

Coeliac disease gene test - analysis and reporting

A letter to the editor published on 21 August 2017 in the Medical Journal of Australia has raised concerns about inaccurate analysis and reporting of genetic testing for coeliac disease.

The main utility of coeliac HLA typing is to assist with diagnostic risk-stratification by excluding coeliac disease when typing is negative for HLA susceptibility genotypes.

The letter: *Discrepancies in genetic testing results for coeliac disease highlight an urgent need for standardised testing and reporting and national assessment*, was prepared by Dr James Daveson, a Queensland based gastroenterologist and member of Coeliac Australia's Medical Advisory Committee (MAC) after he became aware of discrepancies in genetic testing results after some patients with negative gene test results were found to have coeliac disease.

The discrepancies reported in the letter were discovered as a result of a small audit in Queensland; however, as some of the HLA typing was outsourced to other states this issue could potentially affect patients across Australia. A study in collaboration with the Royal College of Pathologists of Australasia Quality Assurance Program (RCPAQAP) is underway to examine the extent of the problem.

Patients who have previously tested negative for the HLA-DQ2/8 genotype associated with coeliac disease may approach you regarding the validity of this test result.

Repeat investigations that may or may not include repeat HLA typing may be warranted in some cases, depending on the specific clinical history. If you are uncertain, please contact the treating gastroenterologist or the pathology lab performing the test to determine the best approach for your patient.

Please note that a positive HLA gene test result is NOT diagnostic for coeliac disease. You can find more information about diagnosis on our website:

<http://www.coeliac.org.au/uploads/65701/ufiles/Brochures/FamilyScreeningLetter.pdf>

Members of Coeliac Australia's MAC have developed an evidence-based approach to guide clinically appropriate use of HLA typing and established a reporting template for pathology providers to improve communication of results, which you may find of use: *Appropriate clinical use of human leukocyte antigen typing for coeliac disease: an Australasian perspective*.

<http://onlinelibrary.wiley.com/doi/10.1111/imj.12716/abstract>