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Commission suitable NHS services in Wales for people with EDS or hypermobility spectrum disorders

Y Pwyllgor Deisebau | 03 Gorffennaf 2023
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Petition Number: P-06-1348

Petition title: Commission suitable NHS services in Wales for people with EDS or hypermobility spectrum disorders

Text of petition: Historically those showing signs of Ehlers-Danlos syndromes (EDS) or HSD have been referred to rheumatology departments. In 2021, they were directed to stop seeing these patients in favour of their diagnosis and management in primary care, which is not currently equipped for this role. A unique tertiary service in England has also closed to out of area patients. This situation has led to inequalities in access to healthcare for those with EDS and HSD in Wales resulting in unacceptable suffering.

1. Background

Ehlers-Danlos syndromes (EDS) and hypermobility spectrum disorders (HSD)

Ehlers-Danlos syndromes (EDS) are a group of genetic disorders which affect the body's connective tissue. Connective tissue links and supports other tissues and organs of the body. There are 13 types of EDS, most of which are rare, with hypermobile EDS (hEDS) being the most common. The condition can be inherited but an individual with no family history can also possess the genes responsible.

People living with EDS can experience a variety of symptoms, due to connective tissue being positioned throughout the body. These include excess joint mobility, known as hypermobility, and stretchy or fragile skin. Symptoms can also extend beyond the skin and joints, with some patients experiencing chronic pain and fatigue, dizziness and digestive problems, amongst others. Some types of EDS, such as vascular EDS, can be life-threatening.

Additionally there are other conditions which affect joint mobility but do not meet the clinical criteria for EDS. These are referred to as hypermobility spectrum disorders (HSD) and have similar symptoms to hEDS.

A study from 2019 which investigated the prevalence of these conditions in Wales found that there was an estimated 1 in 500 people in Wales with a diagnosis of EDS or HDS on their medical records in Wales in 2016/2017. 70% of these cases were women. However, men in Wales were diagnosed with EDS an average of 8.5 years earlier than women.

This petition is part of Ehler-Danlos Support UK's #EnoughIsEnough campaign, calling for governments across the UK (through petitions and community engagement) to fund suitable NHS services for diagnosis and treatment of hypermobile EDS and HSD.

Support and treatment for EDS and HSD

There is no test available for hEDS or HSD, however the rarer types of EDS can be diagnosed using genetic testing through a genetics specialist. The NHS 111 Wales website states that patients who are suspected to have rare EDS types can be

referred to a specialist EDS service in England, either Sheffield or London, for diagnosis.

There is no specific treatment for EDS or HSD, instead care is focussed on helping people manage their symptoms. For example, physiotherapists can help patients with symptoms like joint pain whilst counsellors can offer help dealing with long-term pain.

2. Welsh Government action

The Minister for Health and Social Services, Eluned Morgan, states in response to this petition:

> The Welsh Government is committed to improving the lives of those impacted with a Rare Disease in Wales and recognises the substantial challenges, including equity of care, management and if available, treatment faced by people.

The Minister notes that the Welsh Health Specialised Services Committee (WHSSC) plans and secures services for people with rare conditions and it is the responsibility of health boards to manage the access and referral to services as appropriate.

The Welsh Government has developed Wales Rare Disease Action Plan 2022-2026 which forms part of the UK Rare Diseases Framework. This plan aims to help provide the best care to and improve the lives of people living with rare diseases, of which EDS and HSD are included. The four key priorities of the plan are:

1. Helping patients get a final diagnosis faster.
2. Increasing awareness of rare diseases amongst healthcare professionals.
3. Better conditions of care.
4. Improving access to specialist care, treatment and drugs.

The plan has a strong focus on the use of genome sequencing to improve genetic testing for rare diseases. It announces the development of a three-year genomics delivery plan (2022-2025) for Wales, which intends to roll-out whole genome and exome sequencing to patients with a suspected rare disease.

The Rare Diseases Implementation Group oversees the delivery of the Rare Disease Action Plan in Wales and monitors its progress annually. A senior clinician was appointed in 2022 as Clinical Lead and Clinical Champion for rare diseases to work with the group "to raise the profile of rare diseases and initiate appropriate workstreams in discussion with partners".

The Minister reported in her response to the petition that the Rare Diseases Implementation Group has "made progress with all priorities" in the last year. Looking to the future, she also said:

> […]there are ongoing investigations regarding the opportunity for a virtual health hub/Wales Rare Disease centre that will provide remote support to those with Rare Diseases.