

CCR3 C/T POLYMORPHISM

ORDERING INFORMATIONS

REF: GEN-034-50 RDM Code: 1768994/R Tests: 50 Reactions: 62 CND Code: W0106010499 Manufacturer: BioMol Laboratories s.r.l.

CONTENTS OF THE KIT

The kit consists of: reagents for Real-Time PCR amplification *reagents for the extraction of genomic DNA are not supplied in the kit

IVD





PRODUCT CHARACTERISTICS

Detection of C/T polymorphism of the CCR3 gene (rs6441961) by Real-Time PCR technique. Kit optimized for Real-Time PCR instrumentation Biorad CFX96, Biorad Opus Dx, Agilent AriaDx.

Celiac disease (CD) is a chronic enteropathy, triggered by the presence of gluten proteins contained in wheat, barley and rye. The evidence of a strong genetic component is suggested by a considerable family aggregation: the prevalence of celiac disease is, in fact, 10 times higher in first-degree relatives (10%) than in the rest of the population (1%) and a very high (80%) is present in monozygotic twins.

Celiac disease is, therefore, a multifactorial disease in which the genetic predisposition contributes, together with environmental factors, to the onset of the pathology.

- § Improving the estimation of celiac disease sibling risk by non-HLA genes. PLoS One. 2011; 6 (11):e26920. doi: 10.1371/journal.pone.0026920. Epub 2011 Nov 7.
- § The Inter-Relationship of Platelets with Interleukin-1β-Mediated Inflammation in Humans. Thromb Haemost. 2018 Nov 19. doi: 10.1055/s-0038-1675603.
- § The Role of Human Leukocyte Antigen in Celiac Disease Diagnostics. Clin Lab Med. 2018 Dec; 38 (4):655-668. doi: 10.1016/j.cll.2018.07.007. Epub 2018 Oct 5.
- § Systematic review and meta-analysis of the association between IL18RAP rs917997 and CCR3 rs6441961 polymorphisms with celiac disease risk. Expert Rev Gastroenterol Hepatol. 2015; 9 (10):1327-38. doi: 10.1586/17474124.2015.107588. Epub 2015 Aug 8.

CLINICAL SIGNIFICANCE

Celiac disease is, therefore, a multifactorial disease in which the genetic predisposition contributes, together with environmental factors, to the onset of the pathology.Susceptibility to celiac disease is largely determined by class II molecules of the major histocompatibility complex (HLA-Human Leukocyte Antigen Complex), in particular by HLA-DQ2 and HLA-DQ8, cell membrane glycoproteins encoded by the HLA-DQA1 HLA-DQB1, located genes and chromosomal region 6p21.3. The HLA, however, identifies only 30-40% of the genetic risk considering that the presence of DQ2 and DQ8 does not automatically lead to the onset of celiac disease although their absence implies the impossibility of developing the disease. Recent data have demonstrated that IL18RAP rs917997 and CCR3 rs6441961 are potential risk factors for celiac disease in European populations. Other possible genes involved in the onset of celiac disease have been identified through association studies. These genes can be grouped into 5 large classes:

- 1. T cell differentiation genes (IL2, IL21, IL18RAP, IL12A);
- 2. immune activation signaling genes (SH2B3, TAGAP, CTLA4, PTPN2,ICOS);
- 3. immunity and TNF signaling genes (TNFAIP3, TNFSF14);
- 4. cytokine receptor genes (CCR1, CCR3);
- 5. cell compartment genes (LPP, RGS1, SCHIP1, REL, KIAA1109, CUTL1, VIL2)

The CCR3 gene encodes a G protein-associated receptor that responds to a variety of chemokines including eotaxin (CCL11), eotaxin-3 (CCL26), MCP-3 (CCL7), MCP-4 (CCL13), and RANTES (CCL5). It is expressed not only in eosinophils but also in basophils, Th2 lymphocytes, macrophages and epithelial cells of the respiratory tract.







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| DESCRIPTION | LABEL | VOLUME | STORAGE |
|--------------------------------------|----------------------|------------|---------|
| | | GEN-034-50 | |
| Mix oligonucleotides and probes | Mix CCR3 10X | 1 x 170 µl | -20°C |
| Mix buffer and Taq polymerase enzyme | Mix Real-Time PCR 2X | 1 x 850 µl | -20°C |
| Deionized H ₂ 0 | Deionized H₂0 | 2 x 1 ml | -20°C |
| Genomic DNA or recombinant DNA | Control + 1 | 1 x 22µl | -20°C |
| Genomic DNA or recombinant DNA | Control + 2 | 1 x 22µl | -20°C |
| Genomic DNA or recombinant DNA | Control + 3 | 1 x 22µl | -20°C |

TECHNICAL CHARACTERISTICS

COD. GEN-034-50

| STABILITY | 18 months |
|--|--|
| REAGENTS STATUS | Ready to use |
| BIOLOGICAL MATRIX | Genomic DNA extracted from whole blood, tissue, cells |
| POSITIVE CONTROL | Recombinant DNA for at least 3 analytical sessions |
| VALIDATED INSTRUMENTS | Biorad CFX96 Dx, Biorad Opus Dx e Agilent AriaDx |
| TECHNOLOGY | Real-time PCR; oligonucleotides and specific probes; 2 FAM/HEX fluorescence channels |
| RUNNING TIME | 85 min |
| THERMAL CYCLING PROFILE | 1 cycle at 95 °C (10 min); 45 cycles at 95 °C (15 sec) + 60 °C (60 sec) |
| ANALYTICAL SPECIFICITY | Absence of non-specific pairings of oligonucleotides and probes; absence of cross-reactivity |
| ANALYTICAL SENSITIVITY: LIMIT OF DETECTION (LOD) | ≥ 0,016 ng of DNA |
| ANALYTICAL SENSITIVITY: LIMIT OF BLANK (LOB) | 0% NCN |
| REPRODUCIBILITY | 99,9% |
| DIAGNOSTIC SPECIFICITY / DIAGNOSTIC SENSITIVITY | 100%/98% |



