

CELIAC DISEASE (HLA-TYPING: DQ2 AND DQ8)

PATHOGENESIS

Although the pathogenesis of celiac enteropathy is not yet fully understood, it is now established that the triggering of the disease requires both endogenous (genetic predisposition) and exogenous (gluten) elements. The role of the hereditary component is demonstrated by the observation that 8-10 percent of first-degree relatives of celiacs are affected silently by the disease and that there is a 75 percent agreement in monozygotic twins. The predisposition to gluten intolerance is transmitted through the alleles of the major histocompatibility complex of class II. The sum of the frequencies of the HLA genes DQA1 * 0501, DQB1 * 0201 (DQ2); DQA1 * 0310 and DQB1 * 0302 (DQ8) and DRB1 * 04 are found in 95-100% of celiac patients. The DQ2 complex is also present in 25% of the general population. The typing of HLA genes is therefore to be considered an important diagnostic tool, in addition to the classic ones: anti-gliadin and anti-transglutaminase antibodies and intestinal biopsy. In fact, it has been known for years that celiac disease, if not adequately treated, predisposes to several problems, including infertility, the tendency to spontaneous abortions in women, osteoporosis but also cancer.

TEST

PCR amplification of HLA DQ2 and DQ8 alleles. The polymorphism on one of the two alleles is compatible with a diagnosis of celiac disease. Tests for anti-gliadin (IgA) and anti-tTG (IgA) antibodies are also performed on the same material (EDTA blood).

PRELIEVO

Blood/EDTA 5 mL

ESECUZIONE

Daily.
