# INFORMATION LEAFLET

## Genetic tests for functional biology

**This leaflet is designed to help you understand the genetic tests offered by the laboratory before you sign the consent form.** It covers the FUT2, DIO-2, MTHFR, COMT, LCT, alpha-1 amylase and APOE tests. Please take the time to read it. If you have any unanswered questions, please ask the healthcare professional who recommended the test or contact the laboratory (contact details at the end of this leaflet).

## 1. WHAT IS A GENETIC TEST ?

Each of our cells contains DNA, which carries our genes. From one person to another, certain genes exhibit small differences, known as variants or polymorphisms. Most of these are common and have no impact on health; some slightly alter the functioning of an enzyme or a protein.

The tests on offer look for a few specific variants in a small number of genes. They do not analyse your entire DNA and therefore cannot reveal information about genes other than those you have chosen.

These variants are present from birth and do not change over the course of a person's life. A test carried out once remains valid: there is no need to repeat it.

## 2. WHAT THESE TESTS CAN TELL YOU, AND WHAT THEY CANNOT

- **A variant is not a disease.** Most people who carry the variants studied here are in good health.
- **A result indicates a tendency, not a diagnosis.** Diet, lifestyle, the environment and many other genes matter just as much, and often more, than the variant being studied.
- **A result showing no variants does not guarantee the absence of any problems :** only the variants being tested for are analysed.
- **Our understanding is evolving.** For several of these variants, research is still limited or contradictory. We make this clear for each test.
- **Your background matters.** These variants have mainly been studied in people of European descent. For other backgrounds, the interpretation may be less accurate.
- **A result is never sufficient on its own to change a course of treatment.** Discuss this with your doctor before making any changes.

## 3. TESTS OFFERED

### FUT2 Secretor status

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| **What is analysed** | The main variant of the FUT2 gene (rs601338). This gene encodes an enzyme that enables certain sugars (blood group antigens) to be attached to secretions and the intestinal mucosa. |
| **What the result indicates** | Two copies of the variant correspond to 'non-secretor' status, which affects around one in five people in Europe. This status is associated with differences in the composition of the gut microbiota, reduced susceptibility to certain strains of norovirus and, on average, higher blood levels of vitamin B12. |
| **Its limitations** | These associations are observed at the level of groups of people. They do not allow us to predict the situation of any one individual. |

### DIO-2 Activation of thyroid hormones

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| **What is analysed** | The Thr92Ala (rs225014) variant of the DIO2 gene. This gene encodes an enzyme which, in certain tissues, converts the thyroid hormone T4 into its active form, T3. |
| **What the result indicates** | In some studies, this variant has been associated with reduced enzyme activity and, in people treated with levothyroxine, with persistent symptoms or a different response to treatments combining T4 and T3. |
| **Its limitations** | Study results are inconsistent. The test does not provide information on how well your thyroid is functioning; this is assessed via a blood test (TSH, free T4, free T3). |

## MTHFR Folate metabolism

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| **What is analysed** | Two variants of the MTHFR gene: C677T and A1298C. This gene encodes an enzyme in the folate cycle (= vitamin B9), which plays a key role in the elimination of homocysteine. |
| **What the result indicates** | Two copies of the C677T variant significantly reduce the enzyme's activity and may be associated with higher homocysteine levels, particularly when intake of methylated folates is insufficient. The effect of the A1298C variant is less pronounced. |
| **Its limitations** | These variants are very common: approximately one in ten people in Europe carries two copies of the C677T variant.<br>Medical genetics societies do not consider testing for these variants to be useful for assessing the risk of thrombosis or recurrent miscarriages. A blood test to measure homocysteine levels provides a better indication of your actual situation. |

## COMT Breakdown of catecholamines

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| **What is analysed** | The Val158Met (rs4680) variant of the COMT gene. This gene encodes an enzyme that breaks down dopamine, noradrenaline, adrenaline and certain oestrogen derivatives. |
| **What the result indicates** | The Met form of the enzyme is three to four times less active than the Val form. This variant has been studied in relation to stress response, pain sensitivity and certain cognitive functions. |
| **Its limitations** | The observed effects are small and vary from one study to another. The result cannot be used to predict behaviour or a disorder. |

## LCT Persistence of lactase

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| **What is analysed** | The -13910 C>T variant (rs4988235), located upstream of the LCT gene. In people of European descent, this variant determines whether the production of lactase – the enzyme that digests milk sugar – is maintained into adulthood. |
| **What the result indicates** | The CC genotype corresponds to a programmed decline in lactase after childhood, which may lead to lactose intolerance. The CT and TT genotypes correspond to lactase that remains active in adulthood. |
| **Its limitations** | A CC genotype does not necessarily cause symptoms: the amount of lactose consumed and the gut microbiota play a role. The test does not detect a reduction in lactase caused by a bowel condition.<br>In people of African, Middle Eastern or Asian descent, other variants may maintain lactase activity; a CC result is therefore less informative. |

## Amylase alpha A1 Starch digestion

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| **What is analysed** | A variant indicative of the number of copies of the AMY1 gene, which codes for salivary amylase, the enzyme that initiates starch digestion in the mouth. This number varies from 2 to more than 15 copies, depending on the individual. |
| **What the result indicates** | The higher the number of copies, the greater the amount of amylase in the saliva tends to be. |
| **Its limitations** | A link with weight and obesity had been reported, but subsequent large-scale studies have not confirmed this or have found only a modest effect. The result cannot be used to predict either weight gain or an individual's tolerance to starchy foods. |

## 4. THE APOE TEST: A SPECIAL TEST

**This test can only be requested by a doctor. The results are provided to you by that doctor and are not published on the patient cyberlab.**

### APOE Apolipoprotein E

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| **What is analysed** | Two variants of the APOE gene, which define three forms: ε2, ε3 and ε4. Each person carries two of these, for example ε3/ε3 or ε3/ε4. |
| **What the result indicates** | The APOE protein is involved in the transport of cholesterol. The genotype provides information on certain blood lipid disorders, particularly in the case of the ε2/ε2 genotype.<br>The ε4 allele is the main known genetic risk factor for late-onset Alzheimer's disease. Compared with individuals with the ε3/ε3 genotype, the risk is approximately three times higher with one ε4 allele, and approximately 10 to 15 times higher with two ε4 alleles, depending on the studies and populations. |
| **Its limitations** | Many people who carry the ε4 allele, even two copies, will never develop the disease, and many people with the disease do not carry it. This test is not a diagnosis. There is currently no cure for Alzheimer's disease. |

### Before taking this test

- Take some time to think about what you would do with the result and how you would cope with it.
- You have the right not to know. You can withdraw from the test at any time before the sample is analysed.
- You can request a genetic counselling session, either before or after the test. Your doctor can refer you for this.
- This result also affects your parents, siblings and children. Talk to your doctor about whether or not to share this information.

## 5. YOUR RESULTS AND YOUR FAMILY

You share some of your genes with your family members. A result may therefore also apply to them. It is entirely up to you whether or not to discuss this with them. The laboratory will never disclose your results to a family member without your written consent.

## 6. HOW IS THE TEST CARRIED OUT ?

- **The sample collection.** This involves taking a swab from the inside of your cheek, to be carried out according to the kit's instructions. Do not eat, drink, smoke or brush your teeth for 30 minutes beforehand.
- **The form.** Enclose the test request form and the completed and signed consent form with the sample. The test will not be carried out without a signed consent form.
- **Turnaround time.** Results are available in approximately 3 weeks.
- **Communication of results.** This follows the rules set out in your consent form: if the tests were requested by a healthcare professional, they will receive and communicate the results to you; in all other cases, they will be made available to you via the patient cyberlab.
- **Your questions.** If you requested the test directly from the laboratory, you can arrange a telephone consultation with a biologist to understand your results.

## 7. CONFIDENTIALITY, DATA RETENTION AND YOUR RIGHTS

- **Vos Sensitive personal data.** Your genetic data is health data, protected by the General Data Protection Regulation (GDPR) and by professional confidentiality. Only those who need it to carry out and validate the analysis have access to it.
- **Insurance.** In several countries, including Belgium and France, the law prohibits insurers from requesting or using the results of genetic tests. Please check the legislation in your country.
- **Your sample.** It is used solely for the tests you have requested. Unless you give your consent, the remainder of the sample and the extracted DNA are destroyed no later than three months after the result has been validated.
- **The remaining sample, if you agree.** The consent form offers you two optional choices: the coded storage of the remaining sample for up to 5 years to ensure the quality of the analyses, and the coded use of your results for internal statistics. The remaining sample is never sold or transferred. Refusing has no impact on your test.
- **Your rights.** You may withdraw your consent to the test before analysis takes place, and your consent regarding the remaining sample at any time, by writing to info@mbnext.be. Your rights of access and rectification, and the contact details of the Data Protection Officer, can be found at https://lims-mbnext.be/privacy-policy/.

## 8. CONTACT US

**Medical Secretariat (Louvain-la-Neuve) :** info@mbnext.be, +32 10 87 09 70.

Monday to Friday from 8.00 am to 9.30 pm, Saturday from 9.00 am to 6.30 pm.

**Website :** lims-mbnext.eu