ISSUE 3

The Bottom Line

Vaughan Endoscopy Clinic (VEC) is a **state of the art** out-of-hospital endoscopy clinic providing Screening colonoscopy and **endoscopy** for the work up of mild gastrointestinal disorders. It is staffed by gastroenterologists**.**

In addition to the endoscopic services, they will provide all the necessary **GI follow-up** and make all the appropriate referrals required due to findings at the endoscopy.

The Medical Director has been an active participant at the CPSO in the development of **standards for out-of-hospital clinics**, all of which VEC adheres to.

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In addition to high quality and convenient access to endoscopy, the Doctors at VEC will provide you with supplemental practical GI advice through this periodic newsletter. This article on Celiac Disease is written by Dr. David Kreaden (a gastroenterologist from Humber River Regional Hospital).

What is Celiac Disease?

Celiac disease is a very common food intolerance especially in the Western population. It was first described, believe it or not, in the 1st and 2nd century A.D! Celiac disease is a chronic small bowel enteropathy induced by gluten proteins from wheat, barley and rye resulting in mucosal injury.

What is the epidemiology of the disease?

It would appear that the incidence of celiac disease is increasing. This is probably based upon a combination of increased recognition and a true increase in the number of cases. Until the 1970s the estimated global prevalence of celiac disease in the general population was .03%. Currently the estimated prevalence is at least 1% and in Europe and North America. However, it should be noted that celiac disease has a worldwide prevalence. In fact the highest prevalence of celiac disease (5.6%) occurs in Algeria. By contrast celiac disease seems to be rare in individuals of Japanese and Chinese ancestry likely because there is a low frequency of HLA-DQ 2. About 90% of individuals with celiac disease carry the DQ 2 genotype and all remaining patients express DQ 8. Without these 2 HLA genes development of celiac disease is unlikely. Celiac disease is more common in women (3:1). It can be diagnosed at any age with a peak in early childhood and then in the 4th and 5th decade of life for women and men respectively.

Are there environmental risk factors?

1. Infant feeding: introduction of gluten abruptly after breast-feeding after 6 months was at least a risk factor in a Swedish study which found an almost threefold higher risk of development of celiac disease. It is now recommended that small amounts of gluten be gradually introduced between 4 and 7 months of age during breast-feeding.
2. Infections: a prospective study showed that frequent rotavirus infection in children represented an independent risk factor for celiac disease in genetically susceptible individuals. HCV is also associated.
3. Socioeconomic factors: being poor might be protective.

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What is the clinical presentation?

Celiac disease has long been considered a pediatric syndrome with classic symptoms of diarrhea, steatorrhea and weight loss. However in older children and adults there appears to be a broad spectrum of clinical presentation. About 50% of patients present with atypical symptoms such as anemia, osteoporosis, dermatitis herpetiformis, neurological problems and dental enamel hypoplasia. Many patients I have seen present with bloating, flatulence and fatigue. Other atypical presentations include isolated elevation of transaminases, infertility, or recurrent miscarriages. There are also many "autoimmune disease" associations. The presence of atypical signs and symptoms may mask the actual diagnosis of celiac disease as typical GI complaints are lacking.

How is the diagnosis made?

Diagnosis is made when the following features are **both** present: typical small intestinal mucosal biopsy abnormalities which include increase in intraepithelial lymphocytes, crypt hyperplasia, villous atrophy and clinical remission on strict gluten-free diet with relief of symptoms. Pathological description is characterized by using the Marsh classification ranging from 0-III. Marsh III lesion includes villous atrophy which is most specific. Serological testing using IGA-TTG (antibody to tissue transglutaminase) is very sensitive but if positive should be followed with small bowel biopsies for confirmation of histological disease. Gliadin antibody is less specific and sensitive and generally I ignore the result. In addition I always check IGA levels as IGA deficiency may give a false negative result.

What are the treatment options?

Dietary exclusion of grains containing gluten (barley, rye and wheat) and supportive nutritional care in the case of various vitamins. Other therapeutic alternatives are being examined. These therapies focus on alteration of gluten by rapid enzymatic degradation or alteration of the immune response but currently not ready for "prime time".

Are there complications?

Some patients with adult onset celiac disease especially those diagnosed above age 50 show a lack of response to gluten restriction. They are diagnosed with *refractory celiac disease*. It might affect about 5% of patients with celiac disease. RCD can be subdivided into types I and II with normal and aberrant intraepithelial T lymphocytes in the small intestinal mucosal respectively. The latter may lack certain surface expression of T cell markers. Flow cytometry is used as well as biopsies and immunohistochemistry. RCD I often responds to prednisone and Imuran whereas there is no standard approach for RCD II. Unfortunately, those individuals with RCD II are at risk of developing lymphoma. The principle malignancy associated with celiac disease is EATL (enteropathy associated T-cell lymphoma) which has a rather low incidence of .5-1 case per million people in Western countries. EATL is one of the main causes of celiac related death in patients with adult onset celiac disease, with 2 and 5 year overall survival rates of 15-20% and 8-20% respectively. This poor prognosis is mainly due to incomplete response to currently available therapies, and high rates of life-threatening complication such as perforation and poor nutritional conditions.

The bottom line is:

Celiac disease is fairly common and can present at any age. It frequently presents in an atypical fashion and can be diagnosed with an anti-TTG blood test. Once diagnosed lifelong gluten restriction is indicated.

This newsletter will be posted on our website (www.vaughanendoscopy.com) thus your patients are able to download a copy for reference. Other GI topics of interest will be published periodically.