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Risk stratification by HLA-DQB and -DQA alleles in patients suspected for Celiac Disease in the Belgian population

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Background: Results of 200 samples, sent to HILA over a period of 2 years for HLA-DQ genotyping in the context of Celiac Disease (CD), were reclassified according to recently described low risk alleles. So far we only reported alleles with a high and intermediate risk for developing CD as positive: DQB1*02:01, DQA1*05:01 (DQ2.5 cis), DQB1*02:02, DQA1*05:05 (DQ2.5 trans) and DQB1*03:02, DQA1*03 (DQ8). We evaluated the change in risk stratification if we also included low risk alleles: DQB1*02:02, DQA1*02 (DQ2.2), DQB1*03:01, DQA1*05:05 (DQ7.5).

Results: 118 (59%) patients had no change in risk. 39 (20%) remained 'negative'. 82 (41%) patients had an increase in risk stratification.

53 (27%) of the patients changed from 'negative' to 'positive'. All of these had DQ2.2 and DQ7.5 alleles, associated with low risk. Of the alleles with high and moderate risk 36% were DQ2.5cis, 3% DQ2.5 trans and 20% DQ8. Some patients carried double predictive alleles. DQ2.5cis was found 2.6-times, DQ2.5trans 3-times (very small population) and HLA-DQ8 2.7-times more frequently in patients compared to the Belgian population. Alleles with low risk accounted for 14% DQ2.2 and 27% DQ7.5, which is respectively 1.8- and 1.7-times more than the Belgian population.

Methods: Since mid2018 HLA genotyping is performed using the NGS MIAFORA 11 loci kit (Immucor), including the linkage to HLA-DRB, allowing us further fine tuning of the risk assessment. Despite the small cohort until now (33 samples) we found, as suspected, the same (change in) risk stratification.

Conclusions: Finding an increased frequency of Celiac Disease associated alleles (three times more than in the Belgian population) in samples sent to HILA in the context of diagnosing Celiac Disease confirms the indication of the test request and the correlation of these alleles with the presentation of Celiac Disease.

The finding that low risk alleles are also more frequent in our Celiac cohort suggests their relevance in the disease prediction and confirms our current strategy to classify them as positively associated.