Age-Related Macular Degeneration – AMD

Age-related macular degeneration (AMD) is characterized by pathological changes of the retinal pigment epithelium (RPE), progressive degeneration of photoreceptors, thickened Bruch's membrane and choroidal neovascularization. These alterations lead to the loss of sharp, central vision. It is an age-related process and usually develops after a person reaches 50 years.

In Western Europe and USA 30% of people older than 75 years suffer from different types of AMD. 85-90% cases of AMD are dry AMD, which have no treatment. 10-15% cases of AMD are wet AMD, which have number of treatments available (injection into the eye to stop further development) and early diagnosis can save vision.

AMD increased risk assessment enables prevention and early diagnosis of the disease. The early diagnosis is vital to delay progression of disease and vision loss.

Indications for genetic testing:

1. Risk determination of at-risk individuals for early diagnosis and prediction of disease progression
2. Risk assessment of individuals with family history of AMD
3. Genetic counseling

Alzheimer Disease, Recovery from Traumatic Brain Injury, Coronary Heart Disease

Apolipoprotein E (ApoE) function is to transport cholesterol and other lipids in blood circulation and the central nervous system. ApoE is essential to maintain the structural integrity of **lipoproteins**, stabilize and solubilize lipoproteins in the blood, and to serve as cofactor in enzymatic reaction.

ApoE mediates **cholesterol metabolism** in an isoform dependent manner. The human ApoE gene exists as three polymorphic alleles – isoforms ε2, ε3 and ε4. Differences between the three ApoE isoforms are limited to amino acid residues 112 and 158, where either cysteine or arginine is present. The single amino acid differences at these positions affect the structure of ApoE isoforms and influence their ability to bind lipids, receptors and amyloid-B peptides in brain.

Patients with the ApoE4 genotype recover from **brain injury** much more slowly, have an increased danger of **postoperative complications** in cognitive functions, and also have a much worse general prognosis. The ApoE4 genotype is also associated with an elevated risk of late onset **Alzheimer's disease** and **coronary heart disease**.

Indications for genetic testing:

1. Risk assessment for Alzheimer disease
2. Determination of prognosis for recovery of the brain injury
3. Risk assessment for
3. Genetic counseling

Athletic performance test of strength, speed and endurance

Nowadays it is possible to investigate an individual's genetic prerequisite to aspects of athletic**strength, speed and endurance**.

This genetic test has been especially designed for the serious sports enthusiast to study two genes that are connected with athletic performance: **ACE** and **ACTN3**. ACE influences cardiovascular performance and adaptation to physical strain, ACTN3 regulates muscle performance.

Identification of ACE and ACTN3 gene variants allows to determine whether the individual is successful in **sprint and strength sports** (sprint distances, hurling and throwing sport, bodybuilding etc), **endurance sports** (running, skiing, rowing, triathlon etc) or whether the person is a so-called**universal sportsman**.

In addition to genetic factors, success of a sportsman depends on several factors as nutrition, living and training conditions, psychological factors.

It is easier to choose the most suitable sport for you, if you are aware of your hereditary prerequisites. The best results are achieved by those who does not only have favourable environment conditions and sufficient motivation, but also has the most suitable gene set for the specific sport.

argeted mutation analysis enables testing of 2 variants in genes **ACE**, **ACTN3** associated with athletic performance.

Turnaround Time:
10 working days

Specimen Requirements:
Buccal cell sample
The sample collection kit and instructions for the sample collection will be sent by Asper Biotech.

Celiac Disease

Celiac disease (CD) is chronic **gluten-intolerance** that primarily affects small intestine in genetically predisposed individuals. Complaints are resolved by exclusion of gluten from diet. CD is characterized by nutrient malabsorption resulting from inflammatory injury to the mucosa of the small intestine after ingestion of wheat gluten or similar proteins in rye, barley and triticale (hybrid of wheat and rye). In contrast rice, buckwheat, millet and corn have generally been considered safe grains for celiac patients. Oats can be introduced into the diet of most people with CD (>95%) but only if oats are confirmed to be uncontaminated by other gluten-containing grains.

The prevalence of CD is estimated at about 1:100 in Caucasian population. Although CD has long been considered a pediatric syndrome, it has been increasingly diagnosed in older children and adults.

CD has a multifactorial inheritance. Pathogenesis of CD is caused by a combination of environmental factors, immunologic factors and variations in multiple genes. It has been shown that viral infections altering intestinal permeability, gut microbiota, breast-feeding and timing of gluten introduction in infant diet influence development of celiac disease. The most important genetic risk factors for CD are presence of **HLA-DQ heterodimers DQ2** (encoded by alleles A1*05 and B1*02) and **DQ8** (encoded by alleles A1*03 and B1*0302).

> Celiac Disease – read more

Indications for testing:

1. Testing patients with symptoms, such as chronic diarrhea with weight loss, steatorrhea, postprandial abdominal pain, and bloating
2. Testing seronegative patients with equivocal small-bowel histological finding
3. Evaluation of patients on a gluten free diet (GFD) who have not been tested for CD before GFD
4. Testing patients with discrepant celiac-specific serology and histology
5. Screening of risk groups (e. g. first-degree relatives, specific syndromes and diseases)

Lactose Intolerance

Lactose intolerance is a widespread metabolic disorder caused by the inability to digest lactose due to a shortage of the lactase enzyme. The typical symptoms of lactose intolerance (inactive lactase enzyme) include abdominal pain, bloating, flatus, diarrhea, and occasionally nausea and vomiting.

Indications for genetic testing:

1. Confirmation of clinical diagnosis
2. Differentiation between primary and secondary hypolactasia

RFLP (restriction fragment length polymorphism) based test detects the variation (-13910C>T) near the **LCT gene** which is associated with **lactose intolerance**.

The test is available with diagnostic package service (includes DNA extraction, genotyping, interpretation, hard copy of the results report).

Turnaround Time:
Diagnostic package service
1-2 weeks

Specimen Requirements:
2-4 ml of blood with anticoagulant EDTA
Send blood samples at room temperature. Blood samples can be preserved at 2-8°C before shipping. Blood samples are recommended not to freeze and not to store longer than one week.

200 ng DNA in TE, AE or pure sterile water at 100-250 ng/µl
Send DNA samples at room temperature or frozen. To avoid sample loss and contamination, please use 0,5-2,0 ml screw cap tubes, tubes with safe lock lid or wrap the caps of each microtube with parafilm.

The A260/A280 ratio should be 1.8-2.0. DNA sample should be run on an agarose gel as a single band, showing no degradation, alongside with a quantitative DNA marker.

2 ml of saliva
Preferred saliva collection kits are Oragene®DNA and SalivaGene Collection Module II.

Nutri inCode

Nutri inCode test studies and integrates genetic and clinical data, as well as information about individual's lifestyle in order to estimate the **predisposition to obesity** and **related diseases**.**Osteoporosis, diabetes** and **hypertension** are among other conditions, which risks are assessed with the test.

The test also enables personal evaluation of factors that influence the prevention or treatment of obesity, such as appetite regulation, caloric expenditure, expected response to hypocaloric diet and exercise.

In addition to detected genetic variants and risk scores, the results report includes nutritional and lifestyle recommendations adapted to the person's genetic profile.

Nutri inCode test details

Nutri inCode test analyses 88 genetic variations in more than 60 genes related to different **nutritional aspects**. The test is performed in the laboratory of Ferrer inCode.

The test is available with **diagnostic package service** (includes DNA extraction, genotyping, interpretation).

Turnaround Time:
4-6 weeks

Specimen Requirements:
2-4 ml of blood with anticoagulant EDTA
Send blood samples at room temperature. Blood samples can be preserved at 2-8°C before shipping. Blood samples are recommended not to freeze and not to store longer than one week.

1 µg DNA in TE, AE or pure sterile water at 100-250 ng/µl
Send DNA samples at room temperature or frozen. To avoid sample loss and contamination, please use 0,5-2,0 ml screw cap tubes, tubes with safe lock lid or wrap the caps of each microtube with parafilm.

The A260/A280 ratio should be 1.8-2.0. DNA sample should be run on an agarose gel as a single band, showing no degradation, alongside with a quantitative DNA marker.

Resistance to norovirus infections

Noroviruses are RNA viruses of the Caliciviridae family, and are also called “Norwalk-like” viruses. This virus causes approximately 90% of non-bacterial epidemic outbreaks of gastroenteritis around the world. Norovirus affects people of all ages. About 20% of the population is resistant to most norovirus infections. Such individuals are called non-secretors. Several polymorphisms in the FUT2 gene are associated with non-secretor status and resistance to norovirus infection.

Indications for genetic testing:

1. Determination of resistance status to infection by the most common type of norovirus

Resistance to norovirus infections test details

RFLP (restriction fragment length polymorphism) based test detects the SNP (rs601338) in the **FUT2 gene**.

The test is available with genotyping service (includes genotyping, electronical copy of the results report) only.

Turnaround Time:
Genotyping service
1-2 weeks

Specimen Requirements:
2-4 ml of blood with anticoagulant EDTA
Send blood samples at room temperature. Blood samples can be preserved at 2-8°C before shipping. Blood samples are recommended not to freeze and not to store longer than one week.

200 ng DNA in TE, AE or pure sterile water at 100-250 ng/µl
Send DNA samples at room temperature or frozen. To avoid sample loss and contamination, please use 0,5-2,0 ml screw cap tubes, tubes with safe lock lid or wrap the caps of each microtube with parafilm.

The A260/A280 ratio should be 1.8-2.0. DNA sample should be run on an agarose gel as a single band, showing no degradation, alongside with a quantitative DNA marker.

2 ml of saliva
Preferred saliva collection kits are Oragene®DNA and SalivaGene Collection Module II.

Venous Thrombosis

The annual incidence of venous thrombosis ranges from approximately 1 to 3 per 1000 people. Venous thrombosis events often occur when multiple risk factors, including genetic and environmental, are present simultaneously. Acquired risk factors of venous thrombosis are age, immobilization, surgery, trauma, malignancy, myeloproliferative disorders, obesity, pregnancy, postpartum period, hormone replacement therapy or use of oral contraceptives. Genetic risk factors are related to a 30- to 80-fold higher risk for developing a thrombotic episode.

Indications for genetic testing:

1. Vein thrombosis before the age of 50
2. Recurrent vein thrombosis in family
3. Identified genetic variant for higher risk of venous thrombosis in family
   For women in addition to the above named:
4. Myocardial infarction in 50-year-old women who are smoking
5. Vein thrombosis in the period of taking oral contraceptives
6. The presence of pregnancy complications, for example multiple miscarriages, preeclampsia and stillbirth.

Venous Thrombosis test details

RFLP-based test is carried out to determine the predisposing genetic factors for thrombosis such as the **factor V Leiden mutation**, the **prothrombin G20210A mutation**, and mutations **C677T** and**A1298C** of the **MTHFR gene**.

The test is available with diagnostic package service (includes DNA extraction, genotyping, interpretation, hard copy of the results report).

Turnaround Time:
Diagnostic package service
Standard: 1-2 weeks

Specimen Requirements:
2-4 ml of blood with anticoagulant EDTA
Send blood samples at room temperature. Blood samples can be preserved at 2-8°C before shipping. Blood samples are recommended not to freeze and not to store longer than one week.

300 ng DNA in TE, AE or pure sterile water at 100-250 ng/µl
Send DNA samples at room temperature or frozen. To avoid sample loss and contamination, please use 0,5-2,0 ml screw cap tubes, tubes with safe lock lid or wrap the caps of each microtube with parafilm.

The A260/A280 ratio should be 1.8-2.0. DNA sample should be run on an agarose gel as a single band, showing no degradation, alongside with a quantitative DNA marker.