

**Athlete Report** 

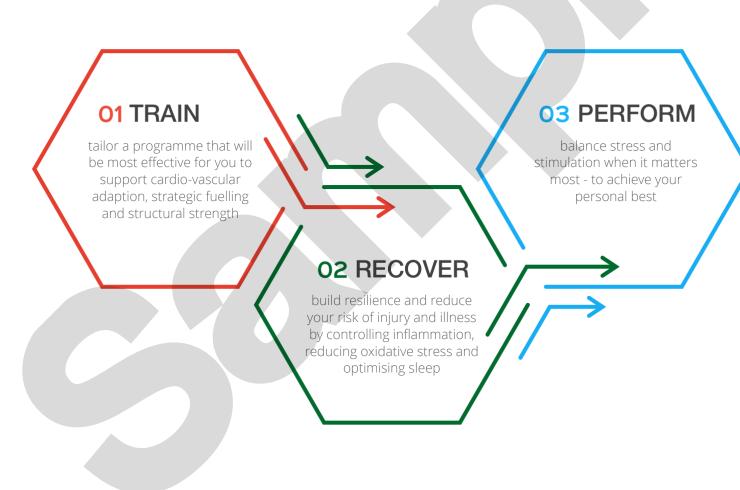


# Welcome to your unique, personalised athlete DNA report!

DNA sequencing has opened the door to personalised approaches to health and fitness, enabling a more intelligent approach to training, recovery and performance. In this report we present elements of your unique DNA profile that have been shown to affect athletic performance.

It is well understood that athletic performance depends on interactions between genetic and environmental factors. Your genes are not your destiny. By aligning your training, nutrition and sleep practices with your genetics you can realise your athletic potential.

Whether you are an elite athlete, a 'weekend warrior', or just want to get healthier, your athlete DNA report will enable you to:



# How to Read Your Report - Your Results in Detail

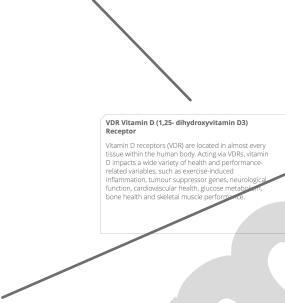
#### Genes

Genes are listed in alphabetical order, by name. The short name (e.g. VDR) is usually the acronym of the long name (e.g. Vitamin D Receptor). The description explains the gene function (purpose).

# **Personalised Result**

The 'rs' number is the reference sequence number that identifies a specific location on the genome. It is also known as a SNP (Single Nucleotide Polymorphism) pronounced 'snip', polymorphism or mutation.

Your genotype result is shown as two letters (A,G,T or C) which represent the DNA bases present at that location.



rs1544410 ▼
Potential lower respo¶se to Vitamin D and increased

To reduce risk of stress fractures and shin splints, and muscle fatigue and screness ensure sufficient Vitamin D intake and UV expo ure. Training should include weight bearing exerci es. Dietary sources of vitamin D include oily fish salm on, mackerel, sardines and tuna, milk and egg yolks.

# **Highlight Colour**

The genotype result highlight indicates the potential effect of the SNP on gene function in a particular context.

**RED** the effect of the variant is negative

**AMBER** the effect of the variant is neutral (no effect) or is subjective

**GREEN** the effect of the variant is positive

# **Arrow Direction**

The direction of the arrow indicates the potential effect of the SNP on gene expression - it can increase or decrease activity, or neither.

- ▲ up-regulates or increases the activity and effect of the gene
- ▼ down-regulates or decreases the activity and effect of the gene

No arrow - no effect on the activity of the gene

# Train



## **Cardiac Output**

ACE ▲

AGT 🛦 🛦

Your cardiac output potential is **elevated**. This can be beneficial for sports that require high-intensity power such as sprinting or weight-lifting. However, high sodium levels and fluid retention can elevate blood pressure.

Monitor and control for high blood pressure. Potassium, magnesium and calcium can moderate the blood pressure raising effects of sodium (salt). Maintain a healthy body weight and exercise regularly.

#### **Blood Circulation**

BDKRB2 ▲▲ VEGFA NOS3 ▼

Your circulation potential is **normal**.

Aerobic exercise will stimulate blood vessel growth and improve circulation of oxygen and energy needed for training. Increasing intake of foods rich in nitric oxide, such as beetroot, can improve blood vessel growth.

# Cardio

The cardiovascular system is essentially made up of two parts - the heart ('cardio') and the blood vessels ('vascular' or 'circulation'). Its primary functions are to transport nutrients, hormones, gases, and waste to and from our cells and to regulate body temperature and fluid balance.

Exercise places a greater demand on cardiovascular functions. As working muscles require more oxygen and nutrients than normal, they produce more waste products and generate more heat. The good news is the body is designed to adapt to these demands. The extent of adaptive response is determined by training, lifestyle, and genetic predisposition.

Although the cardiovascular system is integrated, we present the genes and variants according to their impact on the 'cardio' or 'vascular' components - described in terms of cardiac output and circulation.





## **Cardiac Output**

Cardiac Output is the amount of blood pumped by the heart, measured in litres per minute. It ranges from 5 l/min at rest up to 40 l/min during maximal effort. Cardiac output increases during exercise in order to meet demands for more oxygen and nutrients and removal of waste. It has two components:

- Heart Rate is the number of times the heart beats per minute (bpm). A normal resting heart rate range is between 60-100bpm.
- Stroke Volume is the volume of blood pumped by the left ventricle with each heart beat.

#### **Blood Pressure**

Blood Pressure is a measure of the force being exerted on the walls of arteries as blood is pumped out of the heart. Systolic (the top number) is measured as the heart contracts, and diastolic (the bottom number) as it relaxes between beats. A blood pressure reading of 120 (systolic)/ 80 (diastolic) measured in mmHg is considered normal.

High blood pressure (hypertension) occurs when resting systolic blood pressure readings consistently exceed 140mmHg and/ or diastolic readings exceed 90mmHg. Increases in cardiac output, blood volume, blood viscosity (thickness) or systemic vascular resistance can all increase blood pressure.

Low blood pressure (hypotension) and low heart rate are very common amongst athletes, and are usually a sign of good cardiovascular efficiency.

#### **Cardio Genes**

The **ACE** and **AGT** genes form part of the reninangiotensin-aldosterone system (RAAS) which plays an important role in regulating blood volume and blood pressure.

Variants on the ACE and AGT genes can increase RAAS activity, including levels of angiotensin II. This has multiple potent effects which can contribute to raised blood pressure:

- Constricting (narrowing) blood vessels increasing blood pressure directly.
- Triggering the release of aldosterone, causing the kidneys to retain sodium and water and excrete potassium increasing blood volume.
- Increasing thirst and appetite for salt which can also lead to increased blood volume.

Higher RAAS activity is also associated with greater muscle growth (hypertrophy) and left ventricular size which has been linked with elite status in anaerobic/high-intensity sports.

**Potassium** is an essential nutrient that can counter-balance the blood pressure raising effects of sodium (in salt).

Apricots, bananas and coconut water are good natural sources of potassium and easy to eat on the go!

#### Circulation

The circulatory, or 'vascular' system, is made up of vessels that carry blood and lymph through the body.

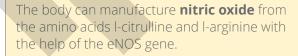
Arteries carry oxygen rich blood away from the heart. The arteries break down into smaller and smaller branches, called capillaries. As blood moves through the capillaries, the oxygen and other nutrients move out into the cells, and waste matter from the cells moves into the capillaries. Blood is then moved from the capillaries to the veins, which carry it back to the heart.

The circulatory system directs blood to where it is needed most. During exercise blood flow is directed away from core systems and organs to skeletal muscle. The body adapts to exercise by growing new blood vessels - this is called 'angiogenesis'.

#### **Circulation Genes**

As its name suggests, vascular endothelial growth factor (coded for by the **VEGFA** gene) promotes the growth of new blood vessels. VEGF also stimulates endothelial nitric oxide synthase (**eNOS**) which supports the production of nitric oxide from l-arginine. Nitric oxide relaxes and dilates blood vessels, thereby increasing blood circulation.

Variants on the **BDKRB2** gene increase sensitivity to bradykinin, lowering RAAS activity and blood pressure and increasing circulation. Athletes with lower RAAS activity and good circulation tend to perform better at endurance sports.



Dietary nitrates can also be turned into nitric oxide via a separate oxygen-independent pathway. Beetroot juice has a particularly high nitrate content.



# Your Results in Detail

## **ACE Angiotensin I Converting Enzyme**

Angiotensin I Converting Enzyme is able to cleave (cut) proteins. By cutting angiotensin I at a particular location it converts it to angiotensin II.

Angiotensin II causes blood vessels to constrict (narrow), raising blood pressure. As a growth factor, it is associated with greater muscle volume and strength which may be beneficial to high intensity (power) oriented sports.

rs4343 AG 🛦

Some increase in ACE activity and angiotensin II levels. Mixed impact - greater cardiac strength (positive) however angiotensin II is associated with water and salt retention which may elevate blood pressure.

Likely to excel at, and benefit from sports that require a mix of endurance (low-medium intensity) and power (high intensity) activity. Monitor blood pressure and ensure adequate potassium intake to balance the effects of sodium.

## **AGT Angiotensinogen**

Angiotensinogen is the primary controller of the reninangiotensin-aldosterone system (RAAS) which regulates blood pressure and the balance of fluids and salts in the body.

Variants are associated with higher activity and more rapid conversion to angiotensin I and II. Although higher angiotensin II is associated with higher blood pressure and sodium retention, it is also linked to increased cardio output which is beneficial for power (high-intensity) performance.



Increased AGT activity and angiotensin II levels. Mixed impact - greater cardiac strength (positive) however water and salt retention may elevate blood pressure (negative).

Likely to excel at, and benefit from, power (high intensity) oriented activities. Monitor blood pressure and ensure adequate potassium intake to balance the effects of sodium.

## **BDKRB2 Bradykinin Receptor Beta 2**

Bradykinin causes blood vessels to dilate (widen), and blood pressure to fall. It also stimulates uptake (storage) of glycogen by skeletal muscle.

Variants on the Bradykinin Receptor Beta 2 gene increase sensitivity to bradykinin, so less bradykinin is needed to stimulate vasodilation, and improve blood circulation.



Increased sensitivity to bradykinin. You need a smaller amount of bradykinin for its effects to be felt.

This genotype is associated with the most efficient circulation and transport of oxygen and other nutrients - beneficial for endurance (low to medium intensity) activities

# **Phosphocreatine**

MTHFR ▼

**MTRR** 

Your potential to generate a lot of energy, quickly, is **reduced**. This may affect your ability to excel at high intensity activities that require short bursts of energy such as sprinting or weightlifting.

To improve your potential, ensure adequate intake of creatine (red meat and fish are good natural sources). Vitamins B9 and B12, magnesium and zinc will help to support methylation.

## **Glycolysis**

ADRB2 ▲▲

Your potential to generate energy anaerobically from glucose is **elevated**. This may be advantageous for speed-oriented activities such as short distance swimming or sprinting.

Excess glucose will be stored as glycogen or fat. To avoid weight gain, limit your intake of simple carbohydrates.

## **Aerobic**

AMPD1 PPARA ▼ PGC1A

Your potential to adapt to using fat for fuelling endurance activities is **good**. This can be beneficial for sports that require sustained energy release such as long distance running or cycling.

Good sources of healthy fat include avocados, nuts, seeds, fatty fish and organically farmed meats.

# Fuel

The body uses a mixture of carbohydrates, fats and proteins as sources of energy. The balance at any point in time depends on intensity of activity, availability of fuel type, and genetics.

Carbohydrates are the favoured fuel source as they are quickly and easily converted, providing immediate energy for high-intensity activities. Fat takes longer to convert and supplies energy to support low to moderate intensity exercise. Protein can provide energy when other sources are scarce.

There are three systems for generating energy, each of which has a different biochemistry and rate of ATP production:

- Phosphocreatine
- Glycolysis
- Aerobic



## **Phosphocreatine**

The phosphocreatine system uses creatine to supply a phosphate group to recycle ADP to ATP and release energy. It is able to generate energy quickly, but only for a short period of time. This is useful for high-intensity activities such as weight training or sprinting.

Creatine synthesis can be impacted by variants on the **MTHFR** and **MTRR** genes and supply of vitamins B9 (folate) and B12. Creatine can also be obtained directly from the diet.

**Creatine synthesis** is heavily dependent on methylation and uses up significant amounts of a substance called SAMe. Variants on the MTHFR and MTRR genes can also impact SAMe levels.

To support methylation you need a good supply of B vitamins including B9 (found in green leafy veg, citrus fruits, nuts and beans) and B12 (found in meat, eggs and fish).

# **Glycolysis**

Glycolysis is the break down of glucose to generate energy. It can be anaerobic or aerobic. Anaerobic glycolysis is the major source of energy during strenuous exercise without the use of oxygen.

Glycogen is broken down into glucose and rapidly converted to ATP. However, as each glucose molecule only produces a small amount of ATP, it is inefficient. After a few minutes of exercise, the body starts to switch over to the aerobic system.

Variants on the **ADRB2** gene can affect the amount of blood glucose available to fuel glycolysis.

## Aerobic

When oxygen is available, the body is able to generate ATP by breaking down carbohydrates and fat aerobically.

Initially, most energy production is fuelled by muscle glycogen. After about two hours of high-intensity exercise, the fuel source switches to carbohydrate and fat (lipolysis). Although fat is energy dense its conversion to ATP is slow compared to carbohydrate.

Variances on **AMPD1**, **PGC1A** and **PPARA** genes affect your aerobic capacity and adaptability to utilise fat as fuel alongside carbohydrate.

**Fat adaption** is the process of training your body to break down fat for energy. As well as improving endurance ability, this can have many health benefits including weight loss, balanced energy levels and cardiovascular health.

Oily fish, avocados, olive oil and coconut oil are good sources of healthy fats.

#### Muscle

ACTN3

Your muscle fibre type is **fast twitch** dominant. This is the 'speed' version of the gene which is beneficial for sports that require quick bursts of power, such as sprinting.

You can increase the size and strength of fast twitch muscle fibres, and power capacity, by regular strength training, however you ability to develop slow twitch muscle fibres may be limited by your genetic make-up.

#### **Connective Tissue**

COL1A1

Normal (healthy) collagen synthesis. Your risk of tendinopathy type injuries is **not elevated**.

Your diet should include collagen rich nutrients, including vitamins C, E, manganese and zinc.

#### Bone

VDR

VKORC1 ▼

Variants in on the Vitamin D Receptor gene may **reduce** your sensitivity to Vitamin D, increasing your need. Variants on VKORC1 may impact your ability to convert Vitamin K to its active form. The body needs vitamin D, vitamin K, calcium and phosphorus to build strong bones.

Your body can make vitamin D directly if you have sufficient exposure to sunlight. Vitamin K is abundant in leafy green vegetables, brussels sprouts, cabbage and broccoli. Weight bearing exercise will also help to improve bone strength.

# Structure

The musculoskeletal system gives the body structure and stability and enables movement. It is made up of bones, skeletal muscle and connective tissues - tendons, ligaments and cartilage.

Whilst genes may determine our physical limits, we all have some potential to alter aspects of our size and strength through physical training and lifestyle choices. Although they are interconnected, we look at genetics affecting the following three components:

- Muscle
- Connective tissue
- Bone



#### Muscle

Skeletal muscles are made up of a combination of two main types of fibre, called slow-twitch (type I) and fast-twitch (type II), which influence how muscles respond to training and physical activity.

Slow-twitch muscle fibres can work for a long time without tiring so are good for endurance activities, like long distance running. Fast-twitch muscle fibres fatigue more quickly but support rapid, bursts of movement needed for power activities, such as sprinting or weightlifting.

Humans are genetically predisposed with a larger percentage of one type of muscle group over another. The **ACTN3** gene has a significant influence on the number of type II (fast twitch fibres).

Although the number of muscle fibres cannot be increased, cells can be induced to grow larger through hormone signalling and strength training, supported by high quality protein intake.

Amino acids, the building blocks of proteins, are needed to build and repair muscle tissue, to increase strength and reduce soreness. Nine of these amino acids are categorised as 'essential', as the body cannot make them. The complete range of amino acids can be sourced in one hit from animal proteins (meat, fish, eggs) or by consuming a variety of plant-based foods.



Collagen is the main structural protein in connective tissue and the most plentiful protein in the body. It is mostly found in fibrous tissues, tendons, ligaments and skin, and in bone. It also constitutes a small proportion of muscle tissue.

There are more than 30 genes associated with synthesis of collagen, each coding for a different type. The **COL1A1** gene codes for type I collagen, the most abundant form, found in tendons, skin, artery walls, muscles, bones and teeth.

Collagen is made from amino acids (principally, glycine and proline) and requires vitamin C as a co-factor. Longterm deficiency in vitamin C results in impaired collagen synthesis and scurvy-like symptoms.

**Vitamin C** is essential for the formation of collagen. Athletes may be at greater risk of vitamin C deficiency due to the wear and tear associated with training.

Good sources include citrus fruits, kiwi, strawberries, broccoli and tomatoes.



#### **Bone**

Bones are made from a matrix of collagen into which bone building minerals - calcium, phosphorus, magnesium and potassium are deposited. Bones are in constant flux, endlessly being broken down (by osteoclasts) and re-created (by osteoblasts).

Weight-bearing exercise combined with sufficient protein, mineral and vitamin intake stimulates bone growth. Vitamins D and K work together to support the absorption and distribution of calcium - increasing bone formation and preventing bone loss. Sensitivity to vitamin D can be impacted by variants on the Vitamin D Receptor (VDR) gene. Variants on the VKORC1 gene can impact your ability to convert vitamin K to its usable form.

Vitamin D

Vitamin D is called the 'Sunshine Vitamin' because the body can make its own vitamin D when skin is exposed to sunlight.

Once in the body, vitamin D is converted to the activated form of vitamin D3 (also called calcitriol, or 25(OH)D)) - which acts via the vitamin D receptor (VDR).

Vitamin D is essential for the maintenance of bone mineralisation through the regulation of calcium and phosphorus levels. Deficiency can increase the risk of lower bone density - broken bones, shin splints and osteoporosis. Other symptoms of vitamin D deficiency include muscle weakness, difficulty thinking clearly and unexplained fatigue.

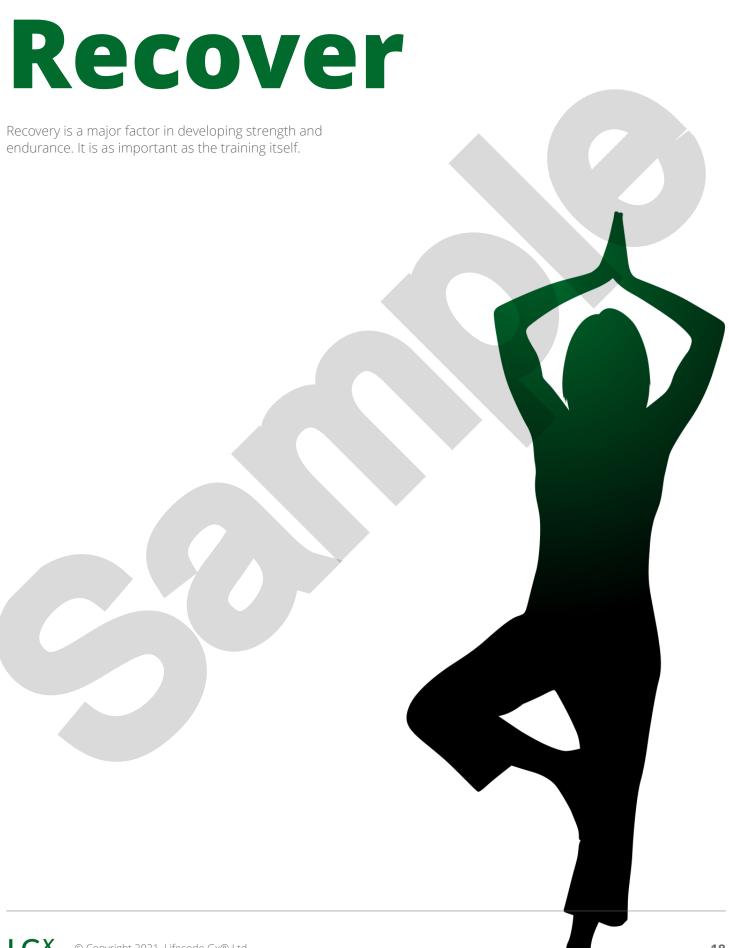
Sun exposure, diet, skin colour, age and body weight, have varied and substantial impacts on vitamin D levels. Although sunlight is the best source, small amounts of vitamin D are available in food - mackerel, salmon, tuna and sardines are particularly good sources.

Vitamin D supplements are widely available, however you can have too much of a good thing. Vitamin D toxicity can cause abnormally high blood concentrations of calcium, leading to over-calcification of bones, soft tissues, heart and kidneys.

Bone is a living organ that is continuously being reshaped. **Vitamin D and vitamin K work together** to support absorption and distribution of the calcium needed to build strong bones.

Vitamin K can be found in dark green, leafy vegetables and brassicas (broccoli, brussels sprouts and cabbage). Oily fish, such as mackerel, sardines, salmon and tuna are good sources of vitamin D.

**A vitamin D blood test** is the only way to categorically determine if you are deficient in vitamin D.



#### **Inflammation**

L6 Ti

Your predisposition to exercise induced inflammation is **reduced**. You may be less susceptible to muscle damage and soreness in response to training and benefit from shorter recovery times.

Ensure adequate recovery times and follow an anti-inflammatory diet including omega 3 fatty acids.

# **Inflammation**

Athletes who don't recover adequately from workouts become vulnerable to injury or illness. To help prevent these problems the body relies on its natural inflammatory and anti-inflammatory mechanisms.

When injured, the body creates natural inflammation around a wound, making it reddish, swollen and hot. This is called acute inflammation, and is the body's way of recovering from the daily wear and tear of physical and chemical stress.

Chronic inflammation, on the other hand, develops when the body is unable to control the inflammatory mechanism. This is often due to nutritional imbalances, stress, overtraining or insufficient sleep.



Although a certain amount of muscle damage is needed to promote adaptions to muscle size, strength and efficiency, excessive damage or inadequate recovery can increase inflammation, muscle fatigue and risk of injury. As we get older we are likely to experience more muscle damage and take longer to recover.

Exercise reduces expression of inflammation-promoting cytokines, such as tumour necrosis factor (**TNF**) and promotes the expression of the anti-inflammatory cytokine interleukin 6 (**IL6**).

Gene variants that influence inflammatory response have been linked to differences in exercise induced muscle damage and recovery times.

C-reactive protein (CRP), which is secreted by the liver, circulates in the bloodstream and is a commonly used **biomarker for inflammation** 

## **Pro-inflammatory**

TNF is a pro-inflammatory cytokine that plays a vital role in the body's immune response. However, over-production of TNF can result in chronic inflammation and autoimmune and immune-mediated disorders.

The inflammatory effects of TNF are inhibited by several natural compounds including curcumin (found in tumeric), catechins (in green tea), omega 3 fatty acids (in oily fish) and echinacea. More recently, exercise has also been shown to blunt the effects of TNF.

## **Anti-inflammatory**

Plasma IL6 levels increase exponentially (up to 100 fold) with exercise depending on intensity and duration, muscle mass, and endurance capacity.

IL6 increases ATP generation, induces lipolysis and fat oxidation and maintains blood sugar homeostasis. It also has strong anti-inflammatory effects including inhibition of TNF activity and has consistently been shown to protect against muscle damage.

Knowing how someone is likely to respond to a particular type of exercise could help practitioners individualise the exercise training of their athletes; maximising recovery and reducing the risk of overtraining.

The Omega-3 fats found in fresh, 'oily' fish such as salmon, sardines and anchovies, have powerful **anti-inflammatory** effects.

Ginger, turmeric, citrus peel and foods in the onion family (shallots, chives and garlic) all have anti-inflammatory effects.

#### **Oxidative Stress**

NOS3 ▼

SOD2 ▼

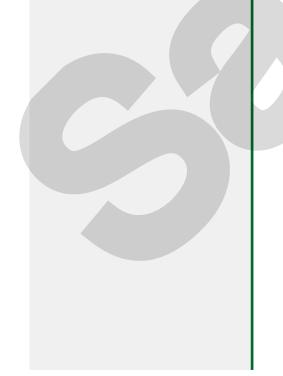
Your genetic predisposition to oxidative stress is **normal**. This means your susceptibility to cellular damage caused by exercise is not elevated.

Antioxidants protect cells against free radicals and help repair cellular damage. Ensure your diet is rich in antioxidants including vitamins A, C and E, found in many fruits, vegetables, nuts and some fish, and meats.

# **Oxidative Stress**

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 anti-oxidation is critical to maintaining healthy biological systems.

Dietary anti-oxidants such as vitamin C, vitamin E, carotenoids and polyphenols are free radical scavengers that interact with free radicals to deactive them. However, the major role in anti-oxidant defence is fulfilled by the body's own 'master antioxidant', glutathione.



Oxidative stress can be managed by addressing both cause and effect.

Causes of oxidative stress can be psychological, emotional and social, as well as physical. Activities like meditation, walking in nature and yoga can help to relieve the sources of some of these stressors.

As an athlete, a certain amount of oxidative stress due to physical activity is inevitable. Individuals who have variants on the Nitric Oxide Synthase (**NOS**) or Superoxide Dismutase (**SOD**) genes may be more susceptible to oxidative stress than others.

Increasing intake of antioxidant foods and herbs will help improve antioxidant status and reduce the effects of oxidative stress. However, the most prolific antioxidant in the body is glutathione, which is made from the amino acid cysteine, with help from the Glutathioine Synthetase (**GSS**) gene.

**Glutathione** is the most prolific antioxidant in the body. It is often referred to as the 'master antioxidant'.

The amino acid, **cysteine** is needed to make glutathione. Cysteine is found in high protein foods, including meat, eggs and dairy, and in lesser amounts in sulphur containing foods, such as broccoli, Brussels sprouts, peppers, onions and garlic.

# Foods High In Antioxidants:

Goji berries, Blueberries, Cranberries, Blackberries, Dark chocolate, Pecan nuts, Coriander, Kidney beans

# **Herbs High In Antioxidants:**

Basil, Cayenne pepper, Cloves, Cinnamon, Cocoa, Garlic, Ginger, Green tea, Oregano, Parsley, Turmeric, Thyme

# Sleep

CLOCK

PER1 ▼

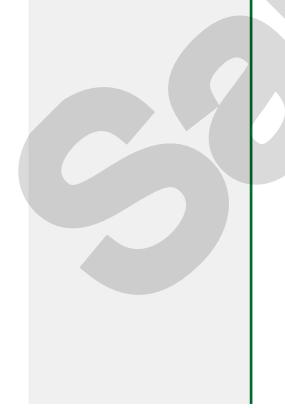
The **in-betweener** may find it easier to adjust their sleep and wake routine to fit their environment.

Try to maintain a regular sleep routine and avoid stimulants and bright light close to bed time.

# Sleep

Sleep is vital to overall health and enabling repair and recovery from exercise. Like other aspects of biology sleep is individual - sleep and wake patters are influenced by genetics.

The term chronotype describes someones sleeping characteristic - if they are naturally a morning person (early bird) or evening person (night owl) or, the most frequent type, an 'in-betweener'. Understanding someones chronotype can enable sleep patterns to be adjusted to optimise recovery and performance.



Genetic testing can indicate which chronotype - 'early-bird', 'night-owl' or 'in-betweener' - an individual is most likely to associate with.

An early birds body clock runs slightly fast, and their circadian rhythm is just short of 24 hours. They find it easier to wake naturally and focus and perform better in the morning. Early birds are less likely to feel tired in the day and go to bed reasonably early.

A night owls body clock runs slightly slow, and their circadian rhythm might be twenty four and a half hours long. They find it difficult to wake and get up early and are more likely to peak around late afternoon or early evening.

The majority of people are 'in-betweeners' with a circadian rhythm just over or just under twenty four hours. Almost no-one has a 'perfect' twenty four hour cycle!

If you know your chronotype you may be able to schedule activities that require most focus and energy to coincide with your 'peak' performance time. Alternatively you can adjust your circadian rhythm by adopting regular sleep and wake times.

Foods that contain tryptophan can help the body make the sleep hormone, melatonin. Good sources include bananas, almonds and turkey.

Having a **regular wake time** is one of the most effective ways of re-training your body clock. If you are tired from getting up early, and exercising, your 'going-tosleep' time, should adjust naturally.



#### **Balance**

HLA-DQA1 ▼ LCT ▲ HTR2A ▲

Your susceptibility to symptoms of GI (gastrointestinal) distress is **elevated**. You can lower the risk of experiencing symptoms by avoiding generic and specific triggers. For some people, foods that contain gluten, dairy or tryptophan (which can raise serotonin levels), may trigger adverse GI responses.

# **Balance**

The last thing you want to affect your performance on the big day is stomach problems, or worse! However, gastrointestinal (GI) symptoms, are common in athletes, and certain individuals or groups (elite athletes, women and endurance athletes) are more likely to be affected than others.

The impact of high-intensity physical activity on gut blood flow, motility and permeability can affect anyone. However, there are some dietary triggers that affect people quite differently and are reliably linked to genetics.



Athletes are more likely to experience symptoms relating to GI (gastrointestinal) damage and distress than non-athletes. Whilst mechanical effects, such as bouncing of organs, are difficult to avoid, habitual aspects, including nutrition and supplementation, can be pro-actively managed.

Genetic variances can significantly impact an individual's response to specific nutrients, or food groups, such as gluten and dairy.

Whilst most babies can digest lactose, the sugar found in milk and other dairy products, a variant near to the **LCT**, lactase, gene determines if you can retain this ability as an adult. If you have the 'wild'/default version of the gene you are unlikely to be able to digest lactose. In this case, the mutated version of the gene is beneficial.

The **HLA** gene variant that can lead to celiac disease is thankfully quite rare. However, many people develop non-celiac gluten sensitivity which means eating gluten, found in wheat and other grains, can cause Gl damage.

Whilst serotonin is commonly known as the 'happiness' neurotransmitter, it can also increase nervousness and anxiety, and gut motility. Serotonin levels can be adjusted by eating tryptophan rich foods such as almonds, white meats and dairy products.

Many brands of **dietary supplements** include buffering agents, or 'fillers', such as sodium bicarbonate which may cause GI irritation with no nutritional benefit.

**Symptoms of GI distress** or injury include, abdominal pain and/or distention, nausea, flatulence, belching, constipation, loose stools, heartburn, acid reflux, hunger pains or stomach rumbling.

#### Non-steroidal anti-inflammatories

(NSAIDs), such as ibuprofen, are amongst the most commonly used drugs worldwide, and their pain-relieving and anti-inflammatory effects are thoroughly accepted. However, they can also cause GI perforation and bleeding. Risk increases with concurrent use of other drugs, including aspirin, alcohol use and H Pylori infection.

# **Caffeine Sensitivity**

ADORA2A 🛦

ADRB2 🗚

**Increased** sensitivity to caffeine. Likely to experience greater adrenergic effects - increases in heart rate and improved mental focus - with relatively low dosage. High caffeine intake may cause feelings of anxiety and jitteriness and negatively affect performance.

#### **Caffeine Metabolism**

CYP1A2 ▲

**Faster** metabolism of caffeine may shorten the duration of its effect.

# Motivate

To optimise performance on competition day you need to be physically and mentally on top form at the moments when it matters most. Many athletes achieve this with the help of caffeine. Caffeine has proven benefits including alertness, reaction times, concentration and endurance capability. It is used extensively in sport as a legal and safe performance booster.

Caffeine acts mainly on the central nervous system, stimulating adrenaline release and the classic 'fight or flight' response; and increasing sensitivity to dopamine. It also has a metabolic effect, increasing energy expenditure. The overall effect of caffeine is reduction of fatigue, and improved ability to tolerate stress and pain.

Now, with the benefit of genetic testing, it is possible to further personalise the use of caffeine.



Genetic testing can enable personalisation of timing and dosage of caffeine to optimise performance. An individual's sensitivity to caffeine can be impacted by which variant of the **ADRB2** and **ADORA2A** genes they carry. Variants on the **CYP1A2** gene will impact the time it takes for caffeine's effects to wear off.

The ADORA2A gene controls the response to adenosine which acts to block the effect of dopamine. By inhibiting ADORA2A, caffeine enables dopamine to exert its effects, such as mood enhancement, better memory, and feelings of pleasure.

When caffeine triggers the release of adrenaline, the adrenaline receptor, ADRB2, stimulates increases in heart rate, blood pressure, muscle strength and blood sugar as a means to get you ready for strenuous activity, or 'flight or fight'.

CYP1A1 is the main deactivator, or detoxifier, of caffeine. Variants on this gene, are reported to increase its activity and metabolise caffeine more quickly.

To get the best results from caffeine, consider environmental aspects - body size, habitual caffeine intake, and timing and duration of the event, alongside genetic analysis.

Adrenaline can increase focus so much that **pain is forgotten**.

Coffee, tea, citrus fruits, bananas, chocolate, cocoa and vanilla can all raise adrenaline levels.

Take care not to overdo it, particularly if you have the more sensitive version of the ADRB2 gene.

# References

#### **ACE** Angiotensin I Converting Enzyme

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#### **ACTN3** Alpha 3 Actinin

Eynon, N., Hanson, E.D., Lucia, A. et al. Genes for Elite Power and Sprint Performance: ACTN3 Leads the Way. Sports Med (2013) 43: 803. doi:10.1007/s40279-013-0059-4 (https://www.ncbi.nlm.nih.gov/pubmed/23681449)

Hogarth M.W., Garton F.C, et al; Analysis of the ACTN3 heterozygous genotype suggests that -actinin-3 controls sarcomeric composition and muscle function in a dose-dependent fashion, Human Molecular Genetics, Volume 25, Issue 5, 1 March 2016, Pages 866-877, https://doi.org/10.1093/hmg/ddv613 (https://www.ncbi.nlm.nih.gov/pubmed/26681802)

Yang N, MacArthur DG, Gulbin JP, et al. ACTN3 Genotype Is Associated with Human Elite Athletic Performance. American Journal of Human Genetics. 2003;73(3):627-631. (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180686/)

#### **ADORA2A** Adenosine A2A Receptor

GoldsteinER, ZiegenfussT, KalmanD, et al. (2010) International Society of Sports Nutrition position stand: caffeine and performance. J Int Soc Sports Nutr 7, 5 (https://jissn.biomedcentral.com/articles/10.1186/1550-2783-7-5)

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#### **ADRB2** Beta-2-Adrenergic Receptor

Association of the ADRB2 Gly16Arg and Glu27Gln polymorphisms with athlete status Marek Sawczuk, Agnieszka Maciejewska-Karlowska, Pawel Cieszczyk, Bogumila Skotarczak & Krzysztof Ficek Journal of Sports Sciences Vol. 31, Iss. 14,2013 (http://www.tandfonline.com/doi/full/10.1080/02640414.2013.786184?src=recsys)

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#### **AGT** Angiotensinogen

Aleksandra Z, Zbigniew J, Waldemar M, et al. The AGT Gene M235T Polymorphism and Response of Power-Related Variables to Aerobic Training. Journal of Sports Science & Medicine. 2016;15(4):616-624. (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5131215/)

#### **AMPD1** Adenosine Monophosphate Deaminase 1

Ginevičienė V, Jakaitienė A, Pranculis A, Milašius K, Tubelis L, Utkus A. AMPD1 rs17602729 is associated with physical performance of sprint and power in elite Lithuanian athletes. BMC Genetics. 2014;15:58. doi:10.1186/1471-2156-15-58. (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4032451/)

#### **BDKRB2** Bradykinin Receptor Beta 2

Associations of polymorphisms of eight muscle- or metabolism-related genes with performance in Mount Olympus marathon runners Georgios I. Tsianos, Evangelos Evangelos, Arnoud Boot, M. Carola Zillikens, Joyce B. J. van Meurs, Andre G. Uitterlinden, John P. A. Ioannidis Journal of Applied Physiology Mar 2010, 108 (3) 567-574; DOI: 10.1152/japplphysiol.00780.2009 (https://www.ncbi.nlm.nih.gov/pubmed/20044476)

Colleen J. Saunders, Stavroulla L. Xenophontos, Marios A. Cariolou, Lakis C. Anastassiades, Timothy D. Noakes, Malcolm Collins; The bradykinin 2 receptor (BDKRB2) and endothelial nitric oxide synthase 3 (NOS3) genes and endurance performance during Ironman Triathlons. Hum Mol Genet 2006; 15 (6): 979-987. doi: 10.1093/hmg/ddl014 (https://academic.oup.com/hmg/article-lookup/doi/10.1093/hmg/ddl014)

#### **CLOCK** Circadian Locomotor Output Cycles Kaput

Micó V., Díez-Ricote L., and Daimiel L. (2016). Nutrigenetics and Nutrimiromics of the Circadian System: The Time for Human Health, Int J Mol Sci.; 17(3): pp. 299 (http://europepmc.org/articles/PMC4813163)

Sleep, The Myth of 8 Hours, the Power of Naps... and the New Plan to Recharge Your Body and Mind by Nick Littlehales. Penguin Life Published 27th October 2016 (https://www.penguin.co.uk/books/291128/sleep/9780241975985/)

Zhang L., Ptáček LJ., and Fu YH. (2013). Diversity of Human Clock Genotypes and Consequences. Progress in Molecular Biology and Translational Science; Volume 119, pp. 51–8 (http://europepmc.org/articles/PMC4169291#R78)

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