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Copy to

Requested 02/08/2023

Clinical Notes 43M, persisting fatigue and GIT symptoms Refer to request for Clinical History.

Collected 05/09/2023 10:01

Received 05/09/2023 10:02

Genotyping for Coeliac Disease

Specimen type
Method

EDTA blood
Real-time PCR

Result:

Potential susceptibility genotype DETECTED
(DQA1*05+, DQA1*02-, DQB1*02-, DQB1*03:02/05-)

Interpretation:

Not consistent with the presence of HLA-DQ2 or HLA-DQ8 antigens. However, HLA-DQA1*05 was detected in the absence of HLA-DQB1*02 or HLA-DQB1*03:02/05. This combination most commonly indicates the presence of the HLA-DQ7 antigen.

This may be associated with susceptibility to coeliac disease, but confers a lower risk than DQ2 and DQ8 antigens. In the appropriate context, further clinical work-up for coeliac disease should be considered.

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. Reported by Douglass Hanly Moir Pathology(2178), a member of the Sonic Healthcare Group.

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LAUNCESTON PATHOLOGY NATA ACCREDITATION NO. 2208

Reported on 11-09-2023 22:07