Ehlers-Danlos Syndrome: An Overview

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Ehlers-Danlos Syndrome is a rare disease with many types that may present differently. In some cases, there may be dilation and even rupture of major blood vessels. It was discovered by Edvard L. Ehlers from Denmark and Henri-Alexandre Danlos from France. It is alternatively called: Chernogubov's syndrome, Danlos’ syndrome, Meeker-Ehlers-Danlos syndrome, Sack’s syndrome, Sack-Barabas syndrome, Van Meekeren’s syndrome but Ehlers-Danlos Syndrome is the most common, and most correct, name.

Human collagen cannot be regrown by the human. When you reach a certain age, all the collagen you have is all the collagen you’ll ever have (which is also a reason that older people have more joint problems if you wanted an example). There are currently only 3 Ehlers-Danlos specific research projects, none of which involve stem-cell research. The Hypermobility Type of Ehlers-Danlos Syndrome does not have a specific gene mutation recorded, while the Vascular Type does (COL3A1). Currently one of the only treatments is physical therapy but that does not undo the problem it only makes pain more manageable and helps to prevent dislocations. There is no genetic test for EDS Type III. Due to the fact that there is no test medical professions use the diagnoses of Ehlers-Danlos Syndrome Hypermobility Type and Joint Hypermobility Syndrome interchangeably, even though that is incorrect.

Ehlers-Danlos Syndrome causes many symptoms including but not limited to: overly flexible joints that can dislocate, and skin that's translucent, elastic, and bruises easily. Carriers may be undiagnosed due to being asymptomatic or the general unawareness of many medical
professionals. Many people with Ehlers-Danlos, especially the Hypermobility Type (III), have chronic musculoskeletal pain & fatigue and end up diagnosed with fibromyalgia or chronic fatigue syndrome.

Due to the thin veins and vessels caused by Ehlers-Danlos many patients also have Postural Orthostatic Tachycardia Syndrome, or “POTS”. POTS is a form of dysautonomia, which is a dysfunction of the autonomic nervous system. This secondary syndrome causes lightheadedness, low blood pressure, and increased heartbeat upon standing. There is no cure for dysautonomia either. There are few medications for dysautonomia but the most common are midodrine, which is a vasopressor/antihypotensive drug, and salt.

In addition, Ehlers-Danlos Syndrome can cause gastroparesis which is partial paralysis of the stomach. Gastroparesis symptoms can include vomiting, nausea, fullness after eating a small meal, abdominal pain, and weight loss. Many patients who have gastroparesis get a feeding tube. For patients that only need a feeding tube temporarily doctors usually give either a naso-gastric tube (NG) or a naso-jejunal tube (NJ), they both go in through the nose and through the esophagus while the NG stops in the stomach and the NJ goes to the jejunum. When patients require a long term feeding assistance a more permanent tube is surgically placed through the skin into either the stomach (g-tube) or the jejunum (j-tube).

Moreover, patients with Vascular Ehlers-Danlos Syndrome (VEDS) can have many significant or life threatening problems. Due to the lack of collagen around the heart the mitral valve in a syndrome called Mitral Valve Prolapse, which can cause an irregular heartbeat, chest pain, and panic attacks. This Mitral Valve Prolapse can be helped with testing to see if the problem may also be ventricle and/or supraventricle, a series of beta blockers, and abstinence
from things that may make the condition worse. A common death in VEDS patients is organ or arterial rupture. The vessels in a patient’s body can rupture with little force or spontaneously.

Also, patients who are affected by Ehlers-Danlos Syndrome may have significant difficulty with anesthesia. Most common in those affected with the hypermobility or vascular type. These issues are attributed to the thin blood vessels and veins of the patients. This immunity causes yet another case of accused hypochondria.

Now, you may ask: Why should I care? There is a very simple answer, most people don’t know what it is until someone tells them that they have it. A person sitting next to you may be undiagnosed and silently suffering until the day that they finally see a doctor who knows what it is, or until they have problem such as a simple dislocation or an organ rupture that kills them. Studies show that 1 in 5,000 people has Ehlers-Danlos Syndrome but less than 200,000 cases are diagnosed, are you one or someone you know one of those people? People may be unaware and therefore cause themselves harm by doing possibly unnoticeable things such as locking their joints which causes pain and dislocations. As well as patients who don’t have anyone pursuing cures or treatments due to little general knowledge by medical practitioners.

To conclude, Ehlers-Danlos Syndrome is a genetic disease with no cure and no real treatment options. There are many different forms of EDS ranging from Classical to Arthrochalasia. Patients may present with no symptoms, or severe symptoms such as chronic musculoskeletal pain or organ/arterial rupture. Ehlers-Danlos can cause many other syndromes such as Postural Orthostatic Tachycardia Syndrome, which is a form of dysautonomia. Awareness of this disease is very important because it helps with treatments and diagnosis. Very few people are looking into cures and treatments because very few people know what EDS is.
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