

PE2038/I: Commission suitable NHS services for people with hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders

Petitioner written submission, 20 February 2025

We would like to challenge the response from the Rare Diseases, Genomics, Diagnostics and Participation Unit of 15 November 2024. As previously detailed in our evidence there are 13 types of Ehlers Danlos Syndrome (EDS), 12 of which are rare and one which is far more common. That is hypermobile EDS (hEDS) and hypermobility spectrum disorders (HSD). We need to be clear that given the published studies and growing body of evidence that the prevalence of this condition is common, not rare; with studies suggesting a prevalence of between 1 in 227 and 1 in 500; and respectfully that perhaps this is outwith the Rare Diseases, Genomics, Diagnostics and Participation Unit's remit. This petition is focused on hEDS and HSD which do not have a known genetic basis, which prevents these conditions from being identified by genetic testing. Therefore, our community is faced with incredible barriers to accessing diagnosis and care equitably.

We were pleased to hear that the Rare Disease Unit is producing awareness videos for the rare types of EDS and looking to reform diagnostics.

While we are pleased that guidance on the assessment and diagnosis for hEDS is available on the RDS website, we believe that this guidance would benefit from revision by a knowledgeable multidisciplinary team, should include information on HSD, and could make use of the EDS UK GP Toolkit which has been based on the relevant evidence base. We would be pleased to see this advice readily available on RefHelp pages for each Health Board to ensure that all healthcare professionals in Scotland have access to the same guidance, while we await a national care pathway.

We would like to understand how the information provided on RDS is used by primary care across all Health Boards and if it is being accessed then why are our members still reporting problems in getting the diagnosis and care that they desperately need?

The response from NHS Wales states that 'Hypermobility spectrum disorders (HSD) and Hypermobile EDS (hEDS) were highlighted by several patient, charity and clinical forums as an area which lacks guidance and consistency in delivery.' As a result of our petition in Wales, we were invited to join the working group as subject matter experts to co-create a pathway for 'advice on diagnosis and treatment for primary care clinicians, direct access to therapies services for primary and community care rehab and supported self-management interventions, and clear referral guidance for those with suspicion of the rarer forms of EDS that require genetic testing and speciality involvement.' We used our GP toolkit <https://gptoolkit.ehlers-danlos.org/> as a basis for the work and we are pleased to report that the final draft has now gone out to wider consultation in NHS Wales. We are hopeful that this will be in use in the coming months and that we will start to see improvements in equitable access to healthcare in Wales.

Our questions for the Scottish Government are as follows:

How is the Right Decision Service used by healthcare professionals in NHS Scotland? Are there any statistics to show the use and impact of the hypermobility/hypermobility Ehlers Danlos syndrome guidance? Is the guidance widely used in primary care? If not, what systems are used, like the Community Pathways system, that could be adapted with the right information on current best practice?

Given the work that NHS Wales has done in collaboration with us, would NHS Scotland be prepared to commit to a similar project? Would the Scottish Government also support the development of SIGN guidance given recent evidence?