Case study of Ms BM – Coeliac Disease

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Presenting history
Ms BM was a 28-year-old European lady who presented with a 4-year history of left cheek pain. The pain was continual, rated 6/10 in severity and could be associated with nausea and vomiting. The pain was said to have started following an episode of infective sinusitis. She mentioned that she might have been iron deficient at the time. She had had extensive dental work done looking for a dental cause of the pain. Three dentists and three oral surgeons had been unable to find a cause for the pain. She had no neck pain, shoulder pain or low back pain. There were no irritable bowel symptoms or period problems. There was no past history of anxiety or depression. The pain was poorly controlled with conventional anti-inflammatory drugs and Amitriptyline. She had had normal ultrasound and MRI scans of the area. Previous blood tests had shown an iron deficiency, but no anaemia. She was married with two children. She had tertiary and post-graduate qualifications.

Examination
Height 155 cm, weight 42kg. BMI 16. Tender left masseter muscle; palpation of muscle replicates pain. No other significant physical findings. Trigger point in left masseter muscle.

Initial management
1. Injection of the left masseter muscle with local anaesthetic. This completely relieved the pain for 6 days.
2. She was advised regarding heat and self-massage techniques to the masseter muscle.
3. Investigations were initially performed firstly to exclude metabolic causes for the trigger point and secondly looking for other possible causes. At this time, she had had a normal ultrasound and MRI scans of the area.

Results of initial investigations
Iron 12g/L, ferritin 1.5μg/L, vitamin B12 528pmol/L, zinic 11μmol/L and random urinary iodide 50μmol/L, tissue transglutaminase (tTG) 2 and antitissue transglutaminase (AGA) 2, Vitamin D 69nmol/L, MCV 86fl and MCH 27pg.

Initial management upon receipt of investigations
She was advised to take vitamin D, zinc and iodine. A gluten-free diet was also advised to avoid any dairy-free food she was already avoiding.

Subsequent management
When placed on a gluten-free diet her pain resolved almost completely and she was able to stop all pain medication. She put on 2kg in weight. Despite my advice to also go dairy free she was still unable to lose weight.

Discussion
This case illustrates the difficulties in diagnosing CD. Classic textbook symptoms include abdominal bloating, vomiting, diarrhoea, weight loss, and in children, stunted growth. However abnormal bowel symptoms, particularly in adults and also in this case, may be limited or even absent. With this patient, the clue was the repeated low iron levels. There was very little on blood testing alone that caused one’s suspicions to be raised. There were minimal other clues to suggest other nutritional deficiencies. MCV and MCH were in the lower half of the normal range. While she was zinc deficient, zinc deficiency is common in the New Zealand population. New Zealand soils are frequently low in zinc and selenium. Blood testing for zinc status is also notoriously unreliable. While this lady had a low BMI in keeping with CD, this is not necessarily a reliable clinical sign.

Investigations
Antibody testing for transglutaminase IgA antibodies has a reported sensitivity (diagnosis of disease) of greater than 90% and a specificity (exclusion of disease) of greater than 95%. HLA testing is more useful in excluding CD. Duodenal biopsy remains the gold standard due to the false-negative rate associated with serology alone. This was the situation with this patient. Multiple samples (ideally 6) need to be taken from the second part of the duodenum and beyond on endoscopy. Not all areas may be equally affected; if biopsies are taken from healthy bowel tissue, the result will be a false negative.

Pathophysiology
CD is a complex multifactorial condition in that while genetic factors are important, other factors are necessary for the disease to manifest itself. The vast majority of coeliac patients express the HLA-DQ2 or HLA-DQ8 genes, which are part of the MHC class II antigens presenting receptor system distinguishing cells between self and non-self in the immune system. However in the West, 25-30% of the population express HLA DQ2 and are exposed to gluten, but only a minority develop CD. Around 5% of patients with CD do not have the HLA-DQ2 gene. The HLA-DQ2 molecules bind to and present gluten derived peptides to T lymphocytes, initiating the autoimmune process. Most coeliac patients have a two gene HLA-DQ2 haplotype referred to as DQ2.5 haplotype. While most coeliac patients inherit only one copy of this DQ2.5 haplotype, some inherit it from both parents. This latter group is especially at risk for CD, as well as being more susceptible to severe complications. The frequency of the HLA-DQ2 genes varies geographically. DQ2.5 has a high frequency in peoples of North and Western Europe; Ireland particularly has a high frequency in peoples of North and Western Europe; Ireland particularly has a high frequency in peoples of North and Western Europe; Ireland particularly has a high frequency in peoples of North and Western Europe; Ireland particularly.

Muscles and Coeliac Disease
Myopathy in the context of gluten sensitivity exists, but has not been well characterized. The myopathy normally presents as a proximal muscle weakness. Endomysium-specific antibodies (antibodies that bind with high affinity to cell-surface tTG) are thought to be involved, but there are also the most sensitive markers for the presence of enteropathy, which were not present in this patient. A spectrum of abnormalities has been described on muscle histology in gluten sensitivity-associated myopathy. Muscle inflammation is a common feature. IgA deposits against tTG have been demonstrated in extra intestinal target sites such as muscle in a patient with coeliac disease and myopathy and cerebellar tissue in a patient with gluten ataxia. Myopathy may be a marker of gluten sensitivity and may have an immune mediated pathogenesis.