Disappointments in medicine

Lack of effective treatment for neurological disorders like ALS

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One of my greatest disappointments in medicine during my long career as a general practitioner has been our inability to deal with certain horrific illnesses, in particular neurological illnesses like amyotrophic lateral sclerosis (ALS). While patients with ALS are best cared for by an interdisciplinary team of professionals familiar with ALS, it is most helpful when family doctors are involved and educated about ALS care, especially when patients do not have convenient access to a specialty clinic in their geographic location.

About ALS

To refresh your memory, ALS is considered to be a rare disease owing to its low incidence of approximately 2 in 100 000 patients per year. This is the same incidence as multiple sclerosis in Canada, but the mortality rate for ALS is equal to the incidence, resulting in a much lower prevalence of 6 to 8 in 100 000 patients. For unknown reasons ALS attacks either the central nuclei of the brain or the anterior horn cells of the spinal tract, causing a wasting of the motor muscles initially, then eventually leading to muscle death. From the time of diagnosis, patients live about 3 to 5 years on average, but 20% of patients live beyond this period and some live a year or less. There is no treatment to effectively slow the progression of ALS or repair damaged nerves. Death is most commonly due to respiratory failure caused by severe deterioration of the muscles involved in breathing.

Prime mover

The problem with ALS is that it can be difficult to diagnose because there is no single definitive test—other conditions have to be ruled out, and it can present differently among individuals. For instance, the part of the body in which it starts and the rate of progression vary. Some things are typical though, such as asymmetrical presentation. The criterion for meeting an ALS diagnosis is evidence of both upper and lower motor neuron involvement. I encountered 2 very different ALS presentations in my own practice.

My first encounter with ALS came when a friend of mine, who was also a long-time patient, came to my office complaining of increasing weakness of his legs. While examining him, the first thing I noticed was the severe loss of muscle in his legs. I asked him to squat, which he did, but he needed to use his hands and arms to stand again. As he lay on the table I saw early fasciculation of his thigh muscles. He had very little leg strength and was not able to raise his legs perpendicularly. In keeping with his disease there were no sensory changes. I knew the diagnosis but I sent him to a colleague for confirmation. He died a few years later.

The second case was not so simple. Twelve years ago a patient presented with severe burning of his mouth. I examined him on several occasions and sent him for a battery of tests. I found nothing of note. I referred him to different neurologists, but there was no definite diagnosis made. He was prescribed topical liquid medications, but nothing helped him. After I retired he was referred "out of province," and a diagnosis of ALS, bulbar type, was made. I was curious about how this diagnosis was arrived at, and because I knew him and his wife well, I visited him at his home.

I was surprised to see what had happened to him during the interim. He now had speech difficulties; he drooled; his mouth was sore and painful; and all his food had to be puréed because of swallowing difficulties. His legs were now spastic, and he was hyperreflexic.

Despite all his problems he is always in good spirits. I visit him regularly. He walks inside and outside, with the aid of a walker. His hands and arms have been spared. He is very alert. He fights this illness with all the strength he can muster. He knits to preserve his hand function. In his basement he has a system of pulleys that he uses to help keep his arms strong. I believe his will to live has put him well outside the typical life expectancy range. He is plagued with severe coughing spurts leading to near-choking episodes, especially when eating. He might soon need a feeding tube. These episodes are terrifying to his wife, who has a few comorbid illnesses herself.

Seldom happens

Because the incidence of ALS is low and the population of Newfoundland and Labrador is approximately 500 000, a doctor practising in this area might never see a new ALS case in his or her practice. I was just a little unlucky in one way; however, these patients taught me a great deal and I cannot say enough about their caregivers—but that is a whole other story.

Dr Yarn is a retired general practitioner in Corner Brook, Nfld.

Competing interests
None declared