Patient Perspectives

The process of medicines optimisation places patients at the heart of the process. It seems only right, then, to seek the views of patients about their experiences with medicines, their medical condition in general and their contacts with health professionals. Understanding what it is really like for a patient to live with a particular clinical condition will hopefully assist healthcare professionals to become more effective with their interactions and communications with patients and improve the healthcare services provided.

This has been done by providing patients identified through healthcare contacts with a template of questions to be completed anonymously by the patient on the basis that no individual be named or identifiable from the content. What some people have to cope with and the way they do it will amaze you.

Hypermobile Ehlers-Danlos Syndrome (hEDS)

Abstract

Title
Patient Perspective: Hypermobile Ehlers-Danlos Syndrome (hEDS)

Summary
A patient's perspective of living with Hypermobile Ehlers-Danlos Syndrome (hEDS) is described. The way contacts with healthcare professionals might have been better are outlined. The medicines taken, elements of service provision found to be most helpful and steps to improve the ongoing management of the condition are outlined. Key messages for healthcare professionals that have arisen from the patient experience are indicated.

Keywords: medical condition, medicines

About your medical condition

What is the medical condition most important to you that is being presented here?

Hypermobile Ehlers-Danlos Syndrome (hEDS).

Can you please explain the problems you experience with this medical condition?

Chronic pain and fatigue, functional gut disorder, joint dislocations and subluxations, easy bruising, poor wound healing, poor proprioception.

Can you please say how the medical condition was first diagnosed?

Diagnosed in clinic by a specialist rheumatologist, following presenting with symptoms since childhood.

Can you please say when the medical condition was first diagnosed?

February 2015.

If you look back, what would you say have been the main things you would have liked to have been different in terms of contact with health professionals?

- Earlier detection at primary care level.
- Shorter waiting times to see specialist clinicians.
- More specialist clinicians and clinics to make an accurate diagnosis.
- Being listened to and valued as a patient, presenting with numerous physical symptoms, particularly at primary care level and with community pharmacists.

About your medicines

Please list the medicines you taking for your medical condition.

- Lansoprazole, mebeverine for functional gut problems: daily
- Opiate painkillers for musculoskeletal pain: PRN

Have you had any particularly bad experiences with regard to your medicines? If so, please explain and indicate how this could be avoided in future.

I suffer from Serotonin Syndrome with SSRIs, TCAs and tramadol. I have trialled these drugs to help treat pain, depression and anxiety, but cannot tolerate them. It is therefore important that I explain to clinicians why these drugs should be avoided and other options explored.

I developed a physical dependence on opiate painkillers at one point, which led to unpleasant withdrawal symptoms. I felt that primary care healthcare professionals and community pharmacists didn't support me well during withdrawal. I now recognise the need, when I need to take opiate painkillers, to be more aware about becoming physically dependent again, and discuss with the prescriber.

It is my opinion that primary healthcare professionals and community pharmacists should provide better support and care for patients who are experiencing opiate withdrawal symptoms.

Many patients may be over-prescribed and/or be given inappropriate drugs, leading to problems with interactions and adverse effects instead of more effective ways to control and manage symptoms (such as pain/fatigue management).

Have you had any particularly good experiences with regard to your medicines? If so, please explain.

Functional gut issues can be miserable. Taking lansoprazole and mebeverine daily allows some considerable relief. These drugs are well tolerated.

Short-term (no longer than 4 weeks) use of opiate painkillers has helped, though I recognise the importance to review prescriptions for these drugs more actively.

About the services you received

What have you found to be most helpful to you in terms of the services you have received?

- Pain Management course (facilitated by psychologists, physiotherapists and a pain management nurse).
- EDS education seminar (facilitated by a senior, experienced physiotherapist).
- Prompt GI physiological investigations leading to effective treatment.
- Peer support meetings, provided by Ehlers Danlos Support UK, a national charity: https://www.ehlers-danlos.org/ .

To what extent have the health professionals you have come in contact with appreciated what it was like from your position as a patient?

At primary care level, I feel like my position, as someone with my condition, is largely unappreciated. On many occasions before diagnosis, and sometimes after diagnosis, I have felt worse after speaking to a primary care health professional or community pharmacist regarding symptoms and worrying pharmacological adverse effects. I have frequently felt 'fobbed off'.

Due to poor understanding and appreciation of hypermobility, local MSK physiotherapists have issued the incorrect exercises to strengthen and stabilise joints leading to increased pain and deconditioning, and secondary low mood/anxiety.

Specialist clinicians, and those aware of the condition, have been understanding and much more willing to listen and provide appropriate care.

To what extent was the information you were given about your medical condition sufficient for you?

The particular hospital where a diagnosis was reached provides an education seminar for new patients. This was useful as it gives an opportunity to ask more questions. In my case, I was able to make an informed choice about planning my treatment. Other patients are frequently given a diagnosis, without any information for themselves and local healthcare services, leading to problems in favourable outcomes.

I was also able to turn to Ehlers-Danlos Support UK for more information about my condition once given a diagnosis.

To what extent did the health professionals you came in contact with communicate effectively with you?

Health professionals who are aware of the condition have communicated effectively and compassionately.

What have been the best experiences you have had with the services you have received?

- Peer support, which helps to alleviate isolation.
- Specialist healthcare professionals who are aware of hEDS have provided excellent understanding and care which is reassuring.
- Learning how to accept the diagnosis, and use techniques such as 'pacing' to help manage chronic pain and fatigue, provided by a pain management clinic.

About other medical conditions

Do you have any other medical conditions that make life problematic for you? If so, please list them and explain the main problems you experience with each one.

Medical condition	*Main problem experienced*
Migraine associated vertigo	Persistent vertigo
Vestibular dysfunction	Persistent vertigo
Anxiety	Powerful physical symptoms of anxiety
Depression	Low mood leading to social isolation

About going forward

What would you like to happen at this stage that would make living with your condition easier for you?

- More understanding and awareness at primary care level, including local MSK physiotherapy services and community pharmacists.
- Awareness of local support services such as peer support meetings provided by Ehlers Danlos Support UK, by way of recommendation from healthcare professionals, especially primary care healthcare professionals, local MSK physiotherapy services and community pharmacists.
- Prompt access to support during flare-ups, which can appear from nowhere and last for months (some patients can debilitate to such a level whereby they need wheelchairs and/or walking aids to get around. This could be prevented by early diagnosis and prompt treatment to manage flare ups, preventing muscle deconditioning).
- Waiting lists can be in the region of 18 months to see an EDS specialist. This could be avoided if local services had more knowledge and could manage the symptoms locally.

If you could give a brief message to healthcare professionals, what would it be?

Good communication between patient and healthcare professionals is paramount. Patients frequently present with multi-systemic problems, and some patients may be sensitive to typical pharmacological treatments. Doctors must connect multi-systemic problems together - treating EDS symptoms individually is ineffective. Symptoms are connective in nature and should be treated appropriately.

Make yourselves aware about EDS and reach out to specialist clinicians and organisations for more information.

Please do not assume our symptoms are of a psychiatric nature – hEDS is a physical condition. Secondary psychiatric conditions may be present such as anxiety and depression.

Please add any other comments or observations that would be helpful to health professionals who are responsible for providing services for you.

Ehlers-Danlos Syndrome – Hypermobility Type (hEDS) is the most common type of Ehlers-Danlos Syndromes (EDS). There are 13 types of EDS, which is a hereditary connective tissue disorder (connective tissue is abundant in the human body). EDS affects women, men and children at any age. Men with EDS are in the minority, possibly because men are less likely to seek medical attention amongst other reasons. Diagnosing children can be particularly challenging, since children have naturally hypermobile joints. Some types of EDS are very rare, however hEDS is considered to be reasonably common.

EDS is poorly recognised in the medical community. I would, for example, often hear from doctors that "You don't have EDS because you don't have stretchy skin"but stretchy skin is only associated in two types of EDS out of the 13.

Patients may present with very mild symptoms, though some

may be severely affected, suggesting there is a wide spectrum of severity within each type of EDS. Some patients report reaching a diagnosis has taken decades, and during that time, they have been told by healthcare professionals their symptoms are "all in their head". Patients are often misdiagnosed, and given incorrect labels such as Fibromyalgia.

Some patients may also present with co-morbid associated conditions such as Postural Orthostatic Tachycardia Syndrome (PoTS), Mast Cell Activation Syndrome (MCAS), Chiari Malformation and Cervicocranial Instability. These conditions are also poorly recognised in the medical community at large. They should be taken seriously, and appropriately investigated.

At present, hEDS is the only type, which is not diagnosed with genetic testing. Diagnosis is reached clinically, on presenting symptoms, past medical history and family medical history. Other types may be diagnosed based upon genetic testing and clinical features.

There are no specific pharmacological treatments licensed for EDS as far as I am presently aware, nor are there any guidelines provided by NICE.

With appropriate management, many patients may eventually lead fulfilling lives. It may take some time to stabilise and strengthen joints through activity and physiotherapy. Maintaining a good diet is usually recommended.

I would recommend that healthcare professionals who are interested in learning more about EDS contact the Ehlers-Danlos Support UK helpline advisor: helpline@ehlers-danlos.org .

In 2017, the Medical Journal of Medical Genetics published a range of papers with the new nosology of EDS, following a symposium made up of international medical experts. The papers can be seen here: http://onlinelibrary.wiley.com/doi/10.1002/ajmg.c.v175.1/issuetoc .

What are the three most important things that health professionals should learn from your experiences?

1) The patient is giving you the diagnosis – please listen to them. Do not lead the patient to believe it is psychiatric/somatising symptoms.

2) Early diagnosis is important – some patients suffer for decades before reaching a diagnosis.

3) It may take a long time to manage symptoms, and symptoms may flare up from time to time – please be patient with us, and help us access appropriate care promptly.

Declaration of interests

You will have been offered a fee for your contribution to be submitted within a specific timescale. In the spirit of being open and transparent, would you please disclose any other payments, interests or activities that could be perceived as influencing what you have written or state 'none'.

I am the Ehlers Danlos Support UK men's coordinator and an expert patient.

KEY LEARNING POINTS FOR HEALTHCARE PROFESSIONALS IDENTIFIED AT THE EDITING/PEER REVIEW STAGES

- Be alert to the possibility of a patient developing opiate dependency and ensure appropriate support is available during withdrawal.
- Without a diagnosis, it can be difficult for healthcare professionals to provide appropriate support and patients can feel that they that their difficulties are not being appropriately considered.
- Specialist support can be invaluable but the route to obtaining that that needs to be improved.
- If you have a patient diagnosed with a relatively rare condition, then it is important to learn about that condition if you are to support the patient in the best possible way.