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Clinical Notes WGT LOSS FOR INVESTIGATION

Collected 29/02/2024 12:11

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Genotyping for Coeliac Disease

Specimen type	EDTA blood
Method	Real-time PCR
Result:	Coeliac susceptibility genotype DETECTED (DQA1*05+, DQA1*02-, DQB1*02+, DQB1*03:02/05-)
Interpretation:	Genotype consistent with the presence of HLA-DQ2.5 antigen. This result is associated with increased risk of coeliac disease. However, >50% of the general population has an at-risk genotype, and <1% of these individuals will develop biopsy-confirmed coeliac disease. Supportive evidence from coeliac serology and smallintestinal biopsy is therefore necessary to make a diagnosis of coeliac disease.

Comments

Test Information:

Qualitative detection of HLA-DQA1*02:01, HLA-DQA1*05:XX, HLA-DQB1*02:XX, HLA-DQB1*03:02/03:05 and HLA-DRB1*04:XX alleles is performed using the GeneFinder HLA-DQ2/DQ8 RealAmp kit (Osang Healthcare). This assay is designed to identify DQ2 (2.2 and 2.5) and DQ8 antigens that are present in more than 95% of individuals with coeliac disease. Some additional rare genotypes consistent with HLA-DQ8 antigen may be detectable by this assay though indistinguishable from HLA-DQB1*03:02/05. False positive results due to cross-reactivity with rare subtypes are possible. Rare subtypes, the presence of additional heterodimers, and zygosity of detected alleles cannot be determined by this assay. A full list of alleles to 4-digit HLA nomenclature detectable by this assay is available on request. References: PMID 25827511; 23981538.

NATA ACCREDITATION NO 2178

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