. This results in the absence of enzyme activity, leading to reduced capacity for hepatic detoxification and reduced metabolism of quinones. GST enzyme activities are induced in part by the products of cruciferous and allium vegetables. These should be increased significantly in the diet to increase activity of other GST enzymes to compensate for decreased or loss of activity. Daily intake is recommended. When dietary intake is inadequate, a high quality supplement containing diindolylmethane (DIM) may be required. We also recommend a diet rich in antioxidants and to avoid exposure to dietary and environmental toxins.



Glutathione S-transferases (GSTs) are a family of multifunctional enzymes involved in the metabolism of a variety of xenobiotic compounds, including mammary carcinogens. These enzymes are involved in the conjugation of estrogen quinones to glutathione.

#### YOUR RESULT: indeterminate

The absence of a genotype at the GSTT1 15G>C indicates that the gene is deleted. A deletion result is an absence of the enzyme, leading to reduced capacity for hepatic detoxification and reduced metabolism of quinones. GST enzyme activities are induced in part by the products of cruciferous and allium vegetables. These should be increased significantly in the diet to increase activity of other GST enzymes to compensate for decreased activity. Daily intake is recommended. When dietary intake is inadequate, a high quality supplement containing diindolylmethane (DIM) may be required. We also recommend a diet rich in antioxidants and to avoid exposure to dietary and environmental toxins.



NADP(H:) quinone oxidoreductase 1 (NQO1) often referred to as Quinone Reductase is primarily involved in the detoxification of potentially mutagenic and carcinogenic quinones derived from tobacco smoke, diet and estrogen metabolism. NQO1 also protects cells from oxidative stress by maintaining the antioxidant forms of ubiquinone and vitamin E.

#### YOUR RESULT: C/C

The analysis identified no genetic variation at the 209 C>T locus.



Sulfotransferase 1A1 (SULT1A1) is involved in the inactivation of estrogens and bio-activation of heterocyclic amines and polycyclic aromatic hydrocarbons.

#### YOUR RESULT: A/A

A allele carriers have a substantially lower activity of this enzyme, which has been associated with increased with higher BMI, and longer exposure to endogenous hormones. For individuals with the A allele, beneficial modulation of estrogen metabolism can be accomplished through dietary and lifestyle modifications. Key interventions include increasing insoluble fiber, avoiding refined carbohydrates, increasing phytoestrogen intake, losing weight, and increasing exercise. In addition, select nutrients and micronutrients effectively reduce estrogen load by supporting preferred estrogen pathways. These are included at the end of the report.



The SOD2 gene encodes the superoxide dismutase 2, a mitochondrial enzyme involved in the elimination of free radicals which are normally produced within cells and which are damaging to biological systems. The enzyme thus has important antioxidant activity within the cell, especially within the mitochondria.

**YOUR RESULT: C/T**

Individuals with the C allele and with a lower consumption of fruits and vegetables may have an increased risk of experiencing the damaging effects of oxidative stress resulting from free radicals. These effects are also enhanced by the presence of additional risk factors such as smoking or the use of hormone replacement therapy. Therefore, it is important for individuals with the C allele to ensure adequate fruit and vegetable intake. Supplementation with antioxidant nutrients can also reduce the oxidation of catechols and promote greater excretion of these metabolites through the methylation pathway.

## NUTRITION AND ESTROGEN

If a moderate or high impact gene variant is present for COMT, SULT1A1 or CYP17A, the following nutritional support is recommended to effectively reduce estrogen load while supporting preferred estrogen pathways:

- For breakdown of estrogen to the beneficial 2-OH metabolite, supplement with a bio-available form of 3,3'-Diindolylmethane (DIM), or substantially increase intake of cruciferous vegetables (cauliflower, broccoli, cabbage, brussels sprouts).
- Include phytoestrogens in the diet for their many beneficial influences on estrogen synthesis and metabolism. These include isoflavones and lignins. Isoflavones are found most commonly in soy products, but also include legumes, alfalfa, clover, licorice root, and kudzu root, and include genistein, daidzein, equol and puerarin. Lignins are an insoluble dietary fiber found in flaxseeds, whole grains, beans and seeds.
- Ensure adequate intake of magnesium and vitamin E.
- Other beneficial micro and phyto-nutrients that impact estrogen metabolism include calcium D-glucarate, curcumin, green tea polyphenols and D-limonene.

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