Petitioner submission of 20 September 2023

PE2038/B: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders

Hypermobile Ehlers-Danlos syndrome and hyper mobility spectrum disorders (hEDS/HSD) are not rare types of Ehlers-Danlos syndromes, they are just rarely diagnosed. Demmler et al 2019 found that 1 in 500 people had a diagnosis of hEDS/HSD.

In 2018 Professor Stuart Ralston (Rheumatologist) put in a bid for a specialist centre for the treatment and diagnosis of patients with hEDS / complex EDS. The aim being to establish a centre for the assessment, and management of patients whose needs are not being adequately met at a local level. The centre would accept referrals from secondary care in NHS Boards throughout Scotland, where it was felt that the patients' needs could not be adequately addressed locally. This was backed up by consultants, therapists, and patients but was not supported by Scottish Society for Rheumatology; they did not see a need for a specialist centre, as they believed the services were already available to patients. At this time there was also a commitment from National Services Division to produce a paper for regional directors of planning highlighting which issues and service gaps people with EDS and HSD encounter. To our knowledge this still hasn't been written.

There is a lack of people being seen by rheumatology, nearly all referrals are bounced back to GPs. Therefore, most people if they can afford it, pay to see a rheumatologist privately. This is the same for physiotherapy and other Musculoskeletal (MSK) services. It is our understanding that Rheumatology have been directed not to see people with non-inflammatory conditions. EDS is not an inflammatory condition, but it is a connective tissue disorder (CTD) and musculoskeletal (MSK) condition causing multi-systemic issues. Both MSK and CTD conditions normally fall under the remit of rheumatology.

A 2020 report on behalf of EDS Support UK discusses the experience of young people with EDS/HSD in Scotland. The report titled Ehlers-Danlos Syndrome and Young People in Scotland: The Agency Needed to Prosper (by Anne-Marie Martin) states that young patients in Scotland perceive healthcare professional attitudes to be negative. This is particularly prevalent in young females who report that their symptoms are not believed. The diagnostic journey for EDS/HSD patients is

problematic and the report revealed an inequity of service access in Scotland, with most patients surveyed reporting that they had to travel to London to receive a diagnosis. The report advocates for better education and training of health professionals in Scotland and the need for better service access geographically.

We have 850 members in Scotland located across every health board. The majority of whom are living with hEDS/HSD. Meetings are run twice monthly online and in person and we provide 1:1 support. We have lived experience from members and our volunteers who have 7+ years of experience supporting our members. EDS Support UK are currently represented at Cross Party Groups for Rare, Genetic and Undiagnosed Conditions, Arthritis and Musculoskeletal conditions as well as Chronic Pain.

In our experience very few people with hEDS/HSD have been accepted for an out of country referral and through our experience of supporting members there is no specialist centre for people to be sent to.

There is clearly still work to be done. In our experience 4 out of 5 of the people we speak to are undiagnosed and seeking help in getting a diagnosis and / or help to manage their condition.

We had hoped to obtain Scotland-wide data for patients diagnosed with hEDS/HSD but ICH-10 coding only found 321 patients from across Scotland, which suggests hEDS/HSD is not being recorded. In 2019 the old classification of EDS 3 was still being used. If the Scottish Government is basing its budgets on coding, it will be almost impossible to determine accurate data about who has hEDS/HSD which in turn leads to the issue of funding/commissioning of services for hEDS/HSD which is so badly needed.

Csecs et al 2022 found there appears to be a greater prevalence of neurodiversity for people with hypermobility compared to those in the general UK population. This often leads to patients being misdiagnosed and or dismissed entirely. This will account for a large number of our members waiting years for a diagnosis. Researchers and scientists in Scotland and across Europe are looking at changes to the connective tissue in the brain, as well as the body, to help understand the prevalence of neurodiversity.

A specialist service can provide patients with the confidence that the treatment options being offered are rooted in research, knowledge, experience and understanding of their condition. It will ensure that the impacts of surgery are well understood. This is rarely the case at present. We believe a specialist service will lead to more research

opportunities from within the UK and internationally. Through our volunteers we already collaborate with researchers at the universities of Edinburgh and Strathclyde. We also have contacts with a hEDS specialist service and a genetics laboratory in North America.

We hope you can see why we are calling for services for hEDS/HSD to be funded or commissioned in Scotland. Our members have nowhere else to go.