Gastrointestinal problems in hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders

Laura Brockway, Specialist Registered Nurse, Wingate Institute of Neurogastroenterology, London

Please note: The following text cannot and should not replace advice from the patient's healthcare professional(s). Any person who experiences symptoms or feels that something may be wrong should seek individual, professional help for evaluation and/or treatment. This information is for guidance only and is not intended to provide individual medical advice.

What is EDS?

The Ehlers-Danlos syndromes (EDS) is the name given to a group of non-inflammatory hereditary conditions, which are all disorders of the connective tissue. Connective tissue is a supporting structure in our body and is made up of a number of proteins including collagen, which is the protein most often thought to be involved in EDS. Collagen is abundant in skin, muscles and ligaments where it gives these structures strength and support. Changes in the collagen proteins can mean that these structures are more 'stretchy' in EDS.

There a number of types of EDS, each affecting the body in a different way. This article will focus on the most common type of Ehlers-Danlos syndrome – the hypermobile type (hEDS, formerly also described as EDS type III or joint hypermobility syndrome), as this is the type that most commonly presents to gastroenterology services. However wherever possible or relevant this leaflet will give additional information about how the other types of EDS can affect the gut. Many specialists in this field consider hEDS to be a continuum of joint hypermobility syndrome (JHS) – or what are now called the 'hypermobility spectrum disorders' (HSD). In this article hEDS will also refer to HSD. In hEDS the main identifiable factors are the presence of skin elasticity or 'stretchiness' and hyper-flexible joints, which means that joints can move beyond the 'normal' limits. These and other signs and symptoms of hEDS can be present in varying combinations and/or degrees of severity from person to person. The true incidence of EDS, particularly hEDS, is unknown. Although hEDS is characterized by musculoskeletal symptoms, it is often the non-musculoskeletal symptoms that cause the most difficulty for some patients.

This article gives you more information about how the digestive system can be affected by EDS and the symptoms that can occur as a result, as well as mentioning the way that autonomic dysfunction (such as postural tachycardia syndrome (PoTS)) may contribute to this, and identifies some of the tests and treatment strategies your doctors may suggest.

How does EDS affect the digestive system?

As connective tissue is present throughout the body, many different structures around the body including the digestive tract can be affected by EDS. Connective tissue is present in the digestive tract and is essential to the passive mechanical movements needed to complete digestion. It has been suggested that any abnormalities in the connective tissues in the digestive tract are likely to alter the way in which it moves, which could contribute to the range of symptoms experienced by people with hEDS. Connective tissue is also present around the nerves of the digestive tract and abnormalities of this can potentially make the gut more sensitive. It is important to remember that whilst differences in the digestive tract function are likely to be present in hEDS, as yet diagnostic biomarkers have not been identified and more research is needed to better understand the nature and impact of connective tissue within the digestive system, particularly in the other subtypes of EDS.

The digestive tract starts at the mouth, and ends at the anus. Many aspects of the digestive tract can potentially be affected, including both the upper digestive tract (oesophagus, stomach and duodenum) as well as the lower digestive tract (small intestine, large intestine, colon and rectum). We frequently see patients who mainly have symptoms related to either the upper or the lower digestive tract only, and some research studies have found that a significant proportion of people with hEDS experience some kind of gastrointestinal symptoms.

What sort of gut problems can occur?

The type, frequency and severity of digestive symptoms can vary greatly from person to person as everyone with hEDS is different. The most frequently reported problems affecting the upper digestive tract are acid reflux and chronic/recurrent indigestion with pain or discomfort and early fullness after meals. The lower digestive tract can present problems such as constipation, abdominal pain, bloating, diarrhoea and a feeling of general abdominal discomfort. Nausea and vomiting can occur alongside any of the symptoms described above.

Delayed gastric emptying / dysmotility

The term dysmotility is often used to describe abnormal movements (e.g. sluggish movements or spasm) of the gut. Some hEDS sufferers can have a sluggish stomach, which means that there is a delay in the emptying of stomach contents into the small bowel, and this is often referred to as delayed gastric emptying. Delayed gastric

emptying can range in severity from mild to severe, with the most severe form called gastroparesis (paresis = paralysis). A portion of hEDS sufferers do have delayed gastric emptying, however only a few will be severe enough to be diagnosed with gastroparesis. Patients with a lot of bloating and/or fullness after meals or nausea and vomiting can be tested for delayed gastric emptying, but it is important to note that so far a link between hEDS and gastroparesis has not been categorically established.

In other patients increased sensitivity of the stomach may be a more common problem. Both dysmotility and increased sensitivity of the stomach can be associated with symptoms such as acid or bile reflux, bloating, early fullness during meals/extended fullness after meals and nausea.

Heartburn / reflux

There is some preliminary research that suggests that people with hEDS are slightly more likely to have a small hiatus hernia at the lower end of the oesophagus. This means that the upper end of the stomach slips into the chest cavity through a small hole (hiatus) in the diaphragm (the large muscle that separates the chest cavity from the abdominal cavity). This is quite a common finding and is usually not dangerous, but it can mean that the muscle that closes to stop food or liquid contents of the stomach from escaping back up into the oesophagus is somewhat inefficient, resulting in the acid reflux and/or heartburn symptoms, and this is called gastroesophageal reflux disease (GERD or GORD). However, it is also possible to experience reflux and/or heartburn symptoms without having a hiatus hernia. These symptoms can be associated with dysmotility, increased sensitivity of the oesophagus, or be experienced in isolation with none of these underlying causes.

Bloating

Abdominal bloating is a common symptom in people with hEDS, and although the underlying causes are not fully understood, it is thought that dysmotility may be a contributing factor. Overgrowth of bacteria of the small bowel can occur if there is stagnation within the bowel (i.e. constipation) and this can lead to excessive fermentation of food leading to production of gas, which can also be associated with bloating. A link between hEDS and bacterial overgrowth has not been categorically established and further research is required.

Constipation

Chronic constipation in adults is a common and debilitating problem and it is estimated that around 12 to 19% of the general population experience this, with females and the older population being more prone. Constipation is also common in patients with EDS and it is thought that a sluggish colon and difficulty with evacuation of the bowel are key causes. However there are often many factors inter-linked which can contribute to constipation such as diet, metabolic (hormone) or neurological (nerve) conditions, side effects of prescription medications, particularly opioid-based painkillers, or physical disorders such as prolapse of the bowel.

Rectal and genital prolapse are recognised as potential problems for some people with hEDS, and can be a factor contributing to constipation. Prolapse of the rectum means that the lining (mucosa) of the rectum (called a partial prolapse) or the entire rectal wall (called a complete prolapse) protrudes into the rectum, which interferes with the ability for a stool to be passed. Prolapses of the rectum usually occur during bowel movements, and then recede, but more advanced rectal prolapses can occur upon standing as well. However, in most cases prolapses tend to be small and do not require any active interventions. If a significant prolapse is diagnosed upon testing, and it is thought to be contributing to your gastrointestinal problems, your physician will refer you to a surgeon.

Functional gastrointestinal disorders

Sometimes people with hEDS who have symptoms such as reflux, heartburn, constipation or nausea may not have an identifiable cause of their symptoms on any medical testing and these patients are then given a diagnosis of functional gastrointestinal disorder (FGID). Patients who have symptoms with no underlying cause found account for more than a third of new referrals to gastrointestinal specialists, and so this is a common occurrence. A preliminary study amongst patients who were referred to a specialist because no cause of their symptoms could be found, demonstrated that over a third of those patients met the criteria for joint hypermobility and many of them had previously received a diagnosis of irritable bowel syndrome (IBS) or functional dyspepsia. IBS is the most common example of a FGID, and is characterized by recurrent abdominal pain and frequent changes in bowel habits. Functional dyspepsia is another type of FGID and relates to symptoms of upper abdominal pain, fullness, nausea and bloating, frequently following meals.

Complications

Opioid therapy

It is generally considered best to avoid strong painkillers such as opioids, as they can make the gut even more sluggish due to the effect they have on slowing the gut down, therefore worsening dysmotility. In more severe cases opioids can lead to the development of narcotic bowel syndrome, which subsequently increases abdominal pain and constipation.

Perforation

Although extremely rare, the serious complication of spontaneous perforation of organs can occur in vascular EDS (vEDS) and perforation of the digestive tract, for example the small bowel or colon, have been reported. Vascular EDS sufferers who develop sudden-onset stomach pains should seek the advice of a doctor and inform them of the vEDS, so that pre- and post-operative care can be planned appropriately.

Peptic Ulcer

Reassuringly it has been found that the incidence of stomach (peptic) ulcers is not higher in EDS patients.

What tests can be done?

The decision regarding which diagnostic tests are appropriate for you will depend greatly upon a few factors, such as the nature of your digestive problems, what makes them worse or better, the type and location of pain or discomfort you are feeling and any tests or treatments you have already had.

The most common tests used in gastroenterology include:

- Manometry – to study movements of the oesophagus (gullet), small bowel or rectum (back passage).
- 24 hour pH study – to determine if you have excessive acid reflux.
- Gastroscopy and/or colonoscopy – camera tests (endoscopy) on the upper and lower gut respectively, to rule out any inflammation or structural abnormalities. Currently there is no evidence that the risks of endoscopy for hEDS sufferers are any greater than in the general population, as direct comparative studies have not been carried out. Other EDS subtypes should discuss their risk of complications on an individual level with their doctor should an endoscopy be requested.
- Stomach emptying tests – to assess for dysmotility/delayed gastric emptying.

- Hydrogen breath test – to determine if you have excess bacteria in the gut.
- Barium swallow – an x-ray using barium contrast to study the structure and function of the oesophagus (gullet).
- Proctography – x-ray tests of the rectum to assess the function of the rectum and anus when passing a stool. This test can identify any underlying mechanical cause, aiding in the direction of further management of constipation.

Each hospital will have detailed information sheets available for each of these tests. If you are advised that you will be referred for one of these tests, ask your doctor or nurse for more information.

What can be done to help me?

Management options depend on the type of symptoms experienced and to what degree they are bothersome. Treatments for people with hEDS are based on the general principles of managing these symptoms. In our clinical practice we may from time to time use diet-orientated treatments. For example, in patients with bloating and diarrhoea the low-FODMAP diet (diets low in fermentable carbohydrates) may be used. For those with severe constipation, colonic stimulants such as prokinetics can be considered.

It may be that you have already tried many different treatments for your digestive symptoms, yet never found anything that works effectively for you. Sometimes there will be other medications you can try and, when medications are not completely effective at helping to relieve your symptoms, non-medication methods of relieving digestive symptoms can prove very effective for many people. Physical therapy, psychological methods (such as cognitive behavioural therapy), specialist pain management and dietary guidance can all play a part in better symptom management. But just like medications, these methods must be undertaken upon the advice of your doctor and under the appropriate supervision of a professional familiar with your condition. It is also worth remembering that no treatment or therapy is going to ‘cure’ you and not every option may be of benefit for everyone. After assessment your doctor will develop a plan for your treatment which is most suited to you, based on the current available evidence. Often a combination of medicines and non-medicine approaches are used.

Does postural tachycardia syndrome (PoTS) affect the digestive system too?

The autonomic nervous system is the part of the nervous system that controls and regulates many of the organs and functions of the body, such as body temperature,

breathing rate and digestion. Some patients with EDS have symptoms suggestive of involvement of the autonomic nervous system, and the most common of these is PoTS.

In PoTS, the autonomic nervous system can be dysregulated (a bit out of synch), which may be felt as dizzy spells, facial flushing and palpitations (fast heartbeat). These symptoms are often due to a sudden change in posture, but sometimes occur after eating as well. A recent study showed that autonomic symptoms and gastrointestinal symptoms are the two areas most likely to have an impact on the quality of life for EDS sufferers (hypermobile, classical and vascular types).

Preliminary experience continues to suggest that autonomic dysfunction may be a contributing factor to upper digestive tract symptoms, but further research is needed to understand if this is the case. Furthermore, a recent study suggests that stomach emptying may be more rapid in patients with PoTS, but the precise significance of this is currently unclear.

Peer reviewed by: Professor Qasim Aziz, Professor of Neurogastroenterology, Barts and The London School of Medicine and Dentistry and Dr Asma Fikree, Consultant Gastroenterologist, Royal London Hospital

Date of last review: 01/04/2016