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

Scottish Government submission of 15 November 2024

I refer you to your email of 18 October asking for the Scottish Government's views on the actions called for in petition PE2038, following its consideration at the Petitions Committee meeting of 9 October 2024. I have addressed the Committee's requests contained in the bullet points below.

- **The Committee is interested to know whether the Scottish Government accepts the Petitioner's view that Ehlers-Danlos syndrome and hypermobility spectrum disorders are not necessarily rare diseases as they often go undiagnosed and, in light of that, is keen to understand what action is being taken to improve diagnostic services beyond the Rare Disease Action Plan.**

- **The Committee heard that NHS Wales has now committed to co-creating a hypermobility pathway for primary care to help GPs diagnose and manage these conditions. The Committee is interested to know whether a similar exercise could be undertaken in Scotland.**

The Scottish Government recognises that rare conditions may be individually rare to greater or lesser degrees, but collectively more common, and the number of individual conditions under the umbrella of Ehlers-Danlos syndrome, as well as the spectrum of hypermobility disorders, make them collectively more common than some of the rarest conditions. We recognise, and have noted several times in the process of our engagement with the rare community, that delayed and missed diagnoses are a fundamental concern, and that awareness raising is key to addressing this.

As set out in our Progress Report on Scotland's Action Plan for Rare Diseases, published in August 2024, we have worked to understand in the first instance what level of knowledge healthcare professionals currently have of rare conditions, where they would access supportive resources, and identify what they would need to help them improve the care they give to people with rare conditions. This work was conducted in the form of a survey by Genetic Alliance UK and the Office for Rare Conditions, Glasgow. We are using the results of this survey to shape our approach to awareness raising going forward.

We are also developing awareness raising videos, in conjunction with NHS Education for Scotland, to encourage healthcare professionals to 'think rare'. The petitioner's website notes that healthcare professionals can often be trained to 'think horses not zebras when you hear hooves' (i.e. consider the most common diagnosis), and we recognise that a different approach is required when presented with a set of symptoms that, together, can indicate a rare condition. This will be a core message within the professional-facing videos we are developing, and will be

complemented by videos intended for families experiencing a rare condition. We have made faster diagnosis and awareness raising our key priorities for the coming year.

Our work to improve the lives of people with all rare conditions will remain an important commitment. In this instance however, the petitioner has specifically asked what action is being taken outwith our work for rare conditions to improve diagnostic services.

The Scottish Government recognises that demand for diagnostics has been growing progressively, and that reform is necessary to ensure that we have sustainable diagnostics that are able to deliver the best possible services for patients. The Scottish Strategic Network for Diagnostics is currently supporting work to strategically plan this reform across Scotland.

We have also been considering other resources that could be promoted for use by healthcare professionals to diagnose more effectively. One such resource, mentioned in the Progress Report on Scotland's Action Plan for Rare Diseases, is the Right Decision Service (RDS) hosted by Healthcare Improvement Scotland. RDS is a suite of digital tools that enable convenient and quick decision-making, and also hosts useful information for healthcare professionals, including information developed by NHS Boards. As the petitioner may already be aware, assessment and referral guidance for hypermobile Ehlers Danlos syndrome was developed by NHS Dumfries & Galloway last year, and is hosted on the RDS website. We will continue to look for ways to promote such guidance among healthcare professionals.

The Scottish Government is also engaging with clinicians, stakeholders, and those with lived experience, to develop an integrated strategy on long term conditions. This will include a consultation in the new year, to ensure a wide capture of views.

Scottish Government officials engage regularly with our counterparts in the UK government and the other devolved nations in order to share knowledge and progress in our work to improve the lives of people living with rare conditions. We will certainly be interested in learning more about the hypermobility pathway for primary care that NHS Wales will be developing. We will speak to our Welsh counterparts about their project plan, funding practicalities and resourcing required, and consider if it is feasible, within current resources, for a similar exercise to be taken forward in Scotland and by what means.

I hope this response is helpful to the petitioner.

Rare Diseases, Genomics, Diagnostics and Participation Unit