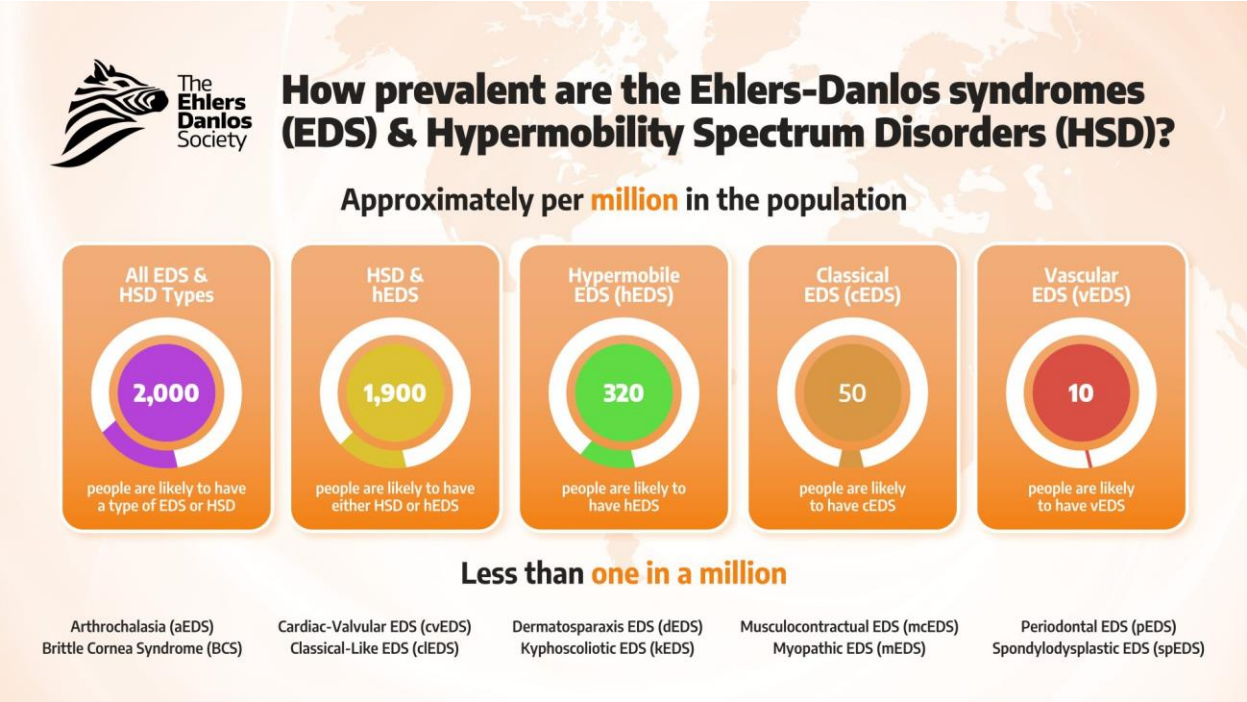
Are the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders Rare or Common?



The prevalence of the Ehlers-Danlos syndromes (EDS) and hypermobility spectrum disorders (HSD)

Our community may read different commentaries about prevalence figures for EDS and HSD. We hope the following is a helpful summary.

What makes a condition “rare”?

A condition is considered rare if it affects less than 1 in 2000 people. Figures may be presented in different ways, but they mean the same thing. For example, a document might say that 5 per 10,000 people, or less than 50 per 100,000 is rare.

Statements about prevalence of the Ehlers-Danlos syndromes

The **overall prevalence** of Ehlers-Danlos syndromes (EDS) is cited as 1 in 5000 based on a publication by Pyeritz (2000), and further supported by population data from Denmark (Kulas Søborg et al., 2017), and data extrapolated from a Swedish study (Cederlöf et al., 2016). In a study of healthcare records in the Welsh population, Demmler et al. (2019), found an EDS prevalence of approximately 1 in 3100. However, as more is learned about the most prevalent type of EDS, hypermobile EDS (hEDS), it is now thought that figures for the prevalence of hEDS are an underestimate. Population studies to clearly show this still need to be done. This is explained further in the section below on joint hypermobility syndrome (JHS), hEDS, and hypermobility spectrum disorder (HSD).

Aside from hEDS, the other types of EDS are rarer than 1 in 5000. Classical EDS has a prevalence of 1 in 20,000–40,000. Vascular EDS has a prevalence of 1 in 100,000–200,000. Other types of EDS affect less than 1 in a million or are ultra-rare in that they affect small numbers of individuals and families (Malfait et al., 2017).

At The Ehlers-Danlos Society we believe that whichever way prevalence figures are discussed, it should always be clear which type or types of EDS are being considered. Using one figure for an **overall prevalence** or a combined prevalence masks the fact that distinct types of EDS have different prevalence figures. To avoid doubt, we would like to see a move away from overall/combined prevalence statements to ones that are explicit about which type of EDS is being discussed. We think that this should also apply to studies and commentaries about HSD.

What do we know about the prevalence of joint hypermobility syndrome, and how is this relevant to the hypermobility spectrum disorders and hypermobile Ehlers-Danlos syndrome by the 2017 criteria?

Two large population studies have looked at the prevalence of joint hypermobility syndrome (JHS) and Ehlers-Danlos syndromes (EDS). One specifically looked at the prevalence of these conditions (Demmler et al., 2019); the second looked at mental health and neurodiversity but the data can be used to determine the prevalence of JHS and EDS (Cederlöf et al., 2016). A third study also gives a sense of the prevalence of JHS by extrapolating observations from rheumatology clinics in the UK (Hakim & Grahame, 2006).

These studies showed similar findings in that the overall / combined prevalence of JHS and EDS was between 1 in 500 and 1 in 800. However, breaking this down into JHS or EDS the studies showed that:
· the prevalence of JHS was approx. 1 in 600 to 1 in 900, and
· the prevalence of all types of EDS was approx. 1 in 3000 to 1 in 5000.

JHS was found to be 5 times more common than EDS.

After 2017 the term JHS was dropped. Instead, this population in the community is now described as having either hypermobility spectrum disorder (HSD) or hypermobile EDS (hEDS) based on the 2017 International criteria for hEDS and descriptors for HSD. Prevalence figures for JHS are now used to describe combined prevalence of HSD and hEDS. The combined prevalence of HSD and hEDS is in the order of 1 in 600 to 1 in 900. Expert opinion is that HSD is common and that hEDS is likely to be common. However, at this time it is not possible to say what the prevalence figure is for each of HSD and hEDS separately because this has not been studied yet.

Experts also believe that all these prevalence figures are underestimates because many people with EDS or HSD do not get diagnosed, or it takes many years to get diagnosed, or they are misdiagnosed with another condition.

Knowing the prevalence influences the way healthcare resources are allocated.

In providing healthcare resources, it is equally important for distinct reasons to know whether a condition is rare or common.

It is important to recognize that most types of EDS are rare or ultra-rare. Healthcare services and health research funding organizations make specific decisions about how they support people with rare conditions. It remains hugely important to acknowledge that within the EDS family there are those types of EDS that benefit from the resources put in place for rare diseases.

However, equally important, if the whole family of these conditions including hEDS and HSD is thought to be rare, then healthcare professionals, organizations that provide care, and policymakers may think that hEDS and HSD are not relevant to them. Priority is given to common conditions that affect more people.

Expert opinion is that HSD is common. The prevalence of hEDS is now thought to be greater than previously described, but by how much is not yet clear. Healthcare professionals and organizations that are not aware that they are likely to see people with HSD or hEDS risk missing the diagnoses, not knowing how to care for people with these conditions, and not having the resources to care for them. For too many in our community, this is not an imaginary state; it is a lived experience (Halverson et al., 2021).

In summary

At the Ehlers-Danlos Society we believe that EDS and HSD are two of the most misunderstood conditions of our time. We, and many expert clinicians believe that HSD and hypermobile EDS (hEDS) are more common than currently recognized. However, it is also important to remember that most types of EDS are rare to ultra-rare.

We know how important it to raise awareness about HSD and EDS. We are committed to our mission to increase education, support, and research into all types of EDS and HSD. Access to early diagnosis, management, care, and validation is at the heart of what we do, and we want to ensure that the wider medical community gives these conditions the respect and understanding they deserve.

Finally, in 2021 Drs. Hakim, Tinkle, and Francomano wrote the following in an editorial for the American Journal of Medical Genetics which we feel sums it up for us:

"In both community care and across a broad range of hospital specialties, many healthcare professionals (nurses, midwives, physical and other therapists, psychologists, and doctors) will likely attend to people with Ehlers-Danlos syndromes (EDS) or hypermobility spectrum disorders (HSD). The belief that EDS is rare or ultra-rare is true for several forms of the syndrome, but increasingly the hypermobile type of EDS (hEDS) is thought more common, and HSD much more common….Yet often the journey to diagnosis and treatment is challenging for patients, who may receive no diagnosis or misdiagnoses. In addition, misconceptions that the issues are solely mental health concerns are commonly reported. Too often there is more than a

decade delay in diagnosis, and the negative impact on quality of life from living with under-managed disease is significant.…

With substantial growth in knowledge in the field, keeping up to date with advances in epidemiology, pathophysiology, and management of this heterogeneous group of conditions is challenging. No individual healthcare professional can truly manage all aspects of these conditions at an expert level. Care typically requires a holistic and multi-disciplinary approach. Yet such care, ideally embedded in the community, supported by subspecialty medical, therapies, and social care is lacking for EDS and HSD internationally. Currently, there are small numbers of expert clinics, centers, and networks striving to support patients and colleagues. Access to care is profoundly limited, yet management of many of the common concerns is well within the ability of most generalists with specialist support when needed."

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