

INTERPRETATION OF HLA RESULTS RELATED TO COELIAC DISEASE

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POSITIVE RESULT

GENOTYPE DRB1*XX:XX, *XX:XX / DQA1*05:01, *XX:XX / DQB1*02:01, *XX:XX

SEROLOGICAL EQUIVALENT

DQ2.5 POSITIVE, DQ8 negative

GENOTYPE DRB1*XX:XX, *XX:XX / DQA1*02:01, *05:05 / DQB1*02:02, *03:01

SEROLOGICAL EQUIVALENT

DQ2.5 POSITIVE, DQ8 negative

GENOTYPE DRB1*XX:XX, *XX:XX / DQA1*03:01, *XX:XX / DQB1*03:02, *XX:XX

SEROLOGICAL EQUIVALENT

DQ8 POSITIVE, DQ2 negative

INTERPRETATION

DETECTED HLA GENOTYPE IS ASSOCIATED WITH THE RISK OF COELIAC DISEASE.

This result cannot be separately interpreted as a confirmation of celiac disease because of its low specificity.

COMMENT

*XX:XX = any allele;

INCONCLUSIVE RESULT – most common genotypes

GENOTYPE DRB1*XX:XX, *XX:XX / DQA1*02:01,*XX:XX (except *05:XX) / DQB1*02:02, *XX:XX

SEROLOGICAL EQUIVALENT

DQ2.2 POSITIVE, DQ8 negative

INTERPRETATION

DETECTED HLA GENOTYPE IS ASSOCIATED WITH THE RARE RISK OF COELIAC DISEASE

The diagnosis is not possible to be excluded

GENOTYPE

DRB1*XX:XX, *XX:XX / DQA1*05:05,*XX:XX / DQB1*03:01, *XX:XX (mimo *02:XX a *03:02)

SEROLOGICAL EQUIVALENT

DQ2 negative, DQ8 negative

INTERPRETATION

DETECTED HLA GENOTYPE IS NOT ASSOCIATED WITH THE RISK OF COELIAC DISEASE

The result excludes this diagnosis with a high probability.

QUESTION

Is it possible in this case report, that the result excludes this diagnosis with the high probability?

NEGATIVE RESULT

GENOTYPE

DRB1*XX:XX, *XX:XX / DQA1 *XX:XX, *XX:XX (all alleles except *03:01 and *05:XX) / DQB1*XX:XX, *XX:XX (all alleles except *02:XX and *03:02)

SEROLOGICAL EQUIVALENT

DQ2 negative, DQ8 negative

INTERPRETATION

DETECTED HLA GENOTYPE IS NOT ASSOCIATED WITH THE RISK OF COELIAC DISEASE

The result excludes this diagnosis with a high probability

EFFECT OF A DOSAGE OF RISK ALLEL

GENOTYPE DRB1*XX:XX, *XX:XX/ DQA1*05:01 homozygote/DQB1*02:01 homozygote

SEROLOGICAL EQUIVALENT

DQ2.5 POSITIVE, DQ8 negative

COMMENT

cannot be clearly detected using CD specific kits

GENOTYPE

DRB1*XX:XX, *XX:XX / DQA1*03:01, *05:01 / DQB1*03:02, *02:01

SEROLOGICAL EQUIVALENT

DQ2.5 POSITIVE, DQ8 POZITIVNÍ

GENOTYPE

DRB1*XX:XX, *XX:XX / DQA1*03:01 homozygote/DQB1 *03:02 homozygote

SEROLOGICAL EQUIVALENT

DQ8 POSITIVE, DQ2 negative

POZNÁMKA

cannot be clearly detected using CD specific kits

this genotype is highly unlikely due to the frequency of particular alleles

INTERPRETACE

SAME AS FOR THE STANDARD POSITIVE RESULT

REPORT OF RESULTS

COMPLETE RESULT CONSISTS OF:

- **Detected alleles** (according to the actual HLA nomenclature)
- **Serological equivalent**
 - In case of DQ 2 it is necessary to distinguish between DQ 2.5 and DQ 2.2
 - For more accurate results it is desirable to mention also DQ2, DQ8 negativity
- **Interpretation of results related to CD including information about predictive value of the test (see slides before)**
 - **DETECTED HLA GENOTYPE IS ASSOCIATED WITH THE RISK OF COELIAC DISSEASE**
This result cannot be separately interpreted as a confirmation of celiac disease because of its low specificity.
 - **DETECTED HLA GENOTYPE IS ASSOCIATED WITH THE RARE RISK OF COELIAC DISSEASE**
The diagnosis is not possible to be excluded.
 - **DETECTED HLA GENOTYPE IS NOT ASSOCIATED WITH THE RISK OF COELIAC DISSEASE**
The result excludes this diagnosis with a high probability