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GENETIC AND SEROLOGICAL CONCORDANCE IN PATIENTS WITH SUSPECTED COELIAC DISEASE

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Abstract

Introduction: Coeliac disease (CD) is an autoimmune gastroenteropathy strongly associated with Human Leukocyte Antigen (HLA) DQ2 and DQ8 genes. The DQ2/8 alleles are also associated with other autoimmune diseases including type 1 diabetes (T1D), which puts these patients at a higher risk of developing CD. CD serology is used to screen these patients but has limitations. Intestinal Fatty Acid Binding Protein (I-FABP) and CX3CL1 (Fractalkine) are two promising biomarkers for CD but have yet to be examined in patients at a high-risk (HR). This study, therefore, aimed to examine genetic and serological concordance in South African patients with T1D and high-risk CD alleles.

Methods: A total of 38 paediatric and adult participants were recruited from an endocrinology clinic and gastroenterologists in the Johannesburg region. Clinical details were collected, and all patients were HLA typed at the DQ loci. I-FABP and CX3CL1 serum levels were then determined using ELISA and Luminex® technology respectively.

Results: Of our patients with T1D, 63.6% had at least 1 HLA allele associated with CD. I-FABP serum levels were significantly higher in CD-positive patients compared to CD-negative individuals ($p=0.03$). No significant differences in the CX3CL1 serum levels were detected although this may reflect the impact of the comorbid autoimmune diseases on the CX3CL1 levels.

Conclusions: This study provides preliminary insight into the HLA-DQ2/8 type frequency in a South African population with T1D and is the first to assess the levels of I-FABP and CX3CL1 in a multi-ethnic population with comorbid autoimmune diseases. It determined I-FABP to be the more promising biomarker in such clinical contexts compared to CX3CL1.