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Awakening from a medical mystery: one patient's experience of being undiagnosed

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Abstract

This personal narrative pleads for a supportive and comprehensive system or sub-system similar to that which exists for cancer patients, to deal with undiagnosed illnesses. By describing the torment of living with a debilitating illness that medicine could not easily recognize, then by contrasting this experience with my wife's experience of the cancer care system, and by referring along the way to lessons learned many years ago from reading the works of the inciteful neurologist, Dr. Oliver Sacks, I hope to inspire the medical system to develop a separate, supportive and comprehensive system to deal with the undiagnosed. As it turns out, my mystery illness was a spontaneous cerebrospinal fluid (CSF) leak. This condition is all too often missed in diagnosis or dismissed, and treatment is often significantly delayed, such as it was in my case for nearly two years. At the same time, my wife experienced the cancer system, allowing us to draw contrasts between the two experiences and leading me to implore the system to draw the two approaches together by identifying some of the missing elements for those with undiagnosed illnesses. To conclude, I make reference to the relatively new Undiagnosed Diseases Network as one possible model to accomplish the above goal across the medical system or within local medical systems.

Keywords

Patient experience, patient-centered care, quality of life, continuum of care, undiagnosed illnesses, cerebrospinal fluid leaks, Oliver Sacks, system change, Undiagnosed Diseases Network

Background

In this retelling of my recent experiences with the medical system, I plead for a carved-out system to deal with medical mysteries, with the undiagnosed, by describing the torment of living with a debilitating illness that medicine could not easily recognize. I refer along the way to lessons that I learned many years ago from the inciteful and eloquent neurologist, Dr. Oliver Sacks, whose writings about anomalous neurological disorders informed much of my early years and have prophetically come back to me as I experienced my own neurological anomaly.

On September 11, 2017, I awoke with random pains and symptoms that would evolve over time but continue to disable me for nearly two years. My general practitioner and I embarked immediately on a quest that through luck and researching would lead to a diagnosis of a cerebrospinal fluid (CSF) leak, but only after nearly two years of living in a limbo that CSF leakers, and others with difficult-to-diagnose illnesses, know all too well.

A few months before I fell ill, my wife was diagnosed with an aggressive carcinoma. In this scenario, we observed two separate medical systems, one for those with clear diagnoses and one for those without.

Her system reached out to her, cocooned her, guided her through diagnosis, prognosis, surgery, chemotherapy, radiation, and subsequent treatments. I, on the other hand, struggled with all of these as the system clearly was not designed for "cocooning" those without a diagnosis.

For example, after repeated denials of my disability claim, my employer's insurance company eventually sent me to their "independent" neurologist, paid by them, who – amazingly – said I could work, based exclusively on the symptoms that he deemed neurological, that being a "headache." All my other symptoms he deferred for comment to other, non-neurological, areas of specialty. Unfortunately, this response to a sufferer of a CSF leak is not at all uncommon among neurologists, as attested to in the medical literature itself.¹⁻⁸

A Deficit of CSF

CSF is fluid that supports our brain and is contained by a usually durable sac called the *dura mater*. When this sac has a hole, the volume of CSF drops, pulling the brain down, causing traction on the nerve endings and head pain² (I resist calling it *headache* as it has little in common with this common ailment).

But there can be many more debilitating symptoms to this condition, and the response of the body to a leak can vary

significantly among individuals.³⁻⁸ I experienced nearly all the *classic* symptoms of a CSF leak,^{3,4} especially head pain that worsened when upright. Not a typical headache, it was as if a flat iron were pressing down on my brain. This was accompanied by constant nausea, vomiting, neck pain and stiffness, sensitivity to sound and light, pain in my cheek, constant high-pitched ringing in my ears, and difficulty maintaining my balance, causing frequent falls.

Additional, rarer symptoms added further impediments to my functioning. I had severe difficulty thinking and focusing, fatigue and sleeping long hours, motor tics, vocal tics, gagging, stuttering, slurring, difficulty swallowing, and difficulty finding words. Although rare, all of these additional symptoms have been evidenced in the literature as related to CSF leaks.³⁻⁸

As a result, I was unable to work, to go out regularly with friends, or to engage in all the activities I would have liked to with my family.

The Medical System and Patients with Mystery Illnesses

With the absence of my income, financial issues were just one enormous challenge that we faced. Far more insidious, however, was the potential for the medical system to sap our hope and energy. In the book *Awakenings*, Oliver Sacks quotes an English poet in making a point about what illness can do to a sufferer:

As Sickness is the greatest misery, so the greatest misery of sickness, is solitude... Solitude is a torment which is not threatened in hell itself. - John Donne⁹

In the face of this risk of isolation, the medical system needs all the more to support patients. While trying to obtain a diagnosis for my condition, we encountered supportive doctors but also some who did not play the role of supporter. Rather, these doctors pushed me further toward solitude, away from the consolation of a medical system that could unite me with others.

I say that these doctors “did not *play the role* of supporter” because, despite the anger that I felt toward them at the time, I did not see in them an inherent malevolence or ignorance or even arrogance that suggested they were *always* or even *often* impediments to their patients; rather they seemed human and giving, and, under other circumstances, I could imagine them being compassionate and helpful.

One neurologist I was sent to early in my quest was particularly unhelpful. In a barely 15-minute interview, he immediately dismissed my symptoms as “not neurological.” I cried on the way home and I cried for a day after the appointment. My wife and I then resolved

not to let this setback stop us from pursuing a solution. Our family depended too much on finding a solution to this mystery, to the pain, to the isolation.

In subtle contrast to this unhelpful doctor were the many doctors who tried to help but showed frustration when they could not. I felt their frustration but felt that at least they spent the time and commiserated with us. I have noticed, however, that many of these well-meaning doctors often feel that telling patients about a negative test is a happy thing, and they expect us to be over the moon with joy that we do not have this or that diagnosis. But someone who has an undiagnosed illness, who is in pain and continues to suffer, hears that negative result as another blow to their quest, rather than a relief, even when the diagnosis could have been life-threatening. What doctors need to understand is that knowing *what* we are fighting is often more important than eliminating a potentially serious adversary. Not knowing pushes us back into a dark solitude, stranded in a limbo that we fear will last the rest of our lives.

For me, these experiences all reminded me of something that Oliver Sacks had to teach us in his book *Awakenings*:

One must drop all presuppositions and dogmas and rules - for they only lead to stalemate or disaster; one must cease to regard all patients as replicas, and honor each one with individual attention [...] and, in this way, with the patient as one's equal, one's co-explorer, not one's puppet, one may find therapeutic ways which are better than other ways, tactics which can be modified as occasion requires.⁹

A Doctor Willing to Co-Explore

One day, after nearly a year of being undiagnosed and in misery, and having followed many leads that led nowhere, one of the members of an online connective tissue disorder support group (a diagnosis I had been given along the way by a geneticist) complained that he had “SIH.” I googled this foreign term, as I had done so many times before with other terms and discovered the medical condition Spontaneous Intracranial Hypotension and its inevitable cause, the “rare” (but as it turns out not-all-that-rare-but-often-overlooked) CSF leak. I recognised the symptoms immediately, especially the positional nature of my head pain and other symptoms.

This coincidence led me to more googling, and I discovered a blogger living in my area who had had a leak and who, after we connected by phone, recommended her neurologist to me. This neurologist was very welcoming, spent an hour and a half of his valuable time interviewing and examining me, and proposed a course of action that included further brain scans and an epidural blood patch (an EBP).

An EBP is a strange treatment that involves taking a patient's own blood and injecting it into the area just outside the *dura mater*. The blood pushes on the sac, causing the CSF to flow into the cranial area and coagulate at the point of the leak, helping the body form scar tissue to seal the leak in the long term.¹⁰

Awakened

"It Has been Explained to Me that I have been Away. I am Back." (Leonard Lowe in the 1990 movie, *Awakenings*).¹¹

As I lay on the operating table, not for surgery, but in anticipation of this procedure, I had abandoned hope that this strange intervention could work. It would not work for me, given all my previous disappointments. My numerous imaging tests had all shown nothing and I was convinced this EBP would be the same. Most other neurologists would not have pushed for the procedure to be done in the absence of supportive imaging, but mine did. He knew that many CSF leaks did not show on imaging, as reflected in Schievink's recently suggested diagnostic criteria.¹²

Fortunately, the procedure produced immediate results. My tinnitus, the much-more-than-annoying but rather soul-shattering, high-pitched tone in my ears, disappeared instantly. I cried. I explained to the doctor that I was not reacting to the pain of the procedure, but rather spilling tears of joy because of a long-missed silence in my head.

However, as monumental as any immediate positive reaction to this procedure was – after all, it meant I did have what they said I did; it was finally proof, a diagnosis! – I still did not at that point have absolute confirmation of a positive result. After all, early positive results from an EBP can be short-lived.⁸

It was only after the positive results were confirmed in the ensuing weeks that I realised that I was reawakening into the world – possibly, hopefully, for good, just like one of Dr. Sacks' patients:

All post-encephalitic patients (all patients), in their individual degrees and ways, suffer from defects and distortions of attention: they feel, on the one hand, cut-off [sic] or withdrawn from the world, on the other hand immersed, or engrossed, in their illness. [...]

Awakening, basically, is a reversal of this: the patient ceases to feel the presence of illness and the absence of the world and comes to feel the absence of his illness and the full presence of the world.⁹

A sufferer loses contact with the world — that world we knew and had carved out for ourselves, that world which,

though often recalcitrant to our will, was nonetheless somehow ours in its making. It was *my* world at work, *my* world in my pool league, *my* world among my family and friends, *my* world going to the movies.

Awakening back into this world is an indescribable experience. For me, it was a sudden and complete rebirth. Many who saw me before this rebirth, and then saw me after, wept or beamed with joy. It was *me* again. At my neurologist's office they called me the "miracle man" as I had gone from a slumped-over, slow-thinking, depressed, off-balance, slurring and word-searching slug to a confident, competent, upright and smiling human being again. I beamed happiness, a happiness that welled up from deep in my soul and that was uncontainable, as Sacks describes:

This return-to-oneself, resipiscence, "rebirth," is an infinitely dramatic and moving event, especially in a patient with a rich and full self, who has been dispossessed by disease for years....⁹

Now

My wife and I are daintily proceeding with our lives. My wife has, so far, no recurrence of her cancer, but we know that it can recur very easily with grade 3 tumours of the aggressive type that she had. We continue to see her doctors and other practitioners at the cancer centre, who co-explore with us and continue to monitor and adjust to issues as they arise. I, for my part, fear a failure of the EBP but am slowly trying to resume normal activities, including slowly getting back to work.

Let's Make a Cocoon for Patients with Undiagnosed Illnesses

Sacks stated, "My aim is not to make a system, or to see patients as systems, but to picture a world, a variety of worlds – the landscape of being in which these patients reside."⁹ During our overlapping experiences of the two medical systems, my wife and I experienced helpful and unhelpful approaches. The approach to cancer was to cocoon the patient in an enveloping and well-defined blanket of diagnostic procedures, supportive counseling, and insulated treatment – to create a world in which the patient could subsist and survive, possibly even thrive.

From the point of suspicion of a tumour to diagnosis, my wife only faced a few weeks of doubt; I waited nearly two years for a diagnosis that was accurate, but only after multiple diagnoses had been queried and discarded like trash – or not even discarded, which would have been preferable, but simply left to hang in the air as if neither true nor false, neither real nor ephemeral – fibromyalgia, chronic pain syndrome, amyloidosis, chronic fatigue, psychosomatic syndrome, and a host of others.

From diagnosis to initial treatment, my wife waited only a few weeks, while I waited over six months. But, most importantly, the local cancer care centre reached out with competent hands on behalf of the medical system and provided us with reassurance, information, avenues for support, emergency numbers, specialists in each area to deal with her particular situation, *a plan*, and, beyond all this, with human beings able to co-explore with us, to grapple with us around tough decisions, and to reassure us that they knew what needed to be done or at least knew enough to plot a potentially successful course.

In stark contrast, individuals who have no clear diagnosis, when capable, must do their own research, seek out their own supports, rely on word of mouth, and fend off the medical professionals who pre-emptively discount their experience, who quell their hopes or share with their patients their own frustration of not being able to provide a diagnosis.

A Network for the Undiagnosed

A movement in the hospital sector in the United States is attempting to deal with this discrepancy in approach by establishing a network of centers, dubbed the Undiagnosed Diseases Network (UDN), specializing in the assessment and diagnosis of undiagnosed illnesses.¹³ While I cannot personally attest to the efficacy of this approach, carving out a part of each hospital as has been done in the cancer sector or establishing networks within a group of hospitals dedicated to those patients with serious but undiagnosed illnesses would seem to be a wise and compassionate way to proceed. As one study¹⁴ suggests, identifying chronic conditions at earlier stages could also produce dramatic savings to the health care system. The approach could be modeled on the successful approach of the hospitals' cocooning of cancer patients and other such well-defined and well-supported groups. In Sacks' words, it is a matter of creating a *world* for undiagnosed patients and patients with anomalous diagnoses to live in, rather than to submit them to a system that simply shuts them out.

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