Benson *et al.*
Orphanet Journal of Rare Diseases (2022) 17:130
https://doi.org/10.1186/s13023-022-02270-4

Orphanet Journal of Rare Diseases

RESEARCH

Open Access

Development of a patient journey map for people living with cervical dystonia

Monika Benson[1,2], Alberto Albanese[2,3], Kailash P. Bhatia[4], Pascale Cavillon[5], Lorraine Cuffe[6], Kathrin König[7], Carola Reinhard[2,8] and Holm Graessner[2,8*]

Abstract

Background: Patient journey maps are increasingly used as a tool that enables healthcare providers to refine their service provision to best meet patient needs. We developed a cervical dystonia patient journey map (CDPJM) that describes the holistic patient experience from pre-diagnosis through to long-term treatment.

Methods: The CDPJM was developed in 2 stages; a patient survey (open questions and multichoice) of 15 patients with CD was conducted to inform the design of the CDPJM, which was then refined and validated by an expert-patient focus group.

Results: Qualitative analysis of the patient survey supported five key stages of the patient journey: symptom onset, diagnosis and therapeutic relationship with healthcare professionals, initiation of care for CD, start of CD treatment, and living with treated CD. Following symptom onset, survey respondents described having multiple visits to their family doctor who prescribed strong pain killers and muscle relaxants and referred their patient to up to 10 different specialists for diagnosis. Over half (53.3%) of respondents had received ≥ 1 misdiagnosis. Respondents reported relief at having a diagnosis but a lack of understanding of the prognosis and treatment options; 46.7% said their neurologist did not spend enough time addressing their concerns. Survey respondents reported using a variety of alternative sources of information, including the internet (86.7%), self-help groups (66.7%) and information leaflets provided by health care professionals (60.0%). While botulinum toxin (BoNT) was consistently discussed as the main treatment option, some neurologists also mentioned physiotherapy, counselling, and other complementary approaches. However, patients were often left to seek complementary services themselves. Patients reported a 'rollercoaster' of relief with BoNT treatment with symptoms (and subsequent impact on daily life) returning towards the end of an injection cycle. *"When BoNT works well I can return to an almost normal life … when the injections stop working so well, I have to rest more and avoid going to work and experience life restrictions."*

Conclusions: We present the first patient journey map for CD that can be used to guide local service mapping and to compare current provision with what patients say they want and need.

Keywords: Cervical dystonia, Patient journey, Rare disease, Patient survey, Patient communication, Access to treatment

Background

Cervical dystonia (CD) is a is focal dystonia of the cervical region primarily characterized by involuntary contractions of the neck muscles, resulting in twisting and repetitive movements, and abnormal postures of the head. CD may also present with tremor [1, 2]. It is one of the most common forms of adult-onset dystonia

*Correspondence: Holm.Graessner@med.uni-tuebingen.de
[2] European Reference Network for Rare Neurological Diseases, Tübingen, Germany
Full list of author information is available at the end of the article

with a recent estimated incidence of about 1.18 per 100,000 person-years [3]. The average age of CD onset is around 41 years old [4, 5], and many patients are working and have young families when they are diagnosed [6]. Disability with functional impairment, pain and embarrassment with social withdrawal are common and bring significant quality of life burdens [7–9]. Treatment with botulinum toxin (BoNT) injections is considered first line therapy [10, 11].

In recent years there has been a shift towards 'patient engagement', broadly defined by World Health Organization (WHO) as *"the process of building the capacity of patients, families, carers, as well as health care providers, to facilitate and support the active involvement of patients in their own care, in order to enhance safety, quality and people-centeredness of health care service deliver"'* [12]. However, such a shift requires an understanding of the patient experience [13], and while there have recently been some important patient surveys to better understand how CD and its management impacts patients, they have tended to focus on daily burden [14] and specific aspects of CD management [15, 16]. Another way to visualize the patient experience is to develop a patient journey map, which describes the processes that patients go through when they undergo diagnosis and treatment. This consists of several stages, where each stage comprises one or more healthcare touchpoints [17]. The insights gained from the patient mapping process can help a service designer optimize the experience and generate value for both the user and the healthcare organization providing the service.

Patient journey maps are increasingly used as a tool that enables healthcare providers to reconfigure their approach to the treatment and care, seen from the patients' point of view [17–19]. Through patient journey mapping, a healthcare provider and other stakeholders can identify unmet needs, the barriers and potential gaps in service provision, and work on the solutions to these problems, as well as identifying potential new opportunities for improvement and innovation [17, 19]. Additionally, patient journey tools are increasingly used as a baseline for designing and improving treatment algorithms and developing costing models that can be used to audit the impact of service improvements [20]. As part of the ongoing European Reference Networks for Rare Neurological Diseases (ERN-RND) program [21–23], we aimed to develop a patient journey map for CD that describes the patient experience from pre-diagnosis through to long-term treatment. The CD patient journey map (CDPJM) is presented from the perspective of a 'typical' patient (Lilly), a persona developed based on the feedback of the patient survey.

Results

The CDPJM was developed between March and June 2021 by a patient experience research company (PARTNERSEITZ) in collaboration with patient representatives from Dystonia Europe and affiliated national societies, and was sponsored by Ipsen. The CDPJM was developed in two stages. First, a patient survey of 15 patients living with CD was conducted to inform the design of the map, and secondly, an expert-patient focus group met to review and validate the map and suggest any refinements.

Online patient survey

Fifteen patients living with CD (five each from France, Italy, and the UK) completed the online patient survey between the 24th and 31st March 2021. Key respondent characteristics are presented in Table 1; three quarters of respondents were female and the mean age at diagnosis was 41.5 years. All of the survey respondents were living with chronic CD ($\geq$ 5 years), with 53.3% having been diagnosed more than 10 years ago.

Figure 1 shows an abbreviated version of the CDPJM. The full version is given in Additional file 1. Qualitative analysis of the patient survey supported five key stages of the patient journey:

1. Symptom onset
2. Diagnosis and therapeutic relationship with healthcare professional (HCPs)
3. Initiation of care for CD
4. Start of CD treatment
5. Living with treated CD

Table 1 Respondent characteristics for patients completing the online survey

Characteristic	Survey response
Female/male, n (%)	12/3 (80%/20%)
Age (years), mean ± SD	54.1 ± 10.9
Age at diagnosis (years), mean ± SD	41.5 ± 9.6
Employed, n (%)	8 (53.3%)
Time since diagnosis, n (%)	
Within past 5 years	2 (13.3%)
Within past 5–10 years	5 (33.3%)
> 10 years ago	8 (53.3%)
First symptoms experienced at onset*	
Head and/or neck tilting/twisting	11 (73.3%)
Neck spasms	3 (20.0%)
Pain	6 (40.0%)
Tremor	5 (33.3%)

* Patients cited the first symptoms they recalled experiencing at onset (open question allowing multiple symptoms to be identified)

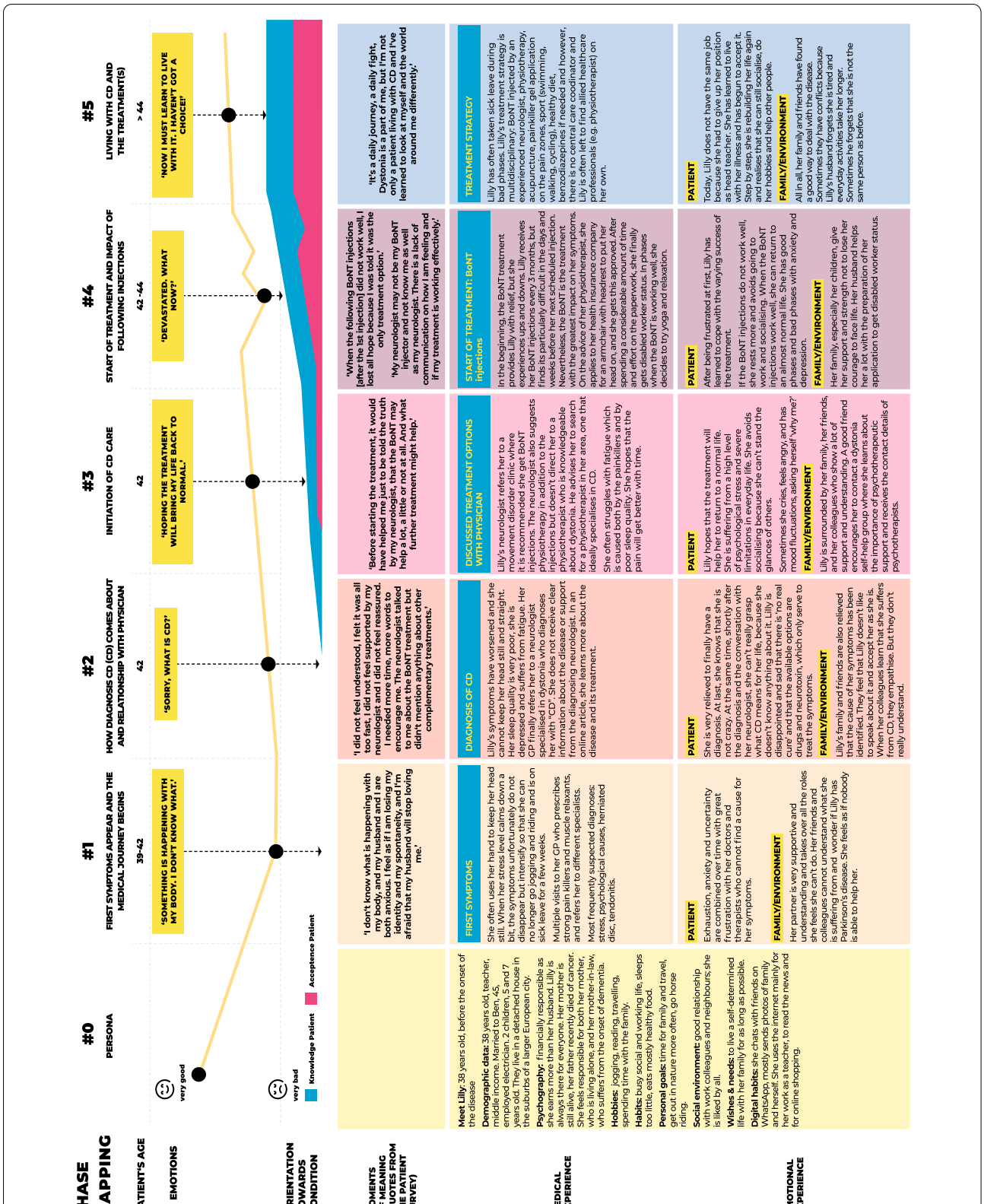


Fig. 1 Abbreviated CD patient journey map

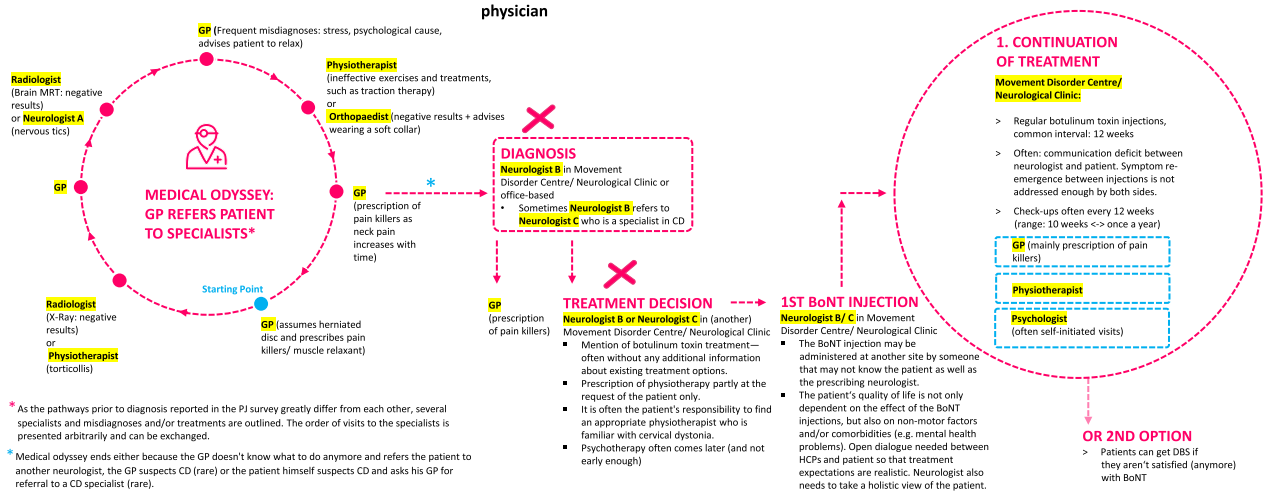


Fig. 2 The intersectoral pathway

At symptom onset, most survey respondents (n = 12, 80%) reported abnormal head and/or neck positions as their first symptom of CD. The other three respondents reported tremor (n = 2) and pain (n = 3) as their earliest symptoms. At this time, 60% of survey respondents already described impact on their daily activities (e.g., eating, drinking, walking, any physical activities) and 10 (66.7%) reported an impact on sleep. Survey respondents described having multiple visits to their family doctor who frequently prescribed strong pain killers and muscle relaxants, and referred their patient to various specialists including neurologists, rheumatologists, orthopedists, psychologists, radiologists, physiotherapists and chiropractors. Misdiagnosis was common (53.3% of survey respondents had received ≥ 1 misdiagnosis) with suspected diagnoses including: stress and other psychological causes, muscular sclerosis, herniated discs, tendonitis and stiff neck due to air conditioning. Without a diagnosis, survey respondents cited feeling even more anxious and stressed, and shame/embarrassment about their condition which was often obvious to their family, friends, and co-workers.

By the time of diagnosis and initiation of CD care, survey respondents had already seen up to 10 specialist and non-specialist HCPs [range 1–10] before being diagnosed with CD by a neurologist. Figure 2 shows a typical intersectoral pathway, from diagnosis through to treatment. Some of the respondents described meeting more than one neurologist before receiving their diagnosis, with many respondents finally being referred to movement disorder specialists (e.g., dystonia expert) who gave the diagnosis and offered treatment. Respondents reported relief at having a diagnosis but a general lack of understanding of the prognosis and possible options for long-term management; 46.7% said they felt their neurologist did not spend enough time discussing their diagnosis and addressing their concerns. The majority of patients (11/15, 73.3%) said receiving their diagnosis of CD impacted their mental health and eight patients (53.3%) said they had received/were receiving mental health interventions (medication and/or counselling). Survey respondents reported using a variety of alternative sources of information, including the internet (86.7%), self-help groups (66.7%) and information leaflets provided by HCPs (60.0%).

Respondents reported that their neurologists often discussed chemodenervation with BoNT as the main treatment option. While some, but not all, neurologists mentioned complementary treatment approaches such as physiotherapy, they did not consistently refer the patient to the allied services and respondents sought the additional treatments themselves. Survey respondents reported a 'rollercoaster' of relief with BoNT treatment with symptoms (and subsequent impact on daily life) returning towards the end of an injection cycle. Respondents noted that *"When BoNT works well I can return to an almost normal life … when the injections stop working so well, I have to rest more and avoid going to work and experience life restrictions."* A few respondents described experiencing their best effect after their first BoNT injection, and that their CD changed over time *"When the following botulinum toxin injections [after the 1st injection] did not work as well, I lost*

all hope because I was told it was the only treatment option." Three of the survey respondents had opted for a surgical intervention (deep brain stimulation or selective peripheral denervation) because of inadequate relief with BoNT. Finally, survey respondents generally reported acceptance about living with their CD, with some fears for the future – especially as they continue to age. Many respondents described strategies such as looking for social and emotional support (from family, friends, patient groups and professionals), physical exercise, and relaxation strategies as helpful in their day to day lives.

Expert patient focus group validation of the CDPJM

The expert patient focus group, comprised of patient society representatives living with CD, generally agreed with the findings of the patient survey and the design of the CDJPM. Select quotes from the focus group can be found in Additional file 2. Briefly, the focus group agreed that it can take 2–3 years before a patient receives their diagnosis of CD from a movement disorder specialist. This was discussed as a result of lack of awareness of CD and other rare diseases in primary care. There was general agreement on the importance of patients with CD being referred to a movement disorders expert neurologist for optimal CD management. However, discussion focused on gaps in the communication between the HCP and patient, particularly about the full range of treatment options and what a diagnosis of CD might mean for the patient.

Another potential gap was the lack of a central coordinator between neurology and other (e.g., physiotherapy and psychosocial) support services. In terms of long-term management with BoNT injections, the expert patient focus group agreed with the description of treatment as rollercoaster, where the patient experiences relief following (re)injection and then symptom re-emergence once the effects start to wear-off. The expert patient focus group noted that many clinicians operate injection clinics which are typically too busy to allow for HCP/patient reflection and re-evaluation of treatment. Here, the significant time restraints of a busy injection clinic hinder active participation of the patient who, for example, can worry that if they are perceived as 'complaining' that their symptom relief doesn't last the full injection interval, the injections will be taken away. The busy injection clinic was generally discussed in terms of perpetuating the paternalistic model of medicine – and hindering patient centered care. The worldwide lack of neurologists [24] and the need for better trained injectors [25] were identified as key problems that limit patient access to the right doctors at the right time.

Discussion

Patient experience and satisfaction have been demonstrated to be the single most important aspect in assessing the quality of healthcare [17]. Accumulating evidence shows the importance of patient engagement and attention to patient expectations in the healing process and it is increasingly accepted that patient involvement in the design of healthcare services improves the relevance and quality of the services [26–28]. This is especially important in the design of services for rare diseases, such as CD, where the knowledge base is often restricted to small numbers of expert doctors. To our knowledge, we present here the first patient journey map for patients living with CD. Importantly, the map was primarily informed by patient experience (in the form of a survey and expert patient focus group) supplemented with clinical guidance and the existing literature.

The mapping process identified five key stages of the patient journey, each with specific gaps in service provision and barriers to optimal care. In stage 1 (symptom onset) family doctor education and awareness were considered the biggest hurdle to diagnosis and, as reported for other rare diseases [29], survey respondents had already seen up to 10 different (non-specialist and specialist) HCPs before being diagnosed with CD by a neurologist. Given the number of rare diseases a family doctor may come across in their daily work, potential solutions to this are difficult but an easier target audience for specific education might be the HCPs to whom the patients are often misdirected (e.g., osteopaths, orthopedics, spine surgeons, physiotherapists etc.). A key gap identified through stages 2 and 3 was the need for improved communication between patient and physician. Here, the mapping process clearly highlighted the need for HCPs to provide their patients with more detailed information on the disease and on the full array of treatment options, including complementary therapies such as physiotherapy and psychosocial support. In such situations, the CDPJM can be used as a tool to help explain a typical clinical pathway to patients and help patients identify their specific needs and raise any issues with their treating team.

With respect to treatment (stages 3–5), survey respondents initially responded they had 'great hope' at the start of treatment. This resonates with the results of a prior patient survey which also identified high patient expectations of BoNT treatment, with a majority expecting freedom from spasms and pain and over half expecting to return to a normal routine [14]. Both respondents and the focus group reported a rollercoaster of relief with BoNT treatment with symptoms (and subsequent impact on daily life) returning towards the end of an injection cycle. This strongly aligns with a recent

patient survey where Comella and colleagues found that 88% of patients living with CD experience symptom re-emergence that impacts their daily life before the next scheduled injection [15]. Such findings highlight the importance of empowering patients to explain how treatment affects their daily life such that the clinician can work to optimize injection and other treatment parameters for best effect. For example, the early re-emergence of motor symptoms may prompt a reassessment of injection parameters (muscles injected, doses used), while the development of non-motor symptoms such as depression or anxiety may prompt referral to an allied professional. Patients also described that they experienced their best response to BoNT during the first injection cycle(s). This phenomenon is well described, and recent observational studies have shown the greatest symptom relief in newly treated patients [30–32]. However, this does not mean that the treatment is ineffective in chronic patients, and the same studies showed a clinically significant effect and high patient satisfaction (> 80%) across repeat cycles [30]. It has been suggested that the phenomenon we observed in our survey may reflect patient perceptions of their disease and how they self-rate their condition [32]. Our own survey results showed that patients gradually become accustomed to their condition, and it might be that by the time patients are into > 5 years of treatment it may be very difficult for them to remember what it was like before.

The CDPJM identifies several common gaps in service provision. The lack of clear clinical pathways for referrals to physiotherapists and psychologists was identified as a key gap in all participating countries. Here the patient journey map can be used as a baseline tool to understand which HCPs patients find useful, so that movement disorder centers can develop and reinforce links with the allied services such that the long-term management plan for CD becomes much more multidisciplinary. Lessons can be learned from Parkinson's disease, which is another neurological condition but is far more common than CD [33]. In the UK, Parkinson's services have traditionally followed a common model of diagnosis by a movement disorder specialist with routine follow-up with a Parkinson's disease Specialist Nurse who refers back to the specialist as required. However, the importance of a multidisciplinary team approach has been increasingly recognized in this area, and has led to the development of local 'hub' services in which a care coordinator serves as the central point of contact coordinating care between established services, including the neurologist, the specialist nurse, physiotherapists, occupational and speech and language therapists etc. [34]. Such integrated care pathways took years to develop but all started with mapping processes based on patient involvement and feedback, similar to the mapping process we present for CD [33, 35].

Although the idea of patient journey maps are gaining traction [17–19], there are no standard approaches to performing the steps of the mapping exercise and it has been suggested that the lack of consistent methodology may contribute to the low adoption rates in healthcare [36, 37]. It is important that our mapping process was patient driven. Although clinicians were involved in the survey design and interpretation of the findings, the CDPJM was purposely designed to reflect the patient perspective; future mapping could look to integrate the healthcare provider point of view. We chose a method which gives the typical patient a 'persona' that clinicians and patients can relate to rather than a data set. Although personas are a commonly used tool to help service designers make decisions, they will not capture every individual patient's needs [38, 39]. In this pilot mapping exercise, we chose to work with smaller groups of patients for ease of communication and because we wanted to collect and collate qualitative feedback. This follows the current recommendations for obtaining deep experiential insights from patients during the mapping process [19, 40]. While larger patient surveys are preferable for collecting quantifiable data, they often miss the unique, connective links that direct patient feedback based on their lived experiences can give. Following this pilot, future work could consider international expansion for a more global approach, or perhaps more pragmatically, a similar mapping process at the national level could also be very informative.

Another possible limitation of the select group of patients involved is the chronicity of their disease. Over half of the patients who responded to the patient survey had been living with their condition for over 10 years, which might have made it more difficult to remember how they felt in the earlier stages of the disease. It is also conceivable that certain processes have changed in the years since their diagnosis. However, we did not observe any obvious quantitative or qualitative differences in the patient experience dependent on the time since diagnosis and the experience of expert patient representatives suggests that little has changed in past decades. Survey respondents and focus group members were all recruited via the participating Dystonia Europe affiliates, which may have introduced bias since people who engage in support groups are often female, younger, more highly educated, of a higher economic status and more anxious about their disease than those who do not [41]. CD is at least twice as common in women than in men [42] and a recent retrospective study at one center found sex differences in the age of onset and treatment response (men were diagnosed younger, had a less robust response to

treatment, and were more likely to discontinue care) [43]. As such the male perspective on the patient journey may differ from females and future mapping may benefit from specific subgroup analyses with a larger sample. Another limitation is the small number of participating countries (Italy, France, UK) all of which have public healthcare provision. As such the tool should be considered a baseline that can be tailored to a local hospital, area or region.

In summary, we present the first patient journey map for people living with CD. It is hoped that clinicians interested in the management of CD can use the map as a tool to guide their own service mapping process and compare their services with what patients say they want and need. Similarly, patient societies (including Dystonia Europe and their affiliates) can use the tool to identify gaps in patient education and support networks and identify potential programs in their local areas as potential solutions to unmet needs. A plain language summary of the paper is provided in Additional file 4. These observations have to be carried forward to the relevant organizations devoted to improving patient care (e.g., NIHR in the UK and similar organizations in other countries). As services and treatments evolve, the CDPJM should be re-evaluated and refined over time.

Methods

Patient survey

An online survey of 15 patients living with CD was conducted using LamaPoll (see Additional file 3 for patient survey questions). The study was conducted in compliance with relevant codes of conduct from the European Pharmaceutical Market Research Association and the Insights Association (formerly known as CASRO).

The structure and contents of the survey were based on a generic patient mapping survey tailored to CD in collaboration with patient representatives from Dystonia Europe, representatives from the European Reference Network for Rare Neurological Diseases, sponsor representatives and experts from the patient experience company. All 15 patients were recruited by the participating Dystonia Europe affiliates (France: AMADYS; Italy: Associazione per la Ricerca sulla Distonia A.R.D.; United Kingdom: Diagnosis, Education and Research [ADDER]). Other than having a diagnosis of CD (self-reported), there were no formal inclusion or exclusion criteria for participation in the mapping process. The survey was conducted in English and included 45 questions. Questions were a mix of multiple choice and free entry formats, and data analysis was purely descriptive.

Developing and validating the CDPJM

To support the development of the CDPJM, a broad literature review was performed using PubMed and Google Scholar to identify relevant literature, search terms included cervical dystonia OR spasmodic torticollis AND diagnosis, treatment, patient. References were limited to those published between 2000 and 2021 and those published in English or German. Using the survey results, a first draft patient journey map was developed, and sense checked against the current literature. This first draft map was then shared with expert patients from Dystonia Europe and its participating affiliates, and an online focus group meeting was convened in May 2021 to come to consensus on each of the stages identified in the mapping process. Focus group participants reviewed each stage of the CDPJM and had an open discussion on how well the map reflects the care pathway in the countries they represent. The focus group meeting was recorded, and the meeting minutes were used to refine the map into the final CDPJM.

Abbreviations

BoNT: Botulinum neurotoxin; CD: Cervical dystonia; CDPJM: Cervical dystonia patient journey map.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s13023-022-02270-4.

Additional file 1.. Full version of the CD patient journey map.

Additional file 2. Verbatim quotes from the expert focus group.

Additional file 3. Patient survey.

Additional file 4. Plain language summary.

Acknowledgements

This work is generated within the European Reference Network for Rare Neurological Diseases—Project ID No 739510. The authors thank all the people living with CD who participated in the patient survey and in the expert-patient focus group. We also thank Dystonia Europe, AMADYS, Associazione per la Ricerca sulla Distonia A.R.D., and Action for Dystonia, Diagnosis, Education and Research [ADDER]) for their assistance in recruiting patients to the survey and participation in the expert-patient focus group. Medical writing support was provided by Anita Chadha-Patel, PhD, of ACP Clinical Communications Ltd (Hertfordshire, UK), which was funded by Ipsen (Paris, France) in accordance with Good Publication Practice guidelines.

Authors' information

Monika Benson is executive director of Dystonia Europe. Monika has cervical dystonia and was a board member of the Swedish Dystonia Association for 10 years. Monika has been working as a coordinator of workshops, courses and lectures at a school in Lund, Sweden. **Alberto Albanese** is Professor of Neurology and Head Neurologist at the at Humanitas Research Hospital, Rozzano, Milan. He has authored over 200 publications covering an ample spectrum of movement disorders, including dystonia, Parkinson disease, choreas and tics. He was a leading pioneer in the introduction of botulinum toxins in Italy and has pioneered the use of deep brain stimulation in movement disorders. **Kailash Bhatia** is a professor of clinical neurology in the Sobell Department of Movement Neuroscience at the Institute of Neurology, UCL, Queen Square, London and an honorary consultant neurologist at the affiliated National

Hospital for Neurology, Queen Square. He has published over 500 publications in the field of movement disorders and is the current chairman of the Movement Disorders subcommittee of the European Neurological Society (ENS). His main research interest is in movement disorders, merging clinical, electrophysiological and genetic methods to study the pathophysiology of conditions like dystonia, tremor and parkinsonism. **Pascale Cavillon** is Global Patient Centricity Director for Ipsen where she oversees patient support programs designed to help patients with chronic disease requiring chronic and or complex medication therapy living with their disease and improve patient outcomes. **Lorraine Cuffe** is Medical Director for Ipsen. **Kathrin König** is Senior Project Manager for PARTNERSEITZ, a patient experience research company specializing in the development of patient journey maps. **Carola Reinhard** is a research manager at the Institute of Medical Genetics and Applied Genomics at the University Hospital Tübingen. She is project manager of the European Reference Network for Rare Diseases (ERN-RND). **Holm Graessner** is managing director of the Centre for Rare Diseases Tübingen, Germany. He is Coordinator of the ERN-RND.

Authors' contributions

MB, PC, LC, CR and HG identified the need for a patient journey map for patients living with CD and advised on the development of the patient survey. KK was responsible for the development of the patient journey map. MB participated in the expert patient focus group. AA, KB and HG advised on the clinical management aspects of the patient journey. All authors provided critical review of the manuscript and approved the paper in its final version.

Funding

The development of the Cervical Dystonia Patient Journey Map was funded by Ipsen.

Availability of data and materials

The CD patient journey map is available on request from the corresponding author.

Declarations

Ethics approval and consent to participate

Clinical Research Ethics Committee or Independent Review Board approval was not required for the patient survey.

Consent for publication

Not applicable.

Competing interests

Monika Benson, Carola Reinhard and Holm Graessner are members of the European Reference Network for Rare Neurological Diseases and have no further conflicts to report. Alberto Albanese reports research support from Ipsen, Merz. He is the Specialty Chief Editor Frontiers in Neurology – Movement Disorders Section. Kailash P Bhatia has received grant support from EU Horizon 2020. He receives royalties from publication of the Oxford Specialist Handbook Parkinson's Disease and Other Movement Disorders (Oxford University Press, 2008), of Marsden's Book of Movement Disorders (Oxford University Press, 2012), and of Case Studies in Movement Disorders–Common and uncommon presentations (Cambridge University Press, 2017). He has received honoraria/personal compensation for participating as consultant/scientific board member from Ipsen, Allergan, and honoraria for speaking at meetings from Allergan, Ipsen, and the International Parkinson's Disease and Movement Disorders Society. Pascale Cavillon and Lorraine Cuffe are employed by Ipsen. Kathrin König is employed by PARTNERSEITZ GmbH which was contracted to develop the CDPJM (funded by Ipsen).

Author details

[1]Dystonia Europe, Brussels, Belgium. [2]European Reference Network for Rare Neurological Diseases, Tübingen, Germany. [3]Department of Neurology, IRCCS Istituto Clinico Humanitas, Rozzano, MI 20089, Italy. [4]Department of Clinical and Movement Neuroscience, Institute of Neurology UCL, London WC1N 3BG, UK. [5]Patient Centricity, Ipsen, Boulogne Billancourt, France. [6]Medical Affairs, Ipsen, Slough, UK. [7]PARTNERSEITZ GmbH, Ludwigshafen, Germany. [8]Centre for Rare Diseases and Institute of Medical Genetics and Applied Genomics, University Hospital Tübingen, Calwerstr. 7, 72076 Tübingen, Germany.

Received: 9 December 2021 Accepted: 20 February 2022
Published online: 21 March 2022

References

1. Grütz K, Klein C. Dystonia updates: definition, nomenclature, clinical classification, and etiology. J Neural Transm. 2021;128(4):395–404.
2. Albanese A, Bhatia K, Bressman SB, Delong MR, Fahn S, Fung VS, et al. Phenomenology and classification of dystonia: A consensus update. Mov Disord. 2013;28(7):863–73.
3. LaHue SC, Albers K, Goldman S, Lo RY, Gu Z, Leimpeter A, et al. Cervical dystonia incidence and diagnostic delay in a multiethnic population. Mov Disord. 2020;35(3):450–6.
4. Chan J, Brin MF, Fahn S. Idiopathic cervical dystonia: clinical characteristics. Mov Disord. 1991;6(2):119–26.
5. Stacy M. Epidemiology, clinical presentation, and diagnosis of cervical dystonia. Neurol Clin. 2008;26(Suppl 1):23–42.
6. Skogseid IM, Roislien J, Claussen B, Kerty E. Long-term botulinum toxin treatment increases employment rate in patients with cervical dystonia. Mov Disord. 2005;20(12):1604–9.
7. Ben-Shlomo Y, Camfield L, Warner T. What are the determinants of quality of life in people with cervical dystonia? J Neurol Neurosurg Psychiatry. 2002;72(5):608–14.
8. Pekmezovic T, Svetel M, Ivanovic N, Dragasevic N, Petrovic I, Tepavcevic DK, et al. Quality of life in patients with focal dystonia. Clin Neurol Neurosurg. 2009;111(2):161–4.
9. Muller J, Kemmler G, Wissel J, Schneider A, Voller B, Grossmann J, et al. The impact of blepharospasm and cervical dystonia on health-related quality of life and depression. J Neurol. 2002;249(7):842–6.
10. Albanese A, Asmus F, Bhatia KP, Elia AE, Elibol B, Filippini G, et al. EFNS guidelines on diagnosis and treatment of primary dystonias. Eur J Neurol. 2011;18(1):5–18.
11. Simpson DM, Hallett M, Ashman EJ. Practice guideline update summary: Botulinum neurotoxin for the treatment of blepharospasm, cervical dystonia, adult spasticity, and headache. Neurology. 2016;86:1–9.
12. World Health Organization. Patient Engagement: Technical Series on Safer Primary Care 2016. https://apps.who.int/iris/bitstream/handle/10665/252269/9789241511629-eng.pdf.
13. EFNA, Optimising patient involvement in neuroscience research. 2020 – European Federation of Neurological Associations. https://www.efna.net/wp-content/uploads/2020/05/EFNA-Report_Patient-Inolvement-in-Neuroscience-Research.pdf.
14. Comella C, Bhatia K. An international survey of patients with cervical dystonia. J Neurol. 2015;262(4).
15. Comella C, Ferreira JJ, Pain E, Azoulai M, Om S. Patient perspectives on the therapeutic profile of botulinum neurotoxin type A in cervical dystonia. J Neurol. 2021;268(3):903–12.
16. Sethi KD, Rodriguez R, Olayinka B. Satisfaction with botulinum toxin treatment: a cross-sectional survey of patients with cervical dystonia. J Med Econ. 2012;15(3):419–23.
17. Bolz-Johnson M, Meek J, Hoogerbrugge N. "Patient Journeys": improving care by patient involvement. Eur J Hum Genet EJHG. 2020;28(2):141–3.
18. Arias M, Rojas E, Aguirre S, Cornejo F, Munoz-Gama J, Sepúlveda M, et al. Mapping the patient's journey in healthcare through process mining. Int J Environ Res Public Health. 2020;17(18).
19. Simonse L, Albayrak A, Starre S. Patient journey method for integrated service design. Des Health. 2019;3(1):82–97.
20. Public Health England. Improvement Leaders' Guide Process mapping, analysis and redesign. https://www.england.nhs.uk/improvement-hub/wp-content/uploads/sites/44/2017/11/ILG-1.2-Process-Mapping-Analysis-and-Redesign.pdf.
21. Post AEM, Klockgether T, Landwehrmeyer GB, Pandolfo M, Arnesen A, Reinhard C, et al. Research priorities for rare neurological diseases: a representative view of patient representatives and healthcare professionals from the European Reference Network for Rare Neurological Diseases. Orphanet J Rare Dis. 2021;16(1):135.
22. Painous C, van Os NJH, Delamarre A, Michailoviene I, Marti MJ, van de Warrenburg BP, et al. Management of rare movement disorders in

Europe: outcome of surveys of the European Reference Network for Rare Neurological Diseases. Eur J Neurol. 2020;27(8):1493–500.
23. Reinhard C, Bachoud-Lévi A-C, Bäumer T, Bertini E, Brunelle A, Buizer AI, et al. The European reference network for rare neurological diseases. Front Neurol. 2021;11(1753).
24. Burton A. How do we fix the shortage of neurologists? Lancet Neurol. 2018;17(6):502–3.
25. Colosimo C, Bhidayasiri R, Fheodoroff K, Bhatia K, Chung TM, Landreau T, et al. Management of spastic paresis and cervical dystonia: access to therapeutic innovations through an international program of practical courses. Clin Ther. 2019;41(11):2321-30.e4.
26. Heckemann B, Wolf A, Ali L, Sonntag SM, Ekman I. Discovering untapped relationship potential with patients in telehealth: a qualitative interview study. BMJ Open. 2016;6(3):e009750.
27. Sharma AE, Knox M, Mleczko VL, Olayiwola JN. The impact of patient advisors on healthcare outcomes: a systematic review. BMC Health Serv Res. 2017;17(1):693.
28. Fønhus MS, Dalsbø TK, Johansen M, Fretheim A, Skirbekk H, Flottorp SA. Patient-mediated interventions to improve professional practice. Cochrane Database Syst Rev. 2018;9(9):Cd012472.
29. EURORDIS. Solve-RD infographic on the patient journey to diagnosis. https://www.eurordis.org/publication/diagnosisjourneyinfographic. Last accessed September 2021.
30. Colosimo C, Charles D, Misra VP, Maisonobe P, Om S. Group IICs. Cumulative effects of long-term treatment with abobotulinumtoxinA in cervical dystonia: findings from a prospective, observational study. J Neurol Sci. 2020;416:117015.
31. Trosch RM, Misra VP, Maisonobe P, Om S. Impact of abobotulinumtoxinA on the clinical features of cervical dystonia in routine practice. Clin Parkinsonism Relat Dis. 2020;3:100063.
32. Colosimo C, Charles D, Misra VP, Maisonobe P, Om S. Group IICs. How satisfied are cervical dystonia patients after 3 years of botulinum toxin type A treatment? Results from a prospective, long-term observational study. J Neurol. 2019;266(12):3038–46.
33. Radder DLM, Nonnekes J, van Nimwegen M, Eggers C, Abbruzzese G, Alves G, et al. Recommendations for the organization of multidisciplinary clinical care teams in Parkinson's disease. J Parkinsons Dis. 2020;10:1087–98.
34. Mace T, Peel C. Parkinson's Hub: an integrated pathway for people with Parkinson's and frailty. ACNR. 2020;19:38–41.
35. Post B, van der Eijk M, Munneke M, Bloem BR. Multidisciplinary care for Parkinson's disease: not if, but how! Pract Neurol. 2011;11(2):58–61.
36. Sijm-Eeken M, Zheng J, Peute L. Towards a lean process for patient journey mapping - a case study in a large academic setting. Stud Health Technol Inform. 2020;270:1071–5.
37. Bharatan T, Devi R, Huang PH, Javed A, Jeffers B, Lansberg P, et al. A methodology for mapping the patient journey for noncommunicable diseases in low- and middle-income countries. J Healthc Leadersh. 2021;13:35–46.
38. Kendall L, Mishra SR, Pollack A, Aaronson B, Pratt W. Making background work visible: opportunities to address patient information needs in the hospital. AMIA Annu Symp Proc. 2015;2015:1957–66.
39. Agapie E, Kendall L, Mishra SR, Haldar S, Khelifi M, Pollack A, et al. Using priorities of hospitalized patients and their caregivers to develop personas. AMIA Annu Symp Proc. 2020;2019:200–9.
40. Bate P, Robert G. Experience-based design: from redesigning the system around the patient to co-designing services with the patient. Qual Saf Health Care. 2006;15(5):307–10.
41. Van Uden-Kraan CF, Drossaert CHC, Taal E, Smit WM, Bernelot Moens HJ, Van de Laar MAFJ. Determinants of engagement in face-to-face and online patient support groups. J Med Internet Res. 2011;13(4):e106.
42. https://rarediseases.org/rare-diseases/cervical-dystonia/. Last accessed January 2022.
43. Supnet ML, Acuna P, Carr SJ, Kristoper de Guzman J, Al Qahtani X, Multhaupt-Buell T, et al. Isolated Cervical Dystonia: Management and Barriers to Care. Frontiers in Neurology. 2020;11.

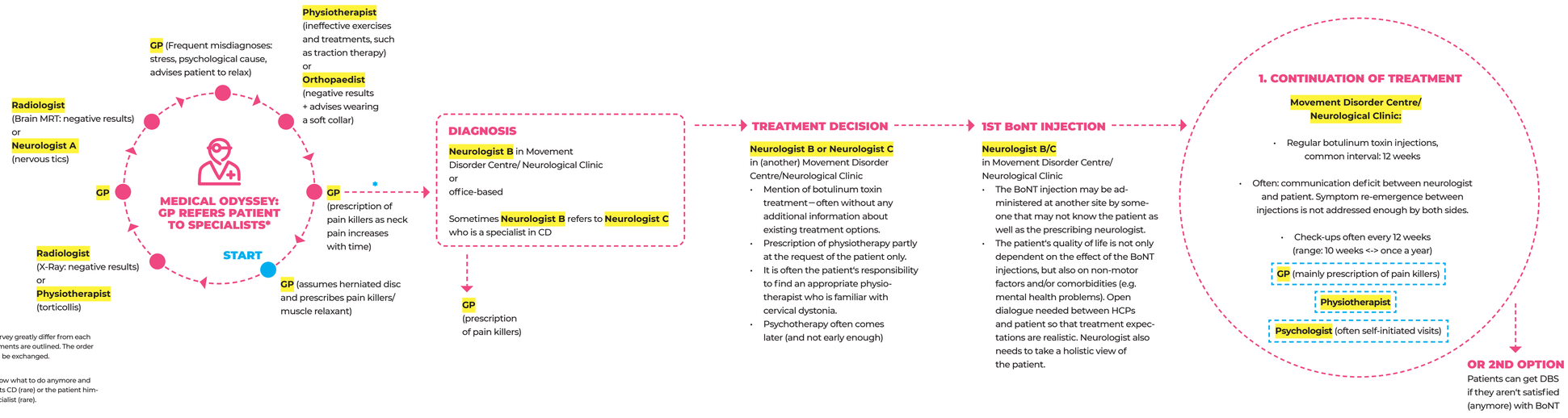
Publisher's Note

PHASE MAPPING (FRAMEWORK)

	#0	#1	#2	#3	#4	#5
	PERSONA	**FIRST SYMPTOMS APPEAR AND THE MEDICAL JOURNEY BEGINS**	**HOW DIAGNOSIS CERVICAL DYSTONIA (CD) COMES ABOUT AND RELATIONSHIP WITH PHYSICIAN**	**INITIATION OF CD CARE**	**START OF TREATMENT AND IMPACT OF FOLLOWING INJECTIONS**	**LIVING WITH CD AND THE TREATMENT(S)**
PATIENT'S AGE		39-42	42	42	42 -44	> 44
EMOTIONS		‘SOMETHING IS HAPPENING WITH MY BODY. I DON’T KNOW WHAT.’	‘SORRY, WHAT IS CERVICAL DYSTONIA?’	‘HOPING THE TREATMENT WILL BRING MY LIFE BACK TO NORMAL.’	‘DEVASTATED. WHAT NOW?’	‘NOW I MUST LEARN TO LIVE WITH IT. I HAVEN’T GOT A CHOICE!’
ORIENTATION TOWARDS CONDITION						
MOMENTS OF MEANING (QUOTES FROM THE PATIENT SURVEY)		‘I don‘t know what is happening with my body, and my husband and I are both anxious. I feel as if I am losing my identity and my spontaneity, and I‘m afraid that my husband will stop loving me.’	‘I did not feel understood, I felt it was all too fast, I did not feel supported by my neurologist and I did not feel reassured. I needed more time, more words to encourage me. The neurologist talked to me about the botulinum toxin treatment but didn‘t mention anything about other complementary treatments.’	‘Before starting the treatment, it would have helped me just to be told the truth by my neurologist, that the botulinum toxin may help a lot, a little or not at all. And what further treatment might help.’	1. ‘When the following botulinum toxin injections [after the 1st injection] did not work well, I lost all hope because I was told it was the only treatment option.’ 2. ‘For the initial years after the diagnosis, I always perceived that doctors did not fully trust me about my condition [...]. ’ 3. ‘My neurologist may not be my botulinum toxin injector and not know me as well as my neurologist. There is a lack of communication on how I am feeling and if my treatment is working effectively.’	‘It’s a daily journey, a daily fight, Dystonia is a part of me, but I‘m not only a patient living with cervical dystonia and I‘ve learned to look at myself and the world around me differently.’
MEDICAL EXPERIENCE	**Meet Lilly** 38 years old, before the onset of the disease **Demographic data:** 38 years old, teacher, middle income. Married to Ben, 45, employed electrician. 2 children, 5 and 7 years old. They live in a detached house in the suburbs of a larger European city. **Psychography:** financially responsible as she earns more than her husband. Lilly is always there for everyone. Her mother is still alive, her father recently died of cancer. She feels responsible for both her mother, who is living alone, and her mother-in-law, who suffers from the onset of dementia. **Hobbies:** jogging, reading, travelling, spending time with the family. **Habits:** busy social and working life, sleeps too little, eats mostly healthy food. **Personal goals:** time for family and travel, get out in nature more often, go horse riding. **Social environment:** good relationship with work colleagues and neighbours; she is liked by all. **Wishes & needs:** to live a self-determined life with her family for as long as possible. **Worries/pain:** Lilly fears that she will not be able to meet her mother’s needs because her mother-in-law needs more and more support. **Digital habits:** she chats with friends on WhatsApp, mostly sends photos of family and herself. She uses the internet mainly for her work as a teacher, to read the news and for online shopping.	**FIRST SYMPTOMS** She often uses her hand to keep her head still. When her stress level calms down a bit, the symptoms unfortunately do not disappear but intensify so that she can no longer go jogging and riding and is on sick leave for a few weeks. Multiple visits to her GP who prescribes strong pain killers and muscle relaxants, and refers her to different specialists. Most frequently suspected diagnoses: stress, psychological causes, herniated disc, tendonitis.	**DIAGNOSIS OF CERVICAL DYSTONIA** Lilly’s symptoms have worsened and she cannot keep her head still and straight. Her sleep quality is very poor, she is depressed and suffers from fatigue. Her GP finally refers her to a neurologist specialised in dystonia who diagnoses her with “cervical dystonia”. She does not receive clear information about the disease or support from the diagnosing neurologist. In an online article, she learns more about the disease and its treatment.	**DISCUSSED TREATMENT OPTIONS WITH PHYSICIAN** Lilly’s neurologist refers her to a movement disorder clinic where it is recommended she get botulinum toxin injections. The neurologist also suggests physiotherapy in addition to the injections but doesn’t direct her to a physiotherapist who is knowledgeable about dystonia. He advises her to search for a physiotherapist in her area, one that ideally specialises in cervical dystonia. She often struggles with fatigue which is caused both by the painkillers and by poor sleep quality. She hopes that the pain will get better with time.	**START OF TREATMENT: botulinum toxin injections** In the beginning, the botulinum toxin treatment provides Lilly with relief, but this effect is unstable with the following injections and she experiences ups and downs. Lilly receives her botulinum toxin injections every 3 months, but finds its particularly difficult in the days and weeks before her next scheduled injection. Nevertheless, the botulinum toxin is the treatment with the greatest impact on her symptoms. Lilly continues struggling with fatigue, but the general situation is much better for her now – thanks to the combination of injections, physiotherapy and psychotherapy. On the advice of her physiotherapist, she applies to her health insurance company for an armchair with headrest to put her head on, and she gets this approved. After spending a considerable amount of time and effort on the paperwork, she finally gets disabled worker status. In phases when the botulinum toxin is working well, she decides to try yoga and relaxation. This also helps her to maintain her posture.	**TREATMENT STRATEGY** Lilly has often taken sick leave during bad phases. Lilly’s treatment strategy is multidisciplinary: botulinum toxin injected by an experienced neurologist, physiotherapy, acupuncture, painkiller gel application on the pain zones, sport (swimming, walking, cycling), healthy diet, benzodiazepines if needed and however, there is no central care coodinator and Lily is often left to find allied healthcare professionals (e.g. physiotherapist) on her own.
EMOTIONAL EXPERIENCE		**PATIENT** Lilly is angry about not being able to control her head movements, especially when she is teaching in front of her students. She attributes the symptoms to her stressful everyday life and her job as head teacher, which she has only recently started. Gradually she realises that the symptoms are not just temporary and that something else must be behind them. Over time, Lilly, a very sociable person who enjoys meetings and eating out with friends and family, becomes introverted. She feels as though she is the only person to have the symptoms as she has never met anyone with the same health problem. Exhaustion, anxiety and uncertainty are combined over time with great frustration with her doctors and therapists who cannot find a cause for her symptoms. **FAMILY/ENVIRONMENT** Her partner is very supportive and understanding and takes over all the roles she feels she can’t do. Her friends and colleagues cannot understand what she is suffering from and wonder if Lilly has Parkinson’s disease. She feels as if nobody is able to help her.	**PATIENT** She is very relieved to finally have a diagnosis. At last, she knows that she is not crazy. At the same time, shortly after the diagnosis and the conversation with her neurologist, she can’t really grasp what CD means for her life, because she doesn’t know anything about it. This unsettles and scares her. Moreover, she is disappointed and sad that there is ‘no real cure’ and that the available options are drugs and neurotoxin, which only serve to treat the symptoms. She often dwells on what her life will be like with the disease, which makes her feel depressed. **FAMILY/ENVIRONMENT** Lilly’s family and friends are also relieved that the cause of her symptoms has been identified. They feel that Lilly doesn’t like to speak about it and accept her as she is. When her colleagues learn that she suffers from cervical dystonia, they empathise. But they don’t really understand.	**PATIENT** Lilly hopes that the treatment will help her to return to a normal life. She is suffering from a high level of psychological stress and severe limitations in everyday life. She avoids socialising because she can’t stand the glances of others. Sometimes she cries, feels angry, and has mood fluctuations, asking herself ‘why me?’ **FAMILY/ENVIRONMENT** Lilly is surrounded by her family, her friends, and her colleagues who show a lot of support and understanding. A good friend encourages her to contact a dystonia self-help group where she learns about the importance of psychotherapeutic support and receives the contact details of psychotherapists.	**PATIENT** After being frustrated at first, Lilly has learned to cope with the varying success of the treatment. If the botulinum toxin injections do not work well, she rests more and avoids going to work and socialising. When the botulinum toxin injections work well, she can return to an almost normal life. She has good phases and bad phases with anxiety and depression. Her challenges are to keep a balanced mind, not to be obsessed over negative thoughts, do not dwell too much on things and avoid thinking about dystonia all the time. **FAMILY/ENVIRONMENT** Her family, especially her children, give her support and strength not to lose her courage to face life. Her husband helps her a lot with the preparation of her application to get disabled worker status.	**PATIENT** Today, Lilly does not have the same job because she had to give up her position as head teacher. She has changed her attitude to life and is relieved that today, she can go back to work regularly with reduced working hours. She has learned to live with her illness and has begun to accept it. Step by step, she is rebuilding her life again and realises that she can still socialise, do her hobbies and help other people. She interacts a lot in patient networks and is involved in the patient association for dystonia in her area. **FAMILY/ENVIRONMENT** All in all, her family and friends have found a good way to deal with the disease. Sometimes they have conflicts because Lilly‘s husband forgets she is tired and everyday activities take her longer. Sometimes he forgets that she is not the same person as before.
STAKEHOLDER INVOLVED		GP, radiologist, physiotherapist, orthopaedist, neurologist, family & friends, line manager & work colleagues	GP, neurologist, physiotherapist, family & friends, line manager & work colleagues	Neurologist, GP, physiotherapist (specialised in dystonia), family & friends, line manager & work colleagues, patient association (PA), psychologist, health insurance	Neurologist, GP, physiotherapist (specialised in dystonia), psychologist, family & friends, line manager & work colleagues, patient association (PA), health insurance/medical service	Neurologist, GP, physiotherapist (specialised in dystonia), psychologist, family & friends, line manager & work colleagues, patient association

PATIENT INSIGHTS

INTERSECTORAL PATHWAY



* As the pathways prior to diagnosis reported in the PJ survey greatly differ from each other, several specialists and misdiagnoses and/or treatments are outlined. The order of visits to the specialists is presented arbitrarily and can be exchanged.

* Medical odyssey ends either because the GP doesn’t know what to do anymore and refers the patient to another neurologist, the GP suspects CD (rare) or the patient himself suspects CD and asks his GP for referral to a CD specialist (rare).

FACTS

PATIENT NEEDS

	GPs are considered the biggest hurdle to diagnosis—the main challenge is to raise awareness and understanding among GPs leading to a rapid diagnosis of cervical dystonia.	**Good communication between patient and physician is essential. HCPs need to provide their patients with more detailed information on the disease, on CD treatment options and sometimes need to show more empathy towards them.** **Patients need to be pro-active and ask about complementary therapies (physiotherapy, psychotherapy, occupational therapy). A holistic and multidisciplinary approach is needed from the medical profession.**	**In some countries, patients trying to gain access to and reimbursement of treatment face great administrative hurdles. Furthermore, HCPs often don’t give their patients any information at all about their treatment options or what to expect from treatment. Again, a multidisciplinary approach is needed and (more) communication between HCPs and patients.**	**Physicians often don’t ask their patients between injections how the treatment is working for them between injections. Patients are afraid to say the botulinum toxin injections are not working well because they are afraid the HCP will stop prescribing the medication.** **More well-trained injectors are required to ensure patients receive effective treatment. Patients also need consistent treatment conditions, i.e. to have the same injector for each session.** **There is a shortage of injectors which leads to a „conveyor belt“ treatment of patients, lacking an individualised treatment approach.**	**Social relationships and contact with other CD patients are considered a high priority along with mindfulness and acceptance. It depends on the individual, but generally speaking psychological support throughout all phases of the patient journey is important.**

NEEDS, LEVERAGE POINTS

LEVERAGE POINTS: HOW CAN WE MAKE THIS JOURNEY BETTER?

	1. **Raise Awareness and understanding among GPs.** 2. Raise Awareness and understanding among medical/ neurology students. 3. Guide patients to relevant online sources and enable them to understand their symptoms.	4. Empower patients to act and to communicate pro-actively with HCPs and their environment. 5. **Ensure that HCPs meet their patients‘ individual information needs about CD and possible treatments options** - not only what CD actually is, but how it will impact their life and what this means for them. 6. Motivate HCPs to refer their patients more often to Patient Associations (PAs).	7. **Support a multidisciplinary treatment approach throughout all phases of the patient journey.** 8. Work towards getting CD treatment recognised and paid for by health insurance in all European countries. 9. **Empower HCPs to providing patients with a treatment (options) perspective.** 10. Convince payers of the importance of sufficient physiotherapy sessions.	11. **Increase number of skilled injectors.** 12. **Establish the conditions for a personalised treatment approach with support from multidisciplinary teams.** 13. **Empower patients to communicate their expectations from and possible dissatisfaction with treatment.**	14. **Facilitate patients’ social contact with PA/other affected persons and motivate them to maintain social relationships.** 15. Motivate neurologists to recommend the services of PAs more often to their patients. 16. Help patients understand the importance of psychotherapeutic support.

Supplementary Appendix 2.

Expert Patient Focus Group Verbatim Quotes

Symptom onset and diagnosis
In France, General Practitioners (GPs) are not well informed of all the rare diseases.
I went to the GP, and she didn't know what it was and then I went home and I Googled things like 'head twisting' and 'muscles pulling' to see if I could get the information on the internet.
It's important for patients to be able to contact patient organizations and where to find good, reliable information online because there is a lot of misinformation around.
Once you get the neurologist of the center which is concerned with movement disorders you are finally in the right place [diagnosis and treatment]
In some parts of the UK, the neurologists are all very knowledgeable about dystonia and there are other areas of the UK where they are not so knowledgeable. What tends to happen in those areas is that the patient does some research to find who they should be referred to for the best treatment.
As soon as you are diagnosed in the UK you have access to treatment, but there is no such thing as a rapid diagnosis.

Initiation of treatment and the therapeutic relationship with HCPs
It is important that the physician gives adequate information at the first time of treatment and diagnosis, maybe not all the details, but the patients have a right and they need to know, because I didn't find out anything when I had my first botulinum toxin injection. My doctor only said 'I hope this will work for you'. I went home and I looked at the internet and there I saw that dystonia was a chronic disease and I started to cry in front of the computer.
Usually, [the neurologists] don't give enough details [at diagnosis]. They just give general information, but they don't spend time in explaining 'Now you are going to be injected, but success depends on injecting the correct muscles and dosing etc.'.
Maybe at the beginning the neurologists feel they don't want to overwhelm the patient with information, so maybe this is a classic miscommunication because the doctor thinks *'I don't want to throw all of this information about what might happen in the future… We need to tell them that we need more information'.*
There is a need for patients to have a helicopter view of how CD is treated generally and what are all the options that are available.
in the UK, we see a specialist for ten minutes once every three months, and in the early days that's just not enough.
In the UK, patients don't have immediate access to something like physiotherapy or any other complementary therapy. The patient has to look for it themselves.

Supplementary Appendix 3

Patient Survey

PERSONAL DETAILS

1. Initials:
2. Sex:
3. Age:
4. Country you live in: France/Italy/UK
5. Are you employed?

   If answer = Yes: full-time, self-employed, part-time, full-time student

   If answer = No : retired, not able to work/disabled, currently not employed
6. Did you have to change your job because of Cervical Dystonia?

   Option box: yes / no / other
7. What is the distance from your place of residence to medical centers (specialists, clinics, university centers)?

   Option box: < 5 km, 5-20 km, 20 -50 km, 50 – 100 km, > 100 km

BEFORE DIAGNOSIS

The following questions refer to your life before diagnosis. We therefore ask you to remember what your life was like before diagnosis, but when you already had symptoms of cervical dystonia.

8. Could you please confirm when you first noticed symptoms that could subsequently be attributed to Cervical Dystonia?

   Month: Year:
9. What symptoms did you have exactly?
10. How often did you experience these symptoms?

    Option box: very frequently, frequently, occasionally, rarely
11. Were any of your bodily functions impaired at the time (e.g. eating, drinking, walking, any physical activities, etc.)

    Option box: yes / no

    If yes, which ones?

    Do these impairments still exist today?

    Option box: yes / no
12. Please describe how the symptoms impacted on your life?

    Family: Partnership: Job: Interests and hobbies: Mental health:
13. Did the symptoms affect your quality of sleep?

    Option box: yes / no

    If yes, do you still suffer from sleep problems today?

    Option box: yes / no

14. How did you feel during this time when you had symptoms but no diagnosis?

DIAGNOSIS

15. When were you diagnosed with Cervical Dystonia?

    Month: Year: Your age at this time:

16. How did the diagnosis affect you? What went through your mind?
17. When your symptoms first started, what healthcare providers did you consult before finally being diagnosed (e.g., GP, neurologist, physiotherapist)?

    Specialty of the healthcare provider → Outcome of the consultation (in terms of diagnosis and treatment)

18. Can you remember after approximately how many visits* at which healthcare provider you were diagnosed with Cervical Dystonia?

    *Please count several visits to the same physician/ therapist e.g. 2 visits to a neurologist and 1 visit to a physiotherapist) = 3 visits.

    Specialty of healthcare provider(s) → number of visits:

19. Were there any misdiagnoses? If yes, which ones?
20. Did you also receive incorrect treatments due to these misdiagnoses? if yes, which ones?
21. What physician/healthcare provider actually made the final diagnosis?

    Option box: neurologist, physiotherapist, other: [free text field])

22. Did you know about Cervical Dystonia before being diagnosed?

    Option box: yes / no /

23. Do you feel your healthcare professional spent enough time discussing your diagnosis and addressing your concerns?

    Option box: yes / no / other

24. Were you satisfied with the information you received from your healthcare professional?

    Option box: yes / no /

    If not, please explain why you were dissatisfied.

25. How did you feel after the appointment? (For example, quite hopeful, lost, left alone, well supported?)
26. What would have helped to make you feel better?
27. Did the diagnosis of cervical Dystonia have an impact on your mental health?

    Option box: yes / no / I prefer not to answer

    If yes: When did you notice a change in your mental health?

    a) Are you still suffering from it today?

    Option box: yes / no / I prefer not to answer

    b) Have you been or are you receiving medical care for this?

    Option box: yes / no

    If yes: medication: yes/no

    Counselling: yes/no

28. Did you suffer from other conditions triggered by Cervical Dystonia?

Option box: yes / no / I prefer not to answer

If yes, which ones?

a) At what stage of your diagnosis did this occur?

Option box: within 1 year, within 1-5 years, > 5 years

b) Do these still exist today?

Option box: yes / no

c) Have you been or are you receiving medical care for this?

Option box: yes / no

TREATMENT

29. Who is/was your attending healthcare provider after the diagnosis? (e.g. general practitioner, neurologist)
30. Which treatment options were discussed with you?
31. What treatment did you receive/are you receiving? (Please list chronologically all therapies received)

    If several treatment options = yes: Did your doctor involve you in the decision for or against a certain treatment?

    Option box: Yes/ no/ other
32. Are you currently receiving a treatment?

    Option box: yes / no

    If yes: which one?
33. Does or did the treatment have an impact on your life/your everyday life? (Family, partnership, job, interests and hobbies...)?

    Option box: yes / no

    If yes: please describe what impact the therapy has or had.
34. Please think back to the time when you were diagnosed and possibly received a certain medication and/or treatment:

    a) How did you feel at that time?

    b) Was there anything that particularly helped you to cope better with Cervical Dystonia?

YOUR LIFE WITH CERVICAL DYSTONIA

35. How often do you go for a check-up?

    a) What is done during these check-ups?
36. What healthcare providers do you see for treatment of the symptoms today?
37. Have your symptoms changed over time?

    Option box: yes / no

    If yes: Please describe how they have changed?
38. What challenges does Cervical Dystonia pose in everyday life?

    Family: Partnership: Job: Interests and hobbies: Mental health:

39. When it was clear that you had Cervical Dystonia: Did you look for further information about it?

Option box: yes / no

If yes, where?

On the internet/ Self-help groups / Patient Organizations/ in patient brochures provided to me by: my physician / my attending clinic / during a stay in rehabilitation / a website

Other, namely:

40. Looking back, what do you wish had been different regarding your experience with Cervical Dystonia?

a) What would have helped you? (e.g. special contacts, special services)

41. Do you have any specific coping strategies to manage your Cervical Dystonia symptoms?

Option box: yes / no

If yes, which ones?

42. Is there anything specific that improves your Cervical Dystonia symptoms?

Option box: yes / no

If yes, please explain

43. Is there anything specific that worsens your Cervical Dystonia symptoms?

Option box: yes / no

If yes, please explain

44. How do you feel about your disease today?

45. What would you like to achieve for yourself? What timeframe?

In Italy, patients must look for physiotherapies themself because the neurologist doesn't give the information. You just feel like a neck to be injected and that's all except they give the next appointment. Some specialist centers do have the connections between the neurologist, the physiotherapist and the psychologist, but they are in just one city with an expert CD doctor.
I am afraid specialists don't know each other between specialties. Most of the time neurologists don't know the physiotherapists and which one is able to take care of dystonia. Neurologists often look to patient organization listings to know the physiotherapists around them able to take care of their patients, so what is the matter? They need to be connected.
In France, there are not many physiotherapists who know about dystonia.
The only time I see my GP in relation to my dystonia is if my consultant has recommended some drug therapy and he would write to my GP. I would then make an appointment to see my GP and they would prescribe the drugs.

Living with treated CD (impact of BoNT-A injections)
Personally, I couldn't move on with my life until I had accepted dystonia into my life, and it was a very important moment for me when that happened.
Injection clinics are planned to inject, 10-15 people per session, so they don't have time to talk. If you want to talk about your disease effects, the evolution, or whatever, you need to take an appointment with your neurologist outside these days and have time to discuss, but this is not well understood by most patients.
Many patients are not fully satisfied with their injections, but they are afraid to tell their consultant in case the consultant says, "I will just stop giving them to you, then." A lot of patients have that fear.
Another problem is there are not enough neurologists around, which limits patient access to the right doctors.

Appendix 4

Lay summary

Development of a patient journey map for people living with cervical dystonia

Monika Benson,[1,2] Alberto Albanese,[2,3] Kailash Bhatia,[4] Pascale Cavillon,[5] Lorraine Cuffe,[6] Kathrin König,[7] Carola Reinhard,[2,8] Holm Graessner[2,8]

Plain language summary

It is known that treatment always work best when the patients actively participate in decisions relating to their own healthcare. Patient journey maps are one tool that healthcare services can use to visualize the long-term care of their patients. To help the professionals understand all the small steps a person with a rare disease goes through, from their first symptoms through to diagnosis and eventual treatment. A collaborative group of experts, including consultants and people living with cervical dystonia (often shortened to CD), set out to develop a map that visually explained the whole disease journey ***from the patients point of view***.

The group found that people living with CD typically go through five key stages. Using detailed feedback from patients they developed a map which described the typical patient journey.

1. Symptom onset
2. Diagnosis and therapeutic relationship with healthcare professional (HCPs)
3. Initiation of care for CD
4. Start of CD treatment
5. Living with treated CD

The process of building this visual map identified several barriers to good treatment in CD. For example, misdiagnoses, lack of care coordination and ineffective communication between patients and their doctors. By developing the first patient journey map for CD, the authors hope that doctors can use the map as a tool to improve the care they offer. Patients can also use the tool as a visual aid to help them describe their experience, including any concerns with long-term treatment plans, to the healthcare professionals using a common language.