Diagnosis, support, and hope: A recipe for managing rare diseases

Consult your family doctor if you or a loved one may be experiencing symptoms commonly associated with hATTR amyloidosis.

CORD Canadian Organization for Rare Disorders

As with so many rare diseases, Canadians with hATTR amyloidosis struggle to both receive a proper diagnosis and access effective treatment.

Anne Marie Carr was 53 years old when her body started giving her signals that all was not well. It was 2011 and she was climbing the stairs of her local Ancaster, ON library when suddenly she got very dizzy and had to sit down. From there her symptoms progressed to weight loss, diarrhea, heart palpitations, and tingling and pain in the hands and feet often mistaken for carpal tunnel syndrome.

She was convinced from the beginning that these symptoms were all connected, but it took her years to finally get an accurate diagnosis. Carr has hereditary ATTR (hATTR) amyloidosis, a rare inherited genetic disease that impacts generations in families.

"In hATTR amyloidosis, a mutation in blood cells and abnormal genes cause the formation of a toxic protein which produces insoluble fibrils that clump together and block up different organs," explains Marsha McWhinnie, Director of the (CASN). "There's no rhyme or reason as to which organs it'll affect. When it affects the heart, the heart walls thicken and you get symptoms of heart failure. The symptoms mimic many other heart diseases, making it especially hard to diagnose."

For Carr, it took four years from the onset of symptoms until her diagnosis in 2015, which is particularly worrying because untreated hATTR amyloidosis is progressive and often fatal within 3 to 15 years of symptom onset.

Keeping hope alive

Some people, however, do live with amyloidosis for extended periods. Spreading this message of hope is part of why McWhinnie founded CASN after her own amyloidosis diagnosis in 2000. Although amyloidosis and hATTR amyloidosis are fundamentally different, McWhinnie has become a valuable and trusted first point of contact for those with the rarer, hereditary form of the disease.

For people living with rare diseases, any ray of hope or contact with others facing a similar struggle can have a huge impact. CASN runs a nationwide toll-free hotline for people living with forms of amyloidosis. It was through this hotline that Carr first contacted McWhinnie, who then connected her with educational resources and the names of leading specialists in her area.

Living with a disease like hATTR amyloidosis isn't easy, but support and education can make the difference between desperation and optimistic living. Carr struggles with her illness every day, but she's able to make the most of her life because she stays well-informed and connected via online message boards, social media, and in-person support groups. "Whenever there's a good day, my philosophy is to just enjoy it," she says. "I try not to think about the things I can't do anymore, whether that's going for a bike ride with my husband, Mervyn, or walking around the neighbourhood with my granddaughter." Despite the financial and emotional challenges brought on by the disease, Carr remains positive. "I focus on the things I can do," she says. "When you focus on the things you can't do, it just brings you down."

And because she has a support network, through CASN and also within her own family, Carr is able to thrive despite the limitations of the disease. "It's an exciting time right now with all of the work that's being done and several new therapies coming down the pipeline," says McWhinnie. "There are a lot of new theories about how to treat this disease."

Becoming your own advocate

The first and most important step, however, is education. "I feel strongly that we need to address the fact that most GPs don't know about hATTR amyloidosis," says Carr. It's with this same tenacity and insistence that she urges all Canadians to embrace their role as the best advocate for their own health and well-being. "Looking back, I remember little things—like tingling in my fingers—that I should have consulted my physician about," she says. "I often wonder if I would've been diagnosed sooner had I reported these symptoms when they first appeared."

The fact remains that Canadians and their health care professionals need more information and better tools to allow them to diagnose rare diseases more promptly. With greater awareness, we would be better equipped to provide the best relief possible for those with hATTR amyloidosis and other rare diseases. And that could allow us to keep people alive and independent while new treatments are approved and access to these treatments is secured. That's a possibility worth fighting for.

Common symptoms include:

- Bilateral carpal tunnel syndrome - numbness and tingling in the hands and arms
- Difficulty climbing stairs
- Progressive weakness, numbness and pain beginning in feet and hands
- Nausea, vomiting and diarrhea
- Cardiovascular symptoms

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