EHLERS-DANLOS

For these complex genetic disorders, multi-disciplinary management and clinical expertise is key to quality of life. ***Hatty Willmoth*** writes

Nutrition often plays a central part of the health management of people with chronic conditions. But it can be a complicated task, often requiring a high degree of expertise from the practitioner — expertise that may be hard to find in the case of less common disorders.

This is certainly the case for people with any of the Ehlers-Danlos Syndromes (EDS). Helen Spriggs, who specialises in nutrition for people with EDS, is all too familiar with the unique challenges it can bring.

What is EDS?

EDS is a group of 13 different inheritable connective tissue disorders, classed as 'rare' by the NHS.[1] Sufferers tend to have joint hypermobility (joints with an abnormally wide range of movement) and skin that's stretchy, fragile, and easily bruised — but there are many other symptoms associated with each individual disorder.

The most common EDS subtype is hypermobile EDS (hEDS). People with hEDS tend to have loose and unstable joints that dislocate easily, digestive problems, fatigue, and dizziness and increased heart rate when standing up. Spriggs says that hEDS is usually diagnosed through an assessment against various criteria by a rheumatologist; the other 12 types tend to be diagnosed through medical genetics (either family history or gene testing).

But hEDS is just one subtype of many. People with classical EDS (cEDS), in contrast, tend to have smooth, velvety, fragile skin; they may experience hernias and organ prolapse; and if they acquire wounds, these will heal slowly and leave wide scars.

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And, as a final example, vascular EDS (vEDS) — a particularly uncommon syndrome — affects blood vessels and internal organs; sufferers may have thin skin with visible small blood vessels. Their blood vessels may bulge or tear, which can lead to internal bleeding. They may also have unusual facial features, hypermobile fingers and toes, and varicose veins.

"Like many conditions, there is a spectrum of symptom severity in EDS and its subtypes," says Spriggs. "Some subtypes of EDS can be more severe or life threatening, such as vEDS. However, less severe types like hEDS may be hugely impactful on quality of life."

Connective tissue

The main characteristic of EDS, bringing these disparate symptoms under one umbrella, is the disruption of collagen and other connective tissue.

Spriggs says: "Connective tissues are found throughout the body, and collagen — the glue that holds the connective tissue together — is a key

WHY A ZEBRA?

One of the main icons of EDS is a zebra: a response to the phrase, "when you hear hoofbeats behind you, don't expect to see a zebra". While medical professionals often search for common diagnoses first, people with rare, chronic diseases know that diagnoses might be unexpected. The Ehlers-Danlos Society website says: "Sometimes when you hear hoofbeats, it really is a zebra."[2]

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component of them.

"Collagen is the predominant protein that is impacted in EDS, yet it is not a lack of collagen in the body but a variation in the production 'recipe' that results in reduced collagen integrity."

That means that cells connecting, binding, separating and supporting other tissues, in people with EDS, function abnormally and/or have an abnormal structure.

Not just hypermobility

Despite the multitude of possible manifestations of EDS, it is often just thought of as hypermobility. But Spriggs is keen to emphasise that EDS and hypermobility are not one and the same. She says that, even if an individual has "hypermobile joints, in one or several areas of the body", this does not mean they have EDS. "The EDS diagnostic criteria differentiates between asymptomatic joint hypermobility, Hypermobility Spectrum Disorder (HDS), a subtype of EDS, or another condition that manifests with hypermobile joints.

"Importantly, individuals can fulfil hEDS criteria but score quite low on the Beighton Scale — a commonly used screening tool for hypermobility." In other words, some EDS sufferers are not even particularly hypermobile.

Complex associations

People with EDS often have symptoms and even co-existing conditions that those unfamiliar with the disorders may not expect.

Spriggs says: "The wider health implications make EDS a complex, multi system-involving condition.

"Several health conditions or manifestations are also recognised as existing alongside the musculoskeletal involvement [bones, muscles and connective tissues], and may include gastrointestinal [digestive system], cardiovascular [heart and blood] or neurological [brain and nerves], amongst others."

Researchers are currently investigating the potential association of EDS with a host of other conditions, she says, including Postural Orthostatic Tachycardia Syndrome (POTs), mast cell disorders, ADHD and autism.

Managing EDS: a multi-disciplinary approach

There is no cure for EDS — they are genetic conditions — but management can make a big difference to quality of life. Spriggs explains that this is often a team effort between professionals in all manner of fields, including nutrition.

She says: "There is no 'fix' for EDS, but there are management strategies for the resulting symptoms and associated conditions of EDS that individuals may find beneficial.

"Care recommendations will be very different, subject to an individual's situation; there is no standard management protocol across specialities for all patients. Multi-disciplinary care is crucial for EDS."

Spriggs says that this care often includes "a form of physical and/or exercise therapy", psychological support and pain management strategies, as a foundation. Beyond this, people with EDS may include a range of specialists from professions including gastroenterology, neurology, occupational health, podiatry, dentistry and nutrition.

"Multi-disciplinary teams or any healthcare professional involved in supporting EDS patients must understand the implications of EDS," Spriggs says. "This is for the patients' outcomes and safety, and may be achieved by adapting care approaches or health goals.

"Experience and understanding allows a practitioner to truly hear the individual in front of them, support them, and not just look at a scan or test result. It allows for improved recognition, referral pathways, diagnosis, and access to care for those with EDS.

"Healthcare professionals often receive little to no training in EDS, in part as EDS is considered rare — although recent research is challenging previous prevalence figures as awareness and diagnosis accuracy improves."

Supporting EDS with nutrition

Spriggs says nutrition can play a major role in supporting people with EDS.

"Nutrition is key for health," she says. "Therefore, it is an important factor for an individual managing EDS or any other health condition.

"An aspect to highlight includes assessing nutrient status, as impacted digestion and absorption is often seen in EDS. However, fundamentally, nutrient intake can also be limited, as some individuals find themselves experiencing food reactions, leading to an ever-decreasing number of 'safe' foods and restricted dietary nutrient intake.

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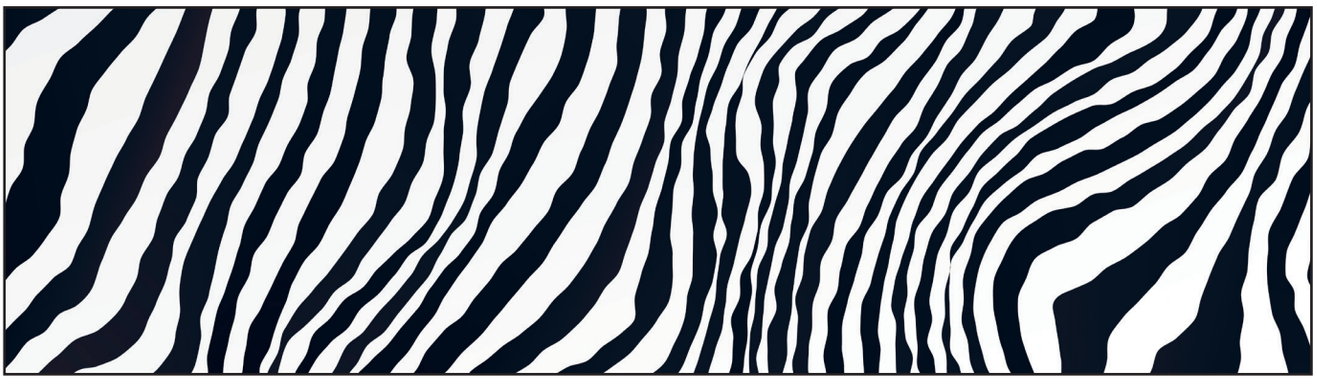

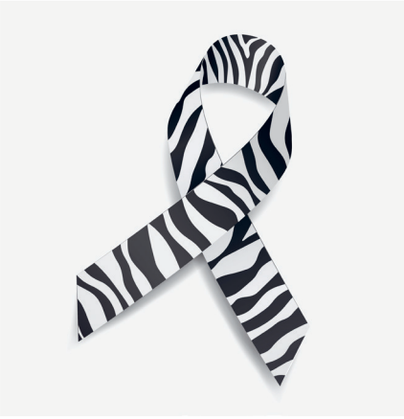

"...an experienced EDS clinician with a clear understanding of the conditions...is crucial"

"Working with an experienced specialist clinician in EDS and the associated conditions, to understand if there are true allergies or intolerances, or if there are other factors at play, is one step to a clearer understanding and a path to increasing dietary and nutrient variety once again.

"Similarly, for some EDS patients, mobility limitations through pain and fatigue or anxiety may impact their access to sunlight, reducing the production of vitamin D."

She explains that other co-occurring conditions may complicate the situation further. "POTs being one example," she says, "where fluid and electrolytes must be assessed and adapted appropriately under clinical guidance to help manage symptoms.

"Yet increasing fluid intake may be challenging if someone has bladder [issues] or if delayed gastric emptying is present. These situations are where an experienced EDS clinician with a clear understanding of the conditions, whilst listening to the individual, is crucial."

EDS: an individualised approach

When tackling EDS — from a nutritional perspective, or otherwise — the personalisation of care is central to its effectiveness, Spriggs says. This is because EDS is so variable and complex, and because every person is unique.

"There is no standard EDS 'diet plan', nor is there a standard EDS supplement protocol as a magic 'fix all'," she says.

"As individuals, with complex health concerns, and other factors such as geographic location, lifestyle, and stress levels, our nutrient requirements are likely to be very different.

"Medical history, current medical diagnoses, symptoms and medications, lifestyle, dietary intake, cultural and socioeconomic factors must first be factored in, alongside the context of an EDS diagnosis, before restrictive diets and large supplement 'stacks' are recommended, if at all."

She says that, currently, nutritional interventions may be held back by the fact that the research behind the role of nutritional changes in people with EDS is limited. However, this is something that the Diet and Nutrition Working Group at the International Consortium for Ehlers-Danlos Syndrome and Hypermobility Spectrum Disorders is working on.

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Ehlers-Danlos Society: www.ehlers-danlos.com
Ehlers-Danlos Support: www.ehlers-danlos.org

References:

1. www.nhs.uk/conditions/ehlers-danlos-syndromes
2. www.ehlers-danlos.com/why-the-zebra