NUTRIGENETICS

MZ-MEDICAL

science with humanity

Precision medicine is at the heart of medicine of the future. Nutrigenetics, a significant strand of precision medicine, focuses on how individual gene variants influence responses to diet.

Nutrigenetic tests can help you to determine your individual needs and consequently adapt your nutrition and lifestyle not only to prevent the development of a disease, but and most importantly, to enhance your wellbeing.

The onset of numerous health conditions, such as food intolerance, weight gain, abnormal cholesterol levels or sub-optimal vitamin values are influenced by genetic factors. Fatigue, depression, insomnia, bloating and abdominal pain can be prevented or improved by nutritional advice based on our individual genetic information.

We aim to empower patients by transforming care from being reactive to being preventive. Waiting until we are sick before acting is an option, but taking a preventive, predictable and participative approach, prioritising our wellbeing may be the option which will allow us to enjoy a better quality of life.

The best investment is in our health. I know it may not always be an easy journey, but "no one is ever defeated until defeat has been accepted as a reality". I look forward to meeting you and working together,

Dr Maria Zalazar
MZ-Medical
Founder and Medical Director

NUTRIGENETIC INTRODUCTION

Today genetic tests enable us to take preventative action by adopting lifestyle changes and personalised nutrition to optimise long-term health.

Some of the genetic variants which can be analysed aim to investigate the genetic predisposition for:

PRIMARY LACTOSE INTOLERANCE

Primary lactase deficiency is the most common cause of lactose intolerance worldwide. Undiagnosed and untreated, these conditions can increase the risk of developing other health problems such as osteoporosis.

Primary Lactose Intolerance is caused by a genetically programmed loss in lactase production, commonly after 5 or 6 years of age.

However, some adults (TT and AA genotypes) have detectable levels of lactase at the surface of the intestinal mucosa while others (CC genotype) have undetectable levels, correlating with lactose tolerance and intolerance respectively.

Genetic tests can help to determine who is predisposed to vitamin D deficiency and should therefore supplement their diet, and those not affected avoid supplementation because excessive intake of vitamin D may be dangerous.

Vitamin D is a fat-soluble vitamin involved in bone formation, immunity, neurological and cardiovascular functions, whose absorption and synthesis are determined by genetic factors.

Vitamin D comes in two dietary forms:

- D3 (cholecalciferol) can be either synthetised in the skin in response to UV light from the sun or absorbed from certain animal-based foods such as fatty fish (mackerel, salmon, tuna, sardines, fish liver oils) and eggs.

- D2 (ergocalciferol) is obtained from plant-based foods.

Both forms need to be metabolized first to the active form, 1,25-hydroxyvitamin D (1,25(OH)2D) and then carried to the target tissues.

LOW VITAMIN D LEVELS

Vitamin D deficiency has been associated with an increased risk of numerous diseases including impaired immune function, osteoporosis, breast and colorectal cancer, all health conditions that can also affect middle-aged women.

Genetic variants associated with low blood levels of 25(OH)D have been found in different genes, including CYP2R1, CYP27B1, and the vitamin D receptor gene (VDR), although the best studied gene is the GC gene, which codes for the vitamin D binding protein (DBP). DBP is the main carrier of vitamin D metabolites in the bloodstream, and it may also serve as a reservoir of vitamin D in periods when intake and synthesis are low. Variance in this gene can have a negative impact on bone formation and lead to other health problems.

VITAMIN D SELF-ADMINISTRATION

Vitamin D self-administration, usually in high doses and for a prolonged time, can cause adverse effects, such as hypercalcemia and hypercalciuria.

COELIAC DISEASE

Why is a genetic test a helpful tool in the context of Coeliac Disease?

Because blood tests or biopsies do not always show a disease state if you remove gluten from your diet. Therefore, those studies are only useful in patients on a gluten-containing diet.

Because some patients suffer from minor symptoms such as fatigue or decreased quality of life and the disease can only be recognized after the introduction of a gluten free diet and a positive genetic test.

Because the clinical manifestation can vary significantly between patients, from no evident symptoms to a wide spectrum of them. For instance, some patients experience fatigue and clear gastrointestinal symptoms, while others have severe nutritional deficiencies (anaemia), thyroid dysfunction, cutaneous or neurological symptoms but no gastrointestinal symptoms.

Because physical signs of malabsorption such as weight loss or oedema (secondary to hypoalbuminemia) can be present or not.

Because some patients have laboratory signs such as iron deficiency anaemia or abnormal liver test, or enamel defects or osteoporosis, but no gastro-intenstinal symptoms. And a genetic test can help us to suspect coeliac disease.

Coeliac disease is a chronic small intestinal inmune-mediated enteropathy precipitated by exposure to dietary gluten in genetically predisposed individuals.

Both gene markers HLA-DQ2 and HLA-DQ8 are major risk factors carried by almost all coeliac disease patients.

ALCOHOL DEPENDENCE

Genetic tests may have the potential to identify women at risk of alcohol dependence and help guide therapy.

There is evidence that breast cancer risk is higher in women consuming even low levels of alcohol compared with non-drinkers.

Alcohol consumption is also associated with increased risk of hip fracture due to both osteoporosis and a greater likelihood of falls.

Metabolism of alcohol involves two enzymes (alcohol dehydrogenase and aldehyde dehydrogenase), which are encoded by specific genes. Some people can have genetic variations in these genes, increasing the risk of developing alcohol dependency and alcoholism. Gene-editing approaches have being considered discussed in order to treat alcoholism caused by genetic defects.

CAFFEINE SENSITIVITY

Caffeine represents a particularly interesting biological component with respect to women, given that enzymes involved in its metabolism also play a role in oestrogen metabolism and may alter circulating levels of these hormones.

It has also been shown that the prevalence of urinary incontinence in postmenopausal women can increase with higher caffeine consumption.

Furthermore, excessive consumption of caffeine exerts negative effects on bone health and may increase the risk of hip fracture.

The above examples demonstrate the importance of studying the presence or absence of a genetic predisposition to caffeine sensitivity in women.

Caffeine increases alertness and focus, and reduces fatigue but can also lead to anxiety and sleep disruption.
Both symptoms are also common in middle-aged women.

Caffeine is an alkaloid compound widely found in coffee, tea, soft drinks (including energy drinks) and pharmaceutical products. Its effects vary mainly due to genetic differences in the enzyme involved in its metabolism (CYP1A2).

These differences make some people metabolise caffeine more slowly or rapidly. For example, the AA genotype can tolerate higher caffeine intake without experiencing immediate side effects due to a fast metabolism.

DIETARY Recommendation

As well as considering amount and timing, a genetic test and assessment of bone health can help you enjoy the benefits of caffeine while avoiding some of its adverse effects.

WEIGHT GAIN AND INCREASED APPETITE

Obesity is a risk factor for oestrogen receptor-positive (ER+) breast cancer occurring after menopause, and negatively affects prognosis independently of menopausal status. Importantly, obesity not only increases the risk of breast cancer, but the risk of its recurrence and cancer-associated death as well.

Variants in genes involved in food intake like FTO and leptin gene receptors can increase obesity risk by altering how the body metabolises food.

Genetic variants in the fat mass and obesity-associated (FTO) gene are associated with higher BMI and waist/hip circumference and total cholesterol, triglycerides and fasting glucose levels.

The protein encoded by FTO is an enzyme which repairs DNA and RNA. This protein is highly expressed in the hypothalamus and the pituitary gland, key areas for regulation of energy balance. The FTO has three genotypes.

Both A/A and A/T allele carriers are predisposed to having more body adiposity than those with T/T alleles, which is likely due to increased food intake and hunger, and lower satiety and energy expenditure. It was also found that A allele carriers have elevated cholesterol and triglycerides.

Higher levels of leptin, a hormone synthesised in fat tissues that regulates food intake and energy expenditure are associated with increased fat storage and reduced brain effectiveness in controlling hunger cues and food intake. While these unhealthy serum levels of leptin have been reported in A allele carriers, TT genotype carriers seem to have a decreased risk of developing obesity.

We all know the advice that is often given such as following a Mediterranean diet, reducing the consumption of saturated fats or doing more exercise but here we show that there are genetic factors behind certain conditions and that genetic information can be used to inform evidence-based targeted nutrition preventive recommendations.

DIETARY RECOMMENDATIONS

Following a Mediterranean diet may work for some people but not for others.

Weight loss can be significantly lower with the presence of an A allele of rs9939609 in the FTO gene compared to those homozygous for the T allele. Therefore, following a mediterranean diet will not be completely appropriate nutritional advice for those carrying an A allele.

Limiting overall intake of food, especially saturated fats, and increasing physical activity are general recommendations that can help people with FTO variants to reduce the risk of obesity.

Where reduced sensitivity to leptin exists, this may be overcome by increasing the consumption of oily fish due to the high levels of omega 3 fatty acids it contains.

DIRECT-TO-CONSUMER GENETIC TESTING

Direct-to-Consumer Genetic Testing (DTC-GT) is a method of providing health-related genetic tests directly to consumers without the involvement and assessment of a doctor.

DISADVANTAGES

Unlike clinical genetic tests, DTC tests are not diagnostic, offering risk information for only a limited set of conditions and may include false positive and false negative findings.

False positive test results indicate incorrectly that a certain genetic variant exists while false negative findings incorrectly indicate that a certain genetic variant does not exist.

In addition, these tests are not able to provide a clinical diagnosis and cannot exclude other causal factors which may be affecting our health and wellbeing. For instance, abdominal bloating can be a symptom of lactose intolerance (where a genetic test may be useful) but it can also be a sign of ovarian or colon cancer. Here, a positive DTC-GT may lead to ignoring the warning signs of a health condition which needs urgent attention to increase the chances of a positive outcome.

ADVANTAGES

DTC-GT can provide genetic information to individuals not tested due to circumstances such as, but not limited, poor insurance coverage, prohibitive cost, lack of family history of disease or inaccessibility of clinical genetic testing.

These tests can inform you about the risk of monogenic developing disorders, such as intolerance and sensitivity to foods (e.g. caffeine and alcohol), weight gain or obesity (e.g. FTO), as well as vitamin and mineral requirements (e.g. vitamin D metabolism).

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Personalised hormone replacement therapy & health programmes to support middle-aged women.

LET'S GET STARTED

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