'You don't get told anything, they don't do anything and nothing changes'. Medicine as a resource and constraint in progressive ataxia

Gavin Daker-White PhD,* Helen Kingston MD FRCP DCH,† Katherine Payne PhD,‡
Julie Greenfield PhD,§ John Ealing MBBS DPhil FRCP¶ and Caroline Sanders PhD**

*Research Fellow, Health Sciences Research Group – Methodology, The University of Manchester, †Consultant Clinical Geneticist, Department of Clinical Genetics, St. Mary's Hospital, Central Manchester University Hospitals, NHS Foundation Trust, ‡Professor of Health Economics, Health Sciences Research Group – Health Economics, The University of Manchester, §Research Projects Manager, Ataxia UK, London, ¶Consultant Neurologist, Department of Neurology, Salford Royal NHS Foundation Trust, Salford, and **Lecturer in Medical Sociology, Health Sciences Research Group – Primary Care, The University of Manchester, Manchester, UK

Correspondence

Gavin Daker-White PhD
Health Sciences Research Group –
Methodology
The University of Manchester
Manchester, UK
E-mail: gavin.daker-white@
manchester.ac.uk

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Abstract

Background Progressive ataxias are neurological disorders affecting balance, co-ordination of movement and speech.

Objective A qualitative study was undertaken to discover patients' experiences of ataxia and its symptoms.

Participants Thirty-eight people with ataxia recruited from patient support groups and two hospital outpatients departments.

Design Cross-sectional qualitative study with thematic analysis.

Results These accounts highlight the limits of medicine in the context of a rare, incurable and disabling disorder, and the embodied uncertainties brought by slowly progressive diseases that lie at the boundaries of mainstream medical knowledge. The existential crises faced by people with ataxia are seemingly magnified by sometimes idiopathic aetiologies and the limited number of inherited conditions identifiable by the available genetic tests. Interviewees were drawn into a medical system that was focused mainly on the diagnosis process, with widely varying results. However, when asked, most had rather valued the provision of disability aids and physical therapies. Only one informant reported overcoming the myriad uncertainties of progressive ataxia, and their account supported the notion of 'biographical repair' in chronic illness.

Conclusions Clinical uncertainties in ataxia constrained people's attempts to deal with their condition. The construction of the proactive, informed, medical consumer who is assumed to be a partner in care is problematic in the context of a rare and difficult-to-diagnose disease for which there is usually no cure. Service providers should be mindful of the need to manage patient expectations in relation to diagnosis and cure. More focus might usefully be placed on the provision of physical therapies and disability aids.

Introduction

People with ataxia have disturbances in balance, coordination of movement and speech. Ataxia may be a symptom of another medical condition (e.g. Multiple Sclerosis) or a disorder in its own right.¹ The focus of this article is on progressive ataxias: rare neurological conditions, inherited in approximately 30% of cases.² However, a similar proportion of people with ataxia currently never find a cause for their condition, which is commonly labelled 'idiopathic'. Other definable causes of ataxia are numerous and include metabolic disorders, vascular disease and chronic alcohol consumption.¹ The potential utility of genetic testing contrasts with other neurological conditions, such as Huntington's disease, where inheritance is the sole cause.³ It is estimated that there are c. 10 000 cases of inherited or idiopathic ataxia in the UK.⁴ Inherited forms of ataxia include Friedreich's Ataxia (FA) and Spinocerebellar ataxias (SCAs), of which there are numerous types. People with symptoms of ataxia in the UK would typically be referred to neurologists from primary care. There are just three specialist ataxia centres in the UK.¹ These centres can offer specialist investigations above and beyond those available in general neurology departments, such as access to specific genetic tests that are currently only available on a 'research' basis.

The findings reported in this article were gathered in patient interviews collected as part of the Care of Ataxia Patients (COAP) study, an exploratory programme of research aimed at discerning the impact of ataxia on well-being and highlighting areas for service improvement. The results are compared with previous medical sociological work concerned with the patient experience in chronic illness.^{5,6}

Ataxia is rare,⁴ difficult to diagnose⁷ and usually untreatable. Thus, the limits of medicine are likely to be brought into sharp focus. The primary features of progressive ataxia from the viewpoint of those with the condition are that it is disabling and stigmatising.⁸ Social and technological advances have led to the rise

of the notion of the proactive and informed medical consumer, who is understood as a partner in care alongside medical professionals.⁹ However, it is not clear how this role would work for people with an untreatable illness of uncertain aetiology.

We relate the experiences of people with ataxia to a framework that draws on Mike Bury's concept 'biographical disruption'.¹⁰ Bury uses this term to describe illness as a radical departure from the 'taken for granted' aspects of everyday life associated with the recognition of pain, disability and an uncertain future. The concept has been widely used in studies of chronic illness.^{11–13} However, we draw mostly on Bury's idea that, in the context of uncertain symptoms, a possibly elusive aetiology and a lack of available treatments, medicine may be seen as both a resource and constraint.¹⁰ The analysis was also informed by the literature on the sociology of diagnosis¹⁴ as this emerged as a central issue in the interviews.

Methods

Following university and NHS ethics committee approvals, 38 people with ataxia living in northwest England were recruited via Ataxia UK*; a hospital neurology department; and a department of clinical genetics. According to the protocol, the original aim was to recruit 30–40 adults (aged over 18) with a primary diagnosis of idiopathic or inherited ataxia living in Greater Manchester. Recruitment was initially by invitation letter. Further people were recruited in person by the first author at meetings of two local Ataxia UK branches. Written informed consent was obtained from all participants prior to interview. The first author conducted the interviews. A semi-structured schedule was used as a topic guide (see Box 1)

*Ataxia UK is a registered charity and national support group for people with cerebellar ataxias. Ataxia UK funds research and offers information and advice to clinicians and people with ataxia. There is also a network of local Ataxia UK support groups. For this study, patients were recruited both from the national membership lists and from two local support groups.

although this was flexibly employed according to the circumstances of each interview. Several respondents had speech disturbances. Whilst it was sometimes necessary for the interviewer to ask respondents to repeat words or phrases, over time he found that he was able to 'tune in' to the speech pattern typically characteristic of ataxia. External transcription agencies employing specialist medical transcribers familiar with such work were employed.

Anonymized interview transcripts were analysed thematically. The interviews were coded thematically in batches of five-to-ten as the study proceeded.¹⁵ The first author undertook the coding, although the coding scheme was revised in team discussion following a pilot analysis of the first five interviews. A computer software package (Atlas.ti) was used to facilitate coding of the data. Given a widespread dissatisfaction with medical intervention and the myriad of uncertainties related to the condition, a framework centred on the constraining and resourceful aspects of medical processes and information seemed to offer a useful way of describing the experiences of interviewees.

Box 1: Topic Guide for the Interviews

When were you diagnosed with ataxia?

- When did you first realise there was something wrong?
- Who did you talk to?
- At what point did you go to see your GP?
- When did you see a hospital doctor?
- What sort of information were you given about the condition?

How does having ataxia affect you?

- Ask about symptoms and what is most disruptive
- What do you need to do to look after yourself?
- Are you able to work?
- Did you need to give up work?
- Do you look after anyone else? E.g. children – Has this changed since being diagnosed with ataxia?
- Social activities and relationships – how far have these changed since having ataxia?

Do you need help from anyone else?

- Who helps you?

Box 1: Continued

- Do you have support from family and friends?
- Are you a member of any support group?
- What sources of information have you found helpful regarding your condition?
- Have you received written information from health professionals? – what was it like?
- Have you found information on the internet? – what was it like?

Do you currently use any types of healthcare services to help with your ataxia?

- Who do you consult? – GP, nurse, hospital specialist?
- Do you have a regular review appointment – with any of above?
- Are you currently on any medicines prescribed by your doctor?
- What other types of services have you used? Do you see any other professional? – e.g. occupational/physiotherapist?
- Do you have examples of services that have been especially helpful to you? What was good about these services?
- Have you faced any problems with health services – can you give examples? What could have been improved?
- Thinking about your condition, what would be the most important thing a healthcare service could do for you and help you with?
- If you had to describe the perfect healthcare service for people with your condition, what would it be like?

Findings

Response rates

It was necessary to look further afield than Greater Manchester to fulfil the recruitment targets, and people with ataxia living in Blackpool, Preston and Cheshire were also included. Thirty people were identified on Ataxia UK's membership lists, of whom 11 (37%) agreed to participate. Eight (53% of 15 invited to take part) were recruited via a department of Clinical Genetics and a further 8 via a Neurology department. Eleven people were recruited in person by the first author at local branch meetings of Ataxia

UK. The latter two groups may have included people already invited to take part by other means. People were generally keen to take part as ataxia is a rare condition. Many expressed the wish to help others with ataxia in the future. Only two people preferred not to have the interview tape recorded, which was surprising given that many had disordered speech. Interviews lasted for an average of 44 min.

Those interviewed occupied a wide spectrum of disability and disease onset (results not shown). Demographic and diagnostic characteristics are detailed in Table 1. The time since the interviewees had received a diagnosis ranged from 1 to 38 years (median 5.5 years). There was a wide variation in informants' responses. Attempts were made to undertake sub-group analyses, for example, idiopathic vs. definitive diagnoses and those who had been diagnosed at different points in time.

The experience of symptoms and help-seeking. As in rheumatoid arthritis, the focus of Bury's work,¹⁰ the initial onset of ataxic symptoms was usually described as a slow, insidious process, with initial 'trips' or falls put down to accident, clumsiness or drunkenness. The main problems described were disabilities of balance, walking

or speech. One-third also reported other symptoms, including dizziness, migraines, fatigue, arthritis, circulatory diseases, diabetes, incontinence and depression. When people were asked to describe their symptoms and the effect of these symptoms on their lives, they commonly spoke about the stigmatising effects of a disease where they could be accused of being drunk:

Interviewee: ... when I first got this and my speech sounded like I was drunk, when I answered the phone some people more or less put the phone down because it sounded like a drunkard. So now we've got this answering machine and it will ring and whoever it is will, say it's, like the father-in-law or the kids, then we'll answer it, but if it's someone that I do not know I'll just let them leave a message and get back to them. (Male, 50, SCA 8 diagnosis).

As a result of mobility issues and a fear of accusations of drunkenness, several people reported largely staying in their own homes. Others were still in employment and had added uncertainties about when they would be forced to leave work. Some had lost their jobs because of their ataxia and struggled financially. These issues were exacerbated by having a rare disease, not always identified in insurance policies or the medical retirement component of pension schemes. The length of time taken to get a diagnosis in ataxia compounded these problems further, especially in relation to obtaining invalidity, incapacity and disability benefits. Against this background, interviewees reported having engaged with the health system in an attempt to ascertain what was wrong and what could be done to treat it. Here follows a typical description of the beginnings of a diagnostic journey:

Interviewee: Then one year I went on holiday with my mum and dad as a family and they noticed my walking was terrible ... So when we got home my mum and dad made me go to the doctors and then he thought it was in my mind. I was only sixteen; he thought it was all in my mind.

Interviewer: Was this a GP?

Interviewee: Yes it was ... Mum and dad weren't happy at all, so we went back and said, 'No,

Table 1 Sample Characteristics

	Number
Gender	
Women	18
Men	20
Age	
Mean	52.5
Median	50.5
Range	22–77
Diagnosis	
Friedreich's Ataxia (FA)	7
SCA1	1
SCA2	2
SCA6	1
SCA7	1
SCA8	1
Fragile X	1
Assumed inherited (Unconfirmed by gene test)	11
Idiopathic/unexplained	13

there's something not right.' So he did a few little tests on me, then and there in the GP's office. Then sent me straight away then to the hospital. ... Then I was in for about a week.

Interviewer: What, as an in-patient?

Interviewee: Yeah. And he did lots of tests. But after all the tests I had, he wasn't sure what it was. (Female, 41, FA diagnosis)

In contrast to patients' expectations that a cause and treatment plan would be identified in relation to their symptoms, their experience was more usually of being told that they had a rare and untreatable disease; that they should 'go away and live with it'. Whilst there was some evidence that people diagnosed more recently had fared better in services, the issue was by no means clear cut as whereas some people had spent many years pursuing a detailed diagnosis, others were not as concerned to determine a definitive cause for their symptoms.

As in a previous qualitative study that used internet data,⁷ many interviewees seemed to reflect that an idiopathic designation had not stemmed the uncertainties they faced about their illness:

Interviewer: Does the 'idiopathic' label kind of annoy you?

Interviewee: It does yeah. I'd like them to identify what I've got because every time we go to the consultant I get blood tests and, you know, my blood is on file and apparently they keep testing it. But I just want answers, I just want to know, 'Do you know what type I've got? Do you know if it's going to get worse?' (Female, 32, idiopathic ataxia diagnosis).

Interviewees valued demonstrable clinical expertise in ataxia and praised clinicians who had been able to identify 'ataxia' when others had not. However, the more common experience related by most was of neurologists who 'don't know enough'. It was recognized by some interviewees that not all neurologists can be expected to know about ataxia, especially given its status as a rare disease. However,

this insight did not help them in the face of clinical uncertainty. A common complaint of those who had undergone prolonged periods of diagnostic testing was that neurologists were not giving patients enough information about what they were doing nor what they were looking for: 'I asked, "What do you think it is?" to the junior doctor that I spoke to, and he was like, "Well it could be a number of things, but we are excluding them."' (Male, 26, FA diagnosis). Tests were sometimes not followed by any results, with the patient seemingly left in a sort of anxiety-ridden diagnostic 'limbo land'. Their prior assumptions that clinical tests would lead down a road towards clinical and existential certainty were shattered.

The main criticism of neurologists that they 'did not know enough' remained even when patients had consulted someone they perceived to be a specialist in ataxia. This suggests that it was in some cases the 'ataxia' that was the real problem and the lack of progress in medical advances, rather than the clinicians who could or could not diagnose it. People commonly made remarks along the lines of, 'Nobody knows about it and there is no cure': 'I thought "Oh great, medical science, we can tell you what's wrong, but we can't fix it"' (Female, 61, idiopathic ataxia diagnosis). As another interviewee put it, 'That's all you get, "We don't know, we don't know, we don't know"' (Male, 65, late onset ataxia diagnosis). Another recalled their neurologist telling them, 'You have got ataxia but we don't know how or why, obviously, but that is what it is' (Male, 64, idiopathic ataxia diagnosis).

A small number of those interviewed had been recently diagnosed with 'ataxia', and it was evident in their accounts that they expected that more information, such as the SCA 'type' would follow: 'The neurologist... he said it's probably ataxia. We're just waiting now to find out what strength' (Male, 40, SCA7 diagnosis). One interviewee had received an 'ataxia' diagnosis only days before being interviewed, and he was most unusual in

remarking, 'I've had a diagnosis, but nobody's prescribed treatment as of yet. .. basically waiting and see what happens' (Male, 56, ataxia and multi-system atrophy). Following from the discussion above, the perceptions of this newly diagnosed respondent would suggest that he, as yet, had some way to go before confronting the limits of medicine.

Prognostic uncertainties: Existential and clinical. Many interviewees sought a definitive or 'specific' diagnosis to get a better idea of how fast their disease would progress and how their lives would be affected. However, in this study, only three of the respondents who had been given a definitive diagnosis (21% of 14) seemingly reported any benefit from it. One, a young man who had been diagnosed with FA, said, 'Now I know what's wrong, the relief that I do feel is immense because I can put a name to it and fight it' (Male, 26). This finding echoes those of other studies of rare diseases or hard to treat illnesses. For another person eventually found to have FA, the diagnosis was not perceived as helpful, seemingly because they did not understand the implications:

Interviewer: When [the neurologist] told you that you had [FA], was it a relief to finally know what you had?

Interviewee: It didn't really mean anything [Interviewer], if I'm truthful. It didn't make me sit up and think, 'Oh blimey,' because I'd been told it was a different name, that's all. Because as far as I knew up to then my condition, which I used to tell people, was scoliosis and then it changed to [FA] (Male, 58).

Some respondents had made deliberate efforts to pursue exhaustive diagnostic testing to remove uncertainties about causation and prognosis. However, when this existential uncertainty met the sorts of clinical uncertainty outlined in the preceding section, it was unclear whether the individual was better or worse off following investigation:

Interviewee: ... it was really just for peace of mind for myself to try and investigate and try and pinpoint what was the actual cause. If I had

been born with it, you know, if they could tell me a little bit more, but they couldn't. ... And they told me because they think I was born with it, the chances are that I won't become wheelchair bound until another twenty years time. But they can't say for certain because they don't know what is the true cause. (Female, 40, assumed inherited ataxia, unconfirmed by genetic testing).

For the respondent above, the unknowns related to her condition were described as 'scary.' However, as indicated above, it should be stressed that these uncertainties remained even when people had a definitive diagnosis: 'What's to come? I had absolutely no idea. I still don't know' (Male 26, FA diagnosis). Later in the interview, he said, 'To be hit with this, I think, "I don't know what's coming. I don't know what tomorrow is going to bring."' Following Bury,¹⁰ this seems to suggest a form of biographical uncertainty, as if the person with ataxia has come to embody the sorts of prognostic doubts harboured in relation to their illness. For ill people living with lesser degrees of uncertainty, such as those recovering from stroke, a construction of 'the future,' tied to concerns around 'healthy ageing' has been described with reference to biographical illness narratives.¹⁶ However, illness narratives tied to notions of recovery are problematic in the context of progressive ataxias, which are usually incurable.

One respondent, who was unusual in that she had been given a diagnosis of 'cerebellar degeneration', as opposed to ataxia, alleged that her neurologist had told her that she had 4 years to live when the diagnosis was made. In light of this information, when she was still alive 4 years later, she used the information given at the time of diagnosis to bolster her theory that the lack of disease progression was a result of a trip to China for expensive and untested stem cell therapy treatment (Female, 77, cerebellar degeneration diagnosis).

Whilst it was difficult to disentangle the precise relationship between clinical and existential uncertainties,¹⁷ it was clear that each seemingly magnified the other. A possible downside of

diagnostic labelling is that it may lead to a form of symptom amplification.¹⁸ In our study, this was particularly evident in the case of a woman in her 70s, who only found out that she might have ataxia as result of extensive investigations in her son. For most of her life, she thought she had vertigo and treated her symptoms accordingly:

Interviewee's partner: Since [our son] has been diagnosed, we've all been more focused on the symptoms you've been having. Before, we just ignored them.

Interviewee: Before I just thought it was me, you know, I get tired, I get dizzy.

Interviewee's partner: So in effect we feel they have been worse since [our son was diagnosed], because things that would have been shrugged off before, we now attribute to ataxia. (Female, 76, previously thought she had vertigo).

This passage highlights that a diagnosis can have negative and positive aspects. It has already been stressed that an idiopathic designation is a 'downside' as it offers no certainties as regards disease prognosis. A definitive diagnosis may also be of little use if it does not provide certain information regarding disease progression or, in very limited circumstances in the progressive ataxias, a cure. Thus, the young man with FA introduced at the start of this section commented that he was glad that he was not diagnosed earlier in life, and had not had to live with the (uncertain) knowledge of how his symptoms might progress (Male, 26). Clinical and existential uncertainties in relation to disease progression are thus inextricably linked: 'Obviously you're going to worry and you don't know where it's going to because they can't tell you what you've got' (Female, 32, idiopathic ataxia diagnosis).

One person, unlike the others, did not voice uncertainties about his future, and although he did not know the exact aetiology of his illness, he had chosen not to pursue diagnostic testing. Although confined to a wheelchair, he spoke about what he could do rather than what he could not. With a specially adapted

hand-operated wheelchair he was able, and made a point, of going out every day. In his own words, 'I'm just of the mind, just get on with living and just, you know, whatever it does it does, just carry on.' In his 40s at the time of the interview, he had undergone 5 years of medical investigation into his ataxia as a teenager, largely at his parents' insistence. However, as an adult, he had chosen not to pursue testing, because, as he said, 'I'm not a hunter, I'm not a digger'. He contrasted his approach with that of others with ataxia who had pursued diagnostic testing without a satisfactory result. He described their tenacity in this regard as, 'A little sad actually'. He suggested that his approach stemmed from having 'sorted everything out' in his mind. His account seems to echo the notion of 'biographical repair'.¹³

Views of available services. It was clear in accounts of the use of healthcare resources, mainly from accessing the UK National Health Service (NHS), by people with ataxia, that some of the problems they had faced were not ataxia specific, but reflected more general structural and budgetary constraints. On occasions, it was clear that these sorts of structural constraints had been magnified by the fact that they had an incurable and difficult-to-diagnose condition. However, at other times, it seemed as though their criticisms of health services could have just as easily been made by people with any medical or disabling condition. This point was best exemplified by the following interviewee:

Interviewee: I don't, nothing that was bad, you know what I mean? It was just the NHS. Sitting in the waiting room for 3 h every week. That was about it. You couldn't do anything about that, the amount of doctors to patients. I understand it, I'm not going to get angry about it and have a go at people, because it's noone's fault (Male, 22, FA diagnosis).

A commonly reported response was that at the time of diagnosis, patients were told that there was no cure for their ataxia and they should go away and 'get on with it'. Some of

those interviewed said that they respected this apparently brutal honesty on the part of neurologists. However, others were extremely dissatisfied with what they saw as 'negativity' on the part of consultants who were not able to offer them hope that there was some treatment or therapy that might arrest progression. In a particularly succinct comment on the limits of medical knowledge, one interviewee said, 'They're not interested if they can't help' (Female, 60, idiopathic ataxia diagnosis). The implication here is that if there is no cure, then there can be no role for medicine. However, the idea that nothing can be done, seemingly created problems when some neurologists recommended that their patients attend for regular follow-up or monitoring of their symptoms:

Interviewee: [The neurologist] said, 'See you in a year'. I said, 'Well what's the point?' I said, 'I don't see the point because there is nothing you can do for me, so why don't we just call it a bloody day?' (Male, 65, late onset ataxia diagnosis)

By extension, the above interviewee and others questioned the point of attending specialist ataxia centres when there were no treatments available. As another person put it when asked whether they would consider travelling to attend one of the specialist ataxia centres in London, Oxford or Sheffield, 'Yes, I'd go but it would seem like a waste of time' (Male, 67, late onset ataxia diagnosis). Two respondents had attended a specialist ataxia centre and whilst one (with FA) seemed extremely satisfied and was continuing to travel to attend the centre for follow-up, the other (with an idiopathic diagnosis) had stopped going as they did not perceive that there was anything the specialist centre could offer above and beyond a local neurologist.

Whilst some interviewees saw no benefit in follow-up on the basis that there was no cure for ataxia, others reported a perceived 'uselessness' to these visits on the basis of what happened during the appointment itself:

Interviewee: ... I go approximately once a year.

Interviewer: Okay. And what happens at those appointments?

Interviewee: Have a word with me and come home. Nothing.

... [Later, during the same interview]

Interviewer: ... You don't seem to think that visit is useful for you?

Interviewee: It's not.

Interviewer: It's not?

Interviewee: Well you don't get told anything, they don't do anything and nothing changes. (Male, 67, SCA7 diagnosis)

A minority of respondents would rather have valued ongoing monitoring or follow-up by their neurologist, but were seemingly not offered such appointments.

For a minority of interviewees, waiting for the results of tests was a cause of anxiety. For one interviewee, the worst thing about services, when asked, was waiting for test results. Later in the interview, she added: 'I keep thinking, 'Oh it must be bad news that he's left it so long [to get back to me with the test results]' (Female, 55, late onset ataxia diagnosis). Later in the same interview, she said, 'I suppose it is up to me to ask questions, how much I want to know about this thing'. Once again, this seems to highlight the status of patients as medical consumers who are supposed to proactively fight for information, care and healthcare resources.

The 'proactive medical consumer' is predominantly associated with privatized health systems.⁹ However, in this study focused on the NHS, interviewees' responses pointed to a similar model, tied to the idea that healthcare resources are finite and limited. One of the respondents who said that the best things that they had received from health services was a walking stick, added that they had asked for it themselves (Male, 65, late onset ataxia diagnosis). When asked how to improve services for people with ataxia, one person was initially

unsure about how to answer, and then said, 'Am I just waiting for people or am I doing anything myself about it?' (Male, 58, FA diagnosis). The manner in which patients were sometimes seen to be the 'drivers' of their own care is best exemplified in the following interview extract:

Interviewee: The doctor said, 'I can put you on something to calm you down.' I thought, 'I don't want medication to calm me down'. But I could really do with some sort of, someone to talk to more about it who knows about it. Instead of just going to the hospital and them saying, 'There's nothing we can do'. Well, I have read that you can have physiotherapy.' [Why don't they say] 'We can maybe do that for you?' (Female, 41, assumed inherited ataxia, unconfirmed by genetic test).

During the interviews, respondents who had used health services were specifically asked to name the 'best' thing they had received (Table 2). It was striking that one-third could not name anything and one-quarter named a service or disability aid received from social services rather than the health service.

Table 2 'What is the best thing you have received from the health service?' ($n = 30$)

Service	Number
Care at the eye hospital	1
Counselling from my GP	1
Diabetes Clinic	1
Getting the ataxia diagnosis	1
Home visits from therapists	1
Neuro-rehabilitation team	1
Occupational health	1
Physiotherapy	1
Physiotherapy and occupational therapy	1
Speech and language therapy	2
Total NHS Services mentioned	11 (36.7%)
Bath hoist	1
Electric wheelchair	1
Equipment from Social Services	1
Grab bars around the home	1
Personal care	2
Walking aids	2
Total social services/Disability aids mentioned	8 (26.7%)
'Nothing'	11 (36.7%)

Discussion

The most significant weakness of this study is that the interviews were short by comparison with similar studies, and cross-sectional in nature. However, this finding also reflected the fact that many of those interviewed had experienced little contact with health services beyond diagnostics. It is widely assumed that diagnosis comes as a relief, and indeed a definitive diagnosis was a goal and expectation of many of those interviewed. However, in the case of an elderly woman where diagnosis followed clinical investigation in her son, we can begin to see how in some cases diagnosis may bring considerable psychological burdens and anxieties. This is worthy of further study.

Uncertainty emerged as a central feature in accounts of people with progressive ataxia. An often slow and insidious onset (after Bury¹⁰), characterized by trips or falls caused uncertainty around whether there was an illness at all, or rather whether the person was simply clumsy, or perhaps having a dizzy spell. When symptoms progressed to an extent that people sought help, they usually met a wall of clinical uncertainties that challenged their hopes and assumptions that medicine would provide answers around labelling, treatment and prognosis. Contrary to their expectations, the sample of people interviewed in this study now faced new uncertainties: Is my symptom a disease or rather a symptom of another disease? How long will it take to get worse? Will I find a doctor who is expert enough to find out what is wrong with me? Will I be severely affected? Is it genetic?

Ryle, wrote: 'The three main tasks of the clinician are diagnosis, prognosis and treatment. Of these diagnosis is by far the most important, for upon it the success of the other two depend'.¹⁹ Progressive ataxia presents problems in this context. As a usually incurable disease, diagnosis often does not lead to a care or treatment pathway.⁷ Before the diagnostic process is complete, patients may continue to live in the hope or belief that a cure or treatment will be available at the end of this process. However, because diagnosis can take a long time

within the limits of medical knowledge and healthcare structures, systems and budgets, patient uncertainties around the availability of treatments can seemingly be magnified. The diagnostic moment has been referred to as an occasion of 'theoretical coherence' that gives patients 'badly needed credibility'.²⁰ An idiopathic diagnosis is not 'coherent' and can lead some patients to embody clinical and prognostic uncertainties. Whilst the accounts of people with ataxia often reflected a 'quest'²¹ for a clinical label and a cure for their symptoms, their practical experience rather led them to value therapeutic interventions and practical aids to mobility, as opposed to diagnostic exploration. The problems of achieving a definitive diagnosis in progressive ataxia also present considerable challenges for the informed medical consumer.⁹ It is necessary to ask what effects such a consumer can have in the absence of treatments that would bring a cure, or even help to alleviate symptoms.

Those patients who had been fortunate enough to find an ataxia expert, or attend a specialist ataxia centre, seemingly reported a better experience than most (results not shown). However, people still questioned the value of such services, when they were not able to secure a diagnosis that went beyond 'idiopathic' ataxia. Our findings support those found in a study of people with medically unexplained illnesses, where practical, social and medical supports were valued above diagnostics.²² However, interviewees in our study seemed initially more focused on the diagnosis process, at least until the limits of medicine were encountered. The account of one man who had been through this process and learned to live with his symptoms, highlights that the key for patients in dealing with unexplained and untreatable illness may lie more in psychological coping strategies than in technological investigations.

Progressive ataxias are rare chronic illnesses often characterized by an insidious onset. People with ataxia came to medicine as a potential resource that would provide a definitive diagnostic label, prognostic certainty and treatment to alleviate symptoms. However, when the peo-

ple interviewed for this study met clinical, diagnostic and prognostic uncertainties, the perceived failure of the medical system brought increased existential anxieties around their symptoms. In this manner, the technical procedures around medical diagnosis became a potential constraint in their attempts to deal with the changes to their lives brought by ataxia, as opposed to a resource.

Due to the limitations of current scientific knowledge and lack of effective treatments in idiopathic and inherited ataxia, those living with these conditions face significant challenges in light of their apparent expectations around diagnosis and cure. These experiences are likely to change in light of future progress in medical science. In the meanwhile, the key for service providers may lie in the more effective management of patient expectations in relation to receiving a definitive diagnosis and a treatment or cure. From the clinical experience of some of the authors of this article, it has been found useful to make clear to ataxic patients at the start of their diagnostic journey that an explanation may never be found for their symptoms. Given that the patients interviewed for this study appeared to place most value on practical help in relation to their disabling condition, services might usefully place more emphasis on physical and other therapies than might be the case at present.

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