My Life With Fibromuscular Dysplasia

A Sword of Damocles

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My story begins in December 2007 when I visited my general practitioner (GP) for one of the first times since the birth of my third son in 1992. At the time, I worked as a GP myself and had been at the same busy Scottish practice since qualifying in 1986. I also led an active life outside of work, ran a youth group at church and enjoyed regular gym workouts and several sports. Being a mother of 3 boys kept me constantly on the go! I had never suffered any major illness, was on no medication, and had no obvious risk factors for heart or vascular disease.

Having suffered 3 months of morning headaches and some elevated blood pressure readings (around 170/100) taken at my office, I felt I should seek medical advice. I was also experiencing extra heart beats (ventricular ectopics) fairly frequently and some mild left-sided upper back discomfort. Some basic blood tests were done, along with another blood pressure reading, and I was referred to a cardiologist. The cardiologist performed an EKG, which showed normal patterns other than frequent ventricular ectopics. My blood pressure readings were mildly elevated at 158/100 and later 142/95. No heart murmurs were found, and an echocardiogram was normal. An appointment was arranged for me to have ambulatory blood pressure recordings to see how my blood pressure behaved over a 24-hour period, and an initial diagnosis of white coat hypertension was made. This is defined as blood pressure that is raised when measured by medical professionals but normal outside the doctor’s office. My ambulatory blood pressure recordings were within normal limits, and no further action was taken. My morning headaches and occasional palpitations continued, but I was able to manage the headaches with simple analgesics.

In September 2009, at age 49, I suffered severe pain on the left side of my neck. A month later, when it had persisted, albeit at a lower intensity, I saw my GP, who referred me to physical therapy. I was unsure whether the neck pain had aggravated my headaches this time around but decided to arrange for my ambulatory blood pressure recordings to be repeated as I had checked my blood pressure at my office, not expecting, but more information was needed to clarify the exact diagnosis. Meantime, I was started on a medication for my blood pressure, which had remained high throughout. This same medication was also used to slow my pulse in an effort to reduce the pulsing sound in my head. Magnetic resonance arteriography was done a few days later, and this revealed abnormalities in all 4 of the arteries supplying my brain. The worst affected was the left internal carotid, which showed a narrowing over a 4.7 cm length of the left carotid artery with a bulging area (aneurysm) above this. Again, vasculitis was suspected, but more information was needed to clarify the exact diagnosis. Meantime, I was started on a medication for my blood pressure, which had remained high throughout. This same medication was also used to slow my pulse in an effort to reduce the pulsing sound in my head. Magnetic resonance arteriography was done a few days later, and this revealed abnormalities in all 4 of the arteries supplying my brain. The worst affected was the left internal carotid, which showed a long tight section with a split (dissection) further up and an aneurysm in that region too. There was also a dissection in the left vertebral artery, with an accompanying aneurysm in that area. Further tests revealed that my renal (kidney) arteries were also affected. My consultant, who was surprised at the vascular scan result, discussed my case with several specialists around the country, and the conclusion was that this was fibromuscular dysplasia (FMD). I left the hospital feeling scared of what lay ahead. My arterial system sounded like a train wreck!

FMD is a condition that affects the arterial walls of mainly the renal and cervical (neck) vessels of the body, although any artery can be affected. Abnormal cell overgrowth occurs, which can cause narrowing of the arteries and a reduction of blood
supply to the organs supplied by the affected vessels. Arterial tears or dissections can occur and also arterial aneurysms. Many patients have symptoms of neck pain, headaches, and raised blood pressure at presentation. Patients affected by FMD are also at greater risk of stroke and renal failure and other consequences of reduced blood supply to vital organs. FMD tends to be diagnosed most often in women between the ages of 30 and 50, although it may be present in the system long before then, and it can also affect men and children. The cause is thought to be because of multiple factors, and there may be a genetic element too in some cases. Studies are currently underway to further the understanding of causes and triggers, and there are several patient registries that are collecting information to help this.

At diagnosis, my doctor explained to me that this condition was rare and that my major risk would be of having a stroke. He described the tight section of my left carotid artery as a Sword of Damocles regarding the uncertain prognosis. I was seen by a vascular surgeon also, who explained that the risk of operating on the tight area would outweigh the potential benefits. My arterial abnormalities were extensive and would be awkward to access. During any operative procedure, my brain function could be compromised, leading to stroke or death. My management, therefore, was to be through medication and regular review and scans instead. Before discharge from hospital, as well as the blood pressure medication, I was given a piece of paper with the website of FMDSA (Fibromuscular Dysplasia Society of America) handwritten on it. This turned out to be one of the most valuable scraps of paper I have ever been given. I was told that this website provided the best information on the condition. My doctors encouraged me to contact the hospital immediately if I had any worsening of symptoms or neurological problems. This was of reassurance to me and made a difference in terms of the initial fear and isolation I felt in being diagnosed with such a rare, incurable condition. My hospital doctors were not accustomed to treating patients with carotid FMD. As far as I am aware, I was the first such case they had come across. My colleagues in general practice had never heard of the condition.

At the start, I felt utterly numb and scared. I was emotional at times with a new and deep sense of my own mortality. It took me ≥6 months before I had the courage to look at the FMDSA site to seek further knowledge about my illness. Once I had made contact with Pam Mace, the executive director and herself a sufferer of FMD, she was able to reassure me that there were other patients out there who had this condition and who led reasonably normal lives. She also linked me to a fellow patient in Australia who had similar FMD distribution as mine and who had also been deemed unsuitable for surgical intervention. It was such a reassurance to find people who were living with the same condition and leading stable fulfilling lives many years after diagnosis.

Now, >6 years since my initial diagnosis, I am still alive and well and have had no neurological or stroke events to date. My blood pressure is controlled using 3 tablets daily, along with a statin and an anticoagulant. Unfortunately, my blood pressure can drop to low levels and also can peak in certain situations of stress. This makes fine-tuning of the treatment difficult. I, therefore, choose to manage my life in such a way as to attempt to avoid major stresses on my body. For me, this involves leading a quieter life than before diagnosis. I stopped work after diagnosis because, in my particular role and with these arterial tears and aneurysms, continuing was not compatible with keeping steady blood pressure control, an essential part of maintaining my health and life. Leaving work so abruptly felt like a bereavement because I loved my job and found great fulfillment through it. However, I was acutely aware that if my symptoms had presented in a more sudden and severe way, I could have died suddenly of a stroke. The Sword of Damocles could have fallen and slain me in an instant. My focus now had to be on managing my blood pressure, keeping stress levels low, and just “being” instead of constantly “doing.” Once I had set my affairs in order, my priority was looking after my own health as best I could and being around for my family for as long as I was granted. I have become good at pacing myself and prioritizing things and events to manage my symptoms.

My diagnosis affected the whole family in terms of coming to terms with this independent, busy, working mother suddenly being ill and having to adopt a different kind of living pattern, thus, also giving the family routine a different shape. I had to take things very easy at the start (first 3–6 months) when the dissections were first discovered, in the hope that they may heal. My sense of my own mortality affected all of us and probably brought us even closer together in that sense. My family continue to be very supportive. My husband and I have since had the joy of seeing our boys graduate from university, marry, and even give us grandkids. These are things I never thought I would see and experience when my diagnosis was first given to me.

My symptoms now are mainly long-term left-sided neck pain, possibly as a result of the arterial dissections or perhaps because of my coexisting neck disc wear and tear. I also have paroxysmal atrial fibrillation, which is irregular, nonrhythmic rapid beating of the heart occurring suddenly without any warning or trigger. Again, this may be related to my FMD or perhaps because of the blood pressure treatment causing my heart to short circuit every once in a while. I still get headaches, mainly affecting the back of my head. This may be related to the vertebral dissection or perhaps is linked with the neck disc problems. There is so much more still to learn about the intricacies of FMD and its long-term effects on patients.

I am thankful that once I presented to the hospital with my bruits, the diagnosis was made within 2 weeks. This is much sooner than the journey of many other FMD patients. I am glad that the condition was detected, and I had not had a major neurological event, such as a stroke to herald in my diagnosis. I am also grateful for doctors who listened to me and who were willing to discuss my management with me, treating me as a person rather than a rare condition. I sometimes wonder if my vascular bruits were present back in 2007 and whether detection via stethoscope at that time would have changed my management. Certainly, my own sensation of vascular noises in my head was not present at that time.

It is important for clinicians to consider FMD in their differential diagnosis, even though it is a rare condition. Listening over the carotid arteries with a stethoscope when a patient presents with headaches, raised blood pressure, and neck pain as a cluster of symptoms together could be key to early diagnosis. In medical school, we were taught that common
things are common in terms of diagnosis and always to think of the most likely cause of a patient’s symptoms. I do think it is important to remember the rare conditions too though, especially in patients who rarely present seeking medical attention. Being open to learning about new conditions is paramount in supporting patients, and there should be no shame felt by doctors in admitting when they do not know much about a condition but are willing to learn.

The emotional struggle of being given a rare condition diagnosis is a tough and lonely one. But it is a journey and one that takes time to walk, with supportive doctors, family, and friends. The FMDSA has been a tremendous source of support in terms of providing me with information and also links with fellow FMD sufferers. Although it can feel like an isolating experience, having a rare disease does not always mean being alone. In my opinion, there is really no substitute for experienced interpatient support and understanding. Having a diagnosis of FMD is a double-edged sword, in that it can be frightening living with the uncertainty of the prognosis but, on the other hand, being diagnosed allows best management of blood pressure and monitoring of the condition by clinicians. This helps to lessen the likelihood of the occurrence of permanent neurological events.

There is life after being diagnosed with a rare long-term condition. I appreciate the simple things of life more now and am thankful for each day in a way that I perhaps took for granted before.

Disclosures

None.
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