Reference: UBE2L3 (18BIO03)

METHOD FOR CELIAC DISEASE DIAGNONYS IN THE ABSCENSE OF GLUTEN CONSUMPTION

Mendelian Randomization analysis of celiac GWAS reveals a blood expression signature for celiac disease diagnostic in absence of gluten consumption.

TYPE OF DEVELOPMENT

Diagnosis methodology.

DESCRIPTION

Celiac disease (CeD) is an immune-mediated enteropathy with a strong genetic component where the main environmental trigger is dietary gluten, and currently a correct diagnosis of the disease is impossible if gluten-free diet (GFD) has already been started.

We hypothesized that merging different levels of genomic information through Mendelian Randomization (MR) could help discover genetic biomarkers useful for CeD diagnosis. As a result, we identified *UBE2L3,* an ubiquitin ligase located in a CeD-associated region. We interrogated the expression of *UBE2L3* in an independent dataset of peripheral blood mononuclear cells (PBMCs) and found that its expression is altered in CeD patients on GFD when compared to non-celiac controls.

The relative expression of *UBE2L3* isoforms predicts CeD with 100% specificity and sensitivity and could be used as a diagnostic marker, especially in the absence of gluten consumption.

This approach could be applicable to other diseases where diagnosis of asymptomatic patients can be complicated.

INDICATION

CeD diagnosis in PBMCs from individuals on a gluten-free diet.

NOVELTY/ADVANTAGE

- Method for the diagnosis of CeD in PBMCs isolated from peripheral blood.
- Method for the diagnosis of CeD in individuals that have initiated a GFD before medical consultation.
- No need for gluten-provocation.
- No need for endoscopy.
- Easy PCR-based method for the detection of the relative expression of gene isoforms.

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IPR STATUS

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COOPERATION GOAL

- Licensing Out.
- Collaboration.