

Discovering new ways to improve the quality of life for Canadians with ankylosing spondylitis

AS is a severe, painful form of arthritis that affects more than 400,000 Canadians

Chris Atchison



Gerald Major has undergone surgery seven times to address joint damage caused by ankylosing spondylitis.

Gerald Major was about 12 years old when his body began to change. And not solely in the typical way that a teenager's body transforms and grows, sometimes awkwardly, before settling into adulthood. This was different.

Gerald, a hockey player and top-performing track and field athlete, began to feel that his knees were "loose" after competitions and training. He put it down to an active lifestyle and rigorous athletic training. Soon, recovery times began to lengthen, and his ankles also began showing signs of stress.

"My older brother had knee issues, so I wrote it off," the Oakville, Ont., resident recalls. "Medicine was different then."

It wasn't until he was about 21 years old that Gerald was diagnosed with ankylosing spondylitis (AS), a severe form of arthritis that affects more than 400,000 Canadians.

The disease causes the immune system to become overactive, attacking the joints of the spine and pelvis, and causing the formation of bone spurs. It can also eventually lead to spinal fusion and back curvature, and in severe cases, paralysis – usually from injury.

Common symptoms include pain and fatigue, as well as inflammation of the eyes. A recent study by researchers at University Health Network (UHN) also found that there is a higher rate of cardiac- and stroke-related mortality among AS patients than those without the disease.

And AS is about three times more prevalent in men than in women.

Now in his mid-40s, Gerald, a former hedge fund executive, has been on long-term disability for five years and has undergone seven surgeries to address joint damage from his AS. Another operation is on the horizon.

He says that the stress and discomfort of living with the disease eventually began to take a psychological toll.

"When anything physical hits you like that, it'll eventually affect your mind," he says. "I think eventually my mind took me out of work. My body was just a mess. I was crumbling."

But there is increasing hope for patients such as Gerald, thanks in large part to rapid advancements in the understanding, detection and treatment of the disease.

While there is no known cure for AS, advanced biologic drug treatments – genetically engineered medicines using



Dr. Nigil Haroon, left, and Dr. Robert Inman say AS symptoms, which mirror those of back pain, can be missed by physicians and result in diagnostic delays.

molecular biology – are producing rapid improvements in quality of life for those living with AS. In some cases, the medicines are delaying the progression of the condition.

"These drugs can often be life-changing in terms of quality of life," explains Dr. Robert Inman, a rheumatologist and co-director of the UHN Spondylitis Program.

"AS patients will sleep poorly, they'll wake up with morning stiffness and during the day, the pain and stiffness take a major toll on their functioning. If you look at biologic trials, significant improvement in back pain, which is the primary outcome, is achieved in about 65 per cent of patients."

Those same patients are also displaying significantly better outcomes when treatment is begun early.

But Dr. Nigil Haroon, also a rheumatologist and co-director of the UHN Spondylitis Program, notes that despite major improvements, detection and diagnosis of the disease remain a challenge. That's because AS symptoms often mimic those displayed by people with chronic back pain, and the symptoms can be easily missed by physicians, sometimes resulting in diagnosis delays of five years or more.

"Imagine a busy family physician or orthopaedic surgeon seeing hundreds of back-pain patients. How do you make that diagnosis when there is no dependable distinguishing feature?" he asks.

"Not everyone can be referred for AS. We can talk about young age, morning stiffness and similar AS-like features, but

in practice, they do not help much. Even MRI [magnetic resonance imaging] has its limitations, and early back pain, with its very subtle changes, is very difficult to see in an X-ray."

Then there's the lack of a specific test for AS, leaving the diagnosis up to a clinician's professional judgment and expertise.

"AS-related conditions affect about [up to] 1 per cent of individuals in Canada, compared to the more than 40 per cent of individuals who have chronic back pain at some point in their life," Dr. Haroon points out, underscoring the potential for misdiagnosis.

In addition, many young patients will initially seek assistance from physiotherapists or chiropractors – clinicians who may not be as experienced in AS detection and diagnosis – for what they believe to be simple back pain.

"It's a healthcare system challenge," Dr. Inman comments.

That's why Drs. Inman and Haroon are working with physiotherapists, chiropractors and family practitioners to track a better model for patient referral, with the focus on referring the right patient at the right time to a rheumatologist, while also producing educational materials to boost understanding of the disease among healthcare professionals.

Developing personalized biologic treatments remains another priority.

"To do that," Dr. Inman says, "we use several different modalities, including the clinical profile of the patient, but increasingly genetic and immune profiling.

We look at which aspect of the patient's immune system is turned on at that time that would allow us to target treatment to that particular patient. The hope is that by doing so we can develop drugs which not only control the disease but cure it."

Their team is also working diligently to advance diagnosis and treatment of AS, with a focus on creating change through their research.

The UHN Spondylitis Program is taking a lead role in studying the gene HLA-B27 (a gene present in the majority of AS patients), for example, with a focus on what activates the gene in some patients but not others. Other researchers are looking at what role, if any, Macrophage Migration Inhibitory Factor (MIF) – which is involved in bone formation and is found in much higher levels in AS patients – might play in the progression of the disease.

A better understanding of MIF could also improve AS detection and diagnosis.

For Gerald, the biologic drug Infliximab, which he's been taking for nearly 15 years, has offered relief from AS symptoms and a far greater quality of life. He goes as far as to call its effects "game-changing."

But he feels that the key to managing and living a productive life with a disease such as AS – along with drugs and other therapies – is for patients to educate themselves and become active in the management of their illness. He now volunteers his time to teach a pain-management class for fellow arthritis patients.

"My message is, 'Get engaged and take care of your health,'" he says. ■