

The quest for diagnosis: a narrative analysis of patient journeys

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ABSTRACT

Illness narratives by patients have been compared to hero quests. In a quest the hero seeks a physical or spiritual goal, facing difficulties along the way. Patients with rare disease often face difficulty and delay in their search for a diagnosis, and so are particularly likely to relate their story as a quest. Using the quest structure to analyse patient narratives forces us to pay attention to the elements of the narrative – what is present and what is missing. This paper presents an analysis of narratives written by rare disease patients. The material is taken from a series published in the British Medical Journal titled ‘A Patient’s Journey’, and so these narratives are told to influence doctors. The narratives mostly conform to the structure of a quest, but some elements are strikingly absent.

KEYWORDS

Illness narratives, rare diseases, diagnosis

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INTRODUCTION – NARRATIVE ANALYSIS

Narrative medicine attends to the stories patients tell [1]. To call it a ‘story’ may seem dismissive of the pain and suffering of patients whose disease is overwhelming them, but a careful analysis of first-hand testimony reminds us to focus closely on the lived experience of illness: not the clinical world of tests, investigations and biochemical results, but the world of pain, suffering and difficulty which the patient inhabits. Testimony provides a proper depth and detail to that glib phrase, so widely used and so narrowly understood, ‘quality of life’.

Many authors have suggested that illness narratives can be read as hero quests. Campbell [2] drew on a huge range of stories from many cultures and time periods to suggest common themes and structures in quest narratives. He proposed that there are three main phases of a quest: departure, initiation and return. ‘Departure is the call to quest, which may at first be refused. ‘Initiation’ includes such elements as ‘the road of trials’ and meetings with figures such as the goddess and the temptress before the ‘ultimate boon’ is gained. The boon may be a physical prize or treasure but, more relevantly to narratives by patients, may be a changed understanding of self. Campbell describes the third phase thus:

‘WHEN the hero-quest has been accomplished, ...the adventurer still must return with his life-transmuting trophy. The full round ... requires that the hero shall now begin the labor of bringing the runes of wisdom, the Golden Fleece, or his sleeping princess, back into the kingdom of humanity, where the boon may redound to the renewing of the community, the nation, the planet, or the ten thousand worlds.’

Booker, a literary critic, developed a fuller analysis of quest structure [3]. In Booker’s schema the hero receives a call; he sets off on a journey; he arrives but is met with frustration; undergoes final ordeals; and finally he attains the ‘life renewing’ goal of his quest. Booker drew attention to two further features of the quest: the hero often has companions who accompany him on his journey; and he encounters people who help him on the journey.

Frank [4] suggested that patients’ stories can be grouped under three headings – narratives of restitution, chaos and quest. Of these three the quest narrative is, in Frank’s analysis, particularly important because, unlike restitution and chaos narratives, ‘the quest narrative affords the ill person a voice as a teller of her own story... The quest narrative affords the ill their most distinctive voice’. [115] Frank follows Campbell in identifying three stages of a quest: departure, initiation, and return.

Hawkins [5] also uses the vocabulary of quest in her study of illness narratives: ‘illness is like the hero’s call to adventure, only this is a call that cannot be refused: whether they want to or not, the sick become denizens of a strange land.’ [78]

One other feature of narrative is worth mentioning at this stage. If narrative is structured as a quest, then we expect to find certain elements in the narrative. Alter, in a study [6] of biblical narratives (p69 – 74), has pointed out that, in structured narratives, what is left unsaid may be as important as what is said. If we expect to find a narrative element but it is not there in the tale as told, that is significant. Patients are not, of course, professional authors and are unlikely to structure their narratives deliberately, as do the biblical writers; nevertheless the point stands that if we expect to find an element and it is not there, we should ask why.

In contrast to patients with a common disease, patients with rare disease are particularly likely to face difficulty and delay in obtaining a diagnosis, and hence an extended quest. Their diagnosis may be delayed for years, and a substantial proportion are given wrong diagnoses before the truth becomes apparent. These problems were highlighted by the Chief Medical Officer for England in his 2009 Annual Report [7], and again in the UK strategy for rare disease [8].

Hence using quest as a framing device or organizing principle may be a particularly fruitful way to analyse the narratives of patients with rare diseases. Paying attention to the structure of a narrative – the way the story is organized and told – forces us to pay close attention to what the patient is saying, and in so doing help us to meet the patient’s needs. As Charon [1] puts it ‘the good reader can both understand the text’s content and identify aspects of its structure that lend to its meaning’ (p109).

The present study uses the quest structure to analyse a set of narratives written by patients with rare disease.

METHOD

The material

The source material for this analysis is a set of papers published by the British Medical Journal in a series called ‘A patient’s journey’. Over 100 papers have been published in this series. From this set of material I selected accounts written about rare disease – that is, diseases meeting the standard European definition [9] of a rare disease, ‘affecting fewer than 5 people in 10 000’. I chose to focus on narratives of rare disease partly because this is my area of practice. Also, there are few if any literary analyses of rare disease pathographies. Within these narratives, I focused on the portion dealing with diagnosis. As suggested above, patients with rare disease are particularly likely to face delay and difficulty in diagnosis and I wanted to explore the extent to which they structured their accounts of this search for diagnosis as a quest.

The narratives are all written in English and mostly by native English speakers; contextual clues suggest that one was narrated by a Dutch patient [10]. The narratives were all first hand accounts by patients, with three exceptions: in one case the patient had died of her illness [11]; in another case the patient was too young [12]. In both of these cases the narrative was written by the patient’s mother. The Dutch patient was too disabled to write and so dictated his narrative before it was translated into English.

Also not stated explicitly, but likely from contextual clues such as the authors addresses and text references, most are set in the context of the English National Health Service; there is one probably from the Netherlands [10] and one from Australia [13], and a part of one narrative is set in North America [14]. The papers were published between 2007 and 2013.

All narrative is structured and constrained; and all narrative is structured to a purpose. These narratives are structured in length (about 1000 words) and format by the publishing conventions of the British Medical Journal. They are structured to their purpose of informing or teaching the readership of the Journal. Regardless of the actual readership of the Journal, which we do not know, it seems reasonable to assume that the authors will take the readership to be mainly doctors. This assumption is reinforced by frequent reference to doctors in the advice given to contributors [15]. They were told that ‘articles should demonstrate one or more of the following lessons

- Some doctors take too long to arrive at a correct diagnosis. By reading about patients’ journeys, other doctors might diagnose the condition sooner, which would benefit their patients
- Some aspects of conditions and diseases are very important to patients but doctors may be unaware of these aspects
- Evidence based interventions may exist that could have profound effects on patients’ lives and of which doctors should be more aware’

The present analysis is also structured and constrained by my own background. I have a medical qualification and practice as a public health physician in the English National Health Service. For the past 10 years I have been engaged in planning and monitoring health services for patients with very rare diseases. I have heard or read many narratives from patients, and certain themes stand out. I do not come to the current analysis with a blank mind.

Analytic method

My analytic method was to seek in the narratives evidence of the classical quest structure: hence I start with the structure of a classic quest and see to what extent this structure is reflected or not in the material. There are several other possibilities for analytic method, but there is a strong a priori likelihood that patient journeys will resemble quests. Hence the choice of method. The method is deliberative; unlike grounded analysis, the themes do not arise from the material but are deliberately sought. In Braun and Clarke’s typology [16], the method is deductive rather than inductive.

Specifically, then, I analysed each narrative to see how the elements set out above were represented in the material, using Booker’s schema: call, journey, arrival and final ordeal, goal, companions, and helpers and opponents.

RESULTS

Main analysis

The call

The first point of interest in the narrative is the point at which the patient became aware that something was wrong – that a quest for diagnosis was needed. Sometimes the disease appeared suddenly and unmistakably, over a few days and weeks. For some, diagnosis was made in the first few days of life. For other patients the disease developed gradually, insidiously, over several years or even decades.

Joyce Hobson [17] described the onset thus: there was no mistaking the fact that an illness had started.

“It was during a 280 mile drive to a remote village in the Yorkshire Dales that I was first aware of an ache in my right thigh. Four days later, it had spread to my ankle and buttocks. Next day I was in agony with horrendous back pain. For the next two weeks I barely slept.”

Michael Hart [18] also had no doubt that an illness had started:

“I was 18 and had just returned from a trip to Australia when lumps started to appear on my legs. The doctors thought it was deep vein thrombosis caused by the long flight and prescribed anti-inflammatory drugs, which seemed to calm things down. However, over the next few months I was very unwell—one week with tonsillitis, the next with an infection in my testicles, then back to tonsillitis, and so on.”

A clear sign at birth marked the start for Simon Laxon [19]:

“...within a few days of my birth my parents knew that something was wrong. There were dark purple stains in my nappies, so my mother took me to a consultant at my local hospital for investigation.”

In other accounts, the onset was more subtle and it was not clear to the patient that an illness had started:

“For quite a while I had been feeling breathless. I could feel my lung capacity falling. Eventually, I went to my GP.” [20]

“My mind didn’t listen to my body, ignoring geriatric jogging times, inexpressible fatigue, and increasing dyspnoea.” [21]

Or the patient relates a series of seemingly unconnected problems, none of which in isolation seems to demand a unifying diagnosis [22]:

“The whole picture became clearer in retrospect. Interviews with the medical team began to link many different, non-specific symptoms: the dizziness, low blood pressure, weight loss of about 4 kg. My periods had stopped. I had developed a bald patch on the back of my head. My underarm hair was practically non-existent. So was my libido (for which I had been referred to a psychotherapist). I had strong salt cravings, pigeon-holed as a bad habit best kept under wraps; and constant tiredness was

easily attributed to the demands of two preschool children. Pigmentation, in my case, looked more like a lingering tan.

Such vague and disparate symptoms make early intervention in the case of Addison's unlikely. My own various accounts during the course of several visits to the doctor, were selective, based on what I thought was relevant and what was not (such as not mentioning the salt cravings)."

Diversity is clear in these accounts. Rare diseases are not a single entity with a common set of characteristics. We must attend to the differences.

The journey

And so the patient starts a journey towards diagnosis. Time is an important dimension of the journey, and patients comment on time passing. Sometimes this is measured in days, and sometimes in years.

"Still we had no idea of what was going on. We waited two years to see the geneticist." [11]

"His condition was diagnosed nine months after his first visit to the dermatologist." [12]

"During the two years when I had repeated infections, I was admitted to hospital nine times. My mouth and genitals were ulcerated and lumps kept appearing." [18]

The last patient also commented on time elapsed during the hospital visit which led to his diagnosis:

"I waited nine days as an inpatient to see a professor who immediately diagnosed Behçet's syndrome." [18]

Lack of wait is also worthy of mention in the narrative, particularly when the GP acts promptly. Words such as 'immediately' and 'right away' are used. The words may not be used literally – perhaps the action was done later on the same day - but to the patient the action was urgent.

"Unusually, my general practitioner recognised it and immediately referred me to the Freeman Hospital in Newcastle..." [23]

"On 13 September 2009 I developed double vision. My doctor took one look and asked that I be admitted to Ealing Hospital right away." [17]

Some patients record their own role in delay to diagnosis, perhaps because of some denial of the illness, or perhaps because of the debilitating effect of the illness:

"Something was obviously wrong but I did not have the drive to do anything about it." [24]

Strikingly absent from these narratives is any account of the literal, physical journeys involved – cars, trains or ambulances. These are journeys which have no sense of travel. In a few accounts, but only a few, places are mentioned, usually as the location of a hospital (Ealing hospital, Freeman hospital in Newcastle upon Tyne, Addenbrookes hospital in Cambridge).

Arrival and final ordeal

We might expect that in the quest for diagnosis, the goal is achieved, or the boon is acquired, at the moment when a doctor gives the patient his or her diagnosis. But in many of these accounts, the moment of diagnosis is rendered not as the boon, but instead as the final ordeal. The boon, if it comes at all, comes later as the patient adapts to his or her illness and its prognosis.

"It was all too much of a shock. We went home in silence." [11]

"The physical shock of his throwaway remark preceded its violent emotional impact..." [21]

"No known cure existed and there was no literature. We were told that unless Alex was kept indoors he was unlikely to reach the age of 30. We mourned the loss of our son's future" [12]

"I could not speak or move. My only thought was: 45 – I will be dead by the time I am 45." [20]

"The consultant stayed for only 10 minutes and spent most of this time telling me off for smoking, leaving me to find out the implications of the diagnosis for myself. I was devastated." [11]

Reaching the goal of diagnosis after a very long time may also bring the pain of regret. Two accounts, both by men with low testosterone syndromes, dwell on the problems caused by a long path to diagnosis. In the first account the words used are very strong – "this was what had ruined my life."

"My abiding memory of the period was one of lacking energy and severe muscle weakness, which led to an avoidance of sport. Unlike my peers, I did not shave and was a loner. A lack of concentration and confidence was constantly noted... I was told that I needed testosterone. I perceived that the lack of this was what had ruined my life." [25]

"On a physical level the late diagnosis has left me with osteopenia, which is still present but at least not deteriorating... The lack of testicular development will always be present. The relative lack of penile growth remains a constant frustration..." [24]

"It is on the psychological level that my delayed diagnosis has had the biggest impact in my view... I have not married, never had any serious girlfriends, and have very limited sexual experience. I think this is a direct result of my lack of emotional and physical development while a teenager and young adult." [24]

GOAL

Although several accounts render the moment of diagnosis as a final ordeal, not all of them do. For some patients discovering the diagnosis is itself the boon:

“Receiving the diagnosis gave me an enormous sense of relief because somebody knew what it was.” [18]

“The most important treatment is possibly the diagnosis itself. Being labelled as a “late starter” or “late bloomer” when in your early 20s can be very humiliating. The ability to put a label to your condition and the knowledge that it is a recognised condition are the first steps in coming to terms with a condition that is difficult to describe to others.” [24]

In other accounts, the diagnosis leads rapidly to the boon of effective therapy:

“In hospital, steroid replacement therapy began immediately, resulting in an unforgettable energy surge: I felt that suddenly I had been plugged in and switched on. The relief was tremendous.” [22]

“I was asked to rub on testosterone gel, and this was followed up with blood tests. Within days I felt better, much better, and facial hair appeared. I did not shave; the facial hair now comforted me.” [25]

Or it may be that learning to live with the diagnosis is the lasting boon:

“And I consider myself very fortunate, despite all the anguish my condition has caused.” [26]

Havi Carel, a professional philosopher, gives a fuller exposition:

“In the months that followed I learnt that illness is multifaceted and complex; that it is a process, not a static entity; and that it is possible to go on living well and experiencing wellbeing even within the context of a terrible and incurable illness. This surprised me, as I had always thought of health as the sine qua non of happiness. And yet, all of a sudden, I found myself changing, responding to constraints, learning to make sense of my life in the light of my illness. The work of realigning my life, its values, and the meaning I gave its different elements surprised me.” [20]

COMPANIONS

These narratives all describe the quest for diagnosis as a solo effort: there are no companions of the quest. The word ‘I’ is used extensively; the word ‘we’ almost never. The only exceptions are accounts by mothers seeking a diagnosis for their children.

Family and friends are included in the account after the diagnosis has been found. Sometimes this is as companions of the journey:

“I am determined to live as full and as active a life as possible and am helped by my general practitioner, physiotherapist, friends, and family.” [11]

But in other accounts the tone when companions are mentioned is one of regret, the narrator expressing concern for the effect of his diagnosis on those close to him:

“When I was originally diagnosed, my family faced the real possibility of losing their husband and father.” [26]

“The next 48 hours are spent talking to our four beautiful kids, aged mid-teens to early 20s, whose joyous careers are currently sprinkled through school, part time jobs, and university. I can’t really convey in words the catastrophic hurt my news has inflicted on them, and it is an insult, which at their age they should never have to endure. I will die of this tumour, I say, and we must address that, neither accepting nor comprehending it. This tumour will kill my body, I say, but I will yield my spirit and personhood reluctantly. We embrace. They weep. I weep for them, for fear for myself, and for the unthinkable horror that they will continue to inhabit the world in which I will play no part. Like my wife, they are brave, selfless, and compassionate.” [21]

HELPERS AND OPPONENTS

The main helper in these narratives is the general practitioner:

“I am grateful to my general practitioner, who set the process in motion; she is a compassionate and caring doctor.” [26]

Sometimes gratitude is more general:

“When my friends commiserate and say what a terrible time I’ve had, I tell them that it was quite an adventure which, in a strange way, I enjoyed, and that I’d met lots of dedicated people who restored my faith, if that were needed, in our NHS. I am a fortunate 80 year old woman.” [17]

Opponents are often the very people to whom patients turn for help: their doctors. Consultants may be unhelpful through ignorance:

“The consultants were mystified” [11]

Or because they dismiss the patient’s account:

“You mothers just don’t know what the sun can do” [12]

Malvyn Benjamin expressed his resentment of this ignorance:

“...I do still harbour a strong resentment that so many health professionals seem completely ignorant of amyloidosis. I accept that it is a relatively rare condition, with only 500-600 new cases diagnosed in the UK each year; however, the disorder is almost certainly underdiagnosed. My concern is that it simply never occurred to the consultants in the nephrology and respiratory departments to consider the possibility of amyloidosis. It just was not on their radar. Indeed the nephrology consultant said he did not understand why my kidney function was declining and said, “I will see you in a year.” My response was that I could be dead in a year! My respiratory consultant suggested I have a lung function test and the result was normal. Once again an alternative cause was never considered.” [26]

Most helpers and opponents in this narrative are medical; strikingly absent is any mention of other clinical staff (nurses, midwives, pharmacists or others) who might contribute to a diagnosis. Nor do husbands and wives, family and friends feature,

except where a parent is telling the story. Nowhere does the story say “My husband suggested that...” or “My wife took me to...” In Sweeney’s account a patient, suggests to him (Sweeney is himself a doctor) that it is time to go to the GP. But mostly the quest for diagnosis, as told in these accounts, is a lonely one.

DISCUSSION

Using the structure of a hero quest to analyse this set of narratives has proved informative. The quest structure draws attention to the ways in which patients experience the call, journey, final ordeal and goal. It also highlights the absence of certain features we might expect to read in a quest narrative.

Before attempting some conclusions, we must return to the particularity of the material analysed. These accounts were published in a medical journal for a particular audience, for a particular purpose. Other narratives by patients with rare disease tell a different story.

As Cornwell puts it [21]:

“Patients’ stories are typically rich in detail, complex, and open to multiple interpretations. They are crafted for listeners: this one for a medical audience. Every story tells a truth, but not the only truth. The doctors in the surgical team, the specialist nurse, and the radiology attendant would all have their own stories about this particular patient. Stories are rhetorical (told to persuade): what is it that this storyteller wants his listeners to understand?”

Carel’s account of the moment of diagnosis is instructive because it illustrates this point: all narratives are told in a particular way for a particular purpose. Writing for the *British Medical Journal*, she renders her story thus:

“I was alarmed and asked my father, a director of a medical screening centre, to arrange a computed tomography (CT) scan for me. I had the scan in the morning, and returned to collect the results in the afternoon. The radiologist clearly did not want to break the bad news to me in person. He said, “Sit down. I’ll let you read about what you’ve got,” and handed me a heavy diagnostic manual. It was open at a page headed “lymphangioleiomyomatosis.” I read the description of this strange disease, my illness, and got to the bottom of the page: “Prognosis: ten years from onset of respiratory symptoms.” I could not speak or move.” [20]

In this account, the radiologist appears abrupt to the point of rudeness, lacking in all empathy and communication skill. But as mentioned above Havi Carel has also written a book [27] about her illness (completed before she constructed the *Journal* article). In the story as written in the book (p4) the incident appears in a different light:

“I walked back into the CT department... The receptionist tried to stop me. I pushed past her into the radiologist’s office... The radiologist turned to me, surprised and displeased to see me in his office, normally off-limits for patients.” [27]

In this context the radiologist’s behavior is perhaps more understandable. He has chosen a specialty with little direct patient contact; he has correctly diagnosed an extremely rare condition, one that he has probably never seen before; the diagnosis carries a very poor prognosis and he is sitting with the patient’s father, a professional colleague. At this point the patient herself enters unexpectedly, leaving him no time to prepare how to communicate the diagnosis sensitively. The book was written for her fellow professionals (Havi Carel is a philosopher); but in writing for doctors in the *British Medical Journal*, the tale is told differently. It seems that she wants to emphasise the need for all doctors to handle difficult news well.

Wilkan, writing in a different context, reflected [28] on a narrative she has just written: [226]

“... I realize I have omitted telling you things that might change your ideas of me and my illness. For example, there are few references to my nearest and dearest: my husband is unmentioned, except for his being away; my mother, likewise, no mention; only my son features, in a couple of brief references – when the truth is that my husband was quite beside himself and eventually broke off his engagement in the States to be with me; my mother was desperate to come and look after me; and my son did all in his power to help me.

But my *experience* was of being utterly alone...”

In the present set of narratives, there is a striking lack of references to ‘nearest and dearest’, or indeed any other companions. This does not mean the narrators had no companions, but it does indicate what they want doctors to know about their experience.

Unusually, and against the conventions of medical journals, we know the names of all the patients in this series, because they are authors. (The sole exception is a patient with Klinefelter’s syndrome who remained anonymous. [25]) But the helpers are rarely, and the opponents never, mentioned by name. A name is a powerful thing: it draws attention to the person and not just the function fulfilled. So the lack of names here is significant. Not naming opponents may be a feature imposed by the narrative structure – lawyers at the *British Medical Journal* may fear libel.

But why are the helpers so rarely named? Perhaps because they are not seen as people, with hopes, fears, weaknesses and failings, but rather as instruments towards a function – diagnosis.

And where helpers are named they are all consultants. The GP is mentioned in several accounts, but never by name. This conveys the impression that the GP is less important, or less relevant, to these patients – the named consultant and the anonymous GP. In the *British National Health Service* it is the GP who is supposed to provide lifetime continuity of care, co-ordinate referrals, and organize support from social and other public services. The lack of naming is significant.

There is also no mention of travel – trains, traffic, car parks and so on. This too is significant. Travel is a substantial problem

for patients with rare disease because expertise in their condition is unlikely to be close to home: in a study of the rare disorder MCADD, half of the families who took part had to travel more than 40 kilometres to the specialist centre for confirmation of their diagnosis [7]. Difficulties of travel are mentioned frequently when rare disease patients are asked about their experience of specialist centres (unpublished observations). We return to the point that narratives are told for a particular audience, and for a particular purpose. Perhaps this lack of reference to travelling in these narratives is because they are told for doctors, not administrators or politicians. The narrator wishes to inform or educate doctors, and problems of car parking may not seem relevant to that audience.

Frank [4] asserts that quest narratives may be told as memoir, manifesto or ‘automythology’ [p 119 ff]. Perhaps because of their frame – published in a medical journal – these narratives are almost entirely memoir. In the manifesto ‘the truth that has been learned is prophetic, often carrying demands for social action’. Frank has shown, using narratives of breast cancer and AIDS, how patient narratives mature over time from simple memoirs to angry manifestos which demand social or political action. There is little evidence of manifesto in these narratives, again perhaps because of their frame in a medical journal, or perhaps because rare disease narratives have not yet progressed to that point. We can perhaps see some hint of manifesto in the passage cited above from Benjamin [26] expressing anger at his doctors’ ignorance.

The dominant metaphor of the ‘automythology’ is the Phoenix, reinventing itself from the ashes of the fire of its own body. This is not a strong theme in the narratives analysed. Certainly patients in these narratives have had to come to terms with their new bodies, the functionality of a body with a rare disease, but the tone is one of acceptance rather than triumphant reinvention.

This study of rare disease patient narratives, written for a medical journal, has shown that the narratives conform to certain aspects of the hero quest structure. There are also some interesting absences or omissions. The next phase of this work is to study narratives written for different audiences, such as fellow patients or the general public. There are many websites for rare disease patient groups, often with first hand testimony. Close reading of these materials may further advance our understanding of the lived experience of patients with rare disease.

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