

Translating Research into Change

Reporting the Lived Experience of hEDS and HSD in Scotland



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Foreword

This is an incredibly important report focussed on Scottish data which comes from a comprehensive study providing vital evidence of the lived experience of thousands of people living with hEDS/HSD across the UK.

The case made for the Scottish government to consider cannot be ignored given the facts contained within this. The references to other studies further proves the reality for so many living with a debilitating condition that is too often dismissed and misunderstood.

This report is clear in the recommendations it makes and illustrates the key challenges we need to overcome together. The voices of so many people living with hEDS/HSD are raised quite rightly within it and for once will be heard.

This report is the result of a gigantic effort made by researchers and volunteers collaborating with such passion and dedication to improving health outcomes for all. The EDS UK volunteers who helped to support its production and spent countless hours championing the work should be rightly proud of the results.



An enormous thank you is due to the tremendous dedication of Kathryn and Dervil in leading this project and delivering such a seminal piece of work. We're looking forward to using this as a basis to campaign for change and to turn these woeful statistics upside down.

At EDS UK our vision is that across the UK, people with EDS or HSD will be connected, heard, supported and have equitable access to care. We bring people together, confront misunderstandings and challenge inequalities. We are proud to have been part of this project and will continue to raise the community's voice to the policymakers who need to hear it and take action.

Susan Booth

Susan Booth
CEO, Ehlers-Danlos Support UK

Executive Summary

This report offers a comprehensive examination of the lived experiences of individuals with hypermobile Ehlers-Danlos Syndrome (hEDS) and Hypermobile Spectrum Disorders (HSD) in Scotland. The findings presented are primarily drawn from the “hEDS-START” project, an initiative aimed at highlighting the challenges faced by individuals living with hEDS/HSD in the United Kingdom.

Through a combination of surveys and patient engagement events, this project has gathered invaluable insights into the experiences of patients navigating the healthcare landscape in Scotland and focuses on the urgent need for a pathway of care within the NHS. Currently, there is a glaring absence of such a pathway, resulting in prolonged diagnostic journeys and inadequate access to specialist care for patients. Key findings underscore the pressing need for action:

- **Prolonged Time to Diagnosis:** Individuals with hEDS/HSD in Scotland endure an average wait of 20 years from symptom presentation to diagnosis, significantly impacting their physical and mental well-being.
- **Access Barriers:** A substantial proportion of patients resort to private consultations or travel to England for diagnosis, highlighting the inadequacies of the current healthcare infrastructure in Scotland.
- **Fragmented Care:** The absence of a coordinated approach to hEDS/HSD care leads to disjointed treatment, exacerbating patients' already challenging experiences.
- **Mental Health Implications:** The impact of hEDS/HSD on mental health is profound, with high rates of anxiety, depression, and psychological distress reported among patients. The lack of understanding and support from healthcare professionals further compounds these issues.

To address these pressing challenges, several recommendations are proposed:

- **Establishment of a Pathway of Care:** The Scottish Government is urged to create a dedicated pathway of care for individuals with hEDS/HSD, ensuring timely diagnosis and access to specialist services tailored to patients' needs.
- **Enhanced Education and Training:** Healthcare professionals should receive comprehensive education and training on hEDS/HSD to facilitate early diagnosis, effective management, and empathetic care delivery.
- **Integration of Neurodiversity Considerations:** Given the intersectionality between hEDS/HSD and neurodivergent conditions, healthcare providers should adopt a tailored approach to care that acknowledges and accommodates diverse patient needs.
- **Empowerment through Education:** Patients should be equipped with high-quality self-management tools and resources early in their diagnostic journey to foster agency and independence in managing their conditions.

By implementing these recommendations and promoting collaboration between policymakers, healthcare professionals, and patient advocacy groups, Scotland can take significant strides towards improving the quality of life for individuals living with hEDS/HSD.

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Introduction

The purpose of this report is to provide up-to-date information on the current lived experiences of people with hypermobile Ehlers-Danlos Syndrome (hEDS) and Hypermobile Spectrum Disorders (HSD).

At present, there is no pathway of care for adults living with hEDS and HSD in the United Kingdom. People living with these conditions report feeling dismissed by healthcare professionals, and time to diagnosis currently averages two decades. Many patients in Scotland are forced to travel to England for a diagnosis, and almost one in five find themselves having to pay out of pocket to see private consultants.

The majority of the data presented in the following report has been collected as part of the “hEDS-START” project, which aims to detail the lived experiences of people living with hEDS/HSD in Scotland. This project involved a nationwide survey of EDS UK members living with hEDS/HSD, as well as a patient engagement event, held at the Institute of Genetics and Cancer in February 2024. Unless stated otherwise, the patient voices throughout have been shared with us as part of the 2023 hEDS-START survey.

While we report here the survey data from the Scottish respondents, and the experiences discussed by our Scottish patient engagement group, it is important to bear in mind that these experiences are relevant to all of the devolved nations. Data covering the whole of the United Kingdom will be published in due course.

We call upon the Scottish Government to create an adequate pathway of care for people living with hEDS/HSD, to provide regional services which take into account the many symptoms and co-occurring conditions, and to help provide adequate training and education of healthcare professionals.

Ro-ràdh

Is e adhbhar na h-aithisge seo fiosrachadh as ùire a thoirt seachad mu na h-eòlasan beò a th' aig daoine le Syndrome Ehlers-Danlos hypermobile (hEDS) agus Eas-òrdughan Spectrum Hypermobile (HSD).

Aig an àm seo, chan eil slighe cùraim ann airson inbhich a tha a' fuireach le hEDS agus HSD. Tha daoine a tha a' fuireach leis na cumhaichean sin ag aithris gu bheil iad a' faireachdainn gu bheil iad air an seachnadh le proifeiseantaich cùram slàinte, agus tha ùine gu breithneachadh gu cuibheasach fichead bliadhna. Tha aig mòran ri siubhal a Shasainn airson breithneachadh, agus tha faisg air aon às gach còignear a' faighinn a-mach gum feum iad pàigheadh iad fhèin gus comhairlichean prìobhaideach fhaicinn.

Chaidh a' mhòr-chuid den dàta a tha air a thaisbeanadh san aithisg a leanas a chruinneachadh mar phàirt den phròiseact "hEDS-START", a tha ag amas air mion-fhiosrachadh mu eòlas beò dhaoine a tha a' fuireach le hEDS/HSD ann an Alba. Bha am pròiseact seo a' toirt a-steach sgrùdadh nàiseanta mu bhuill EDS UK a tha a' fuireach le hEDS/HSD, a bharrachd air tachartas conaltraidh euslaintich, a chaidh a chumail aig Institiud Ginteachd agus Ailse sa Ghearran 2024.

Fhad 's a tha sinn ag aithris an seo air dàta an t-suirbhidh bhon luchd-freagairt Albannach, agus na h-eòlasan air an do bhruidhinn ar buidheann com-pàirteachaidh euslaintich Albannach, tha e cudromach cuimhneachadh gu bheil na h-eòlasan sin buntainneach do na dùthchannan tiomnaichte gu lèir. Thèid dàta mu dheidhinn na Rìoghachd Aonaichte gu lèir fhoillseachadh an ceann ùine.

Tha sinn ag iarraidh air Riaghaltas na h-Alba slighe cùraim iomchaidh a chruthachadh do dhaoine a tha a' fuireach le hEDS/HSD, gus seirbheisean roinneil a sholarachadh a bheir aire do na h-iomadh comharran agus suidheachadh co-phàirteach, agus gus cuideachadh le bhith a' toirt seachad trèanadh agus foghlam iomchaidh do phroifeiseantaich cùram slàinte

What are hEDS and HSD?

The Ehlers-Danlos Syndromes (EDS) are a group of heritable connective tissue disorders. Connective tissue lies between other tissues and organs, keeping these separate whilst connecting them, holding everything in place and providing support, like the mortar between bricks.

The 2017 International Classification of EDS refined the criteria for the 13 types of EDS, of which **hypermobile Ehlers-Danlos Syndrome (hEDS)** is a subtype [1]. hEDS is thought to be the most common genetic connective tissue disorder.

Unlike the other forms, hEDS is thought to be polygenic in nature and cannot currently be diagnosed by molecular genetic testing, though work is ongoing to identify the underlying genetic basis.

The hEDS diagnosis is based on three clinical criteria, (1) the presence of joint hypermobility, (2) the presence of other systemic features, family history and musculoskeletal complications, and (3) the exclusion of other heritable and

acquired connective tissue disorders or diagnoses.

Joint hypermobility implies a range of movement of the joint which exceeds what is expected given a person's age and gender. While joint hypermobility is a feature of hEDS, it is also found in the other subtypes of EDS, as well as benignly in the general population.

Many people do not fully meet the new diagnostic criteria for hEDS but their joint hypermobility, systemic symptoms, and co-occurring conditions still cause problems for them.

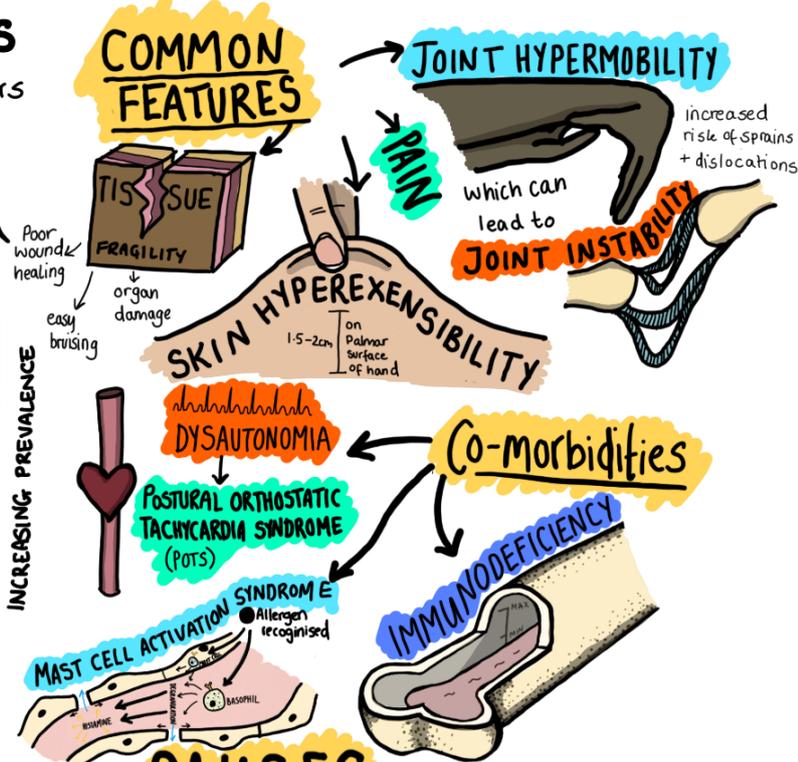
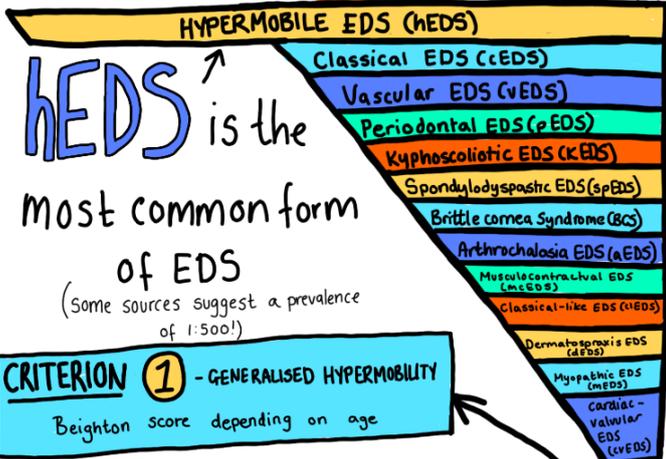
After other possible conditions are excluded, a diagnosis of **Hypermobility Spectrum Disorder (HSD)** may be made.

Although most commonly characterised by joint hypermobility, skin flexibility and tissue fragility, many patients with hEDS/HSD report a much wider range of co-occurring conditions and symptoms, which can be just as, if not more, debilitating than these connective tissue manifestations.

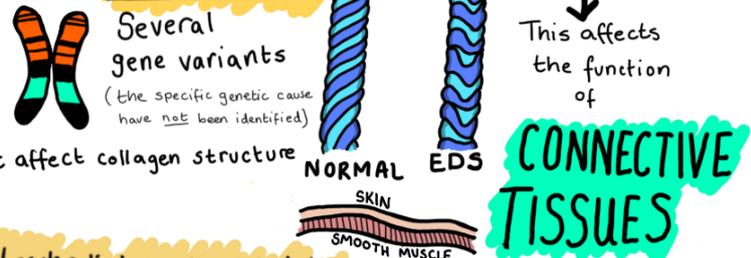
WHAT IS HEDS/HSD?

EDS
Stands for
Ehlers Danlos Syndromes
↳ a group of 13 connective tissue disorders

TYPES OF EDS

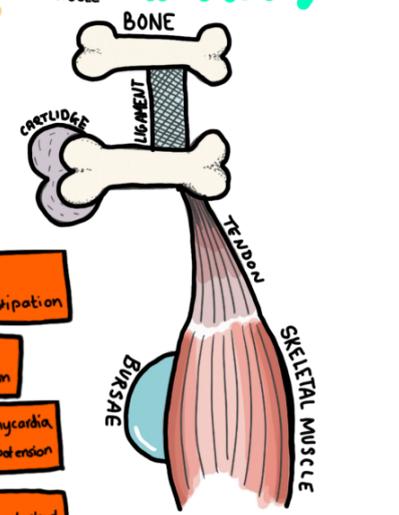


CAUSES



TREATMENT

- *Treat each patient + symptoms individually***
- PAIN** - acute - chronic - complex - neuropathic - fibromyalgia - myofascial
- GI ISSUES** - dyspepsia - nausea - IBS - diarrhoea - constipation
- NUTRITION** - Bespoke diets - reduced nutrient absorption
- CARDIOVASCULAR** - Postural tachycardia - orthostatic hypotension
- MAST CELL ACTIVATION** - poorly understood - H1+H2 antagonists use
- MUSCULOSKELETAL** - joint protection
- HEAD+NECK** - migraines - TMJ dysfunction - cervical disc degeneration



- ### Feature A
- unusually soft, velvety skin
 - mild skin hyperextensibility
 - unexplained striae distensae or rubae without a hx of weight gain/loss
 - bilateral piezogenic papules of the heel
 - Recurrent or multiple abdominal hernias
 - Atrophic scarring in at least 2 sites
 - Pelvic floor, rectal and/or uterine prolapse without other predisposition
 - Dental crowding + high narrow plate
 - Arachnodactyly
 - Arm span to height ratio ≥ 1.05
 - Mitral valve prolapse
 - Aortic root dilatation

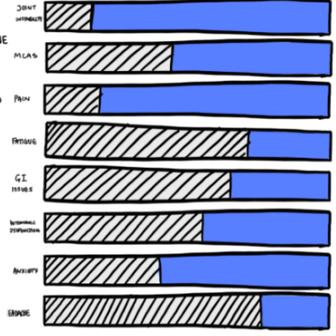
- ### Feature B
- Positive family history
- ### Feature C
- Musculoskeletal pain in ≥ 2 limbs recurring daily for at least 3 months
 - Chronic, widespread pain for ≥ 3 months
 - Recurrent joint dislocations or frank joint instability in the absence of trauma

HEDS SPECIFIC DIAGNOSTIC CRITERIA

CRITERION 3

Exclude other diagnoses

Rather than a linear scale each person with HSD can experience different symptoms.



What if hypermobile but does not fit into any other criteria?

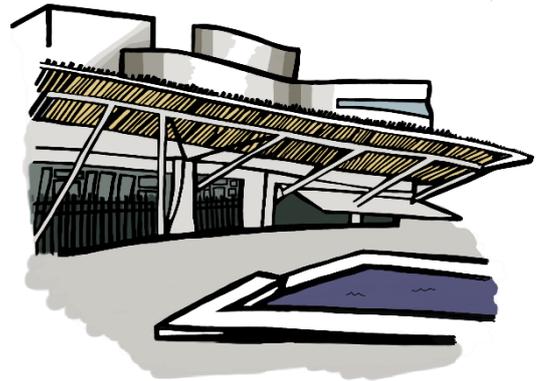
HYPERMOBILITY SPECTRUM (HSD) DISORDERS

- *PAEDIATRICS***
 - Autism Spectrum Disorder
 - Abdominal + bladder issues
 - "growing pains"
- *WOMENS HEALTH***
 - Hormonal + gynaecological issues are common
 - pregnancy can lead to complications

The Scottish Context

Key Messages

- People living with hEDS/HSD in Scotland now face a 20 year wait from symptom presentation to diagnosis.
- 22% of people living with hEDS/HSD in Scotland pay privately for their diagnosis, and more than 1 in 7 have had to travel to England for their diagnosis.
- 24% of people living with hEDS/HSD have had to travel outside of Scotland to receive healthcare.
- There is significant disparity between the potential prevalence of hEDS/HSD in Scotland and the extremely low number of patients officially coded in the health record.



In 2017, Professor Stuart Ralston of the University of Edinburgh submitted an application to the National Services Division (NSD) for the consideration of a specialist centre for the diagnosis and management of patients with complex EDS, and for the training and education of healthcare professionals managing this patient group.

This application was opposed by the Scottish Society of Rheumatology, and rejected on the basis that healthcare provision for patients with EDS was already in place and adequate.

“The notion that Scottish NHS services are being shared effectively with England is not only untrue,

but is actually harmful to Scottish EDS patients, as it leads to a lack of responsibility for Scottish patients within Scotland's NHS, meaning that they are not only being expected to travel for healthcare, but also falling down the cracks because the systems are not joined up. "

At that time, patients in Scotland with hEDS/HSD were reporting an average wait from symptom presentation to diagnosis of **19 years**.

In response to this, the NSD committed to conducting an evaluation of the services available to patients with complex EDS. To our knowledge, this evaluation was never completed.

It has previously been acknowledged that the availability and quality of services for patients with hEDS/HSD in the National Health Service (NHS) varies widely.

A 2020 report, "Ehlers-Danlos Syndromes and Young People in Scotland: The Agency Needed to

Prosper", discussed the experiences of young people in Scotland with an EDS diagnosis [2].

The report found that patients in Scotland perceived the attitudes of healthcare professionals to be negative and their diagnostic journey to be problematic. The author reported that there was an inequity of service access for Scottish patients, with many of the surveyed participants having to travel to London for their diagnosis. The report also advocated for better education and training of health professionals in Scotland.

In 2022, the authors and representatives from EDS UK met with Emma Roddick MSP to discuss the need for a pathway of care for patients living with hEDS/HSD in Scotland. Ms Roddick noted in a Meeting of the Parliament in November 2022 that clear treatment pathways were needed for diseases with chronic pain, such as EDS.



In 2023, EDS UK petitioned the Scottish Parliament (PE2038) to call on the Scottish Government to commission suitable NHS services for those with hEDS and HSD, and to consult with patients on the design and delivery of those services.

This petition was heard on the 4th of October 2023. The convener of that meeting suggested that the Scottish Parliament should write to NSD to ask whether it remains committed to producing the paper highlighting the service gaps, why the proposal for a specialist centre was rejected, and whether they have monitored the

delivery of its commitment to encourage regional expertise.

It was also suggested that the Government be asked how it intends to engage with people living with hEDS and HSD in taking forward actions under the rare disease action plan.

At present, EDS UK have recorded 620 members living in Scotland with hEDS/HSD. Recent data from Wales suggests a prevalence of hEDS/HSD of 1 in 500 [3], with a new study based in Northumberland reporting an even higher prevalence of 1 in 227 [4]. This prevalence data could suggest that between **11 and 24 thousand** people are living with HSD or hEDS in Scotland.

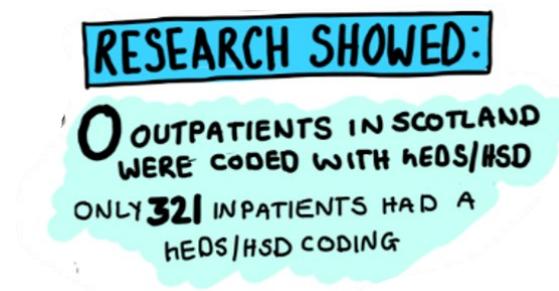
A study looking at a hypermobile population in GP practices in Northumberland found that people living with hEDS/HSD had almost twice as many outpatient and mental health appointments than those without a hEDS/HSD diagnosis. People with

hEDS/HSD were also more likely to present to A&E more than once in a 12-month period. The researchers found that service use and NHS costs were significantly higher for people with hEDS/HSD than for those without [4].

Given that we know that this patient group are heavy service users of the NHS, and despite the potentially high prevalence hEDS/HSD in Scotland, a 2022 review of Scottish health records by the authors of this report revealed that only 321 inpatients had been coded as having a diagnosis of hEDS/HSD or any of their outdated terminologies, and that not a single outpatient had been coded as such.

“It's time to bring water into the Scottish hEDS desert!”

It is abundantly clear that the needs of hEDS/HSD patients in Scotland are not being met. Time to diagnosis has worsened since 2017, and many



patients are still forced to travel to England or pay privately for diagnosis and management. The following pages will provide up-to-date information on the lived experience of these patients.

The Current Situation in Scotland

The hEDS-START study was founded in 2023, with the aim of capturing the lived experiences of people with hEDS/HSD. The study found that the average time to diagnosis in Scotland has increased since the 2017 EDS UK Survey, from 19 years to almost 20 years, despite the Scottish Society of Rheumatology's assertion that healthcare provisions were already in place and adequate.

“There is little support for EDS in Scotland. Traveling is especially difficult due to my CCI/AAI and yet there is no

support for me in A&E during emergencies. I face having to travel to London or Europe for help when I am at my worst, incurring huge medical bills.”

15% of Scottish hEDS/HSD patients have had to travel to England to receive their diagnosis, and more than 1 in 5 hEDS/HSD patients in Scotland are being diagnosed in private practice, forced to pay money out of their own pocket. 24% of respondents reported that they had travelled outside of Scotland for hEDS/HSD-related medical care in the past.

“Most medical professionals here in Scotland don't seem to know what hEDS is, or dismiss it as "just hypermobility". I had to go to London to be diagnosed. I have to pay for private physio since the NHS would only offer 6 months.”

We also sent a survey to EDS UK members based in Scotland, asking them to answer 'Yes' or 'No' to a series of questions which were based on the



1 in 5
are diagnosed privately

Scottish Government's own Health and Social Care Standards [5]. Satisfaction amongst respondents was low, with 95% reporting that they did not think that healthcare in Scotland was adequate, 92% reporting that they don't believe they receive the care that is right for them, and 84% reporting that their ability to live and work has been directly impacted by their care (Figure 2).

In Conclusion

The current situation for people living with hEDS/HSD in Scotland reveals significant challenges that demand immediate attention from policymakers and HCPs. Initiatives to establish a specialist centre for this patient group have faced rejection on the basis that

existing services are adequate – a stance at odds with patient reports of an average twenty-year wait to diagnosis.

The substantial discrepancy between this prevalence and the number of people coded correctly in health records points to a systemic issue of understanding in the healthcare system.

The lack of a unified and knowledgeable approach to hEDS/HSD within Scotland’s NHS forces patients into lengthy and often costly journeys for appropriate diagnosis and care. Recent work suggesting a potential prevalence as high as 1 in 227 people further illuminates the need for specialist services and education.

Average Time to Diagnosis (Years)

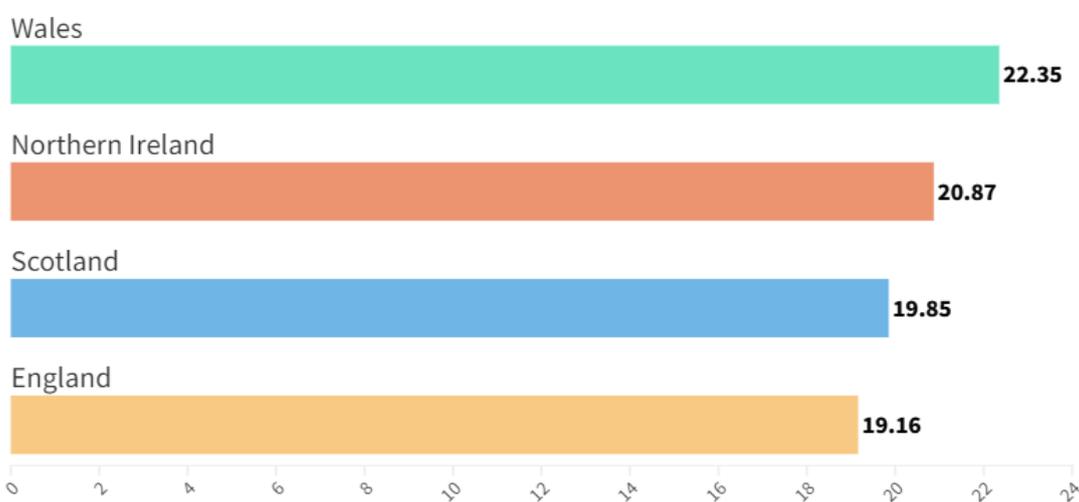


Figure 1: Time to diagnosis as reported by hEDS-START respondents UK-wide

Do you feel the healthcare in Scotland is adequate to support your health needs?



Do you feel the healthcare system in Scotland provides safe, effective, and person-centred care?



Do you believe you experience high quality care and support that is right for you, when you need it?



Do you feel fully involved in all decisions about your care and support?



Do you have confidence in the NHS as an organisation to provide your care and support?



Do you have confidence in the people who provide support and care for you?



Do you experience a high quality, and appropriate environment for treatment, which considers your specific health requirements?

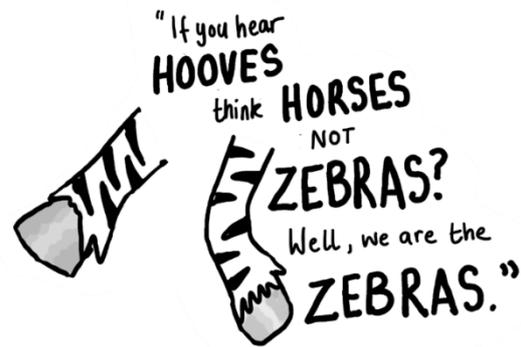


Do you feel your ability to live and work independently has been directly impacted by delays in receiving care or the right level of care?



Figure 2: EDS UK Scottish Member Survey

Ensuring patients get the right diagnosis, faster



Key Messages

- People with hEDS/HSD face almost 20 years between symptom presentation and diagnosis in Scotland. There is an urgent need for a reduction in this time to diagnosis.
- Diagnostic tools do exist, but are difficult to access for those unaware of them. There is a critical need for clearer referral pathways and better access to diagnostic tools.
- Many patients report misdiagnosis of other conditions, often leading to poor management or the experience of other negative impacts on their care.

Time to Diagnosis

The 2023 hEDS-START survey revealed that patients in Scotland are now reporting almost 20 years between symptom presentation and a hEDS/HSD diagnosis, on average one year longer than was reported in the 2017 EDS UK survey, which asked the same question (see Figure 1).

"I'm nearly 44 years old and had to reach a point of severe burnout and desperation to finally be listened to and for a diagnosis and management plan to get under way. The experience has been traumatising and has deeply impacted my mental well-being and confidence."

The purpose of the introduction of the EDS UK GP Toolkit in 2018 was to facilitate diagnosis in primary care. The toolkit can be accessed here:

<https://gptoolkit.ehlers-danlos.org/> [6].

At present, around 60% of diagnoses in Scotland are made in secondary care rheumatology services, with a further 20% being made in other secondary care services.

Only 8% of the Scottish survey respondents had been diagnosed in primary care. This figure increased to just 10% when solely diagnoses made after the introduction of the GP Toolkit in 2018 were analysed.

The most common secondary care speciality for hEDS/HSD patients in Scotland to be referred to in the first instance is rheumatology. The reported median waiting time for rheumatology services - for those fortunate enough to receive a referral to secondary care - is highly varied in Scotland. NHS Fife, for example, has a reported median

waiting time of 5 weeks, whereas people living in the NHS Ayrshire catchment area can expect a median waiting time of 32 weeks [7].

Some hEDS/HSD cases are more complex than others, and may require specialist input. However, it is important to note that the 2017 diagnostic criteria can be easily interpreted by most healthcare professionals.

At present, these criteria - as well as the 2018 GP Toolkit - are difficult to find on the NHS Lothian GP referral guideline homepage, RefHelp. It is presently unknown how accessible these guidelines are in other health boards.

A previous research article from the United States reported that the average number of clinicians a person sees before they are diagnosed with hEDS is 15.6, reducing by half following a diagnosis of hEDS/HSD [8]. Facilitated access to the GP Toolkit, in

combination with the development of a clear pathway of care and easily understood referral guidelines, could empower HCPs to have the confidence to diagnose patients who present with symptoms of hEDS/HSD, reducing burden on the healthcare system.



Misdiagnosis

Many people living with hEDS/HSD report years of misdiagnosis before a HCP recognised the signs and symptoms of hEDS/HSD. When asked about their lived experience of their hEDS/HSD, 9% of respondents specifically spoke of being misdiagnosed with another condition. This figure comes from shared lived experiences and could very feasibly be

under-reported given the open nature of the question asked.

“My diagnosis was very drawn out and full of fighting with my GP who had decided that there was nothing wrong with me. When I fought the incorrect diagnosis I was told I was mentally ill. The more I fought the diagnosis the more the GP practice as a whole doubled down on me being mentally ill”

Common misdiagnoses are fibromyalgia, myalgic encephalomyelitis/chronic fatigue syndrome, and functional neurological disorders.

A misdiagnosis of a functional neurological disorder has previously been reported to be the most rejected diagnosis in a hEDS patient population, with 95% of those who had been given this diagnosis in the past believing it to be inaccurate [8].

All of these diagnoses share similarities with hEDS/HSD, and many symptoms

overlap. However, it is important to patients to have a full understanding of their conditions in order for them to self-manage in the first instance, and so that they know where to turn should new symptoms or co-occurring conditions emerge.

Misdiagnosis can have a harmful social and psychological impact, as well as the physiological costs of mismanagement. People living with undiagnosed hEDS/HSD may not be equipped with the ability to manage their symptoms, and may deteriorate.

“I was referred to Neurology for an assessment for MS after presenting with multiple unexplained symptoms in my mid-20s. The tests came back negative and I was discharged with a diagnosis of a “functional neurological disorder”. I have since learned that all of these symptoms are relatively common in people with hEDS but this incorrect diagnosis now means that my (now very expensive!) life insurance will not pay out if I develop any

neurological condition in the future.”

The impact of late and incorrect diagnoses

It is difficult to quantify the ramifications of an incorrect diagnosis. However, early diagnosis and treatment of people living with hEDS/HSD almost certainly has the potential to improve the trajectory of these disorders, and the lives of those living with them.

“If I had had a diagnosis prior to my pregnancy I truly believe I could have gone on to have more children and would still be in work. The lack of knowledge and support has had a devastating impact on my life and that of my family.”

Previous research has found that an increased delay to diagnosis is more commonly found in hEDS/HSD patients with severe pain, compared to those with moderate pain [9]. It is evident that any delay to diagnosis could compound and exacerbate chronic

pain, a symptom already reported by 87% of the hEDS-START Scottish survey population.

A study from Sweden which explored psychiatric disorders in people living with EDS/HSD found that an earlier diagnosis of EDS actually had a protective effect on later suicide attempt. Though the authors stress this may have been a chance finding, they state that they “cannot rule out that some individuals take comfort in receiving a diagnosis (EDS) for which they may have sought help for many years” [10].

“My anxiety almost vanished once I had to stop fighting to be diagnosed, I was so ill and no one believed me - I felt like I was going crazy”

Patient education and self-management is the optimal treatment strategy for people with non-complex hEDS/HSD. Where people living with these conditions are left waiting

decades to receive a diagnosis, and consequently decades to receive an appropriate management plan, it is common for their ability to self-manage to become impaired:

“The years people spend being left, disbelieved, misdiagnosed and deteriorating has a huge effect on peoples mental and physical health ”

In severe cases, where people with hEDS/HSD have lost mobility or are experiencing chronic pain and lifelong symptoms, it can be incredibly difficult for them to resume a “normal life”, having physically and mentally deteriorated for decades.

“I sometimes wonder if I was diagnosed as a child or adolescent, perhaps I wouldn't have gotten so ill. I'm an electric wheelchair user, I have a gastrojejunostomy feeding tube, I need a lot of help, I even have issues with my throat and larynx which can impact my work.”

The benefit of timely diagnosis

The importance of a timely and correct diagnosis cannot be overstated. A diagnosis of HSD/hEDS can provide a person with validation after several years of feeling disbelieved.

*“The most powerful thing was knowing that I wasn't mentally ill and faking my symptoms as I'd been told *my entire life*, and actually then I could reframe my entire identity.”*

It also equips patients with the ability to manage their symptoms, and to be able to identify when new symptoms or co-occurring conditions appear, enabling them to seek the appropriate care.

“Although there is currently no cure for EDS, earlier diagnosis and management can make a monumental difference to quality of life.”

A paper published by Halverson et al in 2023 describes the journey to diagnosis for hEDS patients. They report that pain and fatigue are the most common reasons that participants sought medical care. Following a diagnosis of

36 YEARS
to get a diagnosis
for hEDS.

hEDS, there was a 52% reduction in the number of clinicians participants accessed [8].

Timely diagnosis undoubtedly benefits the person who is receiving it, but it can also have the effect of reducing service access and use, relieving some of the pressure on the healthcare system.

In Conclusion

The hEDS-START survey has highlighted a critical challenge within the Scottish healthcare system: the excessive time to diagnosis for patients with hEDS/HSD now averaging almost 20 years. This delay significantly impacts patients' physical and mental health.

Despite the introduction of the GP Toolkit in 2018, just 10% of diagnoses since then have been made in primary care. This emphasises the need for enhanced accessibility to diagnostic tools and the development of a clear pathway of care.

Misdiagnosis remains an issue, with almost 1 in 10 respondents reporting unprompted that they had received an incorrect diagnosis in the past. The consequences of misdiagnoses extend beyond delayed appropriate care to include severe social, psychological, and physiological impacts.

Diagnosis is not, and should not be, considered the end of the patient journey. Symptoms and co-occurring conditions are lifelong and can change over the course of a person's lifetime.

HCPs should be knowledgeable and confident enough to educate their hEDS/HSD patients about their diagnosis, equipping them with appropriate information and resources

to underpin an effective self-management strategy.

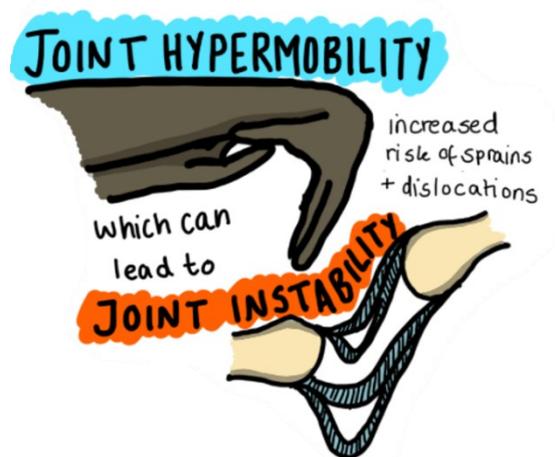
Addressing these challenges requires a multifaceted approach: improved accessibility to diagnostic criteria and tools for healthcare professionals, education on hEDS/HSD to prevent misdiagnosis, and the development of streamlined referral processes.

It is imperative for healthcare professionals and policymakers to collaborate in refining these processes, ensuring timely and accurate diagnoses, and ultimately, enhancing the quality of life for patients with hEDS/HSD.

The clinical characteristics of hEDS/HSD patients

Key Messages

- People with hEDS and HSD exhibit many symptoms beyond those limited to the musculoskeletal system.
- People with hEDS/HSD report significant psychological distress.
- Primary care HCPs should consider which symptoms have the greatest impact on hEDS/HSD patients.
- All HCPs, regardless of discipline and specialty, will encounter patients with hEDS/HSD – diagnosed or undiagnosed – and should have adequate understanding of the physiological and psychological implication of these disorders.



Although hEDS and HSD are most commonly characterised as connective tissue disorders, and as such are primarily handled in Rheumatology clinics in Scotland, individuals with these disorders exhibit symptoms across the full body.

"I have spent as long as I can remember in pain and suffering from Gastro complications, extremely painful periods, headaches, joint dislocations and subluxation, I wasn't diagnosed until I was in my mid 20s and still at almost 35 don't fully understand the condition myself because there is so little help after diagnosis. The diagnosis was a relief to finally know what was causing the pain and symptoms..."

HCPs should be prepared to listen to people with hEDS/HSD to ascertain which symptoms they are experiencing, in order to provide the highest level of care. Patients should be equipped with information about co-occurring conditions and symptoms so that they are able to advocate for themselves should new manifestations appear in the future.

“EDS is the diagnosis but all of the symptoms that are caused by it also require management. It is not just hypermobility, as many practitioners believe. Once you finally get a diagnosis, it's a really long road to figuring out how EDS is impacting all of the different parts of your body - like putting a puzzle together. I'm still trying to figure things out many years later.”

The hEDS-START Study 2023 survey asked participants to record whether they had a diagnosis of a number of symptoms and co-occurring conditions known to be associated with hEDS/HSD.

The following pages will provide information on the prevalence of these symptoms and co-occurring conditions.



Chronic Pain

Chronic pain is common in patients with hEDS and HSD as a result of inherent joint instability, subluxation and soft tissue injuries. Musculoskeletal pain which was at one time occasional and acute, can evolve over time and become chronic and widespread [11], limiting quality of life.

Despite chronic pain being the most commonly reported symptom in the Scottish hEDS-START survey population, with 87% of our respondents reporting this, less than one third of participants had previously received a referral to a pain management service, and only 26% had seen a pain management team within the last 5 years.

Physical therapy is key to pain management in patients with

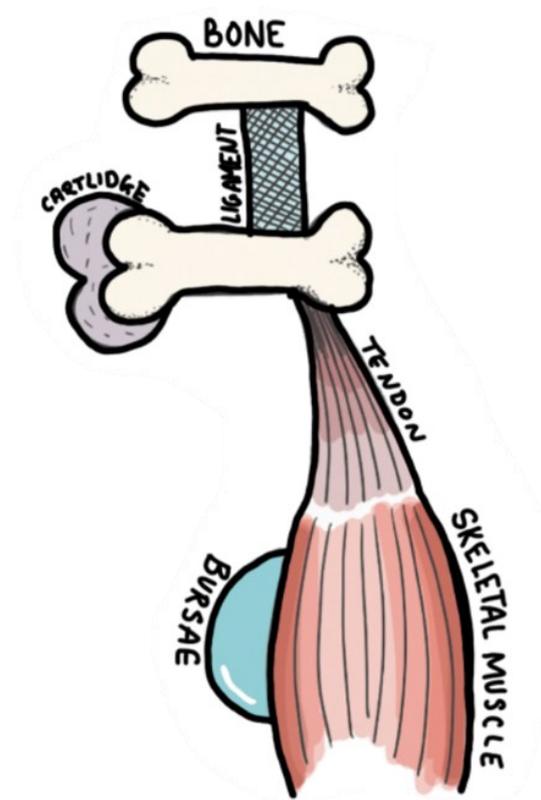
hEDS/HSD, with evidence suggesting that it reduces joint instability and pain, and increases patient mobility [12]. The hEDS-START study found that 75% of hEDS/HSD patients had previously received a referral to physiotherapy.

Of course, pain in patients with hEDS/HSD is not limited to the musculoskeletal system. Patients also report increased abdominal [13], neurological [14], and gynaecological pain [15, 16], to name but a few.

Musculoskeletal Symptoms

The most common musculoskeletal manifestations reported by the hEDS-START Scottish survey population were joint subluxations, reported by 71% of respondents, and joint dislocations, reported by 45% of the respondents.

Joint subluxation is when a bone moves out of the joint, but does not fully separate. This most often occurs in the shoulders, elbows, hips, kneecaps and hands and can cause pain and loss of sensation.



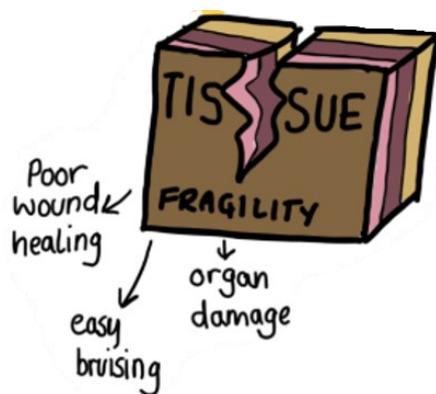
Joint dislocation is the full separation of a bone from its joint. This can cause severe pain, loss of range of motion, and neuropathic symptoms, and often requires medical intervention.

"During lockdown I dislocated my collarbone in my sleep and was not believed by my GP: "Are you sure? We normally only see that in car crashes."

Joint subluxation and dislocation are common in hEDS/HSD because of the

inherent increased laxity in the connective tissue surrounding the joint.

Subluxation and dislocation can present many challenges for people living with hEDS/HSD, such as an increased difficulty with functional activities, mobility and high levels of distress. Subluxation and dislocation of the jaw for example can lead to temporomandibular dysfunction, which can affect eating, drinking, and speaking.



Skin Manifestations

Most of the EDS subtypes have skin involvement. Skin manifestations reported by the hEDS-START Scottish survey population were: poor wound healing, reported by 39%, abnormally

stretchy skin, reported by 37%, and abnormal scarring, reported by 39%.

“I am now currently in a lot of pain after surgeries to remove excess skin has not had good results as my skin isn't strong enough. If I knew I had EDS I would probably never have went for the surgeries.”

Gastrointestinal Symptoms

It is very common for people living with hEDS and HSD to report gastrointestinal manifestations of their disorder. These symptoms include abdominal pain, postprandial fullness, constipation, diarrhoea, oesophageal reflux [17], irritable bowel syndrome [18], and rectal hyposensitivity [19].

66% of the hEDS-START Scottish survey population reported gastrointestinal manifestations of their hEDS/HSD.

Despite this, only 28% of the respondents reported that they had seen a gastroenterologist in the last five years.

In the hEDS-START Scottish survey population, 68% of hEDS-diagnosed participants reporting GI manifestations, compared to 47% of HSD-diagnosed participants reporting the same.

Faecal incontinence is more common in people living with hypermobility disorders. In a study of Hypermobility Syndrome Association members, 14.9% surveyed reporting faecal incontinence, compared to 2.2% in the general population [20].

Psychological Diagnoses

NICE report an estimated prevalence of generalised anxiety disorder of 4-7.9% in the UK [21]. The World Mental Health Survey Initiative reported a lifetime prevalence estimate of 14.6%, and a 12-month prevalence estimate of 5.5% of depression for adults living in high-income countries [22].

The hEDS-START Scottish survey population reported high diagnosis rates of both anxiety (68%) and

depression (63%), much higher than previously reported in EDS populations [23]. Despite these high numbers, only 13% of respondents reported having seen a clinical psychologist in the last five years.

Anxiety and depression are frequently co-occurring conditions. The relationship between anxiety and joint hypermobility has long been established, first reported in 1988 by a clinical team in Barcelona who noticed an overlap of anxiety and joint hypermobility in their patients [24].

The hEDS-START study participants were asked to fill in the Patient Health Questionnaire 4 (PHQ4). This questionnaire provides a score for core symptoms and signs of anxiety of depression within the past two weeks, with a score of 3 or greater in each domain representing a reasonable cut-off for identifying cases of each condition [25].

A combined score of 9 or greater is considered a “red flag” for the presence of severe psychological distress, warranting referral for further assessment [26].

43% of the hEDS-START Scottish survey respondents reported a score of 3 or greater in the anxiety domain, with 44% recording a score of 3 or greater in the depression domain.

Concerningly, 20% of the population recorded a score of 9 or above. These results indicate significant and substantial current psychological distress in this patient group.

Poor awareness of the condition and a lengthy time to diagnosis have been linked to a decrease in self-esteem and bodily autonomy. A diagnosis of hEDS/HSD can have a significant psychological impact on people, due to limitations on activity and socialisation, and in some cases the changes in physical appearance [27].

Musculoskeletal pain and gastrointestinal dysfunction are significantly associated with the presence of psychiatric disorders in patients with EDS/HSD [28]. There is a high rate of both of those symptoms in our study population which may go some, though not all of the way to explaining the high prevalence of anxiety and depression.

Healthcare-induced anxiety should also be taken into account in this patient population. Many people with hEDS/HSD report an exceptionally difficult journey to diagnosis, with disbelief from healthcare professionals being explicitly mentioned by almost two-thirds of the hEDS-START Scottish survey respondents.

hEDS has previously been associated with a substantially increased risk of attempted suicide [10]. A study of adolescents with hEDS in France reported a rate of attempted suicide almost three times higher than the

French average, which they attributed to the distress faced by patients prior to receiving a diagnosis [29].

Neurological Symptoms

Migraine is a series of symptoms most commonly associated with a severe one-sided headache. Symptoms can include malaise, vomiting, altered vision, and light, smell, and sound sensitivity. The Migraine Trust report a prevalence in the UK of between 15% and 23% [30].

Previous research into headaches in individuals with hEDS/HSD in Israel found that migraines were reported by 47% of study population [31]. This is reflected in the hEDS-START Scottish survey population, in which 53% of participants reported a previous diagnosis of migraine.

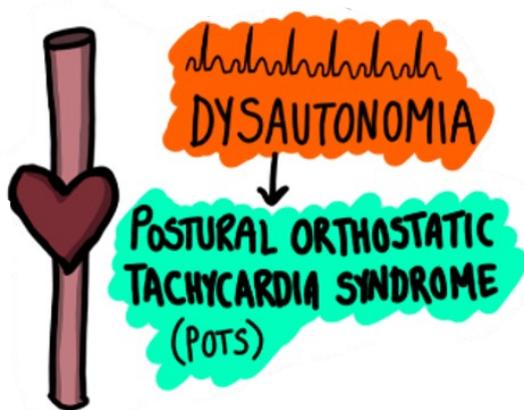
Other neurological symptoms associated with hEDS/HSD diagnoses include craniocervical instability (CCI)



and atlanto-axial instability (AAI). These conditions are similar in presentation, as both cause an instability or excessive movement in the neck. The implication of this is that there can be an increased pressure placed on the spinal cord. 6% of the hEDS-START Scottish survey population report a diagnosis of these conditions.

Chiari malformation occurs when the lower part of the brain bulges through the normal opening in the skull, putting pressure on the spinal cord. Symptoms include headache, neck pain, dizziness,

and numbness [32]. The estimated prevalence of Chiari malformation in the UK is less than 1% [33]. The hEDS-START Scottish survey population reported a Chiari malformation diagnosis rate of 2.3%.



Cardiological Symptoms

Postural Orthostatic Tachycardia Syndrome (PoTS) is a common diagnosis in people with hEDS/HSD. PoTS is a group of disorders which are caused by an abnormal autonomic response to moving into the upright position.

When a person stands up or does exercise, their heart rate increases and their blood pressure drops. This can

result in fainting, dizziness, and nausea, and can be very debilitating.

It has been speculated that PoTS occurs in a hEDS/HSD population as a result of the laxity of the connective tissue, which may prevent the vascular system from effectively carrying blood to the brain fast enough when rising to an upright position [34].

The prevalence of PoTS in the UK is thought to be 0.2% [35]. The hEDS-START Scottish survey respondents reported a diagnosis rate of 36%. This is in line with the 33% of participants who reported having seen a cardiologist in the last five years.

Other orthostatic intolerances are common in a hEDS/HSD population, with some studies suggesting that up to 100% of hEDS patients experience some sort of orthostatic intolerance [36].

Mitral valve prolapse is where the valves of the left ventricle of the heart do not always close, which can

sometime cause the blood to leak backwards into the left atrium [37].

Prevalence of mitral valve prolapse has previously been recorded to be 2.4% in the general population [38]. 5.8% of the hEDS-START Scottish survey respondents reported a previous diagnosis of mitral valve prolapse and 6.1% reported other heart valve issues.

12% of the hEDS-START Scottish survey population reported other bleeding or clotting problems, and 33% reported a diagnosis of Raynaud's, compared to 15% of the UK population [39].

Urological Symptoms

A person with EDS might present to urology with a variety of symptoms, from storage symptoms such as urge and stress incontinence, urinary frequency and urgency, bladder sensitivity, recurring urinary tract infections [40] and difficulties emptying the bladder [41].

"I have had recurring issues with miscarriages and

ectopic pregnancy and I know I have a vaginal prolapse and have had incontinence for a long time because of this but no referral."

The prevalence of urinary incontinence in women with EDS is reported to be between 69% and 84%, compared to 30% in the general population, with a higher risk of incontinence and prolapse after childbirth [20]. Urological symptoms can be due to connective tissue and pelvic floor muscle defect [41], which results in insufficient bladder support [40].

Gynaecological Symptoms

Endometriosis is a chronic disease in which tissue similar to the lining of the uterus grows elsewhere, causing extreme pain and fatigue.

Endometriosis can severely impact a person's quality of life, in some cases preventing them from working, socialising, or completing education.

The condition can have repercussions on a person's reproductive health, sexuality, and relationships.

The UK prevalence of endometriosis is thought to be around 10%. The hEDS-START survey found that 14% of Scottish respondents reported a diagnosis of endometriosis.



Pelvic organ prolapse (POP) is when the muscles and tissues supporting the organs in the pelvis (bladder, uterus, or rectum) becomes weak or loose, causing one or more of those organs to drop into, or out of, the vagina. POP can present in patients with EDS at a young age and in the absence of pregnancy. Management of POP in EDS requires a multidisciplinary

approach. 13% of the female hEDS-START Scottish survey population reported a diagnosis of POP.

Lately there has been a steady increase in research looking into the relationship between hypermobility and pregnancy. Recent work by Pearce et al. has revealed a number of increased risks associated with pregnancy and hEDS.

“I feel even during the pregnancy and birth of my child my health could have been managed better and I wouldn't have suffered as badly as I did during labour. I've since found out the impact EDS can have on labour and delivery.”

Women with hEDS are more likely to develop pre-eclampsia, eclampsia, pre-term birth, postpartum haemorrhage, hyperemesis gravidarum, caesarean wound infection, postpartum psychosis, and post-traumatic stress disorder, as well as reporting that their child was born before arriving at their intended place of birth [42].

Women with hEDS are also more likely to experience miscarriage and multiple miscarriage [43] and to require a C-section [44].

Fatigue

EDS and hypermobility have long been associated with chronic fatigue. In 2011, research was published describing the presence of chronic fatigue in hEDS (then called EDS-Hypermobility Type) patients. This research found that 82.6% of the participants with hEDS met the diagnostic criteria for chronic fatigue syndrome (CFS), compared with 2.1% of the control population [45].

A study of adolescents with CFS also found that 60% of the participants displayed joint hypermobility [46], indicating there may be some clinical overlap between the two diagnoses.

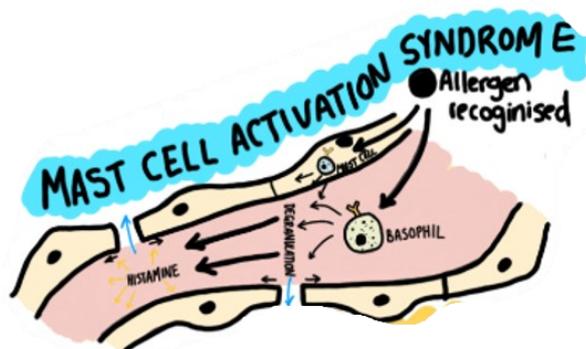
“In my late teens-early 20's I had a period where I could barely get out of bed. No one helped me and no one was interested. This hugely impacted my life and still does.”

Factors that contribute to fatigue in people with hEDS include sleep disorders (reported by 25% of the hEDS-START Scottish survey population), chronic pain (reported by 87% of respondents), muscular deconditioning, cardiovascular autonomic dysfunction (e.g. PoTS), bowel and bladder dysfunction, psychological issues, and nutritional deficiencies [47].

“[hEDS] is basically a lifelong sentence without support, I feel like I have to stop doing anything I enjoy in fear that it will cause me to be bedridden for days afterwards in pain or with extreme fatigue”

Chronic fatigue has a significant impact on quality of life in people with hEDS/HSD, and can have a larger impact on daily functioning than chronic pain [48]. Given the strong overlap between fatigue and many of the symptoms and co-occurring conditions associated with hEDS/HSD, HCPs should be mindful and

understanding when a patient presents with this symptom.



Mast Cell Activation Syndrome

Mast Cell Activation Syndrome (MCAS) is a condition which causes allergic symptoms which range from severe itching and swelling, to anaphylaxis which may be life-threatening. Often this response is in reaction to something not usually considered to be harmful, such as foods or household goods [49].

EDS and PoTS are being increasingly reported in populations who have an MCAS diagnosis, though limited data on the relationship between the three exists. Prevalence of MCAS in the UK is

unknown. 15% of the hEDS-START Scottish survey population reported a diagnosis of MCAS, which is much lower than the reported prevalence of 48% in a previous study of hEDS patients [8].

In Conclusion

The results of the hEDS-START Study have revealed critical insights into the clinical needs of a Scottish hEDS/HSD population. Key findings highlight a significant prevalence of chronic pain, psychological distress, and multisystemic manifestations.

However, there is a noticeable gap in access to specialist services, such as gastroenterology, pain management, and clinical psychology, underlining the disconnect in current care.

For policy makers and healthcare professionals, these findings underscore the urgent need for enhanced awareness, education, and a holistic management strategy for hEDS/HSD patients.

Neurodivergence and hEDS/HSD

Key Messages

- People with hEDS/HSD are more likely to report diagnoses of Autism, Attention Deficit Disorder AD(H)D, dyslexia, and dyscalculia.
- People who have hEDS/HSD in combination with Autism are significantly more likely to report co-occurring conditions and symptoms of hypermobility.
- People who have hEDS/HSD and who are also autistic are also significantly more likely to be out of work, receiving benefits, have had their education interrupted, and require a carer.
- HCPs should consider neurodivergence when communicating with, assessing,

and treating patients with hEDS/HSD.

Recent work by Csecs et al has revealed a significant link between joint hypermobility and neurodivergence, with 51% of a neurodivergent group showing increased levels of joint hypermobility [50].

There is clear interest within the patient community into the link between neurodivergence and EDS. A recent survey of autistic adults in Scotland asked participants to rate and rank what their research priorities were. Several participants explicitly mentioned EDS and MCAS as co-occurring conditions to be investigated further [51].

This was echoed at the 'Translating Research into Change' event. When we asked people with lived experience of hEDS/HSD what they would like to see in future research, participants said that they would be interested in more research looking into the relationship



between hEDS/HSD, PoTS, MCAS, Autism, and AD(H)D.

The hEDS-START study collected information on rates of neurodivergence in a population of people with hEDS/HSD, and found higher rates of Autism, Attention Deficit Disorder AD(H)D, dyslexia, and dyscalculia than the general population.

Autism

Autism is a neurodevelopmental difference which can in some cases present challenges with social interaction and communication.

In the hEDS-START Scottish survey population, 21% reported having had a

clinical diagnosis of Autism. Although it is widely agreed that Autism is underdiagnosed in the general population, particularly in those assigned female at birth, estimates of prevalence in the UK sit at around 1.1% [52], with the highest estimated rates found in adolescent boys, reported at 2.9% [53].

It is important to note that the majority of our patient population (90%) identify as female. Women are typically less likely to receive a diagnosis of Autism, with research suggesting that the male to female ratio of Autism diagnosis is between 2:1 and 3:1 (M:F) [54]. The Autism diagnosis rate was 17% in respondents who identified as women

(n = 309), 20% in those who identified as men (n = 10), and 65% in those who identified as non-binary (n = 26).

As part of the hEDS-START study, the authors asked participants to fill in a fifty-part questionnaire called the Autism Quotient (AQ50). Questions cover five key areas: social skills, attention switching, attention to detail, communication and imagination.

While this questionnaire is not a diagnostic tool, its developers note that a score of ≥ 26 indicates a relatively high level of autistic traits. In this part of the study, 64% of the Scottish survey respondents had a score of ≥ 26 .

Scores of ≥ 32 are particularly significant as only 2% of a control group would score 32 or above, compared to 79% of those with an Autism diagnosis [55]. In the Scottish survey, 47% of respondents scored ≥ 32 , suggesting that people with hEDS/HSD show high levels of autistic traits.

When the full UK-wide hEDS-START dataset was analysed, it was found that people who had a diagnosis of Autism were much more likely to present with hypermobility-related symptoms of chronic pain, joint subluxation and dislocation, abnormal scarring, poor wound healing, and stretchy skin.

People with co-occurring hEDS/HSD and Autism were also significantly more likely to report the following co-occurring conditions: anxiety, depression, gastrointestinal manifestations, PoTS, AD(H)D, Raynaud's phenomenon, ME/CFS, sleep disturbances, dyslexia, dyspraxia, dyscalculia, MCAS, and cranio-cervical or atlantoaxial instability.

People with co-occurring hEDS/HSD and Autism were more likely to report being out of work, receiving benefits, having had their education interrupted by their diagnosis of hEDS/HSD, and requiring a carer.

Attention Deficit Disorder AD(H)D

AD(H)D is a condition which is characterised by periods of inattention or impulsivity, to the point at which those periods interfere with daily activities [56]. NICE suggest that the prevalence of AD(H)D in the UK is between 3-4% [57].

Previous work from Sweden, published in 2021, suggests a prevalence of AD(H)D in a hEDS/HSD population ranging from 16% being diagnosed in childhood, up to 46% being diagnosed by late adolescence [58].

These high numbers were not reflected in the hEDS-START Scottish survey population, with 15% of the respondents reporting a diagnosis of AD(H)D. It is important to note that the hEDS-START study had a much higher proportion of female participants (90%) when compared to the Swedish study, which was 43% male, and males are more likely to be diagnosed with AD(H)D than females [59].



A 2019 paper published in Rheumatology International suggested that AD(H)D prevalence is significantly higher in the HSD population, but not in the EDS population [28].

This is reflected in the hEDS-START Scottish survey population, with 25% of the HSD population reporting a diagnosis of AD(H)D, compared to 14% of the hEDS population, though numbers of participants with a diagnosis of HSD were relatively small.

Dyslexia

Dyslexia is a condition which can affect, but is not limited to, a person's reading and writing skills. The British Dyslexia

Association reports that around 1 in 10 people living in the UK are dyslexic [60]. 17% of the hEDS-START Scottish survey population reported a clinical diagnosis of dyslexia.

Dyscalculia

Dyscalculia represents a severe and persistent difficulty with maths, which is present throughout a person's lifetime. The British Dyslexia Association reports a prevalence of dyscalculia in the UK of 6% [61]. 16% of the hEDS-START Scottish survey population reported a diagnosis of dyscalculia, making it twice as common in this hEDS/HSD population than in the general population.

Dyspraxia

Dyspraxia is a condition which affects a person's co-ordination and movement. 6% of the hEDS-START Scottish study population reported a diagnosis of dyspraxia, which is in line with the UK population prevalence [62].



Looking Ahead

Recognition and understanding of higher levels of neurodivergence in this patient population should be fundamental in the management and treatment of people with diagnosed or suspected hEDS/HSD, and should underpin any pathway of care in the future.

Clinicians and healthcare professionals should consider the appropriateness of materials shared, clinical environment, and treatment strategies utilised on an individual basis. Neurodivergent people experience higher levels of difficulty in accessing healthcare [63], higher levels of illness and decreased life expectancy, and face increased

inequalities within the healthcare system [64].

NHS England have published key points which should be addressed by HCPs when assessing and treating neurodivergent patients [65]. These include, but are not limited to;

- Being aware of diagnostic overshadowing, which occurs when symptoms arising from physical or mental health are mistakenly attributed to neurodivergent diagnoses.
- Ensuring that clinical decisions around care and access to treatment are made on an individual basis.
- Listening to parents and carers who have additional insight into an individual and their health, including comorbidities and medications.
- Communicating with the aim of understanding the person you are caring for, and with the aim

of ensuring that you are understood.

- Understanding behavioural responses to illness, pain, and discomfort.

HCPs need adequate and specific neurodivergence training in order to be able to assess and treat all people appropriately. There is little benefit in treating patients using management techniques which do more harm than good overall, such as hands-on physiotherapy in patients who report tactile defensiveness or touch sensory sensitivity.

HCPs should take into account that a patient presenting with co-occurring hEDS/HSD and Autism might require additional support, both clinically and socially. It is important that HCPs know where to signpost patients who might require this additional support.

Whilst a thorough clinical examination is vital for all HSD/hEDS patients, clinicians should be aware that there is

a higher incidence of symptoms and co-occurring conditions in an Autism-diagnosed HSD/hEDS population and should make every effort to record this. HCPs should be aware that pain can present differently in Autistic people [66].

It could also be worth considering whether people who are diagnosed with hEDS/HSD should be assessed for the presence of neurodivergent diagnoses when they present to clinic.

In Conclusion

The intersectionality between hEDS/HSD and neurodivergence necessitates a tailored approach to care. HCPs should be vigilant for symptoms indicative of neurodivergent conditions in patients with hEDS/HSD, recognising that traditional diagnostic and management strategies may not be sufficient or appropriate.

The implementation of individual care plans, considering the specific sensitivities and needs of

neurodivergent individuals, is crucial. HCPs should ensure environments and communication strategies are adapted to support patients effectively, acknowledging the increased likelihood of co-occurring conditions such as anxiety, depression, and gastrointestinal issues.

Policy makers and healthcare providers must prioritise training and resources to equip HCPs with the knowledge and skills necessary to recognise and address the complex needs of patients with co-occurring hEDS/HSD and neurodivergent conditions, ensuring equitable access to care and support services.

This approach will facilitate a more comprehensive and effective healthcare delivery, improving outcomes and quality of life for this underserved patient group.

Increasing awareness among healthcare professionals



Key Messages

- Education of HCPs is paramount in any care pathway aimed towards reducing the time to diagnosis for hEDS/HSD patients and improving the levels of care they experience.
- 82% of the survey population who spoke of their lived experience of healthcare reported having had an encounter with an unknowledgeable HCP.
- Almost half of the Scottish survey population reported feeling dismissed by a HCP. And 40% reported being made to feel as though they were imagining symptoms.

In 2020, the authors of this report surveyed Occupational Therapists and Physiotherapists working with hEDS/HSD patients in Scotland [67].

This work revealed that the majority of referrals received by therapists did not use the 2017 terminology, suggesting that knowledge of the updated criteria and terminology is lacking in referring HCPs.

Several therapists noted that the referrals they received often did not list hypermobility as a diagnosis at all, despite this being apparent during the assessment.

Less than one third of the respondents reported having previously had external hypermobility-specific training. Commendably, 95% of participants reported that they had undertaken self-directed training, and many reported that they were confident in the management of hEDS/HSD patients.

In contrast to the adult services, paediatric therapists reported better access to services such as pain management and clinical psychology, but half of the paediatric therapists surveyed were not aware of the paediatric guidelines for the assessment and treatment of hypermobility.



The majority of the therapists who were surveyed were in favour of a specialist centre for training and education of healthcare professionals [67]. It should be noted that the survey was distributed in 2019 to gauge the opinions of therapists after the application for a specialist centre for complex EDS patients was turned down.

“A specialist centre for the training and education of Health Care Professionals in Scotland is crucial. It would save hundreds if not thousands of people from years of unnecessary suffering and deterioration. This would save the NHS and Government huge expenses and allow those living with HSD/hEDS to have drastically improved health and quality of life. Left undiagnosed and unsupported people become a bigger drain on services!”

The lack of knowledge of hEDS/HSD in HCPs has been reported elsewhere. A

survey distributed to medical residents in Children's Mercy Hospital in Missouri found that HCPs had often heard of EDS but were not confident in the diagnosis, treatment, and management of patients [68].

A study of Flemish physiotherapists found that hEDS/HSD was considered difficult to manage and not well known, and that therapists desired more education in this subject area [69].



Patient experience of HCP knowledge and training

When asked to describe the lived experience of their hEDS/HSD diagnosis, many of the hEDS-START Scottish survey respondents spoke at great length about the lack of knowledge evident in primary and secondary care.

"I was left in the dark after being diagnosed. GPs know very little about the condition and will try to treat symptoms individually, or look for other underlying conditions, rather than listen to the patient."

82% of the respondents explicitly mentioned encounters with unknowledgeable HCPs when they were asked about their lived experience of diagnosis.

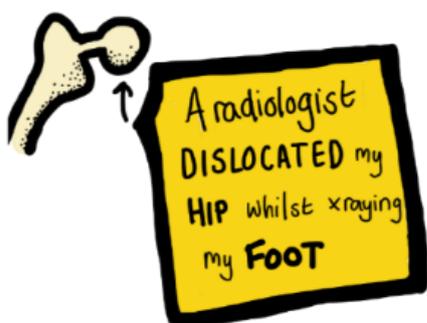
"The two consultants I saw 15 years apart were useless. The first said: "You should have joined the circus," and "don't bother coming back". The second sent me a letter when I was referred to him saying 'don't bother coming in - it's incurable'. Their lack of knowledge or concern is shocking. Even most GPs have no clue."

We asked respondents of the hEDS-START survey to tell us if they had a HSD/hEDS-aware medical professional that they could speak to if they had any questions, or if they had an emergency. The vast majority (88%) of those

respondents reported that they did not have any access to a knowledgeable HCP.

“My GP is very supportive but as he has no previous experience of EDS I ended up buying him a book about hEDS.”

When survey participants were asked if they believe there is value in a specialist centre for training and education of HCPs in Scotland, 98% of the respondents said ‘Yes’.



The impact of limited education and training of hEDS/HSD in healthcare

Lack of training and education of healthcare professionals does not just have the consequence of a lengthy

time to diagnosis for people living with hEDS/HSD.

It can lead to incorrect diagnoses being made, dangerous management techniques being utilised or recommended, and feelings of dismissal being reported by patients [8].

“I had several invasive procedures that probably caused me more pain and discomfort than they should have because I wasn't believed when I said it hurt”

A 2023 paper found that people with hEDS who report lower satisfaction with their healthcare providers experience a marked reduction in their health-related quality of life.

This research also found that the most commonly reported theme when patients were asked about what changes they would make to improve healthcare services was improved education and awareness of hEDS and HSD among HCPs [70].

“It would be preferable to know that regardless of where you live in Scotland that you can attend your local hospital and be met with people understanding, or at least willing to learn, rather than the gaslighting and current misdiagnosis that occurs ”

Previous research has shown that inadequate education and awareness of EDS among HCPs has the potential to contribute to a delay to diagnosis and a delay to access multidisciplinary care [47].

“I have had a terrible experience, even though I have been diagnosed there isn't really anyone who specialises in EDS so no one knows what to do”

Limitations in HCP knowledge and training can lead to significant barriers in patient access to appropriate support and educational materials about their diagnoses.

Clinical Guidelines

A major hurdle in improving the education and awareness levels in HCPs is the absence of appropriate guidelines from bodies such as the National Institute of Healthcare Excellence (NICE), its Scottish equivalent, the Scottish Intercollegiate Guidelines Network (SIGN), and the British Society for Rheumatology (BSR).

The BSR issued a statement in March 2020 concluding that there was insufficient evidence for them to develop a guideline for the management of hypermobility at present, despite a steady increase of evidence in the academic literature in recent years [71].

This statement also reported that NICE were considering developing guidance in response to previous concerns about the diagnosis of these conditions. The last update to the NICE Clinical Knowledge Summary for hypermobility in children was in May 2019 [72].

Equivalent guidance for adult diagnosis does not appear to exist at the present time.

The inaccessibility, and lack of awareness of, the EDS UK GP Toolkit is a further hurdle in the education and awareness of HCPs in primary care. The toolkit is an easily understood and comprehensive resource for HCPs with limited knowledge of hEDS/HSD. Improving access to and awareness of this toolkit would be a first step in improving knowledge levels, enabling HCPs to feel more confident in diagnosis and treatment.

Education and Training

There are some training programmes available to HCPs. The Hypermobility Syndromes Association (HMSA) provide training resources [73] and the Ehlers Danlos Society run a programme called Project ECHO, which is an accredited course for HCPs [74]. The British Society for Rheumatology (BSR) have a 90-minute webinar about HSD

on their website [75]. The main barriers to accessing this training are lack of awareness and management support of protected training time.

Although these programmes are available for the training and education of HCPs working with a hEDS/HSD patient population, enrolment is elective. We cannot, however, expect HCPs to do self-directed training in a subject area of which they have no prior knowledge.

“I often feel that I know more about EDS than the health care professionals I am talking too. Often this leaves me upset as I constantly have to reexplain things when I'm trying to have treatment. How can someone provide me with the right care when they don't even understand how my joints etc function. More education is needed for medical professionals.”

In 2022, the authors of this report wrote to the course organisers of the Medicine, Nursing, Physiotherapy,

Occupational Therapy, and Podiatry departments at twelve Scottish universities, asking if hEDS/HSD was included in their curriculums.

Only two respondents reported that hEDS/HSD was taught as part of their course, and both of those respondents stated that this teaching represented one hour or less of teaching time across the full degree programme.

Challenging Perceptions

One of the most common criticisms from people with hEDS/HSD relates to the lack of understanding, belief, or empathy from healthcare professionals with regards to their condition.

“Up until I hit my 30s I was completely dismissed and told it was just anxiety, laziness, made up, hormones or because I was overweight.”

In fact, 64% of the hEDS-START Scottish survey respondents explicitly mentioned the lack of belief from HCPs



when asked about their lived experience of diagnosis.

“I was told my pain was functional and the way this was described made me feel like they meant I was imagining it. I feel my age and various symptoms meant I was dismissed, I couldn't describe it perfectly or blood tests didn't show anything, so it was all in my mind.”

A further 48% reported feeling dismissed and 40% said that they were made to feel as though they were a hypochondriac or were imagining their symptoms.

This is in line with recent research from the United States in which 41% of participants reported feeling dismissed or made to feel as though their symptoms were in their head [70].

“We want to feel heard and understood, our pain acknowledged rather than constantly dismissed. Getting our pain and symptoms taken seriously, let alone clinically assessed is such a difficult and exhausting battle.”

39% of the Scottish survey respondents reported that they felt as though their symptoms had been ignored by a HCP, and 16% reported that they have experienced medical “gaslighting”, which in relation to healthcare is described as “a behaviour in which a physician or other medical professional dismisses or downplays a patient’s physical symptoms or attributes them to something else, such as a psychological condition” [76].

“The healthcare system in Scotland has let me down badly, in so many ways [...]. From symptoms being ignored for years, to weapons-grade medical gaslighting, to having to pay for years of expensive private medical investigative testing to try to uncover what was wrong with me. When I was eventually diagnosed, I thought that I would finally be able to access proper treatment. How wrong I was.”

In Conclusion

It is important that the education of HCPs underpins any future pathway of care, or regional service, for people living with hEDS/HSD. Education and training should be available to HCPs at all stages of their career. Healthcare professionals have previously expressed overwhelming support for the establishment of a centre dedicated to education and training of hEDS/HSD.

Wider research demonstrates a pervasive lack of confidence amongst

HCPs in the diagnosis and management of this patient population, leading to delayed diagnoses, incorrect and dangerous treatment approaches, and diminished health-related quality of life.

The high levels of disbelief reported may be due to the lack of education available to healthcare professionals, or simply to ingrained negative attitudes, but it is imperative that this is addressed moving forward.

The experiences of healthcare professionals and the healthcare system which have been reported by our study population do not reflect the core values of NHS Scotland [77]:

- Care and Compassion
- Dignity and Respect
- Openness, Honesty and Responsibility
- Equality and Teamwork

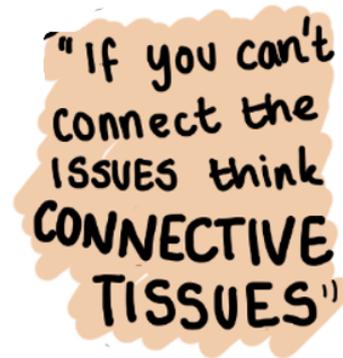
It is clear that experiences of dismissal, medical gaslighting, and disbelief are not limited to Scotland, or indeed the

UK. However, it is imperative that HCPs are adequately trained to engage with patients in a respectful manner, and equipped with an adequate degree of knowledge of these diagnoses in order to reduce the impact on patients.

The inclusion of hEDS/HSD in healthcare education curricula is minimal, necessitating the integration of these conditions into training programs to ensure future healthcare professionals are adequately prepared to diagnose and manage hEDS/HSD effectively.

The absence of comprehensive guidance from key healthcare bodies, and limited exposure to hEDS/HSD in undergraduate education further exacerbate the issue of a lack of understanding. Existing resources, such as the GP Toolkit, remain under-utilised, suggesting a need for improved dissemination and awareness efforts.

Towards an adequate pathway of care



Key Messages

- It is more than likely that all HCPs, regardless of discipline and specialty will come across patients with diagnosed or undiagnosed hEDS/HSD.
- HCP understanding of hEDS/HSD would provide the opportunity for timely and informative diagnosis, patient reassurance, and appropriate, high-quality multidisciplinary care.
- People living with hEDS/HSD believe that primary care would be the most beneficial place to receive a diagnosis.
- People living with hEDS/HSD want to be equipped with high-quality self-management tools, early in their diagnostic journey.

One of the biggest frustrations reported by people living with hEDS/HSD is a feeling of being “passed around” specialities without communication between departments, or being discharged from one specialty who is unable to treat their symptoms, with no onward referral to a specialist who could help.

“The main problem is that doctors only focus on one symptom and not the problem as a whole. It took my years to get a diagnosis and the only reason I got one is because I kept telling my doctors that my various health issues were related.”

At present, there is not an adequate pathway of care for adult patients with hEDS/HSD and there is limited



awareness amongst healthcare professionals about how to treat and manage these conditions.

The vast majority of referrals to secondary care in Scotland for hEDS/HSD patients at present are to Rheumatology, with 56% of the hEDS-START Scottish study population having accessed Rheumatology clinics in the last five years.

The majority of hEDS/HSD diagnoses are made in these secondary care Rheumatology services, and only 8% of diagnoses are made in primary care, despite the introduction of the GP toolkit in 2018.

Given the wide variety of symptoms and co-occurring conditions associated with hEDS/HSD, it might be more appropriate for primary HCPs to ascertain the most debilitating symptoms and prioritise secondary care referrals which will address those.

“Rheumatology is pointless if you’re needing physio, or if gastro symptoms are most prevalent. No need to have rheumatology involved in dealing with PoTS.”

All healthcare professionals should feel empowered in their ability to clinically diagnose hEDS/HSD early. Without a clear pathway of care or clinical guidelines, clinicians are limited in their ability to effectively diagnose and

“There is a big lack of joined-up working when it comes to hEDS that means I have spent years on waiting list and when I do finally see a specialist they more than often have never heard of EDS, which means that I am either completely dismissed or I am placed back on a waiting list.”

“The main problem is that doctors only focus on one symptom and not the problem as a whole. It took my years to get a diagnosis and the only reason I got one is because I kept telling my doctors that my various health issues were related.”

“I have a range of symptoms which are multi systemic and fluctuate in severity. I think many health professionals treat the symptoms in isolation and don't take a holistic or joined up approach.”

“There is no joined up thinking with care [...] My migraines have a huge impact on my life but it is the subluxation of joints which often triggers this [...], similarly with the problems I have faced with anaphylaxis and extreme histamine reactions my whole life, but no one is looking at the potential link with MCAS.”

“I feel that I really benefitted from seeing two specialists in England that worked together [...] The rheumatologist then worked with my genealogist in Scotland [...]. This made my life so much easier and meant that I felt that my care was coordinated and each specialist was able to help knowing what the others were doing. If this could have happened in Scotland on the NHS it would have been so much better”

The hEDS/HSD Patient Pathway Wishlist

At the “Translating Research into Change” patient engagement event, we asked participants to tell us what their ideal pathway of care would be, if they were given an opportunity to help policy makers and healthcare service managers with this. This discussion was illustrated and can be found on page 54.

- **Diagnosis in primary care where possible.** GPs should be willing and able to diagnose hEDS/HSD in primary care using the GP Toolkit. This would reduce time to diagnosis by protecting patients from often lengthy secondary care waiting lists.
- **Consistent coding of hEDS/HSD.** All patients with a diagnosis of hEDS/HSD should be coded as such so that their medical records are accurate and to ensure consistency of care across multiple specialities.
- **Newly diagnosed patients should be given age-appropriate information.** All patients who are given a diagnosis of hEDS/HSD should be equipped with high-quality materials with information on their diagnosis and a self-management toolkit.
- **If required, patients should be referred to a knowledgeable multidisciplinary team.** Not all patients will require referral to secondary care, particularly if diagnosed early. If specialist care is necessary, onward referral should be to an appropriate secondary care service, taking into account which symptoms have the most impact on health-related quality of life in the first instance.
- **When referring to other specialties, symptoms should not be communicated in isolation.** It is important that all

HCPs are made aware of a connective tissue disorder.

- **A coordinated care approach.**

Participants believed that a coordinated care approach would break repeated cycles of referral and discharge. They believed it was important that one specialty had oversight of hEDS/HSD care and that there was a single point of access to healthcare services.

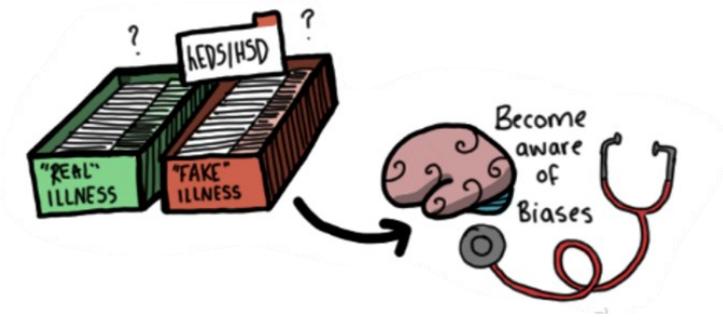
- **HCPs need appropriate and useful guidance to support decisions.** The development of NICE and SIGN guidance for treatment and management of adults with hEDS/HSD should be a priority. Guidance should include updated criteria to recognise hEDS/HSD and give advice on which symptoms can be managed in primary care and which symptoms require onwards referral.

- **All HCPs need better education on hEDS/HSD.** This should include how to assess a patient for hEDS/HSD and where to signpost patients for additional support.

- **Pro-active, not reactive care.** HCPs should be trained to look out for the symptoms and signs of hEDS/HSD and feel empowered to make the connection between seemingly unrelated symptoms.

- **Changing perspective and removing unconscious bias.** All HCPs should be equipped with the knowledge necessary to treat patients with respect and dignity.

- **Awareness of neurodivergence.** HCPs should consider the appropriateness of treatment strategies and environments. For example, telemedicine may not be appropriate for people with Autism or learning difficulties.



which means they also tend to require access to mental health services. Not that those are easily accessible either."

Although we presented this workshop as the development of a "wish list", it is our opinion that all of these points are actionable, and could be easily integrated into current regional services, provided that adequate and appropriate education and training is given to HCPs. Participants were made aware of the current budget limitations in NHS Scotland and therefore the ideas presented were realistic and pragmatic.

Some respondents to the hEDS-START survey spoke of a concern regarding the lack of specialist knowledge and care in rural areas of Scotland.

"The lack of specialist care in Scotland, especially rural areas, is quite frankly disgusting and only means that more strain is put on the NHS, as many patients end up depressed due to their health problems,

Future care pathways should ensure that provisions are put in place for the appropriate diagnosis and management of hEDS/HSD patients in every health board in Scotland.

"I'm worried about my future in a rural environment with no provision for hEDS as I get older, I'm already disabled at 37"

Discussions at the patient engagement event about telemedicine, being appointments with specialised HCPs over video call or telephone call, were enlightening. While some participants were enthusiastic about the option of accessing specialist care without travelling, others expressed reservations, explaining that such platforms may not be suitable for everyone.

In conclusion

Individuals with hEDS/HSD frequently cite frustrations with the lack of coordinated care available to them in NHS Scotland. The frequent narrative of being shuttled between specialties without effective communication or appropriate referrals highlights a fragmented approach to conditions which demand coordinated care.

It is likely, given the prevalence of hEDS and HSD, that all HCPs will come across patients with these conditions, and it is important that there is clear guidance and education available to clinicians in all specialties and disciplines. A considerable majority of patients are being diagnosed in secondary care services, despite a comprehensive and easily understood GP toolkit being available. There is a critical need for primary HCPs to play a pivotal role in early diagnosis and management.

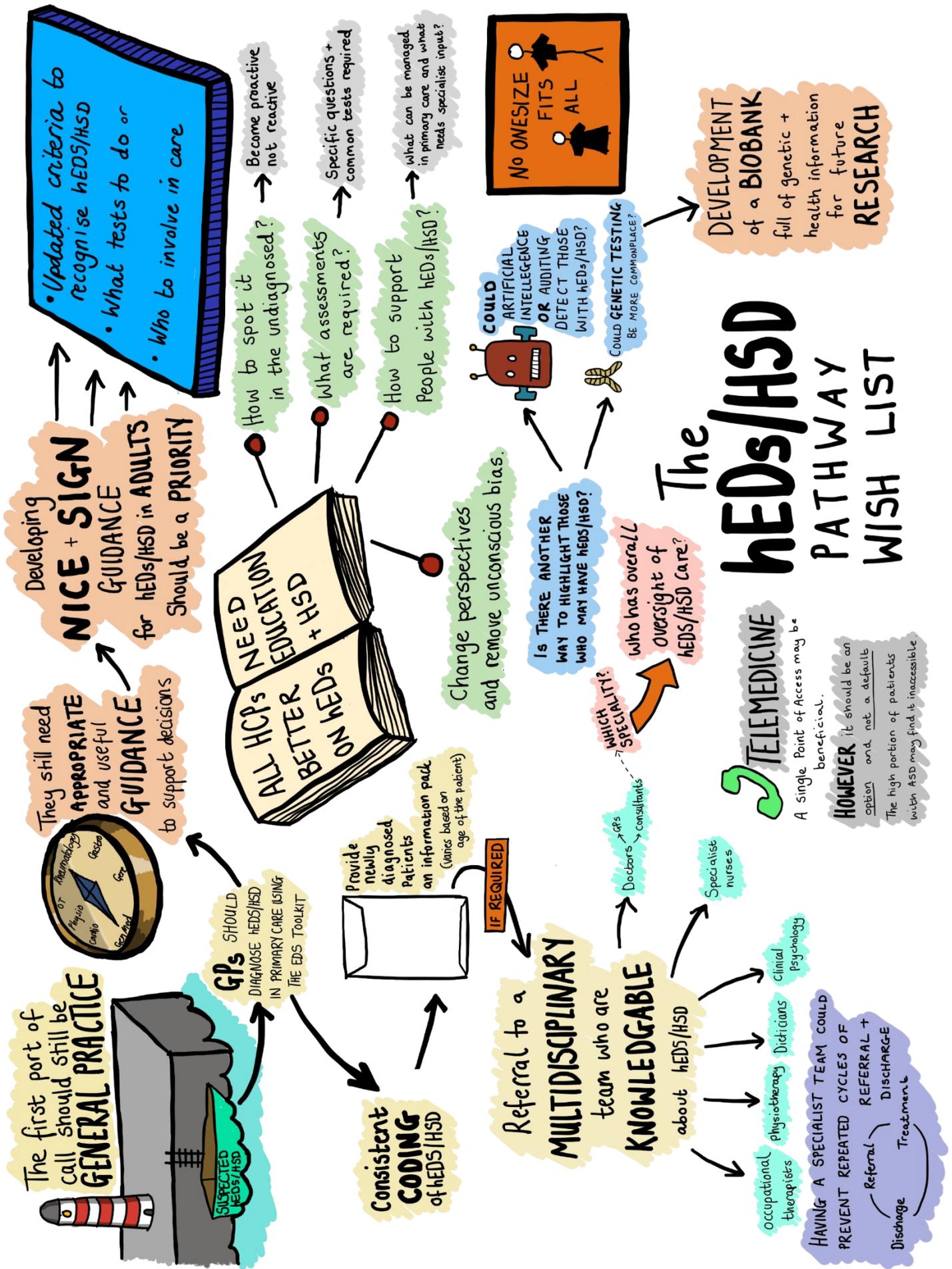
The “patient pathway wishlist” provides a pragmatic roadmap for overhauling

the care of hEDS/HSD patients. It advocates for an approach tailored to the individual’s more debilitating symptoms, challenging the existing siloed treatment paradigm.

For healthcare service managers and policy-makers, the message is clear: the absence of a care pathway for patients with hEDS/HSD demands attention and must be addressed in order to alleviate the prolonged and difficult diagnostic journey faced by patients in Scotland.

Implementing the changes that patients themselves want is manageable within the existing healthcare system.

The HEDS/HSD PATHWAY WISH LIST



The social impact of hEDS/HSD



Key Messages

- There is a significant financial implication of a diagnosis of hEDS/HSD, including the costs associated with private and out-of-country diagnoses, and self-funded equipment and treatment.
- There are high levels of unemployment, underemployment and educational interruption in a hEDS/HSD population.
- Loss of agency and independence causes a significant psychosocial impact on people living with hEDS/HSD.
- The mental health implications of a hEDS/HSD diagnosis in Scotland should be addressed with urgency.

Whilst there is a clear clinical need for improved services for people living with hEDS/HSD, it would be remiss of us not to discuss the enormous social and financial impact of these conditions on the people living with them.

“I have a diagnosis but still no effective treatment or testing unless I'm driving it in the private sector. It's not a good situation for those without private healthcare or the resources to fund treatment. It's wholly unacceptable that a patient has to research, fund and drive their own healthcare.”

It is commonly understood that having a diagnosis of chronic illness means that a person is likely to earn less than their peers, is more likely to be out of

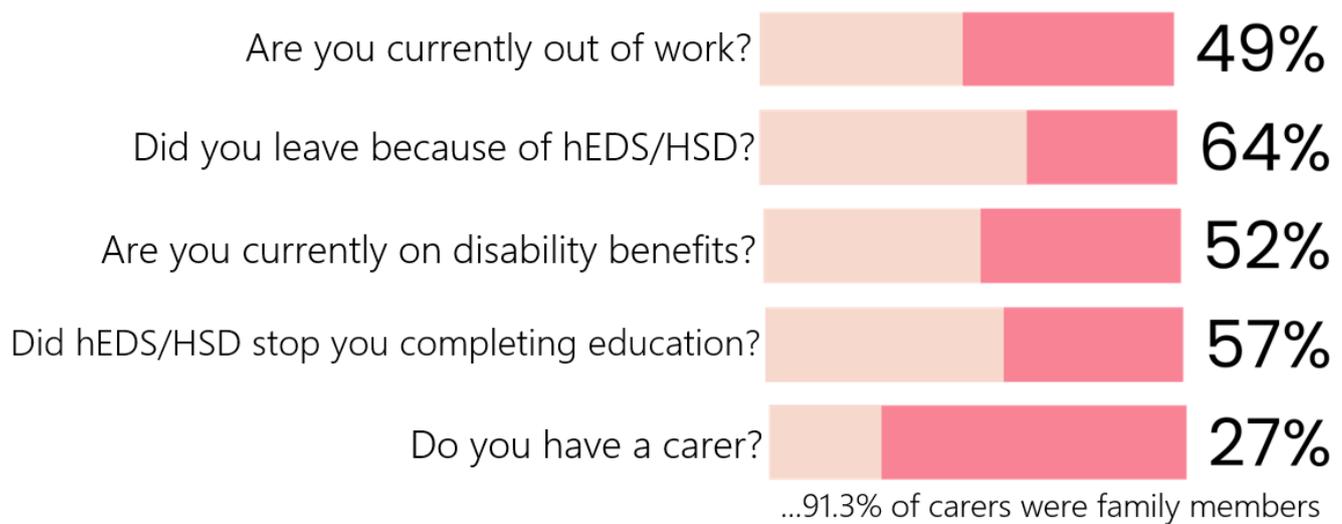


Figure 3: hEDS-START Survey 2023: The social impact of a hEDS/HSD Diagnosis

work, and is more likely to live in a low-income household [78].

The financial impact

One in five people with a hEDS/HSD diagnosis reported that they had to pay a private clinician for that diagnosis, increasing the mental and financial strain of living with a chronic condition.

"I am now pursuing a private referral to a rheumatologist but I'll probably never get past the initial consultation because I cannot afford to pay for further appointments and testing."

"I now have to save up to be seen privately in England which may take a few years to save the money I need, despite working full time (for the NHS) I cannot afford this cost straight away."

Within the hEDS-START Scottish survey population, 15% had been diagnosed outside of Scotland, and 24% had previously received treatment outside of the country.

"Having to travel vast distances to current specialist centres is not viable for everyone, especially when the NHS won't reimburse hotel fees for benefit recipients."

It comes as no surprise then, that many of the survey population reported anxieties related to the costs associated with having a hEDS/HSD diagnosis.

“I had to save up hundreds of pounds to see someone privately to be diagnosed. the NHS has never helped me with my health condition. I have been made to feel like I am lying and making up my symptoms. This has worsened my anxiety and made me have a negative view when seeing a health care professional.”

Of course, financial implications are not limited to the cost of clinical care and diagnosis; for example, 77% of the survey population reported that they required equipment or supports on a



regular basis. Of this number, almost three quarters reported that they had had to pay privately for their equipment, despite supports and equipment often being recommended by HCPs for the management of hypermobility-related symptoms.

One participant of the “Translating Research into Change” event spoke of their difficulty with regards to the wheelchair they had been given by the NHS:

“I was provided a wheelchair by the NHS. It is a manual wheelchair, and one of my major joints that dislocate is my shoulder so I cannot self-propel a wheelchair [...] independently we had to fund a power chair”

Many of the hEDS-START study participants also reported having to pay for private prescriptions and adjustments to their homes.

“I still have issues and pain but I've accessed a medical cannabis prescription

privately that manages the pain so much better than any of the NHS options. I'd grieved my mobility and independence and freedom and all I ever really needed was the right diagnosis and the knowledge that led to."

We asked hEDS-START survey participants about their employment status. Almost half of those who responded stated that they were out of work, with two thirds of those having left as a result of their hEDS/HSD diagnosis.

"I feel abandoned since my diagnosis. I feel like a discarded piece of rubbish. [...] My workplace offered to demote me to a desk position however I wasn't prepared to take a reduction in pay and job title. I'd worked there for over 20 years and progressed up the ladder to be one of their most valued employees so it felt like a slap in the face to me. Ultimately I was dismissed due to medical conditions and retired. I can honestly say I've never felt so alone and low in my life."

"A wrist injury that has not healed due to having hEDS caused me to GIVE UP MY CAREER."

People living with chronic illnesses in the UK are more likely to be unemployed and work part-time, reducing their earning potential [79].

"Earlier diagnosis and support would enable individuals to stay in work, remain independent and contribute to society."

In some cases, people living with hEDS/HSD are forced to change career or give their careers up completely due to unwillingness of employers to engage with flexible working arrangements.

“I am a highly qualified professional working in an investment bank. I have not been able to work for 5 years as I can't get any part time positions in my career [...] 8 years at university, 2 degrees and one professional degree - all for nothing because there is no part-time jobs.”

People with hEDS/HSD may find that their diagnosis causes professional restrictions, such as having to apply for work leave, or adapting their working day to accommodate the limitations imposed by their condition [80].

“The lack of health care means I have lost my job, I've had to drop out of a PgDip (1 day a week commitment), I have developed debt living on inefficient disability benefit and I've had to move home to try and pay back some of this debt, isolating myself further.”

One of the participants of the “Translating Research Into Change” event spoke of being passed up for a

promotion and missing out on professional opportunities, as a result of working part-time - something they have to do in order to preserve their energy and manage their symptoms.

“I only work part-time, so I'm earning less than the full salary per year, and I've also noticed that because I only work part-time [...] I wasn't eligible for a promotion and was passed over for getting managerial experience for a younger member of staff.”

More than half of the respondents reported that they were currently receiving benefits. Many of those surveyed reported the challenges they had faced when negotiating the social security system, such as applying for disability payments and personal independence payments.

“After I see Neurologist my costs will be over £1000 for private care. I have to go through a Tribunal for Adult Disability Payment. I believe Social Security Scotland do not understand how debilitating HSD and

Currently Receiving Benefits (%)

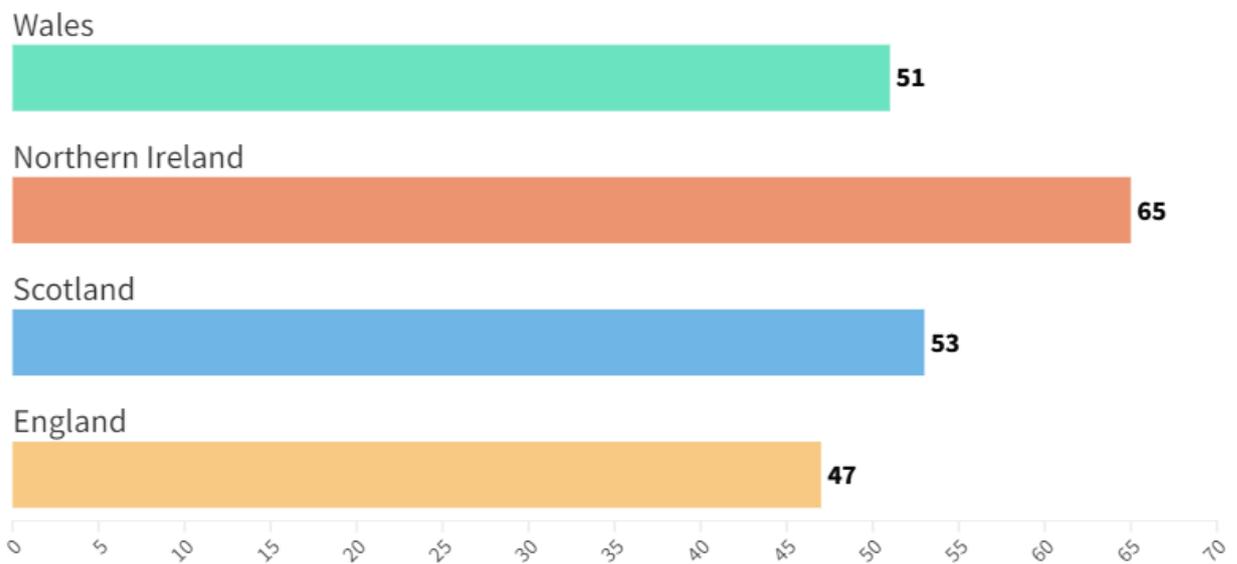


Figure 4: hEDS-START Study 2024: % respondents receiving benefits by country

the co-morbidities can be. I contacted them to point out errors and assumptions they made in their report.”

Some survey respondents spoke of the recent transition from Personal Independence Payments held by the Department of Work and Pensions (DWP) to the new Social Security Scotland Adult Disability Payments. Previous research has highlighted the difficulties experienced by people who face changes to their disability benefits, with Jessica Saffer et al describing the process as “unpredictable and unreliable” [81].

“I'm also scared as I wait to be ported over to the Scottish Social Security System to put myself through speaking to the DWP as they have not even recognised my lack of mobility post-surgery. I have to walk with a crutch and can't manage even walking from a car park to a building.”

Previous work has linked the process of applying or being assessed for disability payments to increased levels of stress and a negative impact on mental health [82, 83]. This is reflected in the hEDS-START survey responses.

“I am currently almost 90 days into my recent review for the transition from the English PIP to the Scottish adult disability system and am quite frankly terrified. I’ve had to battle to remain in my home of 30 years that I worked so hard for all of those years [...] These are examples of the additional stresses an already highly anxious, undiagnosed neurodivergent disabled person faces in the UK in 2023.”

The nature of hEDS/HSD means that people living with the conditions can experience symptoms that fluctuate and vary in severity over time, making it difficult for people who require social security to be assessed at a single timepoint.

“I also think that it's important for there to be UK wide education about dynamic disabilities, they are so poorly understood that disabled and ill people are often called "fakes" because their needs fluctuate. This affects their ability to access resources and it's almost a feature of the UK's disability benefits

system that they penalise folk with dynamic disabilities.”

The financial impact of a hEDS/HSD diagnosis cannot and should not be ignored by policy-makers. It is unacceptable that in a country with a national health service, people living with these conditions are having to pay out of pocket to receive private diagnoses and treatment, are forced to travel outside of the country for specialist treatment, and are paying for their own prescriptions and aids.

In light of the Equality Act 2010, it is difficult to accept that many employers remain unwilling to accommodate reasonable adjustments such as flexible or part-time working hours in order to retain highly skilled employees, and that employees in part-time positions feel they are disadvantaged by this.

Restrictions to benefits based on earning power may prevent highly skilled workers with a chronic illness

HUGE SOCIAL IMPACT

from returning to their profession, should they only be able to work on a part-time basis.

Difficulties in accessing social security and navigating the welfare system cause a great deal of stress and have huge impact on people's mental and physical wellbeing.

Impact on education

Previous research has shown that people living with a chronic illness are more likely to have worse academic outcomes, grade repetition, poorer educational attainment, and higher absenteeism when compared with a healthy control population [84].

"My schooling was badly affected, I had to leave one school and move to another, due to teachers insisting that I attend when I wasn't able to. My parents were reported to social services over my absence, this really affected me and I still feel that I am in trouble and causing trouble for my parents. I feel a burden and if only I could give myself a shake that everyone would be happy."

Work by Hiroki Ishiguro et al suggests that the presence of EDS, diagnosed or otherwise, can have a negative social effect on children's schooling. The authors note that symptoms can cause decreased self-esteem, parental overprotection and bullying from other children [25].

"I feel that with support I could have managed school and life but the obstacles put in my way made this impossible and this isolated me at a time I needed people around me."

More than half (53%) of the hEDS-START Scottish survey respondents reported that their education had been interrupted as a result of their diagnosis. Almost three fifths (57%) of those respondents went on to directly attribute the incompleteness of their education to their diagnosis of hEDS/HSD.

“I first took ill as I hit puberty, I was dismissed as a school avoider and each time I went to A&E made to feel that I was wasting their time and being a nuisance, this still lives with me and affects me daily.”

Difficulties in completing education can occur for a number of reasons. Pain and fatigue are the most common and debilitating symptoms of hEDS/HSD and can limit a person’s ability to sustain concentration and stamina, as well as their ability to engage with physically demanding education programmes:

“Studying to be a nurse is difficult, studying to be a

nurse with EDS is almost impossible.”

Other barriers to education encountered by our study population were related to the challenges implementing reasonable accommodations to their educational environments:

“It took two thirds of the first academic year to put into place my required adjustments so that I could access the course. Not to mention the number of accessible toilets, automatic doors etc, that were never working properly even after I reported them several times. I put a formal complaint in, but the University wouldn't take any responsibility. I did not finish my degree.”

Policy-makers should encourage educational institutions to work toward truly accessible environments, suitable for all students. Support should be easy to access and solutions implemented reasonable timeframes in order to support educational attainment in

EDS takes away your INDEPENDENCE and your DIGNITY to sometimes function fully in society. This also affects any other disabled person. We NEED you all to implement stringent welfare reform in order to protect a huge part of the population from SOCIAL + ECONOMIC DISADVANTAGE.

people living with chronic conditions. Awareness of hidden disabilities is crucial and suitable accommodations should be made for those who need it.

Loss of Agency

Loss of agency in people living with hEDS/HSD can manifest itself in many forms, including experiences of diminished control over their body and healthcare decisions, and experiences of struggling to have their voices heard in the healthcare system.

"I believe that I still suffer from the damage done while trying to figure out what was going on with me and it has affected how I see medical professionals, I see them as a last resort, I will only go to them if absolutely necessary."

An EDS UK report published in 2020 describes an "overwhelming" sense of young people feeling as though they not being helped in Scotland, and attributes much of this to the attitudes of HCPs.

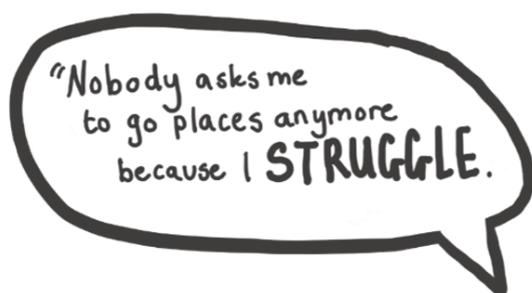
The 2020 EDS UK report describes loss of dreams, loss of self, and deprivation of agency as key themes identified in interviews with young people in Scotland.

Young people grieve the loss of an imagined future as an independent adult, and are "stripped of their sense of self through chronic traumatic experiences of healthcare, exclusion from society, and overdependence on their mothers" [2].

Similar sentiments were shared by Scottish respondents of the hEDS-START Study, with many describing a concurrent loss of confidence in both themselves and the healthcare system.

“It's taken me a long time to rebuild any trust with medical professionals as a result of this, and I have a lot of anxiety and loss of confidence in myself due to the way I was treated.”

Loss of agency is a significant consequence of the inadequate infrastructure surrounding the diagnostic journey for people with hEDS/HSD. People with these conditions report feeling dismissed, ignored, and disbelieved by HCPs, robbing them of their ability to self-advocate.



Loss of Independence

Loss of independence for people living with hEDS/HSD can manifest itself in many forms, including reduced physical mobility, dependence on others for support with self-care and daily activities, and the need for assistive devices such as mobility aids.

“I have had to purchase my own wheelchair, it gave me my independence back but I rely on my mum to assemble it, I have ended up with my mum being my carer in her retirement.”

The reliance on mobility aids and devices is common amongst people living with hEDs/HSD, with 77% of the hEDS-START Scottish survey population reporting the use of these. This can be seen as both a means to maintain independence but also as a marker of lost autonomy, as individuals may not be able to engage in activities without them.

“This condition has affected every single part of my life: my quality of life,

mobility, independence, friendships and relationships, education, career and studying, hobbies and interests, my family. Everything from the clothes I wear, to the food I eat. ”

The variability and severity of the symptoms related to hEDS/HSD can lead to a dependence on others to provide some level of care. 27% of the hEDS-START Scottish survey population reported that they currently had a carer. Of these, 91% reported that their carer was a family member. The loss of independence associated with these conditions can place a significant stress on people with hEDS/HSD and their households.

Loss of Relationships and Social Isolation

Loss of relationships and social isolation can stem from a number of factors associated with a diagnosis of hEDS/HSD. These conditions are often considered “hidden disabilities”, and patients report misunderstandings

from friends and family who do not realise the severity of the pain, fatigue, and physical limitations associated with the diagnoses.

“I have had arguments with colleagues, bosses and friends regarding my diagnosis and pain it causes as they find it hard to understand how it fluctuates in severity.”

Some of the hEDS-START participants described a loss of relationship with friends and family as a result of these misunderstandings.

“I am incredibly isolated and on my own, My friends do not understand either and most have left me. I "ask a lot of them", which I understand, but that it the nature of the condition, especially when you have gone through so much trauma your entire life and no longer have family members to ask for help.”

Social isolation can also be a consequence of the financial implication of a diagnosis of hEDS/HSD

as limited capacity to work and loss of income can impair a person's opportunities to socialise.

"I have no money and cannot enjoy any social interaction due to my pain nor do I have time for it. I am just trying to survive each day."

HCPs can play a crucial role in identifying loneliness and lack of social support in their hEDS/HSD patients, and should be willing and able to "socially prescribe" activities, support groups and services to combat feelings of isolation.



Loss of Self-Esteem

Loss of self-esteem in people with hEDS/HSD can manifest itself in various ways, including feelings of poor self-worth, feelings of societal burden, altered body image, and grief for their past, "healthy" self.

"I had to give up my home, I had to give up my job as a teacher, I had to give up my entire life, I had to give up my relationship."

hEDS/HSD are chronic conditions which often force people to constantly adapt to fluctuating symptoms which can lead to frustration with their bodies. Participants of the hEDS-START study spoke of feelings of guilt and burden for the perceived impact of their diagnosis on others, especially family.

"I have trust issues now and feel a burden and that why would anyone bother with me as I'm not worth it."

These feelings appear to begin in childhood, with many respondents

describing difficulty being believed by schoolteachers, which then persist into encounters with healthcare professionals into adulthood.

“I still feel the guilt of the problems I caused for my parents by not being normal and fitting in, I now dismiss my pain or health issues as not be relevant and have to be reminded that they are and they are real.”

Unrelenting feelings of being disbelieved and dismissed by people in positions of “authority”, such as teachers, parents, and HCPS, undoubtedly have a lifelong impact on a person’s sense of self-worth and agency.

“It was hard as a teenager to be ignored, not believed and let down, I feel I will carry feelings of not being good enough or letting everyone down for my life, this affects my daily life.”

Positive interactions with HCPs and the implementation of support structures for children and young people with

chronic illness would go a long way to reducing the negative social and psychological impact of these “hidden”, dynamic health conditions.

“If the NHS had spent as much time and resources supporting me as they have in gaslighting and denials my outlook would be much better than it is now, the utter short-sightedness has significantly impacted my societal contribution.”

By receiving validation of their experiences, symptoms, and limitations, individuals may experience improvements in their self-perception and be empowered to manage their condition more effectively.

Feelings of Shame and Stigma

One of the core psychosocial challenges faced by many people living with hEDS/HSD is the invisibility of their condition. The most common and debilitating symptoms of pain and fatigue are not immediately apparent to others. This can lead to disbelief and invalidation of their experiences,

leadings of feelings of not being able to fulfil societal expectations.

"I find that there's often a lot of internalised shame and guilt, my life is no longer reliable, an injury or illness can happen in a moment and I am not able to keep every appointment or plan which is difficult."

Attitudes of HCPs can exacerbate feelings of stigma and shame by invalidating a person's lived experience. Almost two thirds of the hEDS-START Scottish survey respondents reported a lack of belief from a HCP. Almost half reported previously feeling dismissed by a HCP, and 40% spoke of being made to feel like their symptoms were all in their head.

"The stigma attached to my illnesses particularly EDS is frustrating, I have so many ailments and no one is joining the dots or helping me, I got told by a doctor "there is nothing physically wrong with you, stop coming to the doctors, it's all in your head!"

Feelings of stigma in this patient population go beyond encounters with healthcare professionals. Bennett et al describe facets of social stigma faced by people with hEDS, such as: (1) difficulty keeping up with family and friends, (2) judgements of others which can lead to feelings of shame and guilt, and (3) hiding symptoms from others [85]. Social stigma can also come from pervasive opinions of "hidden" chronic conditions online:

"I sometimes feel embarrassed by my diagnosis because of the way it is shown online; as an identity, and is associated with faking or dwelling in symptoms."

Stigma and shame can have an exceptional psychological impact on people, and in some cases can lead to increased stress levels [86] and depression [87]. HCPs should be aware of the impact that symptom invalidation and disbelief can have on a person with any chronic condition.

Impact on Mental Health

The impact of the current lived experience of hEDS/HSD on people's mental health is vast and multifaceted.

The rates of mental health diagnoses in this patient population are already high, with 68% of the Scottish survey population reporting a diagnosis of anxiety, and 63% reporting a diagnosis of depression. A further 20% met the criteria for current psychological distress, when assessed using the PHQ4. People have a myriad of symptoms that can affect their mental well-being, including but in no way limited to debilitating chronic pain and fatigue.

Impacts on individuals' mental health are exacerbated by the health and social contexts in which people living with these diagnoses reside. Time to diagnosis in Scotland remains lengthy, which leads to feelings of distress and frustration, and exacerbates chronic illness.

Disbelief and dismissal from HCPs leads to feelings of distrust, and can increase feelings of medical stigmatisation, which in turn can prevent people from seeking and accessing mental health support services when required.

Previous research has suggested that people with hEDS are 2-3 times more likely to attempt suicide than those without the diagnosis [26]. This alarming statistic points to the urgent need for adequate and informed healthcare, and accessible social support.

"I have had two suicide attempts. If there was appropriate health care I would be able to have purpose in my life, I'd be able to manage my EDS enough to keep a job. I would have been able to complete my studies and not have such financial burdens adding to my anxiety and difficulties just trying to live."

"I live a very lonely debilitating life. I am forced to work full time which is causing significant pain and have carers. ...I am suicidal a lot and don't have anyone to talk to. I suffer C-PTSD from medical appointments and would rather suffer and die when the time comes than fight with GP and attend hospital to be told I am wrong and there is nothing wrong with me."

"I'm fighting compulsion to self-harm and draw the pain out"

During the pandemic I lost another job. I had amazing employers and I loved the job. They tried everything they could do to keep me in the position, but after 2 years I was still not getting any appropriate health care from the NHS and the OH had signed me off as unfit for work for the third time and I had attempted suicide. I had to leave.

"I am scared and alone. I cannot manage my condition. I can't clean my flat. I can't afford to get someone to come and help me."

In Conclusion

This work illustrates the vast social and emotional impact of the lived experience of people with hEDS/HSD.

These diagnoses necessitate a multidimensional approach in addressing the clinical, social, and financial burdens held by people with these conditions.

The financial implications of a hEDS/HSD diagnosis extend far beyond the common costs of private healthcare, affecting people's employment opportunities and earning potential.

The reality that a large fraction of hEDS/HSD individuals are unemployed or underemployed as a direct result of their diagnosis underscores the need for policy reforms that accommodate flexible working arrangements.

The academic impact of a hEDS/HSD diagnosis should not be overlooked. Students frequently encounter barriers to learning, exacerbated by physical

limitations and absenteeism directly attributable to symptoms and co-occurring conditions. Policymakers should advocate for and implement policies that address these educational disparities.

The loss of agency and independence reported underscores the importance of patient-centred care and systemic reforms within healthcare and social support systems. The psychosocial impact (isolation, lowered self-esteem, stigma, and shame) necessitates comprehensive support services and training programmes for HCPs.

The mental health implications of these conditions are profound, with high rates of mental health diagnoses and psychological distress, exacerbated by disbelief and dismissal from HCPs, and worsened within the current Scottish context.

These issues require urgent review and attention from policy-makers and healthcare providers.

'hEDS/HSD: Translating Research into Change' Patient Engagement Day

On the 8th February 2024, we invited EDS UK members in Scotland to a patient engagement day at the Institute of Genetics and Cancer at the University of Edinburgh. The following pages capture some of the outputs of this event.

We asked participants to tell us:

- (1) If you could speak to a policy maker, what would you want to say to them?
- (2) Which values should underpin a regional service in Scotland?
What resources would you like to see available to hEDS/HSD patients?

- (3) What else would you like to see in research? What are we not showing that you have experienced?

Some of the responses to those questions are depicted on the following three pages. They offer an insight into what people living with hEDS/HSD want to see in policy, healthcare, and research.

We also asked participants of the event to indicate whether they agreed, disagreed, or neither to the same statements we asked the EDS UK members based on the Scottish Government's own Health and Social Care Standards. Those responses can be found on page 77.

Not one participant of the event indicated a positive response to any of those questions, indicating profound dissatisfaction with their experiences of the healthcare system with regards to their diagnosis.



Give Scotland a proper ALLERGY service

If you could speak to a Policy maker, what would you want to say to them?

We **DESPERATELY** need services + support

Be mindful of the **SOCIAL** aspects of EDS.

The need to support disabled people who may be **ISOLATED** to engage with others and support mental health + wellbeing.



X
OFFER GENETIC SCREENING

IF I HAVE HUMAN RIGHTS WHY DO I FEEL SUB-HUMAN?

LISTEN TO US + LISTEN TO RESEARCH



GIVE ALL HCPS A CLEAR PATHWAY FOR APPROPRIATE CARE

"HAD HEALTH PROFESSIONALS BEEN INFORMED OF HEDS MAYBE IT COULD HAVE BEEN CAUGHT EARLIER RATHER THAN DIAGNOSED RANDOMLY AT PRIVATE HOSPITALS FOR OTHER ISSUES."
- DIAGNOSED AT 43yrs

PATIENTS are more important than **MONEY!**

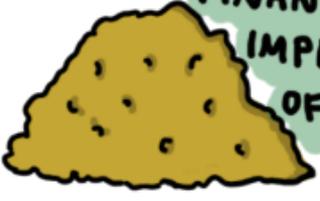
EDS takes away your **INDEPENDENCE** and your **DIGNITY** to sometimes function fully in society. This also affects any other disabled person. We **NEED** you all to implement stringent welfare reform in order to protect a huge part of the population from **SOCIAL + ECONOMIC DISADVANTAGE**.

"GIVE ME MY LIFE BACK"
I want to live, not spend my life waiting for the right medical care.

There is no FOLLOW UP after diagnosis



GIVE CONSIDERATION TO THE FINANCIAL IMPLICATIONS OF HEDS/HSD



MAKE ACCESS TO PIP/ADP EASIER FOR HIDDEN DISABILITIES

ACCESS TO ELECTED OFFICE FUND
Do Demographics **THOROUGHLY** throughout **NOT JUST AT THE END.**



Which values should underpin a regional service in Scotland?

What resources would you like to see available to hEDS/HSD patients?

RESPECT
"Believing the patient"

Respect Alert tags + Medical anxiety

BE PROACTIVE
NOT REACTIVE

"NOTHING ABOUT US, WITHOUT US!"

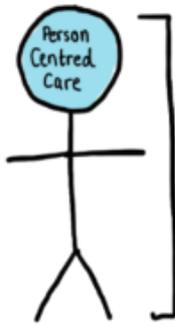
SEE THE PATIENT NOT THE CONDITION

IMPROVE

QUALITY OF LIFE
NOT JUST SEARCHING FOR A CURE



LISTEN
TO PATIENTS +
DO NOT DISMISS THEM



A holistic approach as all systems are affected. **1 on 1**

Practice existing NHS VALUES

Designated **ADVOCACY** workers



DEVELOP CARE PLANS WITH PATIENTS

SUPPORT WITH THE ADULT DISABILITY PAYMENT PROCESS



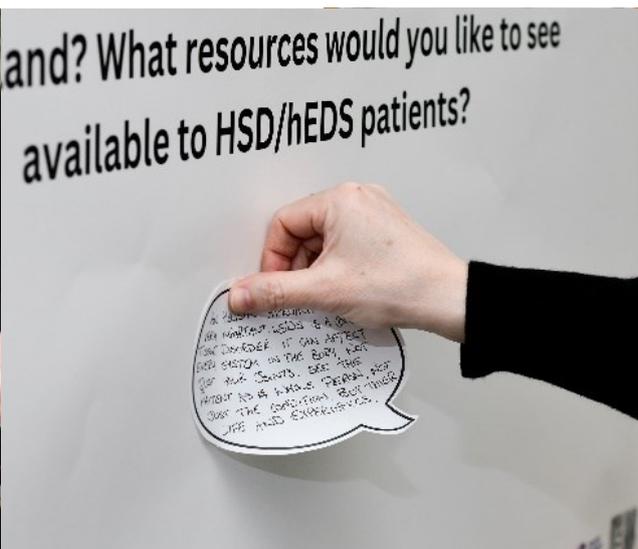
Animal Therapy



Access to **ALTERNATIVE THERAPIES**

- mental health
- pain review
- genetics
- cardiology
- Dietician
- neurology
- occupational therapy
- physiotherapy

A multidisciplinary Team Approach



and? What resources would you like to see available to HSD/hEDS patients?

In a multidisciplinary team approach, it is important to have people from different backgrounds and specialties working together to provide the best care for our patients. This includes not only medical professionals but also support staff, therapists, and other healthcare workers. It is essential to have a good communication and collaboration between all team members to ensure the best possible outcomes for our patients.

What would you like to see in RESEARCH?

♀ issues with women's health + HEDS/HSD including PREGNANCY

USE OF MULTIDISCIPLINARY TEAMS FOR HEDS CARE

AWARENESS OF HEDS IN HEALTHCARE PROFESSIONALS BROKEN DOWN BY HEALTH BOARDS

How we can be Proactive in our care?

THE PREVALENCE + LINK BETWEEN HEDS AND OTHER CONDITIONS (+ how they can help highlight potential sufferers)

The effectiveness of treatments in HEDS + how is ABSORPTION affected due to GASTROINTESTINAL issues

Compression Technologies

EDUCATIONAL PROGRAMMES FOR PATIENTS + HCPs

GENETIC + FAMILIAL TESTING + SCREENING

IMMUNOLOGICAL EFFECTS of HEDS

The mental impact of EDS + misdiagnosis

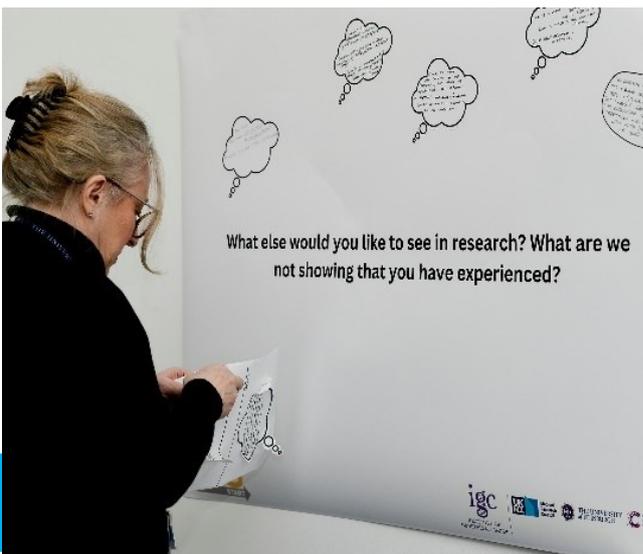
The extent of CONSCIOUS + UNCONSCIOUS BIAS in HCPs.

The rate of MISDIAGNOSIS

IMPROVED DIAGNOSTIC CRITERIA

NEUROLOGICAL ISSUES

- CSF
- neuropathy
- Sinuses
- neural rewiring
- Dysautonomia
- Atlanto-axial instability
- 'Slippy spine'
- Tethered cord



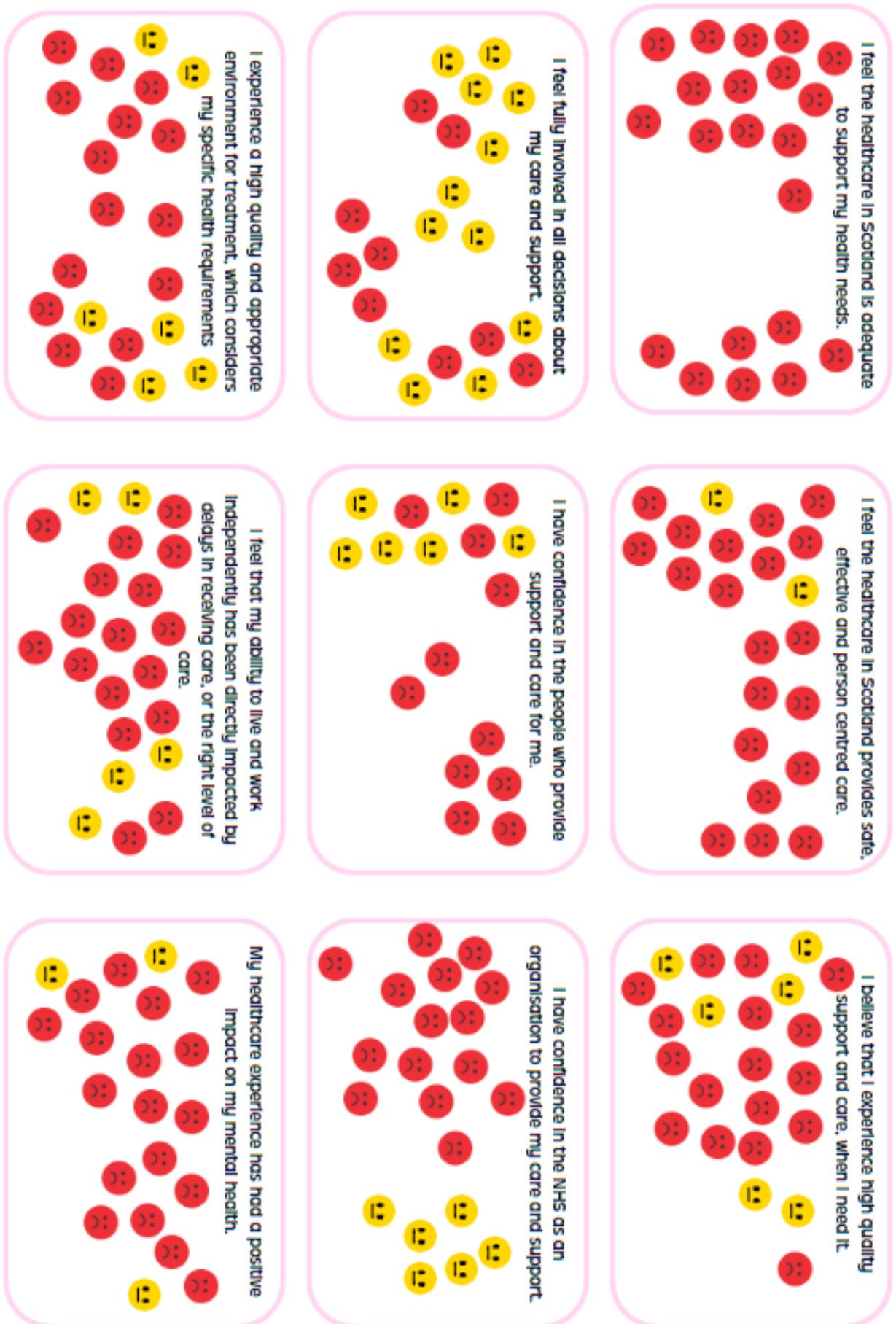


Figure 5: 'Translating Research Into Change' event participant responses to the Health and Social Care Standards Statements. Responses have been digitised for clarity. Yellow faces indicate neutrality, while red faces indicate disagreement.

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This report, and the data it contains, would not have been possible without the input from members of the public with lived experience of hEDS/HSD who volunteered their time to answer the hEDS-START survey and participate in our engagement event,

This work would have been very difficult without the support of our EDS UK colleagues, Anne Sutherland and Michele Goldie. Their commitment to improving the lives of people living with EDS/HSD in Scotland is unwavering, and highly commendable.

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This has been both an inspiring and distressing project to bring to fruition but we hope that our research will initiate a change in the current healthcare system for hEDS/HSD patients to improve health related quality of life.

*Kathryn Berg &
Dervil Dockrell*

Kathryn Berg, Research Manager
Dervil Dockrell, Occupational Therapist

Works Cited

1. Malfait F, Francomano C, Byers P, Belmont J, Berglund B, Black J, Bloom L, Bowen JM, Brady AF, Burrows NP, Castori M. The 2017 international classification of the Ehlers–Danlos syndromes. In *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* 2017 Mar (Vol. 175, No. 1, pp. 8-26).
2. Martin A-M. EDS and Young People in Scotland: The Agency Needed to Prosper, [Internet]. EDS UK; 2020 [cited 2024 Apr 19]. Available from: <https://www.ehlers-danlos.org/wp-content/uploads/2020/12/EXECUTIVE-SUMMARY-EDS-Scot-SINGLE-PAGES.pdf>
3. Demmler JC, Atkinson MD, Reinhold EJ, Choy E, Lyons RA, Brophy ST. Diagnosed prevalence of Ehlers–Danlos syndrome and hypermobility spectrum disorder in Wales, UK: a national electronic cohort study and case–control comparison. *BMJ Open* [Internet]. 2019 Nov;9(11):e031365. Available from: <https://bmjopen.bmj.com/content/9/11/e031365>
4. Frankel B. Healthcare Use by Women with Joint Hypermobility in Northumberland* [Internet]. <https://northeastnorthcumbria.nhs.uk/women-s-health-conference-2023/>. NHS Northeast and North Cumbria; 2023 [cited 2024 Apr 2]. Available from: <https://northeastnorthcumbria.nhs.uk/media/tyznrina/nenc-womens-health-conference-abstract-summary-final.pdf>
5. Health and Social Care Standards: My support, My life,“ [Internet]. Scottish Government; 2017 [cited 2024 Apr 2]. Available from: <https://www.gov.scot/publications/health-social-care-standards-support-life/pages/3>
6. The Ehlers–Danlos Syndromes GP Toolkit [Internet]. The Ehlers–Danlos syndromes (EDS) GP Toolkit. [cited 2024 Apr 23]. Available from: <https://gptoolkit.ehlers-danlos.org/>
7. Waiting Times [Internet]. NHS Inform. [cited 2024 Mar 20]. Available from: <https://www.nhsinform.scot/waiting-times>
8. Halverson CM, Cao S, Perkins SM, Francomano CA. Comorbidity, misdiagnoses, and the diagnostic odyssey in patients with hypermobile Ehlers–Danlos Syndrome. *Genetics in Medicine Open*. 2023 Jan 1;1(1):100812.
9. Kalisch L, Hamonet C, Bourdon C, Montalescot L, de Cazotte C, Baeza-Velasco C. Predictors of pain and

- mobility disability in the hypermobile Ehlers–Danlos syndrome. *Disability and Rehabilitation*. 2019 May 7;42(25):3679–86.
10. Cederlöf M, Larsson H, Lichtenstein P, Almqvist C, Serlachius E, Ludvigsson JF. Nationwide population-based cohort study of psychiatric disorders in individuals with Ehlers–Danlos syndrome or hypermobility syndrome and their siblings. *BMC Psychiatry*. 2016 Jul 4;16(1).
 11. Syx D, Inge De Wandele, Lies Rombaut, Fransiska Malfait. Hypermobility, the Ehlers-Danlos syndromes and chronic pain. *PubMed*. 2017 Oct 3;35 Suppl 107(5):116–22.
 12. Gullapalli PA, Javed S. Multidisciplinary chronic pain management strategies in patients with Ehlers–Danlos syndromes. *Pain Management*. 2022 Oct 28;13(1).
 13. Zarate N, Farmer AD, Grahame R, Mohammed SD, Knowles CH, Scott SM, et al. Unexplained Gastrointestinal Symptoms and Joint hypermobility: Is Connective Tissue the Missing link? *Neurogastroenterology & Motility*. 2010 Mar;22(3):252-e78.
 14. Bendik EM, Tinkle BT, Al-shuik E, Levin L, Martin A, Thaler R, et al. Joint hypermobility syndrome: A common clinical disorder associated with migraine in women. *Cephalalgia*. 2011 Jan 28;31(5):603–13.
 15. McIntosh LJ, Mallett V, Frahm JD, Richardson D, Evans MI. Gynecologic disorders in women with Ehlers–Danlos syndrome. *Journal of The Society for Gynecologic Investigation*. 1995 Jun 1;2(3):559–64.
 16. Carley ME, Schaffer J. Urinary incontinence and pelvic organ prolapse in women with Marfan or Ehlers–Danlos syndrome. *American Journal of Obstetrics and Gynecology*. 2000 May;182(5):1021–3.
 17. Thwaites PA, Gibson PR, Burgell RE. Hypermobile Ehlers–Danlos syndrome and disorders of the gastrointestinal tract: What the gastroenterologist needs to know. *Journal of Gastroenterology and Hepatology*. 2022 Jul 20;182(5).
 18. Choudhary A, Fikree A, Aziz Q. Overlap between Irritable Bowel Syndrome and Hypermobile Ehlers–Danlos syndrome: an Unexplored Clinical phenotype? *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. 2021 Nov 6;187(4):561–9.
 19. Choudhary A, Vollebregt PF, Aziz Q, Scott SM, Asma Fikree. Rectal hyposensitivity: a common pathophysiological finding in patients with constipation and associated hypermobile Ehlers–Danlos syndrome. *Alimentary Pharmacology & Therapeutics*. 2022 Jun 27;56(5):802–13.
 20. Arunkalaivanan AS, Morrison A, Jha S, Blann A. Prevalence of urinary and faecal incontinence among

- female members of the Hypermobility Syndrome Association (HMSA). *Journal of Obstetrics and Gynaecology: The Journal of the Institute of Obstetrics and Gynaecology* [Internet]. 2009 Feb 1 [cited 2020 Feb 15];29(2):126–8. Available from: <https://www.ncbi.nlm.nih.gov/pubmed/19274546/>
21. National Institute for Health and Care Excellence,. 2024 [cited 2024 Mar 22]. Available from: <https://cks.nice.org.uk/topics/generalized-anxiety-disorder/background-information/prevalence>
 22. Bromet E, Andrade LH, Hwang I, Sampson NA, Alonso J, de Girolamo G, et al. Cross-national epidemiology of DSM-IV major depressive episode. *BMC Medicine*. 2011 Jul 26;9(1).
 23. Kennedy M, Loomba K, Ghani H, Riley B. The psychological burden associated with Ehlers-Danlos syndromes: a systematic review. *Journal of Osteopathic Medicine*. 2022 Apr 14;0(0).
 24. Bulbena A, Pailhez G, Bulbena-Cabr e A, Mallorqu -Bagu  N, Baeza-Velasco C. Joint hypermobility, anxiety and psychosomatics: two and a half decades of progress toward a new phenotype. *Clinical challenges in the biopsychosocial interface*. 2015;34:143-57.
 25. Kroenke K, Spitzer RL, Williams JBW, Lowe B. An Ultra-Brief Screening Scale for Anxiety and Depression: The PHQ-4. *Psychosomatics*. 2009 Nov 1;50(6):613–21.
 26. L we B, Wahl I, Rose M, Spitzer C, Glaesmer H, Wingenfeld K, et al. A 4-item measure of depression and anxiety: Validation and standardization of the Patient Health Questionnaire-4 (PHQ-4) in the general population. *Journal of Affective Disorders*. 2010 Apr;122(1-2):86–95.
 27. Ishiguro H, Yagasaki H, Horiuchi Y. Ehlers-Danlos Syndrome in the Field of Psychiatry: A Review. *Frontiers in Psychiatry*. 2022 Jan 11;12.
 28. Wasim S, Suddaby JS, Parikh M, Leylachian S, Ho B, Guerin A, et al. Pain and gastrointestinal dysfunction are significant associations with psychiatric disorders in patients with Ehlers–Danlos syndrome and hypermobility spectrum disorders: a retrospective study. *Rheumatology International* [Internet]. 2019 Mar 28 [cited 2019 Jul 11];39(7):1241–8. Available from: <https://link.springer.com/article/10.1007/s00296-019-04293-w>
 29. B nistan K, Martinez V. Pain in hypermobile ehlers-danlos syndrome: New insights using new criteria. *American Journal of Medical Genetics Part a*. 2019 May 10;179(7).
 30. The Migraine Trust. The State of the Migraine Nation Who is living with migraine in the UK? Rapid research review [Internet]. Migraine Trust. 2020 Jan [cited 2024 Apr 23]. Available from:

<https://migrainetrust.org/wp-content/uploads/2021/08/State-of-the-Migraine-Nation-population-rapid-review.pdf>

31. 87. Yair Zloof, Simchoni M, Derazne E, Tsur AM, Dorit Tzur, Braun M, et al. Hypermobility spectrum disorders and active migraine in Israeli adolescents: A nationwide study. *Headache*. 2023 Jun 14;63(7):934–41.
32. Chiari Malformation [Internet]. NHS UK Conditions. National Health Service; 2022 [cited 2024 Mar 27]. Available from: <https://www.nhs.uk/conditions/chiari-malformation>
33. Tam SKP, Chia J, Brodbelt A, Foroughi M. Assessment of patients with a Chiari malformation type I. *Brain and Spine*. 2022;2:100850.
34. Rowe PC, Barron DF, Calkins H, Maumenee IH, Tong PY, Geraghty MT. Orthostatic intolerance and chronic fatigue syndrome associated with Ehlers-Danlos syndrome. *The Journal of Pediatrics*. 1999 Oct;135(4):494–9.
35. Tidy C. Postural Orthostatic Tachycardia Syndrome [Internet]. Patient. 2021 [cited 2024 Mar 26]. Available from: available: <https://patient.info/doctor/postural-tachycardia-syndrome-PoTS-pro>.
36. Roma M, Marden CL, De Wandele I, Francomano CA, Rowe PC. Postural tachycardia syndrome and other forms of orthostatic intolerance in Ehlers-Danlos syndrome. *Autonomic Neuroscience*. 2018 Dec;215:89–96.
37. Mitral Valve Prolapse [Internet]. PennMedicine. PennMedicine; [cited 2024 Mar 26]. Available from: <https://www.pennmedicine.org/for-patients-and-visitors/patient-information/conditions-treated-a-to-z/mitral-valve-prolapse>
38. Essayagh B, Sabbag A, Antoine C, Benfari G, Batista R, Yang LT, et al. The Mitral Annular Disjunction of Mitral Valve Prolapse. *JACC: Cardiovascular Imaging*. 2021 Jun;14(11).
39. "Understanding Raynaud's [Internet]. 2018 [cited 2024 Apr 19]. Available from: https://www.sruk.co.uk/media/filer_public/5b/25/5b25678b-177c-4ce4-ad2e-4fb1b8ada855/sruk_understanding_raynauds.pdf
40. Patel M, Khullar V. Urogynaecology and Ehlers–Danlos syndrome. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. 2021 Nov 20;187(4):579–85.
41. Mastoroudes H, Giarenis I, Cardozo L, Srikrishna S, Vella M, Robinson D, et al. Lower urinary tract

- symptoms in women with benign joint hypermobility syndrome: a case–control study. *International Urogynecology Journal* [Internet]. 2013 Feb 23 [cited 2020 Jan 12];24(9):1553–8. Available from: <https://link.springer.com/content/pdf/10.1007%2Fs00192-013-2065-3.pdf>
42. Pearce G, Bell L, Pezaro S, Reinhold E. Childbearing with Hypermobility Ehlers–Danlos Syndrome and Hypermobility Spectrum Disorders: A Large International Survey of Outcomes and Complications. *International Journal of Environmental Research and Public Health*. 2023 Oct 21;20(20):6957–7.
 43. Hugon-Rodin J, Lebègue G, Becourt S, Hamonet C, Gompel A. Gynecologic symptoms and the influence on reproductive life in 386 women with hypermobility type ehlers-danlos syndrome: a cohort study. *Orphanet Journal of Rare Diseases*. 2016 Sep 13;11(1).
 44. Alrifai N, Laith Alhuneafat, Jabri A, Muhammad Umar Khalid, Xiarepati Tieliwaerdi, Faris Sukhun, et al. Pregnancy and Fetal Outcomes in Patients With Ehlers–Danlos Syndrome: A Nationally Representative Analysis. *Current problems in cardiology*. 2023 Jul 1;48(7):101634–4.
 45. Castori M, Celletti C, Camerota F, Grammatico P. Chronic fatigue syndrome is commonly diagnosed in patients with Ehlers-Danlos syndrome hypermobility type/joint hypermobility syndrome. *PubMed*. 2011 Jul 5;29(3):597–8.
 46. Barron DF, Cohen BA, Geraghty MT, Violand R, Rowe PC. Joint hypermobility is more common in children with chronic fatigue syndrome than in healthy controls. *The Journal of Pediatrics*. 2002 Sep;141(3):421–5.
 47. Hakim A, De Wandele I, O’Callaghan C, Pocinki A, Rowe P. Chronic fatigue in Ehlers-Danlos syndrome-Hypermobility type. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* [Internet]. 2017 Feb 10 [cited 2019 Oct 13];175(1):175–80. Available from: <https://onlinelibrary.wiley.com/doi/full/10.1002/ajmg.c.31542>
 48. Voermans NC, Knoop H, van de Kamp N, Hamel BC, Bleijenberg G, van Engelen BG. Fatigue is a frequent and clinically relevant problem in Ehlers-Danlos Syndrome. *Seminars in Arthritis and Rheumatism* [Internet]. 2010 Dec 1 [cited 2021 May 5];40(3):267–74. Available from: <https://pubmed.ncbi.nlm.nih.gov/19878973/>
 49. What is Mast Cell Activation Syndrome? [Internet]. *Mast Cell Action*. [cited 2024 Mar 27]. Available from: <https://www.mastcellaction.org/about-mcas>.
 50. Csecs JLL, Iodice V, Rae CL, Brooke A, Simmons R, Quadt L, et al. Joint Hypermobility Links

- Neurodivergence to Dysautonomia and Pain. *Frontiers in Psychiatry*. 2022 Feb 2;12.
51. Cage E, Crompton CJ, Dantas S, Strachan K, Birch R, Robinson M, et al. What are the autism research priorities of autistic adults in Scotland? *Autism*. 2024 Feb 4;
 52. Buckley C. Autism in adults. *InnovAiT: Education and inspiration for general practice* [Internet]. 2017 Mar 28;10(6):319–26. Available from: <https://doi.org/10.1177%2F1755738016683410>
 53. O’Nions E, Petersen I, Joshua E.J. Buckman, Charlton RA, Cooper C, Corbett A, et al. Autism in England: assessing underdiagnosis in a population-based cohort study of prospectively collected primary care data. *The Lancet regional health*. 2023 Jun 1;29(1):100626–6.
 54. Beggiano A, Peyre H, Maruani A, Scheid I, Rastam M, Amsellem F, Gillberg CI, Leboyer M, Bourgeron T, Gillberg C, Delorme R. Gender differences in autism spectrum disorders: Divergence among specific core symptoms. *Autism Research*. 2017 Apr;10(4):680-9.
 55. Autism Spectrum Quotient | Embrace Autism [Internet]. Embrace Autism. 2023 [cited 2024 Mar 26]. Available from: <https://embrace-autism.com/autism-spectrum-quotient>
 56. Song P, Zha M, Yang Q, Zhang Y, Li X, Rudan I. The prevalence of adult attention-deficit hyperactivity disorder: A global systematic review and meta-analysis. *Journal of Global Health* [Internet]. 2021 Feb 11;11(04009). Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7916320/>
 57. Attention Deficit Hyperactivity Disorder [Internet]. NICE. 2023 [cited 2024 Mar 21]. Available from: <https://cks.nice.org.uk/topics/attention-deficit-hyperactivity-disorder/background-information/prevalence>
 58. Kindgren E, Quiñones Perez A, Knez R. Prevalence of ADHD and Autism Spectrum Disorder in Children with Hypermobility Spectrum Disorders or Hypermobility Ehlers-Danlos Syndrome: A Retrospective Study. *Neuropsychiatric Disease and Treatment*. 2021 Feb;Volume 17:379–88.
 59. Fairman KA, Peckham AM, Sclar DA. Diagnosis and treatment of ADHD in the United States: Update by gender and race. *Journal of Attention Disorders*. 2020 Jan;24(1):10-9.
 60. Dyslexia - British Dyslexia Association [Internet]. British Dyslexia Association. 2019. Available from: <https://www.bdadyslexia.org.uk/dyslexia>
 61. Dyscalculia [Internet]. British Dyslexia Association. 2023. Available from: <https://www.bdadyslexia.org.uk/dyscalculia>
 62. Dyspraxia, or Developmental Coordination Disorder (DCD) [Internet]. Cambridge University Hospitals.

- [cited 2024 Apr 19]. Available from: <https://www.cuh.nhs.uk/our-people/neurodiversity-at-cuh/dyspraxia-or-developmental-coordination-disorder-dcd/>
63. Doherty M, Neilson S, O'Sullivan J, Carravallah L, Johnson M, Cullen W, et al. Barriers to healthcare and self-reported adverse outcomes for autistic adults: a cross-sectional study. *BMJ Open* [Internet]. 2022 Feb 1;12(2). Available from: <https://bmjopen.bmj.com/content/12/2/e056904>
 64. Bishop-Fitzpatrick L, Kind AJH. A scoping review of health disparities in autism spectrum disorder. *Journal of Autism and Developmental Disorders*. 2017 Jul 29;47(11):3380–91.
 65. NHS ENGLAND. NHS England» Clinical guide for front line staff to support the management of patients with a learning disability and autistic people – relevant to all clinical specialties [Internet]. [www.england.nhs.uk](https://www.england.nhs.uk/long-read/clinical-guide-for-front-line-staff-to-support-the-management-of-patients-with-a-learning-disability-and-autistic-people-relevant-to-all-clinical-specialties/). 2023 [cited 2024 Mar 27]. Available from: <https://www.england.nhs.uk/long-read/clinical-guide-for-front-line-staff-to-support-the-management-of-patients-with-a-learning-disability-and-autistic-people-relevant-to-all-clinical-specialties/>
 66. Failla MD, Gerdes MB, Williams ZJ, Moore DJ, Cascio CJ. Increased pain sensitivity and pain-related anxiety in individuals with autism. *Pain Reports* [Internet]. 2020 Nov 16 [cited 2021 Jan 25];5(6). Available from: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7676593/>
 67. Dockrell DM, Berg KM, Ralston SH. Mind the gaps: therapists' experiences of managing symptomatic hypermobility in Scotland. *Rheumatology Advances in Practice*. 2021;5(2):rkab046.
 68. Jones JT, Black WR. Provider Knowledge and Experience in Care, Management, and Education of Pediatric Ehlers-Danlos syndrome. *Global Pediatric Health*. 2022 Jan;9:2333794X2211128.
 69. Rombaut L, Deane J, Simmonds J, De Wandele I, De Paepe A, Malfait F, et al. Knowledge, assessment, and management of adults with joint hypermobility syndrome/Ehlers-Danlos syndrome hypermobility type among Flemish physiotherapists. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*. 2015 Mar;169(1):76–83.
 70. Estrella E, Frazier PA. Healthcare experiences among adults with hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorder in the United States. *Disability and Rehabilitation*. 2023 Feb 11;46(4):1–10.
 71. Statement: Hypermobility guidelines [Internet]. www.rheumatology.org.uk. 2020 [cited 2024 Apr 1]. Available from: <https://www.rheumatology.org.uk/news/details/statement-hypermobility-guidelines>

72. Developmental Rheumatology in children: Scenario: Hypermobility in Children, [Internet]. NICE. 2019 [cited 2024 Apr 1]. Available from: <https://cks.nice.org.uk/topics/developmental-rheumatology-in-children/management/hypermobility-in-children/>
73. HMSA Education Model [Internet]. The HMSA. [cited 2024 Apr 19]. Available from: <https://www.hypermobility.org/hmsa-education-model>
74. EDS ECHO Healthcare Professionals [Internet]. The Ehlers Danlos Society. [cited 2024 Apr 19]. Available from: <https://www.ehlers-danlos.com/eds-echo-healthcare-professionals>
75. Hypermobility | British Society for Rheumatology [Internet]. www.rheumatology.org.uk. [cited 2024 Apr 19]. Available from: <https://www.rheumatology.org.uk/events-learning/webinars/hypermobility>
76. Vinney C. What Is Medical Gaslighting? [Internet]. Verywell Mind. 2022 [cited 2024 Apr 19]. Available from: <https://www.verywellmind.com/what-is-medical-gaslighting-6831284>
77. Everyone Matters: 2020 Health Workforce Vision [Internet]. Scottish Government. 2013 [cited 2024 Apr 19]. Available from: <https://www.gov.scot/publications/everyone-matters-2020-workforce-vision/pages/3/>
78. Booker CL, Andrews L, Green G, Kumari M. Impacts of long-standing illness and chronic illness on working hours and household income in a longitudinal UK study. *SSM - Population Health*. 2020 Nov;12:100684.
79. Barnard H. UK Poverty 2017 [Internet]. Joseph Rowntree Foundation. 2017 [cited 2024 Apr 19]. Available from: <https://www.jrf.org.uk/report/uk-poverty-2017>
80. Palomo-Toucedo IC, Leon-Larios F, Reina-Bueno M, Vázquez-Bautista M del C, Munuera-Martínez PV, Domínguez-Maldonado G. Psychosocial Influence of Ehlers–Danlos Syndrome in Daily Life of Patients: A Qualitative Study. *International Journal of Environmental Research and Public Health*. 2020 Sep 3;17(17):6425.
81. Saffer J, Nolte L, Duffy S. Living on a knife edge: the responses of people with physical health conditions to changes in disability benefits. *Disability & Society*. 2018 Nov 9;33(10):1555–78.
82. Barnes MC, Gunnell D, Davies R, Hawton K, Kapur N, Potokar J, et al. Understanding vulnerability to self-harm in times of economic hardship and austerity: a qualitative study. *BMJ Open* [Internet]. 2016 Feb 1 [cited 2021 Jan 2];6(2):e010131. Available from: <https://bmjopen.bmj.com/content/6/2/e010131>
83. McCartney M. The disturbing truth about disability assessments. *BMJ*. 2012 Aug 8;345(aug08 1):e5347–7.

84. Lum A, Wakefield CE, Donnan B, Burns MA, Fardell JE, Marshall GM. Understanding the school experiences of children and adolescents with serious chronic illness: a systematic meta-review. *Child: Care, Health and Development*. 2017 May 23;43(5):645–62.
85. Bennett SE, Walsh N, Moss T, Palmer S. Understanding the psychosocial impact of joint hypermobility syndrome and Ehlers–Danlos syndrome hypermobility type: a qualitative interview study. *Disability and Rehabilitation*. 2019 Jul 18;43(6):1–10.
86. Stangl AL, Earnshaw VA, Logie CH, van Brakel W, C. Simbayi L, Barré I, et al. The Health Stigma and Discrimination Framework: a global, crosscutting framework to inform research, intervention development, and policy on health-related stigmas. *BMC Medicine* [Internet]. 2019 Feb 15;17(1):1–13. Available from: <https://bmcmmedicine.biomedcentral.com/articles/10.1186/s12916-019-1271-3>
87. Van Brakel WH. Measuring health-related stigma—A literature review. *Psychology, Health & Medicine*. 2006 Aug;11(3):307–34.

