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Trouble with ataxia: A longitudinal qualitative study of the diagnosis and medical management of a group of rare, progressive neurological conditions

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Abstract

Objectives: An exploratory investigation of diagnosis and management in progressive ataxias: rare neurological conditions usually affecting balance, mobility and speech.

Methods: A longitudinal qualitative study into the experiences of people with ataxia and neurologists. Thematic analysis and follow-up interviews were used to determine diagnosis and management issues over time.

Results: People with ataxia recruited via two hospital departments and Ataxia UK were interviewed at baseline (n = 38) and 12-month follow-up (n = 31). Eight consultant neurologists were interviewed once. Patient accounts were diverse, but many expressed frustration at having an incurable condition and dissatisfaction with service outcomes. At follow-up, there was variation in their contact and satisfaction with helping agencies. Service issues regarding continuity of care and the primary/secondary care interface were evident. Neurologists' accounts also varied. One-half reported that there is nothing that can be done, and one-half favoured specialist referral to increase the likelihood of finding an underlying aetiology within budget constraints.

Conclusions: Diagnostic uncertainties existing at baseline remained for patients at follow-up interviews, although some had learned to deal with the uncertainties brought by the diagnosis of a largely untreatable condition. Care pathways only seemed to operate in the case of defined conditions, such as Friedreich's Ataxia, the most commonly inherited cause. The findings point to a need to develop the evidence base to inform the relative utility of diagnostic procedures in the context of finite resources for patient care and support.

Keywords

Diagnosis, neurology, qualitative study, patient experience, professional views

Introduction

Cerebellar ataxias are rare and progressive neurological conditions that affect coordination, mobility and speech.[1] More than 10,000 people in the United Kingdom are thought to be living with a progressive ataxia,[2] and the prevalence is likely to increase with an ageing population.[3] Due to an absence of epidemiological studies, the incidence of cerebellar ataxia is currently unclear.[4]

A familial predisposition is observed in about 30% of cases[5] with a definitive genetic diagnosis in around 60% of those that are inherited.[3] It is important to stress that progressive ataxias are a heterogeneous group of neurodegenerative conditions. In clinical practice, distinctions are commonly made between hereditary and sporadic ataxias. Age at onset is an important tool in the diagnostic algorithm, although this can vary considerably, for example, even between spinocerebellar ataxia (SCA) patients of the same genotype.[6] In the majority of cases, there is no available medical treatment for the ataxia, and management focuses on monitoring, treatment of associated symptoms (e.g. spasticity, cardiomyopathy and diabetes) and physical therapies.[1] In the United

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Kingdom, there are four national specialist neurological centres for patients with ataxia (London, Newcastle, Oxford and Sheffield). There is no specialist centre in Greater Manchester, where this study was based.

The longitudinal qualitative study described in this article aimed to explore patients' and doctors' experiences and perceptions of the clinical treatment and management of progressive ataxias. The findings form part of a wider programme of study, which began from a concern with investigating care pathways and resource use issues in a rare and complex condition with limited treatment and management options. The fact that some ataxias are inherited also offered opportunities for considering the value of genetic testing in relation to diagnosis and management. Given the low knowledge base in relation to progressive ataxias, as well as a concern with resource use, we also set out to examine the patient and service experience from a 'micro' perspective. Thus, we adopted a mixed-methods research strategy, the overall aim of which was to triangulate data from different sources in order to learn more about the provision and utilisation of health services by people with progressive ataxia. For clarity, in this article, we report solely qualitative findings from semi-structured interviews. An analysis of statistical data related to resource use will be presented elsewhere.

A recent study utilising data posted to Internet discussion fora found that diagnosis in ataxia was widely regarded as an arduous and frustrating process for patients. An idiopathic designation (anecdotally about 30%–40% of all cerebellar ataxias) was regarded by many as a 'non-diagnosis', and there was some evidence that patients in this category had withdrawn from medical contact. However, people with an idiopathic designation were over-represented in the sample.[7] An analysis of patient interviews at baseline found that people generally had more experience of diagnosis than management, although physical therapies and disability aids were the most valued aspects of health service provision.[8] In this article, we focus on the diagnostic process and also incorporate the views of consultant neurologists. More importantly, we include the results of patient follow-up interviews, which allow a more detailed consideration of the subjective effects on patients of diagnostic processes and outcomes over time. Follow-up interviews took place in 2011, 12 months after baseline interviews. The time period for which we gathered data on service use and provision allows for comparison alongside existing benchmarked data on services for people with long-term neurological conditions more generally.[9]

Methods

Interviews were undertaken as part of a wider study concerned with the provision, utilisation, experiences and costs of health services for people with ataxia. An exploratory study design was adopted that incorporated triangulation of data collected from patient interviews, diaries, hospital case notes and interviews with health professionals. Overall, a longitudinal and mixed-methods approach was adopted, although quantitative findings in relation to costs and care pathways are considered elsewhere (manuscript in preparation). This article focuses on experiences and perceptions as gleaned from semi-structured interviews that were analysed thematically. Patients were interviewed at two time points, 12 months apart, whereas neurologists were only interviewed once.

At the outset, it was expected that patient interviewees would articulate a narrative describing symptom history and health service use. An interview guide was initially constructed in relation to this expectation. Given the exploratory nature of the study, a grounded theory approach was used in the collection and analysis of interview data. However, this grounded theory approach was applied mainly as a means of 'coding' and constantly comparing the data collected, rather than as a means of generating substantive social theory.[10] We were more concerned with generating themes for investigation in future studies and pointers towards ways of improving services for people with progressive ataxia.

The issue of diagnosis rapidly emerged as a core category in the patient interviews and became a research focus in the ongoing collection and analysis of data. While we were mainly concerned with health service use, many people with ataxia interviewed had not used medical services beyond diagnostic investigation. Many perceived that there is nothing that can be done for people with ataxia or reported that they had been told this directly by medical staff. Thus, a key concern of the interviews with neurologists was to compare their views on the diagnosis process and the perceived utility of medical management with those of patients. The longitudinal component allowed us to capture patients who were currently involved in ongoing diagnostic investigations and assess their views on how a diagnosis (or the absence of or changes to a diagnostic label) had affected the management of their condition.

The sample

A total of 38 people with ataxia and 8 consultant neurologists were enrolled and completed an interview. The patient sampling strategy was purposive. First, 22 members of Ataxia UK living in North West England were invited to join the study by letter, following identification on a national membership list. Of them, 10 (45%) agreed to take part. Second, the research worker visited meetings of two local Ataxia UK groups where a further 10 people were recruited following a presentation about the study. A further 8 people were recruited by letter via a clinical genetics department (36% of the 22 asked to take part) and a further 10 through a consultant neurologist (50% of 20 invited to participate by letter). A total of 29 neurologists working in North West England were approached by invitation letters, emails and word of mouth about the study and 7 were recruited (28% response rate). One neurologist from outside the region, who was an ataxia specialist, was also interviewed. The 7 neurologists from

within the region were all notionally 'based' at one regional neurological centre, although 4 of them chiefly ran neurological clinics at other hospital sites across Greater Manchester.

Data collection

The study had ethical approval, and written, informed consent was obtained from all respondents prior to interview. People with ataxia were interviewed in their own homes about the effect of ataxic symptoms on their lives and their views and experiences of services. At 12-month follow-up, they were asked about perceptions of symptom changes and interaction with health services (n = 31). Neurologists were interviewed at their workplace about their experiences of diagnosis and management in progressive ataxia and their views of service provision. Separate semi-structured topic guides were used for patient baseline and follow-up interviews and those with neurologists. Interviews were tape-recorded, except in the case of two patients who did not give consent to be recorded. In these cases, notes were taken by hand. Two external transcription agencies were used to transcribe the tape recordings. Follow-up interviews were transcribed by the first author (G.D.-W.).

Data analysis

Transcripts and notes of interviews were analysed with the aid of ATLAS.ti (version 4.2; Scientific Software Development GmbH, Berlin), a computer software package for qualitative data. The semi-structured interview schedule was employed flexibly and was also grounded in that it was adapted as the study progressed. A preliminary analysis on the first 12 patient interviews was undertaken by the first author (G.D.-W.), who also conducted the interviews. Subsequently, the conceptual framework was expanded in discussion with all the authors of this article. Although the original aim was to describe patient experiences relating to investigation and management of their ataxia, it rapidly became evident as the baseline interviews progressed that most people's contact with health services ended when diagnostic investigations were completed, whether or not an underlying aetiology was found. Accordingly, diagnosis became a focus in the interviews and in the analysis. This article focuses especially on the follow-up data as these experiences are likely to better reflect the current situation for users in services, as opposed to their own historical experiences of health services, which in some cases stretched back several decades. In the results to follow, interview extracts are labelled using two or three fields showing the participant ID, quotation number and (in baseline interviews only) line numbers from the interview transcript (e.g. P14: 6: 74–78 or P37: 12).

Results

Patients' perspectives and experiences at baseline interview

A total of 20 men and 18 women with a mean age of 52.5 years (median = 50.5, range = 22–77) were interviewed at time point one. Of them, 14 (36.8%) had received an aetiological diagnosis for a specific type of inherited ataxia. These included Friedreich's ataxia (FA) (n = 7); SCA (SCA1 (n = 1), SCA2 (n = 2), SCA7 (n = 2), SCA8 (n = 1)) and ataxia linked to the fragile-X (*FMR1*) gene (n = 1). A total of 11 patients reported that their ataxia was linked to a family history, but a specific type had not been identified by genetic testing. Two pairs of respondents were blood relatives. A total of 13 patients (34% of 38) reported that no explanation had been found for their symptoms or that their ataxia had been designated as 'idiopathic'. The time since patients had received a diagnosis ranged from 1 to 38 years (median = 5.5 years), although there was also wide variation in age of onset of symptoms. It was apparent that several had lived with symptoms but without a diagnostic label for considerable periods of time.

Most of those interviewed reported that it had not taken long to achieve a diagnosis of ataxia once they had seen a neurologist. However, given the typically slow and insidious onset characteristics of progressive ataxia, most had been on a long journey between initial symptom onset and diagnosis. A small number reported having initial symptoms dismissed by general practitioners (GPs) and/or spending some time being investigated and treated in ENT (ear, nose and throat) departments before being referred to a neurologist. In these cases, it had taken some years to achieve a diagnosis of ataxia. In all, five patients had previous diagnoses (vertigo, chronic headache syndrome, suspected sinus infection, scoliosis and cerebral palsy) changed to cerebellar ataxia. Of them, three of those with an idiopathic designation, or who had a family history of ataxia unconfirmed by the available genetic tests, had pursued further diagnostic testing, for example, by attending one of the four national specialist ataxia centres in an attempt to find the definitive cause for their symptoms. One would have liked to attend a specialist centre but was unable to secure the necessary funding arrangements in order to do so. In general terms, it was possible to discern two different approaches to dealing with a diagnosis of progressive ataxia: engaging with biomedicine or withdrawing from the health system and coping alone (results shown elsewhere).[8]

The interviews undertaken indicated that the study group was composed of individuals with extremely diverse illness histories in terms of symptoms, onset, disabilities and speed of progression. However, most shared a dissatisfaction with health services and a frustration that their condition was incurable and sometimes of unknown cause. It was also widely reported by patients that the majority of health

workers do not know what 'ataxia' is, which was confirmed by one person with ataxia who was a nurse:

> She was the first doctor after six years of investigation who said, '[Mary] you've got ataxia'. And I said, 'I've got what?' And she said 'You've got ataxia it's biological, genetic'. (Female, SCA2 diagnosis, P14: 6: 74–78)

For some patients, the lack of clarity surrounding their diagnosis led them to characterise 'ataxia' as 'something that [doctors] can't explain' (as in P32: 18: 225–230). Only three patients reported the diagnosis process as being in anyway positive. Two of these had an underlying aetiology identified, and one remained idiopathic.

Patients' accounts of the diagnostic process focused on interactions with medical professionals who were characterised as being either expert or inexperienced with regard to ataxia. Most people who had been for neurological work-up reported having a series of tests, sometimes over a long period of time and on other occasions in more intensive sessions focused on one, two or three hospital visits. One young man with an FA diagnosis, who had undertaken research into his condition on the Internet, wondered why he had had so many 'unnecessary' tests when the diagnosis was proven by a blood test for FA gene analysis. However, more commonly, patients' accounts of these issues echoed those of doctors (see below), concerning structural limitations in the National Health Service (NHS) around appointment slots for follow-ups and the time taken to receive test results.

Although most patients' accounts of the diagnostic process focused on biotechnological tests (e.g. scans, blood tests, electro-conductive studies or genetic tests), they sometimes defined neurologists as being specialists in ataxia by their ability to diagnose ataxia by use of history taking and physical examination alone:

> She gave me a diagnosis; even though she didn't test my blood, she just knew what it was, by examining me and talking to me and that. So that made me feel a lot better. But because I went to see her, I mean, it did take hold. (Female, idiopathic ataxia, P22: 29: 169–174)

In the case described above, the patient reported improved well-being, brought about by having confidence in the ataxia specialist. However, she says that her ataxia 'took hold' after seeing a clinical expert. This would suggest a form of 'symptom amplification',[11] whereby people become more aware of their symptoms as a result of the diagnostic process. Symptom amplification was most evident in the account of an older woman with mild episodic ataxia, who received her diagnosis following investigation in her son:

> Partner of patient: Since [my son] has been diagnosed, we've all been more focussed on the symptoms you've been having, before we just ignored them.
>
> Patient: Before I just thought it was me, you know, I get tired, I get dizzy.
>
> Partner of patient: So in effect we feel they have been worse since [my son was diagnosed], because things that would have been shrugged off before, we now attribute to ataxia. (P17: 19: 346–357).

The preceding two interviewees cited that both had forms of ataxia that were presumed to be inherited, but this could not be confirmed by the genetic tests currently available. Those with a family history seemed to have most to gain from recent technological advances in genetic testing, but perhaps also the most to 'lose' when the promise of biomedicine failed to yield the kinds of certainty expected via the diagnosis process. Many patients were aware that a certain number of types of cerebellar ataxia are currently identifiable,[3] although this knowledge did not seem to provide any solace when they were found to have a type of ataxia not currently identifiable:

> No, the professor thought that it was something I was born with, they couldn't pinpoint what was actually causing it at that specific time … I think with Ataxia you go up to a certain number and they hadn't gone possibly beyond that range where they thought I could have had, I don't know, whatever number it was up to at the time or beyond. (Female, idiopathic ataxia, P30: 7: 159–170)

Patient responses to this clinical uncertainty varied. In some, it provoked fear and criticisms of medical competence, as in the case of a young woman with a presumed genetic diagnosis:

> Patient: I was so frightened of what was happening and nobody could tell me what was causing it.
>
> Interviewer: … [Later, during the same interview] … What's been the worst thing in health services?
>
> Patient: Not getting a proper diagnosis. All those years and years knowing something was wrong, and almost having to tell them myself. Come on I've got these symptoms and they match my father's and brother's, so just the length of time it took. Is that normal? (P12: 19: 154–156 and 64: 1031–1040)

Other patients appeared to be less concerned by the limits of medicine in the context of ataxia diagnosis, as demonstrated by a patient who had had extensive investigations as a child:

Interviewer: So has anybody ever told you exactly what you've got or what the cause of it is?
Patient: No, no. As I say, I'm not that bothered.
Interviewer: Okay.
Patient: I'm genuinely not that bothered (Male, assumed genetic ataxia, P23: 17: 99–106)

Neurologists' perspectives

Eight neurologists were interviewed. Two were specialists in ataxia diagnosis (i.e. they had a larger ataxia patient case load than the 'one or two' patients encountered by most), while the remainder specialised in other fields of neurology (e.g. multiple sclerosis (MS) and movement disorders). As with the patients interviewed, the consultant neurologists interviewed described the diagnosis process in ataxia as consisting of a possible plethora of tests to exclude treatable conditions or identify the cause of the illness. Neurologists regularly complained that most test results come back as 'normal' or 'negative' when investigating ataxia. In relation to this 'negative yield' of diagnostic tests, some consultants also discussed the financial resource implications of ordering a large number of tests that would be unlikely to provide a definitive diagnosis or affect the medical management of the condition. The typical diagnostic pathway is characterised in the following extract:

> It is a presentation which often leads to a circuitous diagnostic process with increasing desperation regarding tests and you're often left with an uncertain scenario … where diagnosis will come to light later on down the line as new symptoms perhaps develop. Sometimes, you know, in a significant proportion of patients, you do all the tests you can do for treatable and [other] conditions … and you're left with a blank concluding some sort of idiopathic or presumed genetic condition for which you haven't been able to identify at the moment. So that often puts them in a sort of situation where they've got this ongoing diagnostic doubt which, you know, some patients and clinicians find difficult to cope with. (Consultant P6: 1: 136–146)

Several consultants pointed to the fact that 'ataxia' is an umbrella term describing a medical syndrome or condition, for example,

> Because we as neurologists feel slightly uneasy of the fact because we know that 'ataxia' is just a word that describes a syndrome and it's not really, it's a level of a diagnosis but it's not the final level which might be a gene test or antibody test or whatever. But patients, 'ataxia' is not a word that they use, so 'ataxia' for many patients is as good a diagnosis as MS or epilepsy or, you know that's the name of the disease 'ataxia'. But others want a more specific diagnosis than that. (Consultant P4: 10: 376–382)

Several consultants alluded to the notion of the 'level of diagnosis' in ataxia, with one ataxia specialist describing a generic 'ataxia' diagnosis as a 'black box diagnosis of ataxia' (Consultant P1: 39: 947–949), which results in inevitable uncertainty about management. However, most of the neurologists interviewed suggested that there is little that neurologists can do in management terms in any case:

> The primary role as a neurologist is actually to diagnose the existence of ataxia and then work out why that might have occurred. So look for an underlying cause such as MS or whatever. Once we have got a diagnosis or excluded as many treatable things as we can and we are left with an idiopathic ataxia, then I am a bit limited really in what I can do. Sometimes in the case of idiopathic ataxias I may continue to follow patients up in case something emerges that helps with the diagnosis at a later date, and as part of that follow-up time I am interested in disability and how that's affecting the patient. Having said that often that is looked after more by Professions Allied to Medicine (PAMs) and even neuro-rehabilitation consultants. So when I see someone with primary ataxia … I have a tendency to refer on to a neuro-rehabilitation consultant so that they can provide more of a holistic approach to sort of, managing the disorder than I can. (Specialist Consultant P2: 4: 27–51)

It should be noted that all of those interviewed, with the exception of one specialist, believed that the primary role of the neurologist in ataxia ends with the diagnostic process. One-half of the non-specialist neurologists interviewed held the view popularly reported by patients that 'there is nothing that can be done' for people with ataxia. One questioned the value of follow-up appointments in this context (Consultant P4: 17: 512–515). Of the six non-specialist neurologists, one-half were explicit that they would refer the patients to an ataxia specialist, in order to increase the likelihood of a 'definitive' diagnosis and to enable more effective targeting of finite NHS resources. One of the ataxia specialists reported that on occasion diagnosis was achieved as if by accident, when a result surprisingly came back positive. Two of the non-specialists suggested that extensive testing in ataxia was wasteful of resources and questionable on clinical grounds, especially when the outcome was seen as having little impact on illness management. One of the specialist consultants also held these views.

For both specialist and non-specialist neurologists, the 'successful' diagnosis of a cause of progressive ataxia was described as a rare event. As in the patients' accounts, there we also regularly reported structural problems associated with working within the constraints of NHS bureaucracy. These issues were seen to exacerbate the diagnostic problems faced by ataxic patients. When one doctor was asked about a specific case of SCA6, her response showed how the number of tests ordered by a neurologist, as well as the proportion of positive results, might be seen to reflect the skills of an individual clinician (Consultant P5: 16: 844–874), thus supporting the notion of referral to specialist centres.

Patient follow-up interviews

Of the 38 patients interviewed at baseline, 7 were lost to follow-up at 1 year for the following reasons: (1) non-contactable (n = 2); (2) withdrew, no reason given (n = 1); (3) withdrew:

'She's never had any treatment, she has it very mild, there's nothing else she can tell you' (n = 1); (4) missed interview appointments (n = 1); (5) moved away (n = 1); and (6) declined, stress over problems at work due to ataxia (n = 1). The follow-up interviews were short in comparison with the baseline interviews, with an average length of only 21 min. Many people had not experienced symptom changes, and/or they had no or minimal contact with health services, which were the primary topics of interest. One patient provided an email follow-up response that was analysed alongside the interview transcripts.

Patient's accounts at follow-up highlighted themes found in other qualitative studies of chronic conditions such as in a review of studies of rheumatoid arthritis[12] and related to acceptance, adaptation and coping strategies. Many patients continued to discuss the stigmatising effects of ataxia, as found at baseline[8] and as highlighted by Boutté in her seminal study of Machado–Joseph disease (now known as SCA3).[13] Accounts of health-care services were focused on the themes illustrated in Table 1. As at baseline, a disproportionate amount of discussion focused on problems negotiating or getting service inputs, with a specific issue around neurological follow-up appointments for people with idiopathic ataxia. Table 1 also reveals a consumerist approach to service provision, with people 'pushing' for provision. One interviewee's carer specifically recommended that people with a diagnosis of a progressive disabling condition like ataxia should be given an 'A4 signpost of helping agencies' (P19: 20). When considering specific positive and negative comments about services (Table 2), it appeared that physiotherapy input was generally viewed favourably and most negative comments were directed towards interaction with mainstream health-care workers or doctors seen for other health problems, including GPs. It should be stressed that in some cases, involvement in the study seemed to have spurred patients into seeking out services and care more proactively. It was noteworthy that most of those who gave more favourable accounts of neurological services had an FA diagnosis, perhaps because there is screening for, and management of, the associated non-neurological complications of the condition.

Scrutiny of Tables 1 and 2 suggests that dealing with ataxia is as much about dealing with a diagnostic process as it is about dealing with symptoms. Rather than increasing certainty in relation to disease label and prognosis, the diagnostic process had in some cases magnified these uncertainties. However, in one case, an interviewee suggested that patients accepted their condition more following a period of extensive diagnostic testing (P30: 12 and Table 1). By extension, it can be argued that recognising that answers to questions may never be found is one of the things that patients have to adapt to in response to a diagnosis of progressive ataxia. Comments from another interviewee at follow-up in this regard (P31: 9 and Table 2) would suggest that patients might benefit from being informed at the outset that diagnosis may take a long time and that answers might never be found. This may be equally applicable to other neurodegenerative conditions of course. However, in the case of ataxia, the health service and illness experience seem to be fundamentally distinguishable according to whether a definitive – as opposed to a 'black box' – diagnosis is achieved, with negative accounts associated with people with idiopathic diagnoses. Fundamentally, many in this group describe being, or feeling, 'lost' to services, and the potential importance of diagnosis as the key to access therapies and clinical follow-up is underlined.

Commonly, people with ataxia – whenever they were interviewed – expressed frustration at having a condition for which either there is no cure or 'nothing can be done'. Against this background and expectations, when people had found therapies or other interventions (e.g. physiotherapy or a prescription for coenzyme Q10), they were highly valued. However, in management terms, there appeared problems in this regard as their care was often undertaken by GPs who are generally seen as lacking knowledge in relation to progressive ataxias. Thus, in broad terms, a key issue for people with ataxia in their encounters with health professionals related to the individual expertise of each health worker encountered. Perceived lack of knowledge about ataxia caused trouble for people with the condition in their encounters with health professionals about other matters and in settings other than clinical neurology or primary care. From the viewpoint of the people with ataxia interviewed for this study, the need for more health workers to be made aware about ataxia appears unequivocal. In the perceived absence of medical interventions, an expert sympathetic ear was highly valued.

Conclusion

Medical sociologists have considered the diagnosis process as a means by which patients' symptoms are 'made intelligible'[14] or rendered 'coherent'.[15] The accounts of both neurologists and patients suggest that ataxia remains a problematic diagnosis of uncertainty. For those who did not manage to secure a 'definitive' explanation for their progressively disabling conditions, the diagnostic process seemed somehow incomplete, engendering what one clinician termed 'ongoing diagnostic doubt': a challenge for patient and doctor alike. Of the 38 patients interviewed for this study, only 3 (8%) perceived anything positive in achieving a medical diagnosis of 'ataxia' for their symptoms, although some patients were coping better with their diagnostic status when seen at follow-up. The findings suggest that the diagnosis of 'ataxia' represents the first step in identifying an underlying cause, which can give more certainty. This was achieved for some patients by the time of the follow-up interview, where, for example, the disease category was defined as being multisystem atrophy in two cases. One interviewee had undergone genetic testing, which identified a specific type of SCA, but

Table 1. Themes in accounts of patients' experiences of health services between baseline and follow-up (n = 31).

Theme	Diagnostics	Management
Adapting with experience	'The future is not as bad as I first thought … When you are first diagnosed nobody can tell you how fast it's going to progress' (P2: 35); 'I have fully embraced now, my condition' (P14: 5); now accepts it more following long period of diagnostic tests (P30: 12)	Fatigue has got stronger over the past year 'to the point that I can't fight it at all' (P22: 16); 'I just can't walk anywhere … To be honest its getting me down … I've tried to carry on' (P38: 5–7); 'My family must come first' (P2: 32); 'I know how to go about things better than I did … Now I know you can go and get things' (P24: 11)
I do not see anyone about my ataxia	Complains the neurologists he knew have all retired. They used to say, 'I'm sorry we can't do anything for you, but we'd like to see you anyway'. Has not seen a neurologist for 6–8 years (P1: 21) 'Just keep an eye on me really' (P33: 9)	'See if they send another appointment' (P4: 18); 'Hospital keep missing my appointments out' (P9: 8 and P16: 6); 'I think I got lost in the system' (P29: 8); 'I thought they would send for me and tell me how it was getting on' (P8: 10 and P13: 5); 'I very rarely visit the hospital nowadays' (P11: 32); does not want regular follow-up with neuro/physio (P14: 9); does not see health-care workers about ataxia (P23: 4); 'Nobody's been to see me' (P10: 6)
Negotiating access to services	'Because I'm helping them' by attending for medical education sessions; hoping to be pushed 'to the top of the list' (P3: 4, 24 and 29); went for a test at an ataxia clinic but has received letters saying the NHS are reviewing whether they will pay for the test or not (P37: 12)	Passes on ataxia info to GP: 'I think I'm educating them'; 'My GP would listen to me because I insist' (P2: 31 and 41); reports funding for physiotherapy being withdrawn. GP exploring other funding options (P12: 8); wife assertive in arranging therapists (P19: 14); 'Everything I get done, I instigate it' (P24: 7); 'You're sort of left abandoned' (P16: 10)
They could not do anything (was *in vivo* code)	Diagnosis changed from FA to Charcot Marie Tooth disease. 'My first reaction was, "Is there a cure for it?" Unfortunately not.' (P3: 10); 'Nothing they can do' (P2: 6); 'He's done all the tests, he can't do anymore' (P24: 6); discharged by neurologist: 'There's nothing I can do for you' (P27: 10) 'The doctor said, "We can't do anything for you, see you in 9 months." It used to be every 6 months, so he must think I'm better'. (P30: 8)	'The doctor says there's nothing wrong with it'; 'Definitely there is nothing he can find'; 'They couldn't do anything' re: cough, GP and speech therapist (P1: 9, 10 and 15); GPs – 'No point in getting involved with them' (P1: 26); 'Doctor prescribed voltarol but they don't seem to do anything' (P3: 37); 'Can't get pain relief from anything [uses cannabis instead]' (P20: 10); when asked what happened at annual neurological appointment: 'Very little' (P7: 13); 'I don't think there is anything much anyone can do' re: hearing (P11: 17)
Value of specialist services (±)	Had genetic tests but declined to know the detail of the diagnosis (P9: 10)	Visit with neurologist perceived as useful now attends larger hospital (P5: 8); 'I don't think the GP is as aware of ataxia as he could be' (P7: 10); 'He doesn't know anything about [ataxia]' (P8: 13); stayed with old GP after moving as he knew about ataxia (P9: 17)

GP: general practitioner.

declined to know details and implications of the final result. On the whole, defining the underlying cause of their ataxia made little difference to patients when there was still no cure for whatever aetiological label they had been given.

As an umbrella term for a group of conditions, progressive ataxias present patients and their doctors with a range of troubles and challenges beyond the debilitating nature of the symptoms. The length of time it took some patients in this study for their condition to be labelled as 'ataxia' points to a need for greater awareness of ataxic symptoms in primary care in order to reduce the length of the diagnostic journey for patients. Despite differences of opinion, one issue that seemed to unite neurologists was the importance of excluding treatable forms of ataxia. However, given that treatable forms are rare (within a group of conditions that are rare as a whole), it is perhaps understandable that neurologists would not explicitly impart the details of these investigations in order to avoid a possibly false hope that a treatment might be found. While the approach taken by neurologists reflects their wide experience that extensive tests will be returned with 'normal' or 'negative' results, the confusion and uncertainty demonstrated in some patient accounts suggest a need for a more structured approach to diagnosis and subsequent referral, such that patients – perhaps especially those with an idiopathic designation – do not get 'lost in the system' following diagnostic investigation. Our results also point to a need for an evidence base to support the clinical utility of current diagnostic processes, including the role of genetic tests.

Table 2. Patient's views of selected health services used between baseline and follow-up (n = 24).

Service	Positive views	Negative views
Neurology	Neurologist seen as useful (P5: FA diagnosis)	Complaints about neurology department missing or not offering appointments (P4: idiopathic diagnosis, P8: idiopathic diagnosis, P9: presumed genetic, P13: Fragile-X linked, P16: idiopathic diagnosis, P29 idiopathic diagnosis)
	Sees a neurologist at a specialist ataxia centre: 'He tries to get [a test] done on my heart every year' (P22: FA diagnosis)	Attended specialist ataxia clinic but felt it was 'a bit of a waste of time' (P37: idiopathic diagnosis)
	'I understand better that it takes time to diagnose this thing and then once they've put the label on it … just try and take the edge off some of the symptoms. And that seems to be working for me'. (P31: 9, presumed genetic ataxia)	
Physiotherapy	Physio taught her Pilates and strategies. 'It's up to me to do [the exercises]'. Feels improvement in confidence; given practical tips (P2: 21 and 39); 'They were very good, they gave me strategies … But it made me feel better. It made me feel as though someone was temporarily interested in me' (P2: 8–11)	'Quite disgusted' that funding for physiotherapy was withdrawn: 'It's the only treatment, isn't it?' (P12: 9)
	Wife links lack of progression with extensive physiotherapy involvement (P13: 9); 'What I find helpful is I see my physiotherapist regularly' (P18: 4)	
General Practice	GP putting her on a 'trial' of coenzyme Q10 was reported as the best thing that had happened in last 12 months (P2: 43)	'I don't think the GP is as aware of ataxia as he could be' (P7: 10 and P8: 13)
	GP arranging another neurological referral after the neurologist discharged her (P27: 11)	Disagreed with GP that back pain was anything to do with ataxia (P14: 7)
		'He just randomly googled it' (P22: 15)
Services used for other medical conditions		'In hospital, they didn't know what cerebellar ataxia was'; 'Oh let's look that up' (P4: 15 and 21)
		Critical of incident involving reinsertion of catheter. Received formal apology from hospital (P5: 11)
		'They hadn't even read my notes … They didn't even know I was in a wheelchair' (P22: 8 and 9)

Complex conditions such as ataxia can affect a range of physical functions, which often means that care needs to be coordinated between different health professionals. From the patient's viewpoint, it is helpful if someone is overseeing their care and that provision of different service is 'joined up'. In ataxia centres, specialist ataxia nurses are able to provide the kinds of general information and support (e.g. how to access mobility aids or social care benefits) needed by patients in addition to medical care and expertise. However, given that such a service would be valued by those with other neurological conditions, a challenge for services is how to provide such generic advice within a specialist model of care.

While analysis of patient baseline[8] material noted that a definitive diagnostic label may not be important in terms of access to treatment and services, the results at follow-up suggested that a genetic diagnosis of FA is useful in terms of accessing a care pathway and management plan. This was reflected in study participants with a diagnosis of FA seemingly reporting greater satisfaction with neurology services than other groups of ataxia patients. FA is the most commonly inherited form of ataxia found in the United Kingdom and has specific clinical features including an increased risk of diabetes and cardiomyopathy, which can be monitored and managed by other specialist medical services.[1] In this respect, there is a better defined clinical pathway for FA than for SCAs and idiopathic ataxia. This could explain why those with an idiopathic ataxia seemed more negative about neurology services (Table 2) and less satisfied with issues relating to clinical management and follow-up. The fact that people with a specific diagnosis report a better experience of neurological services highlights the importance of a diagnosis. However, given an ageing population, the number of people with idiopathic late onset ataxias is likely to increase. In the context of budget constraints and finite resources, the clinical need for an aetiological diagnosis may need to be weighed against the costs of meeting the needs of

patients with a progressively disabling and incurable condition.

Through the unusual case of a woman in her 70s who had lifelong symptoms of 'vertigo' changed to ataxia as a result of investigations for ataxic symptoms in her son, the circumstances under which a diagnosis of ataxia can lead to a form of 'symptom amplification'[11] were revealed. However, the mechanisms of symptom amplification are by no means clear from this isolated case but point to a fruitful area for further research. In the case of ataxia, however, the fact that there are putative treatments for vertigo, whereas there are none for ataxia may form the kernel of the matter. What remains unclear on the basis of our findings is whether people are better or worse off following a diagnostic process in the context of ataxic symptoms. Historically, attention has not been paid to the potential utility of diagnostic processes in the context of conditions for which there are no clear management outcomes. However, in the context of the financial limits of health-care budgets, these issues are receiving increasing attention. As well as highlighting factors that present potentially negative consequences for patients as a result of diagnostic processes, our findings underline the need for better integration of diagnostic algorithms and procedures with subsequent care and support for people with ataxia. Finally, the setting for this study was an English health region, and it would be fruitful for others to compare our findings with those in other countries, settings and health systems in order to ascertain the best way of providing services to people living with the challenges of progressive ataxia.

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