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Molecular biology

Sample origin: Saliva

Genetic polymorphisms associated with various diseases

Gluten intolerance (celiac disease)

Result

Human Leukocyte antigen serotype

HLA DQ2	negative
HLA DQ8	positive

Interprétation

Presence of a genetic predisposition to gluten intolerance. This predisposition may be present in family members of the first degree.

General background to the analysed genetic polymorphism

Celiac disease is a pathology associated with hypersensitivity to gluten. This auto-immune/inflammatory disease is associated with inappropriate intestinal activation of lymphocytes in HLA DQ2 positive individuals and is triggered by antigenic gluten peptides from wheat or by prolamines from rye and barley. Classic symptoms of celiac disease include chronic diarrhea, weight loss, iron deficiency and other symptoms of malnutrition. The prevalence of the disease varies between 1/100 and 1/300 in industrialized countries. This disease is usually identified in children, but the diagnosis can be done in adults when the disease has been asymptomatic during childhood. Especially between the ages of 30 and 40 years clinical manifestation or complications related to gluten intolerance can become apparent in adults. Complications of gluten intolerance can be: late menarche, early menopause, infertility, pre-eclampsia, recurrent spontaneous abortions and delayed intra-uterine growth, secondary hyperparathyroidism, or intestinal lymphoma. Untreated celiac disease is a predisposition to other autoimmune diseases such as type I diabetes. Up to 90% of patients with celiac disease carry the HLA DQ2 haplotype (DQA1*0501 and DQB1*0201 alleles), while others carry the HLA DQ8 allele (DQB1*0302). Other genes outside the HLA complex seem to have only a modest impact. Only about 4% of carriers of the HLA DQ2 haplotype will develop gluten intolerance. The frequency of undiagnosed gluten intolerance is about 50% among family members of the first degree. The only effective treatment is to avoid gluten a lifelong, regardless of the presence or absence of histological features, in order to prevent the development of a glutenenteropathie, to control the symptoms or to prevent complications.

Note

In the presence of a heterozygous or homozygous variant genotype for one or several analyzed genes, it is recommended that the same genetic analysis should also be carried out for family members of the first degree and, depending on medical recommendations, also for the spouse.
Please note: Studies on associations of genetic variants with a certain phenotype or disease or condition have been performed mainly in Caucasian/North American populations. Thus, the interpretation of the results obtained in the present report can not necessarily be extrapolated to other ethnic groups.

The lab test results have been medically validated by: Prof. Dr. med. Bernard Weber