Celiac disease

The road to diagnosis

Celiac disease is one of the most common gastrointestinal disorders that may affect up to 1% of the world population.

Most cases remain undiagnosed.

This is a collection of stories of individuals with celiac disease describing their long and often treacherous road to diagnosis.

Dr. Mohsin Rashid

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The road to diagnosis

Dr. Mohsin Rashid

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Cover photograph: Gros Morne National Park, Newfoundland, Canada
(Taken by M. Rashid)

To all those who are dedicated to improving
the lives of individuals with celiac disease

Preface

My interest in celiac disease (gluten sensitive enteropathy) is both personal and professional. The personal interest developed when a couple of individuals very dear to me were diagnosed with this disease. Professionally, I have been involved with children with celiac disease and their families as a practicing pediatric gastroenterologist for the last several years. More recently, as a member of the national Professional Advisory Board of the Canadian Celiac Association (CCA) and Medical Advisor to the Halifax chapter of CCA, I have had a chance to collaborate with health professionals with expertise in this area and to interact with individuals and families with this disorder.

Of all the gastrointestinal disorders that I have encountered in my practice, none has been as fascinating as celiac disease. Celiac disease is a real “clinical chameleon”. It can be typical, atypical, silent or latent. Its clinical pattern has changed, and atypical

presentations are now becoming more typical. One often finds celiac disease in patients in whom it is least expected. Those classical images of severely malnourished and wasted children with celiac disease seen in many textbooks are now history.

Our understanding of celiac disease has greatly improved over the last decade. The recent availability of serological tests has opened new avenues of screening high-risk individuals who are minimally symptomatic or asymptomatic. There has been an explosion of new diagnoses. However, before ending up at a gastroenterology clinic, many individuals still shuttle between a variety of physicians including family doctors, internists, hematologists, rheumatologists, neurologists and surgeons. This is not only because the disease can present in unusual ways but also due to a lack of awareness amongst health professionals.

A few years ago, an announcement was placed in the quarterly newsletter of the Halifax chapter of CCA inviting its members to send me a brief description of how they were diagnosed with celiac disease

or dermatitis herpetiformis. The information was requested anonymously with only age and gender to be specified. My objective was to learn about the various presentations of these disorders from people with personal experience. Several responses were received, both from within and outside the province. Each respondent had a unique and fascinating story to tell about his or her road to diagnosis. In most cases the road had been long and treacherous. I thought that these case histories could serve as an interesting educational tool to help increase awareness of celiac disease amongst physicians. All stories were worthy of being published, but due to space limitation only a few could be included in this book. However, they represent the wide spectrum of clinical features of celiac disease.

The information is presented without identifying the contributors. In some cases, dates and places have been deleted to maintain anonymity. I have provided titles to the stories and to preserve the flavor of the narrations, I have not altered or revised the text. Other than a few grammatical adjustments and clarification of some

acronyms, the stories are presented in their entirety. At the end of each narration, I have added a few clinical tips and "take-home" messages.

I am very grateful to my peers Dr. Connie Switzer and Dr. Peter Green for reviewing the book and giving their valuable suggestions and comments. I also wish to thank my administrative assistant, Dorothy Williamson for her help in transcribing and organizing this material.

This book will be made available to various health care professionals. I hope they will find it both interesting and educational. I welcome any feedback. The stories should not be taken as a criticism of the health profession; they simply illustrate the widespread lack of awareness of this disorder. My ultimate goal is to improve the awareness of celiac disease and prevent delays in diagnosis. Increasing awareness is so important for this disorder that may affect up to one percent of the world population!!

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Foreword

Sir William Osler once said: “Listen to the patient…he is telling you the diagnosis.” The validity of his statement could not be confirmed more dramatically than by reading the first-person narratives of patients whose long experience with celiac disease has been compiled by Dr. Rashid in this valuable little book. Their stories illustrate, far better than could be done in a medical textbook, the remarkable spectrum of clinical presentations of celiac disease. Even more instructive is the documentation of the intolerably long delays that too often occur between the onset of symptoms and establishment of the definitive diagnosis and the institution of effective treatment.

Our understanding of celiac disease continues to expand at high speed, with new contributions to our understanding of the disorder appearing almost weekly in the medical scientific literature. The development and progressive refinement of serological and genetic testing is already

leading to identification of patients with previously unsuspected disease, and of asymptomatic or minimally symptomatic individuals, some of whom may develop more obvious clinical disease at some later date. It is even within the realm of possibility that, at some future date, such testing might even replace intestinal biopsy as the "gold standard" for diagnosis, at least in some individuals. Meanwhile, as with most medical research, even when research doesn't give us all the answers, it always refines the questions.

But despite these remarkable advances in scientific knowledge, one fundamental fact remains unchanged. The diagnosis of celiac disease, as with most other conditions, is not made by laboratory or other tests. It is made by *people*…by doctors who listen carefully to what their patients are telling them. Those stories, as told by patients in their own words, remain far and away the most valuable diagnostic aids we have.

By letting people with celiac disease describe their long and often frustrating journeys toward a final diagnosis and

definitive treatment, Dr. Rashid has done us all a valuable service.

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1

Diagnosing “transaminitis” can be fun

(Age: 75 years, male)

My first overt classical symptoms of celiac disease appeared when I was 31, in 1966, in ……. I had had one or two severe attacks of what I took to be a bug from the water supply there, or across the border in France, where I worked with the RCAF. I did not pay too much attention, therefore, to the milder symptoms, which persisted thereafter.

Eight years later, I went for my first annual medical. Since I was at that time part of the military liaison staff attached to the

……., I went to a clinic run by the US Army in …., ….. The US Army doctor looked at the results of all my tests, and said. "Your cholesterol levels might be high for an African bushman, but they're off the bottom of the scale for North Americans. You drink a lot though!"

I denied that vigorously, for though I was certainly on the fringe of the diplomatic "cocktail circuit", I kept a strict limitation on my alcohol intake.

His response was essentially. "Well, you can fool yourself, but you can't fool me!"

In 1975, after a very stressful job change (still in the military), my symptoms came back with a vengeance. I lost 5 kg in two months, before regaining some stability. Over the next few years, as I moved from posting to posting, and doctor to doctor, I kept getting the same comments on each annual medical. "You are drinking too much!"

Finally, I demanded to know why the doctors thought that. The current doctor was smug about it.

"You have folic acid deficiency anemia", he said. "That's caused by chronic alcoholism. Besides, your SGOT (serum glutamic oxaloacetic transaminase) is 186. It should be 35. The alcohol is affecting your liver!"

I fussed and fumed to no avail. They did take some stool samples, but nothing came of that. I was given vitamin supplements.

One newly graduated doctor announced that I had pernicious anemia. When I asked her why my vitamin B12 levels were normal, her only response was. "Maybe it's not pernicious anemia!"

My confidence in medical schools was wearing thin.

In 1981, fifteen years after my symptoms had appeared, I was posted to ……, north of Toronto. By this time, I had lost 10.5 kg and was beginning to resemble a concentration camp survivor. My ribs stuck out.

The Base Surgeon was a rumpled, overweight, beer-swigging, cigar-chomping military doctor of the old school. He phoned me in my office one day, and said.

"You have target cells in your blood! Come to see me."

In his office, he proceeded to explain that target cells were caused by lack of folic acid. I spluttered. "I am not an alcoholic!"

To my surprise, he said. "I believe you! But you *are* sick. I'm going to call in a blood specialist from Toronto."

In due course, the blood specialist arrived and took me through an exhaustive interview. "There's something wrong," he said. "But I have no idea what it is. It isn't alcoholism, but it looks like liver damage. We'll have to send you to hospital."

They sent me to the National Defense Medical Centre (NDMC) in Ottawa. For a week, I went through every test imaginable. They took copious quantities of blood, sat me through xylose absorption tests, fed me barium cocktails and took so many x-rays, I was practically transparent.

At week's end, they said, "Nothing so far. We'll have to do a liver biopsy and bone marrow!"

The days leading up to the biopsy were filled with NPO signs. NPO is Latin for "nothing by mouth". The sign appeared each time a blood sample was required, which was several times per day. It went up before breakfast, before lunch and before supper.

Finally, after I had missed several meals in a row, my roommate yelled, "Will you feed this man! He's starving to death in front of me!"

There was a general "Ooops!" among the ward staff, and I got to eat again.

NDMC was a teaching hospital. When my biopsy time came, the internist arrived with a small coterie of students. He instructed them on how to anaesthetize the skin, make a small incision, and line up the biopsy needles on its stainless steel guide.

Then he said to me, "Push".

The idea being that I had to inhale, and hold me breath to push the liver against the rib cage.

I did.

Quickly, he rammed the needle between my ribs, withdrew it, and ceremoniously deposited the sample in a little dish.

"Damn!", he said.

"What do you mean, 'Damn'?" said I.

"I missed your liver," he said. "We'll have to do it again!

He did, three times.

After the fourth "Damn!" he said to the students. "This man has a large rib cage and a small liver. It's moving out of the way. We'll have to open him up for a sample!"

So saying, he shoved a couple of sandbags against my side, and instructed a nurse to take my blood pressure every five minutes until it was stable.

"What's that for?" I asked.

"Oh," he said cheerfully "If the needle accidentally punctured your hepatic artery, you could bleed to death internally. We're just checking to make sure."

I felt really good after that, especially since my blood pressure promptly nose dived to 75 over 37 and stayed there for about fifteen minutes.

A little later, the doctor returned and said. "We'll have to do a mini-laparotomy for the liver sample. We'll make a five centimeter incision just under the sternum and go in for a look."

I wasn't happy about that, but I did have one good idea.

"Could you do the bone marrow while I was under the anesthetic for the operation?" I asked.

He thought for a moment, and said. "I don't see why not!"

At least I was saved one painful procedure.

Shortly thereafter, I went under the knife. I learned later that they had taken four liver samples and four bone marrow samples. I had a neat zipper of stitches on my abdomen, and a nasty bruise on my hip.

After the initial dopiness, I felt pretty good until I tried to rise. I was immediately floored again by agonizing pains in my neck and shoulders. I cornered an intern and demanded to know why.

"Liver wounds bleed a lot," he said, "and the blood pools on the diaphragm. The nerves that serve the diaphragm come from the spinal column high in your neck. That's why quadriplegics can still breathe. When you try to stand, the blood on the diaphragm pulls it down. The cuts are in your liver, but the pain is in your neck."

It was.

Early the next day, the doctor came by.

"Well, there's nothing wrong with your liver," he said, "but you have no iron in your bone marrow at all!"

He was exaggerating, of course, but also somewhat embarrassed at having missed the fact that I was iron deficient anemic as well as folic acid deficient. Folic acid deficiency makes great big red blood cells (target cells), but iron deficiency makes very tiny ones. They had seen the big cells, but missed the little ones. Some pathologist got a rap on the knuckles for that one.

"There's only one test left I can think of," said the doctor.

The next day he was stuffing an endoscope down my gullet.

That, at least, was a painless, if undignified procedure.

I was munching a piece of toast for breakfast the following morning when the doctor came back.

"Flat as a pancake", he said. "You have no villi left!"

I knew what that meant. It was my last piece of toast.

By the end of 1981, I had spent fifteen years of chronic, if usually low-grade illness and three weeks of acute poking, prodding, slicing, and dicing in hospital. Still, those three weeks put me on the road to recovery.

A couple of weeks after I returned to Base, the wife of one of my Majors walked right past me in a store.

When I called to her, she said. “I didn’t recognize you. You’ve turned pink!”

I had indeed. I had been grey-white before.

At the end of six weeks, I had gained 4.5 kg. At the end of a year, I had gained 16 kg (which is about 4 kg more than I wanted!).

Seventeen years later, I’m doing fine.

Clinical Pearls

✓ Anemia (iron or folate deficiency) is one of the most common presentations of celiac disease.

✓ Celiac disease is one of many causes of idiopathic "transaminitis" (elevated liver transaminases). The timely diagnosis of celiac disease can spare the patient a liver biopsy. If the liver enzymes do not normalize after treatment with a gluten free diet, a liver biopsy may have to be performed for other associated conditions like autoimmune hepatitis.

✓ Uncommon presentations of common disorders are more common than common presentations of uncommon disorders. And celiac disease is a COMMON disorder.

2

Scoping the wrong end

(Age: 55 years, female)

In March 1995, at age 50, I had laparoscopic surgery to remove my gallbladder. The following day I remember being very pleased that I had my first post-operative bowel movement after eating oatmeal and toast for breakfast. I was discharged on the third day and had 3-4 bowel movements that day. After about the fifth day post-op, I was becoming a bit concerned as I was then having up to 6 or 7 bowel movements per day!

I decided to phone the surgeon who prescribed Questran 3-4 times per day before meals and told me that things would probably settle down in a few weeks. I

continued to have the problem after two weeks and was losing weight. I made an appointment with my new family physician whom I hadn't seen before the gallbladder surgery. The G.P. told me to continue with Questran, and to take Imodium when necessary and if the frequent bowel movements continued six weeks post-op, I should call the surgeon back again.

The frequent bowel movements continued and I was losing more weight. I phoned the surgeon, and was told that the "only" treatment for my problem was Questran and that I could have diarrhoea up to one year post-op or like 1 in 100 people, "unfortunately" I could have frequent diarrhoea for the rest of my life.

I returned to the G.P. that week and requested a referral to both a GI specialist and a nutritionist. I saw the GI specialist, who did a sigmoidoscopy in the office. He concurred with the surgeon that I continue with Questran 3-4 times per day and Imodium as necessary.

A few weeks later I had the first appointment with a nutritionist who tried by process of elimination to get me on track. I

continued despite a very bland, restricted diet, which included white bread and pasta, to have frequent diarrhea and was losing considerable weight.

The G.P. continued to send me for blood work every couple of weeks.

Over the summer of 1995, I went to three different Emergency departments. Each time I was given IV and discharged again on Questran. As well, the G.P. had referred me back to the surgeon again who did a colonoscopy, which showed "slight inflammation." I was also referred for a CAT scan, which was negative.

By late August 1995, I was becoming very debilitated, literally wasting. During a trip to Emergency, the doctor who saw me phoned the G.I. specialist on call that weekend and was told to give me papers to come in as a priority patient on Monday am. However, I was so debilitated by Sunday of that weekend, weighing 82 lbs., that I was admitted.

On the following Tuesday, after several tests and lastly the gastroscopy, I was told that I had celiac disease.

I remained in the hospital for approximately two and a half weeks on total parental nutrition.

As a registered nurse and daughter of a deceased general practitioner I was and am most grateful to be diagnosed.

Clinical Pearls

✓ Symptomatic celiac disease is sometimes triggered by stressful events like surgery, trauma, pregnancy and infection.

✓ Chronic diarrhoea is a common symptom of celiac disease.

✓ While sigmoidoscopy or colonoscopy is important to look for lower gut disease, an upper gastrointestinal endoscopy for small bowel biopsies (for celiac disease) should also be considered in the workup of chronic diarrhoea.

3

The typical gone atypical

(Age: 40's, female)

My story really began at age 15. I was a high school student, living on a farm in Ontario. The memory I have is of abdominal cramps and bowel inconsistencies. The family doctor had me admitted to hospital. I had a number of x-rays and was diagnosed as having Crohn's disease. The treatment was a pureed diet with rest for six weeks. The discharge menu was a Crohn's regime.

There was improvement on this diet. I was always somewhat anemic and had times of diarrhea or constipation. My nervous system did then and still does play a role.

When I was 44 I had a very severe upset. My stomach was constantly sick, as well as bowel problems and I lost much weight. At this time I was again x-rayed and also had an ultrasound. I was told that I was "on the verge of a duodenal ulcer" and was treated with Sulcrate. The medication was continued for a year.

For the next few years I remained reasonably well.

At Christmas time in 1999, I had a terrible flu involving both chest and stomach. The nausea was constant and I was losing weight. My family doctor told me my whole red cell blood profile was very low. This, along with weight loss and lethargy, got me referred to a gastroenterologist.

This doctor was fairly sure after meeting me that I would be diagnosed celiac. I had a blood test, which was strongly suggestive. The next step was a biopsy, and I was started on a gluten-free diet.

The results of the first biopsy also showed a duodenal ulcer, which was bleeding a bit. I was treated with Pantaloc for two months.

The second biopsy proved that the ulcer had healed and there was some regeneration of the intestine.

I am taking Sulcrate now, but less of it and seem quite well.

The results of the latest blood work are due this week and I am hoping for improvement.

I have gained six pounds and will continue on the gluten free diet. The change of food was harsh at first, but one gets used to it.

I hope my story will be of some help to you and to others.

Clinical Pearls

✓ Abdominal pain, diarrhoea, constipation, nausea, weight loss, anemia are all typical symptoms of celiac disease.

✓ With lack of awareness, even typical symptoms may appear atypical.

✓ Celiac disease is more common than Crohn's disease, ulcerative colitis and cystic fibrosis combined.

4

Everything in the front, nothing in the back

(Age: 13 years, male)

My son is now 13 year-old and was diagnosed at age 5.

He was first seen by a paediatrician in Emergency for suspected appendix at age 3. At that time his distended stomach and anemia were noted. Conclusion: he was constipated!

Seen six months later, it was suggested he do sit-ups for his distended stomach. Referred to a dietitian to look at his diet.

I asked the GP for referral to a new pediatrician.

He was seen at age 4. His projected height as an adult was 5'4". His father, mother and brother were all very tall.

We went back in six months. He was tested for cystic fibrosis and given fatty stool test. All negative.

Finally, at age 5 referred to a pediatric gastroenterologist in a larger centre for biopsy.

During six-week waiting period I asked if I could do a trial of a gluten-free diet. The pediatrician said that would be fine but to consult with a dietitian. Talking to her she strongly urged me not to as it skews biopsy results.

This child had short stature, wasted limbs, distended stomach, anemia, chronic constipation, small buttocks and brittle bones (he broke his arm at age 4 in a simple fall). For three years he said his stomach hurt.

No doctor ever suggested celiac disease until he finally saw a pediatric gastroenterologist. I did because a nursing friend said I should explore that possibility.

The pediatric gastroenterologist took one look at him in the pre-operative

examination and said that it was a very obvious case of celiac disease.

Clinical Pearls

✓ Abdominal distension is a common clinical finding in young children with celiac disease.

✓ Think of celiac disease if there is abdominal distension along with other features like anemia, chronic constipation, abdominal pain, etc.

✓ Abdominal distension + Wasted buttocks = Everything in the front + Nothing in the back!

5

Million-dollar workup

(Age: 63 years, female)

I am a 63-year-old female and I was diagnosed twenty-two years ago in 1978. At that time very little was known about celiac disease. It was good to finally have a diagnosis. I had never heard of gluten or celiac disease, so it was kind of a shock as well.

My health problems began in 1977. I began to lose weight. I was very tired all the time, had a mouth full of ulcers that never seemed to clear up. I was sick to my stomach a lot of the time. I had bruising of

the skin, which took a long time to fade and some diarrhea.

My GP ordered barium x-rays (stomach and bowel) and suggested a bland diet. But when my hemoglobin dropped from 11.5 to 6.0 within three months, she referred me to a hematologist. He immediately did a bone marrow test in his office and had me admitted to hospital that day.

I spent six weeks in the hospital for tests. I had daily stool collection, another bone marrow test, barium swallow, barium anemia and lots of blood work.

When I was discharged, the only diagnosis was that I was deficient in folic acid and was given medication. I remember, it was either the day before or the day I was being discharged, a gastroenterologist came to see me and was very surprised that I was being discharged. If I'd known then what I know now, I would have gotten referred to him after I got home.

My health did improved a bit, but twelve months later in 1978 I was back in the hospital for another five weeks for many of the same tests. This time, finally the gastroenterologist was involved.

I had the biopsy and the diagnosis of celiac disease.

So, in fact, it had taken twelve months to be diagnosed. This does not include the months under my G.P.'s care.

However, twenty-two years later, the diet is routine and I am well.

Before I was released from the hospital in 1978, the hematologist decided to give my hemoglobin a boost by giving me I.V. iron. Well, after only a few minutes into the treatment, I had an anaphylactic reaction to the drug. Thankfully, the intern was still with me and took care of me. I'll never forget that day as long as I live.

PS: I don't think of this condition as a disease, and prefer to call it Celiac Disorder.

Clinical Pearls

✓ Celiac disease should be a top consideration in anyone with iron deficiency anemia especially when no obvious source of blood loss can be found.

✓ A serological screening test for celiac disease is much cheaper than a bone marrow examination, stool analysis for fat or barium enema in individuals with iron deficiency anemia. The diagnosis of celiac disease must be confirmed by a small intestinal biopsy.

6

White as a sheet

(Age: 74 years, male)

In 1993, I had a triple bypass procedure. I had a hemoglobin (Hb) of 8 gm at discharge, and the Hb was very slow to rise despite taking oral iron. The Hb remained lowish, about 12 gm.

In 1994, I was investigated with an upper GI endoscopy and colonoscopy. The former showed a florid gastritis and the Aspirin dose was lowered from 325 mg to 80 mg.

A follow up gastroscopy showed no abnormality. Despite taking oral iron, my Hb remained in the region of 13 gm.

In 1996, I was again colonoscoped. This ?as a follow up as the Hb remained unchanged.

About a year later, I began to notice abdominal discomfort, which was centred largely in the upper abdomen along with slight feeling of nausea. My bowel habit for years had been intermittently costive (*constipated).* The symptoms were considered to be secondary to the diverticular disease.

In 1999, because of the persistent abdominal discomfort and the nausea, I was again referred to a GI specialist. Because of the persistent iron deficiency anemia, he undertook another colonoscopy.The findings were as before, only diverticular disease.

I continued with the oral iron, which I had been taking at this point for a number of years (thought to be possibly secondary to Aspirin). No explanation for the nausea was given. Symptoms of abdominal discomfort and nausea along with headache persisted.

In late 1999, I commenced to have diarrhea with watery brown stools, usually lasting a day and occurring every three weeks or so.

In May 2000, I commenced to have diarrhoea along with moderate abdominal

discomfort. But instead of clearing after twenty-four hours or so, this persisted.

Whilst on a visit to the U.K. the diarrhea became suddenly worse, with watery stools which were now yellowish and very foul smelling. During this period I was taking Lomotil plus Tylenol (for the abdominal discomfort). After nearly three days the diarrhea stopped, but the stools were pale, putty colored and very soft.

A second bout of diarrhea occurred some three days later, with fecal incontinence lasting twenty-four hours and followed by ten days of pale yellow, soft stools. These symptoms precipitated return to this country for investigation.

Prompt referral to a GI specialist resulted in a barium meal and follow-through, which showed some dilatation of the proximal small bowel. Some weeks later a gastroscopy and biopsy was undertaken.

The diagnosis: total villous atrophy. This was at the end of June.

Following a gluten free diet, I have not experienced abdominal pain or nausea and the stools are normal. I am awaiting result of the recent blood results.

Of interest may be the fact that between 1972 and 1980 I had intermittent symptoms of a distal colitis (lower 12 cms, as noted on sigmoidoscopy). These symptoms were controlled for the most part by a combination of salazopyrine and Betnosol enemas.

In 1980, I had a sigmoidoscopy which was normal. I remained symptom free thereafter.

There is no history that I know of anyone having symptoms of celiac disease in my family. Apart from a paternal grandfather who died of carcinoma of the stomach and an elder brother who died of pancreatic carcinoma aged 70, there is no other GI disease.

Clinical Pearls

✓ If gastroscopy is done as part of workup for anemia, duodenal biopsies should be obtained (for celiac disease) even if the mucosa grossly appears normal.

✓ Iron is absorbed primarily in the duodenum, which bears the brunt of injury in gluten sensitivity. A poor response to oral iron therapy should prompt a search for celiac disease.

✓ A lymphocytic colitis sometimes occurs in celiac disease.

✓ Inflammatory bowel disease occurs more commonly in individuals with celiac disease than the general population.

7

More of the same

(Age: 68 years, female)

I was diagnosed with celiac disease at the age of 49, in 1981. I am now a 68-year-old female.

I did not have any unusual symptoms at any time except for the fact that I was always noted to be very pale. I cannot remember ever being overly tired or having diarrhea or any other symptoms.

My gynecologist was concerned about my low iron count for about three years before I was diagnosed. He referred me to an internal medicine doctor for some testing. After having several tests, bone marrow

biopsy and other tests, the doctor could find nothing wrong.

So, another year went by.

When I went back to the gynecologist again, he was still concerned. He had me admitted to the hospital, where I stayed for one month. I had every test one could possibly have, and interviews from doctors over and over again. They finally zeroed in on testing for celiac.

After doing the stool test and then the biopsy, they all paraded into my room with the news that I had celiac disease! I had never heard of it before, but they were obviously very happy.

This diagnosis was made one month after being admitted.

Very soon after going on my diet, people were telling me how good I looked and what good color I had. That was very nice to hear after having always heard how pale I was.

Thank you!

Clinical Pearls

✓ Unexplained iron deficiency anemia should be considered to be due to celiac disease unless proven otherwise.

✓ Serological testing for celiac disease should be undertaken early in the diagnostic workup of iron deficiency anemia. A small intestinal biopsy is required to make a diagnosis of celiac disease.

8

Spare the biopsy, spoil the child

(Age: 68 years, male)

I am a male, 68-year-old and was diagnosed last year (1999) by a small bowel biopsy confirming that I was a celiac. I was put on a gluten-free diet. It changed my life.

Since I was a child, I remember having symptoms, abdominal cramps and diarrhea, sore bones, lactose intolerance, weight loss.

I was referred by my family doctor to other doctors and had many test and x-rays but none ever looked for celiac disease.

I am certain that I had celiac disease all my life.

Clinical Pearls

✓ Many adults with celiac disease recall having symptoms during childhood.

✓ In a child with abdominal cramps, diarrhea, sore bones, lactose intolerance and weight loss serologic screening for celiac disease should be considered and the diagnosis confirmed with a small intestinal biopsy. Serological testing is less reliable in children under three years of age.

9

Itching to be diagnosed

(Age: 66 years, male)

My symptoms began almost two years ago with a breakout of small, very itchy spots along the crease of my buttocks that did not respond to any over-the-counter creams or ointments.

After exhausting all available options I visited my family doctor.

His diagnosis was poison ivy, even though I was quite clear that there was no way that was possible. However, being desperate by that time, I applied the prescribed ointment with no results.

Subsequent visits over several months, many different prescriptions, and a modified

diagnosis of poison oak, proved equally ineffective.

I was then referred to a specialist in dermatology in …….., Ontario.

A biopsy indicated dermatitis herpetiformis.

As I am allergic to sulfa medications, I was prescribed Dapsone 100 mg along with a topical ointment Elocom 0.1%. Trial and error over several months, and countless blood tests, reduced the dosage to half tablet every three days.

I am now balanced and doing well, except for the occasional unexpected bout with foods containing gluten.

From the initial incorrect diagnosis, to the biopsy results taken by the specialist, was about 12 months.

In light of subsequent knowledge of my condition, I do not blame my doctor. He was only working with the training that he had received.

We have both learned the insidiousness of this problem.

Clinical Pearls

✓ Dermatitis herpetiformis (DH) is “celiac disease of the skin”.

✓ All that itches is not eczema. If a chronic, severely itchy, blistering rash does not respond to conventional therapy, think DH.

✓ A skin biopsy will help make the diagnosis of DH and a gluten-free diet will help alleviate the suffering.

10

The “auto” show

(Age: ? years, female)

May not be part of the picture, but my first health problem was in spring of 1993.

I was on a coach trip to New York City (NYC) with friends. They noted how little I was eating. In hindsight, that was a pattern. As I commented, “I have absolutely no metabolism because I never, ever lose weight”. But on that trip I developed edema of both feet and every stop at the hotel I elevated them but the edema continued even after home. So I went to my family doctor who ordered blood work & found a dead

thyroid and extreme anemia. In fact, he was somewhat alarmed and started Synthroid and iron.

I improved and I began eating.

In December 1994, friends and I flew to NYC and I noted I was beginning to have rushed trips to bathrooms notable only because public washrooms are rather scarce in NYC. But after coming home this became more and more troublesome. The doctor tested for food poisoning and by January, I was house bound.

The doctor made an appointment with an internist and he saw me quite quickly and I had been keeping a chart of my bathroom history. He ordered more blood work and a test of consuming tomato juice then blood work.

Another approximately six days of fat consumption followed along with stool collection. I was alone at home during the collection, which of course you know was a good thing.

On my first appointment with the specialist he told me he felt it was celiac disease so it was very hard to continue

eating the food he felt was causing the problem until after the biopsy.

But I was able to go on a planned trip to NYC in April 1995 and had my last ever bread made with wheat at the Russian Tea Room.

I feel perhaps the low hemoglobin in 1993 may have been partly celiac related. But when my thyroid was not active and I was put on up to Synthroid 0.175 mg and did well, I just thought the problem was solved.

I have had no related problems since following a gluten-free diet.

(Good luck with your review.)

Clinical Pearls

✓ Celiac disease is an autoimmune disorder.

✓ Individuals with autoimmune disorders (especially type I diabetes and thyroiditis) are at high risk of developing celiac disease.

Serological screening for celiac disease is recommended in these high-risk groups. A small intestinal biopsy is necessary to make a diagnosis of celiac disease.

11

One end empty, other end full

(Age: 69 years, female)

I am a female, just turned 69 years. I was diagnosed with celiac disease (then known as sprue disease) in 1969.

From very early childhood I had been sickly, pale, underweight and lost a great deal of school due to lack of appetite and vomiting. I recall crying many times with stomach cramps and suffering from cankers on my tongue.

My relief would come when my parents were able to get me to a doctor and he would give me IRON, sometimes a red

liquid, sometimes a thick syrup-like medication. Very quickly I would respond and would be excited as food now smelled and tasted good.

During my teen years this condition seemed much better. Still I was not of average size. At age 18 years, I had my first menstrual period and I grew inches.

I was married at age 24 and felt well, had five children in seven years (all Sections).

Around 1968, I ran into problems; very tired and losing weight. I went often to my doctor seeking help. I was teaching in school at the time. The worst part of my day would begin around 3:30 p.m. Gurgling would start in my stomach, an overall miserable feeling. I could eat a good dinner, but shortly after I would have to change into something loose, as I would become very bloated. At times this was accompanied with severe diarrhoea.

This continued for months, yet I was able to teach.

Finally, I gave in to going to an out-of-town hospital where I could be checked by a specialist. I spent close to three weeks in hospital. During that time, I was asked to sit

before huge room full of doctors to answer questions.

I was later told that my case was borderline rare. I was asked to return in three months. I had gained eight pounds and was feeling fine.

Again, I returned a year later.

My specialist made the comment, "You don't look like the same patient I treated over a year ago."

Good doctors are a gift from God.

Clinical Pearls

✓ The time from first symptoms to diagnosis of celiac disease is often a decade. This unacceptably long period can be reduced by increased awareness and use of screening antibody testing.

✓ Even in children, iron deficiency anemia (especially beyond infancy) should prompt a workup for celiac disease.

12

As classical as it can get

(Age: 65 years, female)

To my knowledge, I did not have any symptoms of celiac disease as a child or as a young teenager.

In first-year university, when I was 18, I went to give blood at a Red Cross blood drive. It took me ages to recover my energy. When I went to the next blood drive they would not accept my donation because the iron content was too low. They suggested that I not give blood in the future.

After that time a number of doctors prescribed iron pills, which I took for more

than 40 years. During that time I would have periods of exhaustion, when just walking up the stairs would necessitate my stopping at the top to let the muscle pain disperse. I also had periods where I felt much better.

In the 1970's, a doctor started me on a series of intramuscular iron injections. I finally developed an aseptic abscess on my hip and the injections were discontinued. The doctor suggested that I start a series of blood transfusions, but I refused (and in retrospect, I realize that I might have saved myself from an even worse fate).

One problem I had during the summer, for many years, was swollen ankles, but this usually righted itself in the cooler weather.

Although I did not recognize it at the time, I suffered from steatorrhoea for many years, and of course had very smelly feces. I tended always to wait until the family had left home in the morning before I went to the bathroom. If I were in a public washroom, I would always choose the toilet that was closest to the exhaust fan. Of course, I regarded all of this as normal and would never have discussed it with anyone in the family or the medical profession. I did

once mention to the doctor that my stools were very large, but there was no discussion about this.

In the early 1980's, a doctor found that my folate levels were exceptionally low and prescribed folic acid supplements, which I took for a time. In the mid 1980's, as part of my …… study on the impact of dietary salt on calcium excretion in seventeen post menopausal women, my 24-hour calcium excretion (on all six tests) was approximately ten times lower than the rest of the women in the study. My serum albumin levels were also very low. But the doctors involved did not identify these as a concern.

In the late 1980's, I went to the doctor with exhaustion and terribly swollen ankles. My iron and folate levels were very low. I had upper and lower barium examinations, tests for liver, kidney and heart function. I had a low thyroid function and was put on thyroxine.

I finally went to an internist who after five minutes of questioning thought I likely had celiac disease.

I was biopsied on …, 1989 and just from visual examination of the specimen, the doctor said I had celiac disease.

I have had two bladder repair operations and wonder if they might be related to connective tissue deterioration. I am developing osteoarthritis, which might be related to old age (heaven forbid) rather than celiac disease. Last year I had a non-malignant tumor removed from my colon.

I discovered two years ago that at 78 years of age my father developed severe diarrhea (two years before his death) and was put onto a gluten-free diet, which helped his condition. I am not sure that he was definitively diagnosed with celiac disease. My brother died of cancer of the intestine when he was 44.

My sister thinks she has celiac disease, and follows a strict gluten-free diet, although she was not biopsy diagnosed.

My husband and I had three boys. They are very healthy, and I now realize how fortunate we are that they did not have neural tube defects, or any other complications. One of our sons (now 32) has just been biopsied for celiac disease and has

shown no flattening of the villi. Another of our sons (now 33) has also had a blood-screening test for celiac disease, and it too was negative. But both have digestive symptoms from time to time and I have a suspicion one or both might develop celiac disease in the future, but I hope not.

Clinical Pearls

✓ In iron or folate deficiency anemia, think of celiac disease.

✓ Peripheral edema in celiac disease is due to hypoalbuminemia secondary to the protein losing enteropathy.

✓ Individuals with celiac disease can develop osteoporosis from malabsorption of calcium and vitamin D. In patients with osteoporosis, celiac disease should be considered as a diagnostic possibility.

13

Choosing the right parents

(Age: 43 years, female)

I was a girl about 4 years old (1962) when I started having problems.

I remember in Grade 1 being sent home from school because I had vomited. I missed a lot of school time. My mother tells me that she brought me to the doctor just about every week. He suggested several options but continued to think that the problem was mental, my mother must have liked my older sister better and I was suffering emotionally.

I continued to have classic symptoms; anemia, weakness, vitamin deficiency,

diarrhoea, steatorrhoea, abdominal pain, bloating, vomiting and weight loss.

Finally, these symptoms reached a point where the doctor had to agree something was wrong. I was hospitalized when I was 9 but no cause could be found. I had vitamin injections every week and my mother fed me the best food in the house.

A couple of months later, I was hospitalized in Halifax. Many doctors and tests later, nothing helped. I had lost so much weight and was dying of malnutrition.

I was about to be sent home to die when a young doctor was consulted. He made enquiries to the Netherlands for more information.

I was put on the gluten-free diet and sent home. My mother followed the diet very strictly.

I improved dramatically. I gained weight and grew. The doctors had predicted that I would always be short, but I grew by leaps and bounds. At periodic checkups, the doctors were astounded at my progress.

As a teenager, I tried to find out as much as possible about celiac disease. After high school, I worked as a lab technologist

and came to know a gastroenterologist there. With his recommendation, I took the gluten challenge (1979). The biopsy proved inconclusive but, in retrospect, the biopsy was performed too early. The pain was caused by the addition of yeast to my diet, not the gluten.

I continued on the celiac diet.

My first child was born and I watched constantly for signs. There were none. My second child started to have problems around three years of age. The biopsy was very difficult to do and proved to be inconclusive. He is now 16 and remains on the celiac diet because he has pain when he deviates. (He will have my consent should he decide to do the gluten challenge to get his diagnosis). My third child has no symptoms.

In 1999, I took another gluten challenge. I had been to a Celiac Association conference and came home very sick. The only thing different was the yeast in the bread products so I wondered if my entire problem was with yeast. This biopsy was positive for celiac disease.

I am now 43 and have been on the gluten-free diet for thirty-four years.

All five of my siblings are symptom free. My parents are from the Netherlands and sometime after I was on the diet, my uncle in the Netherlands was diagnosed with celiac disease. He had always been a sickly child and young adult.

Clinical Pearls

✓ "Whether you develop celiac disease or not depends on how carefully you chose your parents"!

✓ Celiac disease is a hereditary disorder. Both first and second-degree relatives are at high risk for having celiac disease and should be screened.

✓ Highly sensitive serological tests (tissue-transglutaminase antibody and endomysial antibody) are currently available to screen for celiac disease. The definitive test to diagnose celiac disease is a small intestinal biopsy.

✓ The treatment of celiac disease is a strict gluten-free diet for life. The diet should not be started before the confirming the diagnosis with a small intestinal biopsy.

14

Mouth: The doorway to the gut

(Age: 37 years, female)

I am 37 years old and female. I was 35 when I was diagnosed.

I can remember being extremely irritated as a young child (ages 4-5), a lot of dental problems. My baby teeth were removed in pieces by the dentist, not a pleasant experience.

Around age 7, I had a lot of cankers and continued to have episodes of them until I was diagnosed. I remember it was age 7 because they were so severe I was taken to

the doctor and received a medication to try and clear them up.

I was always considered tall for my age and very thin. At age 16, I was diagnosed as "anemic" and was told I would require iron supplements forever. During my late teens, I did seem to finally have some "extra" weight but that was only from age 17 to about age 20.

In hindsight, I suppose I was "late" developing, as I did not get my first period till I was 13 ½ years old. My energy levels seemed OK when I was a teenager, but I recall having a lot of bloating and "stomach noise" while in high school.

During my 20's till the time of diagnosis, I seemed to be fatigued a lot and this was very stressful.

In the year prior to being diagnosed, I had two severe episodes of cankers on my tongue. I could hardly eat for a week each time. The first episode, I saw a doctor who "burnt" the cankers, and prescribed a mouth rinse. The second occurrence was exactly six months later and, on this occasion, a different doctor just prescribed the same mouth rinse. Due to the painfulness of these

episodes, I put myself on a diet of jello and porridge, as these were the only foods I could stand to eat.

Also, in the year before my diagnosis, I had a severe reaction to a hornet sting on my index finger. Having had no prior reaction to any types of stings, I was shocked when my whole hand became hot and itchy and swelled like a balloon. It took a week to recover from this. Benadryl and bed rest was the treatment.

I thought I had the flu in the weeks before I was finally diagnosed. I had diarrhoea for several weeks before I went to my doctor. I was told to have only liquids for a few days (to rest the bowel) and to provide a stool sample for the lab. Ten days later my lab results were fine, but I was much weaker and still having diarrhoea. My doctor got me into the specialist in less than forty-eight hours.

I was diagnosed immediately.

My weight over the last year had gone from 130 lbs to 105 lbs. Presently, my weight is 116 but it seems difficult to keep it up to that level.

During the ten years prior to my diagnosis I had severe leg cramps, especially at night. I wonder if others experience the same thing.

Clinical Pearls

✓ Recurrent oral aphthous ulcers (canker sores) is a common manifestation of celiac disease.

✓ Dental enamel defects can occur in children with celiac disease. An astute dentist can suspect the diagnosis (if aware of celiac disease)!

15

Looks can be deceiving

(Age: 46 years, female)

Hi. I'm Celiac, 46 yr old female diagnosed three years ago.

When I was young, I always had problems going to the bathroom. My teenage years, as I remember, were a nightmare. We walked to school, came home for lunch and were nearly always late getting back. I had constipation so bad I couldn't pass the stool, although I needed to. I've always had a weight problem.

I had several barium x-rays. They are the worst things.

As I got older I started to have diarrhoea a lot. Four years ago, it got really bad, sometimes 6-7 times a day. Soon after eating and at work I would get so tired and bloated, sick with nausea and very severe irritability.

Finally, after being so sick and many visits to my family doctor at age 43, he sent me for the biopsy.

I'm still overweight but so are the other women in my family.

I stick to the gluten free diet and after only one week on the diet, I felt much better.

Some people say I don't fit the bill for celiac disease because of my weight but don't be fooled. Not everyone has anemia. My blood work is good.

I have found out this year that I have fibromyalgia, the widespread pain and fire spots all over. Testing by a doctor in Halifax I had fifteen of the eighteen tender points.

I am also lactose intolerant.

The chronic fatigue is a great cross to bear as well.

I hope this helps, help some one else who is different.

Thank you.

Clinical Pearls

✓ Being normal weight or overweight does not rule out celiac disease.

✓ The classical images of those thin, wasted and severely malnourished individuals with celiac disease seen in textbooks are a rarity in clinical practice.

16

The Long and Short of it

(Age: 85 years, female)

I will make this brief.

I believe I have probably always been a Celiac, especially since my teens for I had frequent bouts of diarrhoea and abdominal pain.

However, at 28 years of age I gave birth to my second child. Five months later, my husband came close to death with polio (He made a reasonable recovery but never was whole again).

Six weeks later I developed continuous diarrhea.

For 16 years I fought this with many remedies and weeks in hospital while doctors tried to learn what was the cause.

My husband had an engagement in Europe. I hesitantly accompanied him.

Two weeks later I was dreadfully ill, was flown to London, UK where after two weeks they determined that I had gluten intolerance. I was then 45.

Gastroenterologists were only beginning to realize that celiac disease was not only an infant problem.

I am now within months of my 85th birthday. I am very strict with my diet for I am also lactose intolerant.

I am small, 5 feet tall and at most 95 lbs.

I feel I have done very well though still frequently have very loose bowel, often with pain. But I feel quite well most of the time.

Clinical Pearls

✓ Children with celiac disease can present with short stature in the absence of any other symptoms.

✓ Celiac disease is more common as a cause of short stature than all other organic causes of short stature.

✓ All children with short stature should be screened for celiac disease. Don't be swayed by "familial" short stature. Others in the family who are short may also be suffering from celiac disease!!

Suggested Readings:

- National Institutes of Health Consensus Development Conference Statement on Celiac Disease, June 28–30, 2004 *Gastroenterology* 2005;128:S1-S9

- Guideline for the Diagnosis and Treatment of Celiac Disease in Children: Recommendations of the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. *J of Ped Gastroenterol Nutr* 2005;40:1-19

- Canadian Celiac Association web-site *(www.celiac.ca)*

“In his book "Celiac Disease: The Road to Diagnosis"; Dr Rashid chronicles the true stories of the journey many individuals with celiac disease make before being diagnosed with their condition. The stories that these individuals share through this book allow us all to better understand the truly diverse presentations of celiac disease and their negative impact on the lives of everyday Canadians. By sharing their stories with us, these individuals have helped us to learn and understand more fully what they have endured before being diagnosed with celiac disease. This extraordinary collection of stories will touch your heart while providing important information to help busy health care providers consider a diagnosis of celiac disease in an individual with similar symptoms.”

Dr. Connie M. Switzer MD, FRCP(C)
Clinical Professor of Medicine
University of Alberta, Edmonton, Alberta, Canada
Chair, Professional Advisory Board,
Canadian Celiac Association

"What a compilation of stories. Sad but true. Long periods of symptoms prior to diagnosis and even hospitalizations! Reading these stories should prompt the health care administrators to introduce routine testing for celiac disease to save the expense of testing and hospital care. The stories are a reflection on the medical education system that teaches celiac disease is rare. In fact, celiac disease is common occurring in about 1% of the population. Simple blood testing can bring the diagnosis to light and reduce the long duration of symptoms and patient misery. Every medical student should read these stories and learn the lesson of celiac disease, the modern imitator. There is currently a hidden epidemic of the disease, hidden by lack of physician awareness of the disease.

Dr. Peter HR Green MD, FRACP
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