CELIAC DISEASE
TEST REPORT

Name: NAME FAMILY NAME
Date of birth: DD.MM.YYYY
Order ID: XXXXX
Sample ID: xxxxxx

Sample collection date: DD.MM.YYYY
Sample arrival date: DD.MM.YYYY
Analysis completion date: DD.MM.YYYY
Sample type: buccal swab

PRINCIPLE OF THE TEST

Human leukocyte antigen (HLA)-typing is used to confirm or exclude the possibility of celiac disease. The DNA test effectively rules out (99%) the disease if no HLA-DQ2/DQ8 risk types are detected.

RESULT OF THE TEST

The result of the DNA test indicates that the individual carries the HLA-DQ2,2 haplotype. The presence of the HLA-DQ2,2 haplotype is associated with a **MODERATE RISK** of developing celiac disease.

Analysis results of genetic markers tested

Marker	Genotype	Zygosity	Allele variant	Detected HLA haplotypes
rs2187668	CC	homozygous	normal	
rs7454108	TT	homozygous	normal	
rs2395182	TG	heterozygous	risk allele	DQ 2,2
rs7775228	TC	heterozygous	risk allele	DQ 2,2
rs4713586	AA	homozygous	normal	DQ 2,2

DQ2.2 HLA haplotype is detected by tag SNPs rs2395182, rs7775228, rs4713586 haplotype T, C, A.
The test parameters: sensitivity 99.1%, specificity 99.6% and positive predictive value 94.8% (Monsuur et al, 2008).

Methods used: Sanger Sequencing

CELIAC DISEASE DESCRIPTION

Celiac disease (CD) is a chronic gluten-intolerance that primarily affects the small intestine in genetically predisposed individuals and resolves with exclusion of gluten from the diet. It is characterized by nutrient malabsorption resulting from inflammatory injury to the mucosa of the small intestine after the ingestion of wheat gluten or similar proteins in rye, barley and triticale (a hybrid of wheat and rye).

Clinical features of CD are influenced by age of onset, gender (the ratio of female to male is 2:1), extent of mucosal injury and dietary habits. CD can be presented with many symptoms, ranging from typical gastrointestinal manifestations to only atypical signs or no symptoms at all.

Clinical features of celiac disease

Typical signs and symptoms	Atypical signs and symptoms	Associated diseases
Abdominal distension	Alopecia areata	Addison disease
Abdominal pain	Anemia (iron deficiency)	Atrophic gastritis
Anorexia	Aphthous stomatitis	Autoimmune hepatitis
Bulky, sticky and pale stools	Arthritis	Autoimmune pituitaritis
Diarrhea	Attention-deficit hyperactivity disorder	Autoimmune thyroiditis
Flatulence	Cerebellar ataxia	Behçet disease
Failure to thrive	Chronic fatigue, weakness	Dermatomyositis
Muscle wasting	Constipation	Inflammatory arthritis
Nausea	Dental anomalies	Multiple sclerosis
Steatorrhea	Depression	Myasthenia gravis
Vomiting	Dermatitis herpetiformis	Primary biliary cirrhosis
Weight loss	Epilepsy	Primary sclerosing cholangitis
	Esophageal reflux	Psoriasis
	Hepatic steatosis	Sjögren disease
	Infertility, miscarriage	Type 1 diabetes mellitus
	Isolated hypertransaminasemia	Vitiligo
	Late-onset puberty	
	Mouth ulcers	
	Myelopathy	
	Obesity	
	Osteoporosis, osteopenia	
	Peripheral neuropathy	
	Recurrent abdominal pain	
	Short stature	

TREATMENT

Strict, lifelong gluten-free diet (GFD) is currently the only effective treatment for CD as there is no medication that can reliably prevent gluten caused damage to the mucosa of the small intestine. Early diagnosis of CD is important for starting a therapeutic diet as soon as possible. Untreated CD significantly increases risk of developing long-term complications such as malnutrition, malignancy, osteoporosis, infertility, and autoimmunity. The diet requires complete elimination of all forms of wheat, barley and rye and their derivatives. Introduction of uncontaminated oats into the diet of people with CD should be followed up by careful monitoring of any signs of clinical and serological relapse.

Gluten-free dietary guidelines

Grains, flours and starches not permitted in a GFD	Gluten-free grains, flours, and starches allowed in a GFD♯	Other foods allowed in a GFD
Barley	Amaranth (grain type) (A. caudatus, A. cruentus, A. hypochondriacus)	Condiments: plain pickles, olives, nature herbs, pure black pepper, vinegar
Bran	Arrowroot starch is mainly obtained from the rhizomes of Maranta arundinacea	Eggs
Bulgur	Bean flours	Fresh meats (all)
Couscous	Buckwheat	Fruits: fresh, frozen, and plain juices (canned fruits no added)
Farina	Corn	Liquid vegetable oils
Farro*	Legumes: lentils, garbanzo beans (chickpeas), peas, beans	Milk products: cream, buttermilk, plain yogurt
Gluten, gluten flour	Millet	Snack foods: plain popcorn, nuts, and soy nuts
Graham flour	Nut flour and nut meals	Sweets: honey, corn syrup, sugar (brown and white)
Malt (extract, flavoring, syrup)	Oats (“pure”)***	Vegetables: fresh, frozen and plain juices (canned vegetables no added)
Oats (bran, syrup)**	Rice, all forms (brown, white, sweet, wild, jasmine, basmati etc.)	
Rye	Seeds	
Semolina (milled wheat)	Sorghum flour	
	Soy flour	
	Tapioca (cassava, Manihot esculenta)	
	Teff flour (Eragrostis tef)	

* A food product that composed of the grains of certain wheat species in whole form. Depending on geographic region einkorn, emmer or spelt wheat is known as farro.
** Commercial oats are most likely contaminated with gluten from other grains.
*** Only pure oats could be safely introduced into the diet of most people (>95%) with CD, but careful monitoring of clinical and serological symptoms is necessary.
♯ Although these gluten-free grains, flours, and starches are recommended in a GFD, there are concerns over cross-contamination with gluten-containing grains. These food products should be labeled as gluten-free.

RECOMMENDATIONS

- Individual with increased risk of CD should consider counseling by health-care practitioner for further testing needed to confirm or exclude diagnosis.
- First–degree relatives of CD patients should be HLA-typed and if CD cannot be excluded, further testing and counseling by health-care practitioner are recommended to determine disease recurrence risks.
- Individual with CD should follow a strict lifelong gluten-free diet.
- Individual with CD should visit an expert nutritionist in order to receive consultation on dietary recommendations and education on the GFD.
- Individual with newly diagnosed CD should undergo testing and treatment for micronutrient deficiencies. Deficiencies to be considered for testing should include, but not be limited to, iron, folic acid, vitamin D, and vitamin B12.
- Periodic medical check-up should be performed by a health-care practitioner and consultation with a dietitian is recommended if gluten contamination is suspected.
- Monitoring of adherence to GFD should be based on a combination of medical history and serology (IgA TTG or IgA (or IgG) DGP antibodies).
- Upper endoscopy with intestinal biopsies is recommended for monitoring in cases with lack of clinical response or relapse of symptoms despite a GFD.

CELIAC DISEASE TEST RESTRICTIONS

CD is highly unlikely when DQ predisposing alleles are absent.

The presence of the genetic risk factor means only a genetic predisposition for celiac autoimmunity and does not mean that patient definitely has/develop CD. Counseling by health-care practitioner and further testing is needed to confirm or exclude diagnosis of CD.

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