These are Ehlers-Danlos syndromes

Ehlers-Danlos syndromes (EDS) are a group of connective tissue gene disorders that produce complex problems across multiple systems of the body. There are no cures.

Connective tissue is the material in the body that binds together, supports, and separates different tissues and organs. Found between other tissues everywhere in the body, it provides strength and flexibility; general functions including transportation of nutrients between cells, immunological defense, and injury repair; and specialized services such as energy storage and blood cell formation. Connective tissue disorders disrupt these most fundamental processes and structures of the body, so resulting problems can be widespread, in a wide range of severities, and affect areas that might seem to be otherwise unrelated.

There are at least six types of EDS; each type of EDS has certain physical traits and with notable exception to the most common form, the hypermobile type of EDS, most types have a known disease-causing gene. The physical characteristics that are common to all types of EDS include hypermobile joints (joints that move in greater amounts than expected) and skin involvement (soft, stretchy, saggy, too thin, easy bruising, easy wounding, poor wound healing, or atrophic scarring). EDS is known to affect more than one in 2,500 men and women, of every race and ethnicity.

Early diagnosis is crucial to positive patient health. Symptoms can be treated as they arise. Care is largely preventative, to support and manage EDS with the intent of keeping damage as minimal as possible. Specifics have to be tailored to those symptoms exhibited in the person with EDS. Even knowing what type EDS one has, each case of EDS is individual.

These are the types of Ehlers-Danlos

Most but not all of the genes responsible are identified, and new genes are still being identified in rare forms of EDS. Current diagnostic information for each type can be found on www.ehlers-danlos.com/eds-types. Diagnostic criteria don't define what the experience of EDS will be, as there are many more symptoms than criteria.

HYPERMOBILE [distinctive cause unidentified]: Most prevalent EDS type; Hypermobile EDS may be the most common heritable disorder of connective tissue, perhaps affecting more than one in 1000 people. Generalized joint hypermobility, skin involvement (possible hyperextensibility or smooth/soft skin), chronic joint pain, recurrent joint dislocations.

CLASSICAL [*COL5A1, COL5A2, COL1A1*]**:** Rare; second-most prevalent. Variable skin hyperextensibility, widened atrophic scars (tissue fragility), joint hypermobility, easy bruising; ± mulluscoid pseudotumors, subcutaneous spheroids; occasional internal organ fragility.

VASCULAR [*COL3A1*]: Rare. Thin, translucent skin, arterial/intestinal/uterine fragility or rupture, extensive bruising, characteristic facial appearance; ± acrogeria, hypermobility of small joints, early onset varicose veins, and pneumothorax. Vascular EDS is the most serious type due to the possibility of arterial or organ rupture. Minor trauma can lead to extensive bruising and skin tears. Arterial rupture is the most common cause of sudden death; life expectancy is shortened. Complications may occur during and after surgery.

KYPHOSCOLIOTIC [*PLOD1*]: Very rare. Generalized joint laxity, severe muscle hypotonia at birth, scoliosis at birth (progressive), scleral fragility and rupture of the ocular globe; ± tissue fragility, easy bruising, arterial rupture and osteopenia.

ARTHROCHALASIA [*COL1A1, COL1A2*]: Very rare. Severe generalized joint hypermobility with recurrent subluxations, congenital bilateral hip dislocation; ± skin hyperextensibility, tissue fragility (atrophic scars), easy bruising, muscle hypotonia and osteopenia.

DERMATOSPARAXIS [*ADAMTS2*]: Extremely rare. Severe skin fragility, sagging redundant skin; ± soft doughy skin texture, easy bruising, premature rupture of fetal membranes, and large hernias (umbilical and inguinal).

Other known rare forms of EDS include, among others, *TNX*-deficient, progeroid, musculocontractual, spondylocheirodysplastic, *FKBP22*-deficient, periodontitis, and brittle cornea syndrome.

These are complications of Ehlers-Danlos

Autonomic Disorders and Fatigue: Many, perhaps a majority, of people with EDS struggle with body processes that are supposed to happen automatically, like breathing, blood pressure and heart rate, body temperature, and digestion. In EDS, this most often is postural tachycardia syndrome (PoTS) — when we stand up, we can find a range of problems like lightheadedness, high heart rate, weakness, even fainting. Fatigue can be a bigger problem than even our constant pain. Many of us have what's called "brain fog" — feelings of confusion, forgetfulness, and a lack of focus and mental clarity.

Cardiovascular Issues: These can include easy, possibly extensive bruising; varicose veins and blood pooling; prolonged bleeding; and spontaneous rupture of large arteries. Vascular EDS has particularly severe effects.

Chronic Pain Disorders: For many of us with EDS, widespread pain is a constant companion. Our bodies never get a chance to heal; when our joints move

too far (which can happen many times in a day), all the tissue connected to those joints is stretched too far, so we endure a chronic acute pain as we injure ourselves over and over – which makes treating our pain a challenge.

Dental and Jaw Issues: The temporomandibular joint connecting the jaw to the skull can dislocate too. A slipping TMJ can be painful, restrict eating, and affect major nerves. Many with EDS are prone to dental problems, some resulting from fragile gum tissue and periodontitis, others from the teeth themselves, including enamel fractures and tooth mobility.

GI Functional Disorders: The gut is one of the largest organs, and is made of connective tissue. EDS can cause functional disorders like delayed stomach emptying and gastric reflex, irritable bowel syndrome, and less obvious problems such as nutrient absorption and food allergies.

Immune System Complications and Allergy: There is an unexplained correlation between EDS and mast cell problems – half of mast cells reside in connective tissue. Mast cell disorders affect functions in potentially every organ system. When they abnormally release chemical mediators, a range of chronic symptoms results: skin rashes, flushing, nausea, vomiting, asthma, vascular instability, and sometimes anaphylaxis or near-anaphylaxis attacks.

Joint and Spine/Orthopedics: Hypermobile joints cause injury and long term wear. This includes the junction where the spine joins the skull, leading to a variety of problems with stability that may include Chiari malformation. Neck muscles and ligaments can irritate, compress, or pull on the nerves and blood vessels around the base of the skull, referring pain elsewhere. Those with EDS often have low bone density, starting much earlier in life than most would suspect.

Obstetrics and Gynecology: Issues are reported by women with EDS at a much greater rate than general. Depending (as almost always) on the type of EDS, these can include irregular menses, rapid progression of labor and delivery, pelvic prolapse, and premature membrane and uterine rupture.

Skin and Tissue: Skin can be soft and velvety; stretch further than it should; be translucent and fragile, tearing and bruising easily; and scar severely. Wounds may heal slowly and poorly.

EDS can also lead to many other complications, including metabolic, neurological, psychosocial, and sleep issues.

The Ehlers-Danlos Society

The Ehlers-Danlos Society is a global community of patients, caregivers, medical professionals, and supporters, dedicated to saving and improving the lives of those affected by the Ehlers-Danlos syndromes and related disorders.

We support collaborative research initiatives, awareness campaigns, advocacy, community-building, and care for the EDS population.

Our goals are worldwide awareness – and a better quality of life for all who suffer from these conditions. Research is at the center of what we do, so that one day we will have a cure.

Our strength begins with hope.

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What are Ehlers-Danlos syndromes?

The **Ehlers Danlos** Society™