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A unique collaborative model providing supportive and self-advocacy tools to the rare disease community

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ABSTRACT

The National Ehlers-Danlos Syndromes (EDS) service is a highly specialised NHS commissioned service for diagnosing and supporting people with a rare, monogenic type of EDS. The service consists of a team of medical specialists and genetic counsellors. The EDS service has strong ties with national and international EDS charities, and together they participate in working and strategy groups alongside patients and carers. Acquired funding for research means that the service contributes to international knowledge regarding diagnosis and management of rare, monogenic EDS types. As a result of being a highly specialised service, there are multiple points of contact with patients, providing frequent opportunities for feedback. Patient data, surveys and collaborative working have enabled the development not only of extensive information resources and self-advocacy tools for patients, but also guidelines for health care professionals on the best ways to support their rare disease patients. The main challenges faced by patients affected by rare disease and the importance of self-advocacy are discussed, with the highly specialised National EDS service and the patients seen in this service as an example. The recommendations from this unique collaborative model include eight specific domains of self-advocacy: increasing knowledge of rare disease, taking care of mental well-being, taking care of physical well-being including routine care, the development of good working relationships between patients and health care professionals, information accessibility, emergency preparedness, taking part in education and outreach, and involvement in research and feedback opportunities. The service aims to be an example of patient-centred innovation and progress, providing a patient-focused supportive model that can be adapted by the rare disease community and other health care professionals.

1. Introduction

A disease is considered rare, as defined by the European Union, when it affects less than 5 per 10,000 people [1]. Having a rare disease often means increased time to diagnosis, decreased access to specialist care, difficulty navigating the healthcare system and accessing resources and information, increased requirement for physical and psychological support, less evidence-based treatments and fewer health care professionals understanding the condition [2–6]. As a result, a person with a rare disease often needs to self-advocate (represent one's interests when managing their own health concerns), and use their own resources in seeking a diagnosis, getting support, and explaining their condition to health care professionals (HCPs). This also raises concern for equity across different communities [7].

There is a paucity of information about how HCPs are currently supporting people with rare diseases. However, Kerr *et al.* explore the role HCPs can have in mentally and physically supporting patients with rare disease through good communication, education and team work [8]. While Spencer-Tansley *et al.* focus on the importance of HCPs empowering their patients who have a rare disease and the need to integrate mental health support into their care [9]. This may not occur in practice however without creating resources to integrate rare disease education into training across a range of HCPs, from medical students to specialists [4,10,11].

There is also relatively little information in the published literature about the different methods that people with rare disease can use to self-advocate. However, Pignolo *et al.*, discuss the importance of a patient accessing management information and specialist services [12], and

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Kerr et al. focus on the patient perspective of self-care; the increased need for mental and emotional well-being and seeking social support [8].

With the recognition of the complex needs of people with rare disease, the National Health Service (NHS) in England funds around 80 highly specialised services providing specific clinical care and management for people with a range of rare and complex conditions [13]. These services usually involve multi-disciplinary teams of HCPs with specialist knowledge and experience and access to innovative approaches to medical care; usually seeing no more than 500 patients a year. A number of these services are for patients with rare genetic disorders. These services can be compared with expert centres that have been established in Europe and form part of the European Reference Networks [14].

The highly specialised National Ehlers-Danlos Syndromes service (EDS service) was established in 2009 for people with or suspected of having a rare, monogenic type of Ehlers-Danlos syndrome (EDS) with two services covering the North (location Sheffield) and South of England (location London). Each service is commissioned to triage approximately 200 referrals a year of adults and children with a suspicion or diagnosis of a rare type of EDS [15]. Rare, monogenic types of EDS are a group of 13 inherited connective tissue disorders that have overlapping features of joint hypermobility, skin and vascular fragility, and generalized connective tissue friability with prevalence ranging from 1/20,000 to < 1/1,000,000 [15–18]. Table 1 summarises EDS service provisions and support for patients with rare, monogenic types of EDS pre-and post diagnosis.

The EDS service collaborates closely with charities that support people with rare types of EDS. For example, the charity Annabelle's Challenge [19], is dedicated to supporting people with vascular EDS (OMIM #130050) [20], the frequency of which is estimated as 1/50,000–1/200,000 [21]. The condition is characterised by fragile blood vessels and tissues which can lead to major complications such as dissections and hollow organ rupture.

To expand and progress clinical care for people with vascular EDS, a 'self-advocacy survey' was developed by the EDS service in collaboration with Annabelle's Challenge to identify the ways in which people advocate for themselves and get their needs met both independently and together with their HCPs, including barriers and challenges to this process. The methods and survey are included in the supplementary files. Sixty responses were thematically analysed. As a result, eight specific domains of self-advocacy were identified (summarised in Table 2). The results have been presented by patients in collaboration with HCPs in the EDS service at national and international EDS conferences (Annabelle's Challenge [19], EDS society [22] and published by Harris *et al.,* [23]). The domains are summarised below from both the patient and HCP perspective.

Table 1
Summary of provisions and support provided to patients pre-and post diagnosis in the National Ehlers-Danlos Syndromes service.

EDS SERVICE PRE-DIAGNOSIS PATHWAY SUMMARY	
Referral into service	Triage, information gathering
Assessment	Family/ medical history, clinical assessment, examination,
Diagnosis/Testing	Diagnostic genetic testing and predictive testing for at risk family members
EDS SERVICE POST-DIAGNOSIS PATHWAY SUMMARY	
Management	Advice, guidelines, recommendations
Mental health support	Resources, short-term counselling, onward referrals
Resources	Leaflets, emergency information, information for family members, relevant charity information, information for education/employers/ disability support
Referrals	To specialists (where needed) such as cardiologists and rheumatologists
HCP liaising	Advice regarding complex cases, surgery, in-hospital stays, through multidisciplinary team meetings (MDT) where applicable
Research	EDS service research opportunities and collaborations
Opportunities for patient feedback	Biennial patient satisfaction survey, patient public involvement for leaflet and study development, surveys and workshops, charity-led strategy / working groups for HCPs/patients/carers

Table 2
Summary of domains of self-advocacy for rare disease from a patient and HCP perspective.

SUMMARY OF DOMAINS OF SELF-ADVOCACY FOR RARE DISEASE		
	Advice for patients/ carers	Advice for health care professionals
Knowledge of rare disease /knowledge-sharing	-Learn about disease (trusted sources/charities) -Share with family, friends, employers, educational providers	-Learn about disease (trusted sources) -Seek guidance from experts -Create multidisciplinary teams/meetings -Provision of supportive documents
Mental well-being	-Self-awareness of mental health needs -Request access to counselling if needed -Seek social support and connections to others (e.g. through charities)	-Discuss and assess mental health -Mental health provision or referral
Routine medical care / physical well-being	-Compliance in routine medical care -Follow life-style recommendations -Seek advice if care is not being met	-Support self-management, make referrals -Provide documentation of medical needs in clinic letters -Enclose letters to other HCPs -Liaise with other HCPs
The development of good working relationships between patients and HCPs	- Request dedicated appointment to discuss condition with HCPs and share knowledge -Share clinic letters from other HCPs to help join up care	-Provide dedicated appointments -Learn about rare disease of patient from patient and trusted sources -Ensure equity in healthcare and support
Information accessibility	-Have easy access to letters (paper/ Apps) -Where possible use technology (information, social connections)	- Create accessible information for all - Use patient public involvement to ensure accessibility of information resources - Offer virtual appointment where suitable
Emergency preparedness	-Have emergency procedures and clinical information to hand always -Find someone who can advocate for you -Know about medical alert bracelet and other emergency resources -Get to know your local teams	-Inform patients about provisions such as medical alert bracelet -Aid placing of flags on medical records -Help create a care plan in routine and emergency setting
Education / outreach	-Involvement in educating others at conferences, webinars, publications, volunteering expertise to charities	
Research / feedback	Opportunities for involvement in research, patient public involvement, patient feedback	

2. Main domains of self-advocacy

2.1. Knowledge of rare disease and knowledge-sharing

People with rare and complex disease can find learning about the disease difficult especially if there are life-threatening or serious complications. However, self-knowledge enables self-awareness, the knowledge of personal limitations, a recognition of when to seek medical help and advice in both routine and urgent care, and when and how to self-advocate. "Knowledge is power" our survey respondents told us.

Knowledge-seeking can take many forms including learning from specialist providers and trusted educational websites or relevant charities. Charities often coordinate support groups and access to a patient community. These sources of knowledge can provide general information, practical advice and resources, recommended specialists and updates of new developments and research. Self-knowledge and sharing enables conversations with friends and families who in turn can provide support and advocate where needed. Self-knowledge is also essential for discussions with HCPs who may be unfamiliar with the rare condition and helps in self-advocating for extra support or needs for example with employers, in education, or access to disability benefits and allowances. Where applicable HCPs should provide documentation to support these efforts.

It is equally important for HCPs (especially primary care physicians such as general practitioners, family physicians or internists) to take the time to learn about rare diseases should a patient be diagnosed with one; exploring resources, guidelines if available, and bona fide online links for example Genetics and Rare Diseases information Center (GARD) [24], the portal for rare disease and orphan drugs (Orphanet) [25], and GeneReviews [26]. Health care professionals should recognise their own knowledge and limitations, seek to liaise with each other, seek guidance if uncertain about management, identify other HCPs with required expertise to advise, guide and collaborate with, and create multidisciplinary teams to meet their patients complex medical needs where needed.

It is important for HCPs to recognise that their patients (and / or their carers) are experts by experience and to show real commitment to learning from them, listening to their perspective and enabling shared decision-making [8,27].

We recognise however that these interventions require an HCP to have time and resources and may depend on adequate funding of services and management support.

2.2. Mental well-being

The impact of having a rare disease on mental health is well recognised [9]. There is another set of challenges for people with an 'invisible' rare disease. For example, people with vascular EDS who are at risk for sudden life-threatening events due to generalised tissue fragility, describe the anxiety caused by 'living under a threatening cloud', having to explain the condition to HCPs, potentially being misunderstood and not knowing if their health needs are being met in both routine and emergency settings.

For this reason, it is important that HCPs assess their patient's mental health, both current and historical, discuss the impact of having a rare disease on mental health for the patient and the wider family, signpost to relevant charities and refer for counselling where needed. For example, people with vascular EDS in the United Kingdom can request counselling through the charity Rareminds [28]. It is also important to recognise the different ways in which mental health can be impacted depending on age (differing in childhood, adolescence and adulthood).

Survey respondents discussed the importance of a person being self-aware as to their mental health needs and seeking extra support and counselling where needed. They recommended focusing on the "things you can control as opposed to those that you can't" and "not allowing a diagnosis to define you." They suggested seeking social support through connecting with other people through disease-specific support groups, trusted online forums and charities. This connectivity can help not only in support and advice but in 'normalising' the condition and reducing isolation; "hearing other people's stories gives you hope and strength to know that you are not alone."

2.3. Routine medical care and physical well-being

It is important for people living with a rare disease to access routine medical care such as attending routine appointments and investigations, taking prescribed medications and following suitable or adapted diet and exercise recommendations to reduce aggravating factors [3]. For example, people with inherited connective tissue conditions and joint hypermobility may benefit from appropriate and bespoke exercise to gently strengthen their muscles, providing support to tendons and ligaments.

Where routine medical care is not being met, advocating for oneself by addressing it directly with the treating doctor, or discussing it with a patient liaison or support service (such as the patient advice and liaison service (PALS in the NHS) is important so that learning can occur. A recent ruling now gives patients and their families in England the legal right to a second opinion from senior medics in the same hospital if they have a serious concern and feel their concerns are being dismissed (Martha's rule) [29].

It is also recommended that HCPs support their patients with rare diseases in self-management, provide resources, make referrals where needed, and enclose letters to other relevant specialists involved in their patient's care to aid learning and shared practice. Clinic letters for people with rare diseases should clearly delineate the disease and its associated risks and management requirements as these letters may be shared with other HCPs in emergency or routine care.

Many people with rare disease may experience non-specific symptoms such as fatigue and pain that also require management support and guidance as these can severely impact general well-being and health.

2.4. The development of good working relationships between patients and HCPs

The way that knowledge-sharing can aid good working relationships between patients and HCPs is discussed in 2.1. In addition, it is important that medical information is explained to patients in an understandable and accessible way, with a meaningful discourse that creates opportunities for clarification, questions, the addressing of any concerns (both physical and mental) and shared decision-making where applicable.

This collaborative effort can create a trusting patient-HCP relationship that is known to improve patient outcomes and well-being especially in the realm of well-informed self-management [30,31].

2.5. Information accessibility

Having clinical information readily available to share is critical for a person with a rare disease to advocate for themselves. It is recommended that relevant clinical letters and patient leaflets detailing the diagnosis are readily accessible both in a paper and online format. Documents can easily be stored in phone Apps that allow health information to be safely stored. This can aid knowledge-sharing with HCPs and decision making both in routine and emergency care.

With increasing access to technology, it is important that people with rare disease are aware of opportunities to connect, learn and knowledge-share through trusted mobile applications, internet, and social media platforms. Health care professionals, charities and the rare disease community should also ensure that there are equal opportunities for people who do not use digital technology [3]. Information should be understandable and accessible for all people and patients should be involved in the writing of resources to ensure accessibility and relevance

[32]. Health care professionals should be aware of the extra difficulties faced by those patients who may find it harder to access or understand medical information, or advocate for themselves, and provide extra relevant support and knowledge sharing where applicable to ensure equity of care for all.

Whereas attending multiple appointments for complex health needs can be difficult and hard to maintain, it is often helpful to have options that increase accessibility and ease, such as virtual appointments. This is particularly important for people with rare diseases where knowledgeable HCPs may be few and far between [3].

2.6. Emergency preparedness

Some rare diseases may require emergency preparedness paperwork and resources detailing emergency procedures and recommendations specific to their condition.

Having clinical information always accessible (via paper or phone Apps for example) is essential for those with a potentially life-threatening condition or for those that need specialty emergency care. It is also essential that people with a rare disease who may present differently or subtly in an emergency advocate for themselves and make their diagnosis known to HCPs during both triage and treatment. Educating a partner, friend or relative to advocate in an emergency is also recommended. HCPs can also help in supporting their patients with rare serious diseases by recommending or aiding in the placing (where the system allows) of emergency medical information in [electronic] medical records. Health care professionals in England and Wales can also help people with a life-threatening condition in the placing of a 'marker' on their address (in their patient record) so that if an ambulance is called out, the attending crew are aware of the diagnosis and relevant emergency medical information. Many patients with a life-threatening disease wear a medical alert bracelet to alert people as to their diagnosis in an emergency situation or carry an emergency wallet card detailing their condition.

It is also helpful in such cases for patients and HCPs to work together to develop an emergency health care plan for different medical settings such as inpatient and emergency situations. Knowledge and introductions to relevant local teams can help in the creation of these plans.

2.7. Education and outreach

A person with a rare disease is better able to self-advocate if more HCPs know about rare diseases and their challenges, and where to access reliable disease-specific information. Both HCPs and people with rare diseases can play a role in education and outreach through attending and contributing to conferences, on-line forums, webinars, trusted social media sites, publishing and writing, volunteering on steering groups for rare disease charities and taking a proactive role in disseminating knowledge about rare disease. It is increasingly common for medical journals and charities to encourage HCPs, patients and carers to publish together (through journals, blogs and podcasts [11] for example) to illustrate complex needs and knowledge-sharing (as is the case with this journal) [33].

2.8. Research and feedback

It is important for rare disease specialists to be able to initiate or collaborate in clinical and translational research to answer key issues that can improve management and therapy. However, time for this is often not built into their clinical role which is often aimed at individual patient care and therefore separate funding needs to be acquired which can be a challenge when it concerns rare disease. Therefore, it would be helpful if funding for highly specialised services or equivalent services outside the UK could take the importance of clinical and translational research for patient benefit into account.

Research study protocols that include patient and public involvement (PPI) from the outset allow for maximal learning about the disease and what patients' needs are; allowing patients to advocate for the things that matter to them the most, ensuring their needs are recognised and form an essential part of the research.

For example, the London EDS service, recognising gaps in knowledge within rare types of EDS, has launched 'The Natural History Exploration of rare EDS types (NEEDS)' study, collecting data both prospectively and retrospectively and using PPI in the development of the research protocol.

A further example of patient-centred research is the VEDS Collaborative, consisting of a group of patients, family members, researchers, and clinicians 'dedicated to developing a patient-centred plan for scientific research to improve the management of Vascular EDS and increase the quality of life for all those impacted by it [34]. Using this approach they have already identified important topics for research such as addressing mental health and quality of life issues, and creating a care team [35].

All research in rare disease is of course reliant on people volunteering to take part. Research in the vascular EDS community showed that most people say that they would be willing to share their medical records for research studies to help find a treatment or cure and to help others learn from their experience [36].

We would recommend that people with rare diseases are aware of the research opportunities that are available to them in both clinical and research trials. Taking part in research can be both rewarding and may even improve outcomes in the long run. Anyone taking part in research should be well-informed and comfortable with the research protocol and should independently arrive at the decision whether to take part or not.

There are multiple ways for HCPs and charities to collect feedback and recommendations from people with rare diseases as these can provide invaluable insights and data into the patient experience and what their needs are. This can also provide opportunities for learning and strategic directions for HCPs and charities alike but are dependent on people with rare disease volunteering their time and expertise.

3. Conclusions

In summary, the main domains of self-advocacy for both patients and HCPs borne out of the work of the EDS service in collaboration with patients and charities is being widely disseminated to people with rare types of EDS and has the potential to have a significant positive impact on both an individual level and in medical practice as well.

Whereas we recognise that a highly specialised service may have the staff and resources to develop, implement and disseminate this patient and HCP model for self-advocacy, it is hoped that through collaborations with charities and patients, other rare disease clinical and support networks can explore and evolve these practices too. And whereas each rare disease has its own unique features, much of this guidance may be widely applicable as it addresses many of the challenges that people with rare disease face. With this unique evidence-based collaborative model, people with rare diseases have the potential to have a faster time to diagnosis, have greater access to specialist care, resources and support in routine and emergency care, and better options for management and treatment. This in turn will have a lasting and positive impact on their physical and mental health, their support systems and society as a whole.

Ethics statement

Ethics statement is not required as there is no experiment done on human or animal subjects and it is an opinion paper with recommendations'

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Data availability

The authors do not have permission to share data.

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Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at doi:10.1016/j.rare.2024.100026.

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