CONFERENCIA

Pitfalls in adult celiac disease diagnosis

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In adults, a diagnosis of celiac disease (CeD), regardless of the presence of symptoms, requires the demonstration, in the course of a gluten-containing diet, of the presence of a specific serology and typical pathological changes of untreated disease in biopsies from the proximal small bowel. The specific serology is now widely available and, as a first screening, the dosage of anti-transglutaminase IgA antibodies + serum IgA has a specificity and sensitivity of > 90%. There are few laboratory tests with such high specificity and sensitivity in predicting disease.

It is not always as simple as that. In some cases the histological changes of the mucosa, although typically associated with CeD, are not accompanied by a positive serology. In other cases, positive serology pairs with the evidence of normal mucosa. While the last case is a condition known as potential CeD, the case of positive histology and negative serology requires further insight.

From a practical point of view, the genetic testing is useful in the cases of absence of HLA-DQ 2/8 alleles. In fact, in the absence of the predisposing genetics, the possibility of CeD is unlikely. Although antibody-negative CeD may also occur, grade 1 mucosal lesion is non-specific and common in some other inflammatory conditions of the GI tract, including infections, IBD, NSAIDs, and food hypersensitivity other than CD.

However, besides other diseases, in some cases, the description of the mucosal damage may be the result of poorly orientated or inadequately handled biopsy specimen, and/or also from the inaccurate counting of IELs. It may be possible that the pathologists receive small specimens, such as few, superficial, stressed biopsies, or only biopsies from the bulb, which is rich of Brunner glands that twist the villi profile (namely, the Brunner glands artefact). In such a case, there is not much to do other than repeat the biopsy, providing adequate sampling and handling of the specimens. According to some reports, up to 20% of all histology reports describe non-specific lesions that are by a diagnosis of CeD only if the specific antibodies are present. At the moment, the significant pitfalls concentrate in the biopsy handling and histology reading. As a consequence, researchers are evaluating the possibility to avoid duodenal biopsy in a subgroup of patients in whom serology, symptoms and HLA status point to CeD.

References