Ehlers Danlos Syndrome

This syndrome belongs to a group of genetic connective tissue disorders.

Connective tissue is any type of biological tissue with an extensive extracellular matrix that supports, binds together, and protects organs.

In biology, the extracellular matrix is a three-dimensional network of extracellular macromolecules, such as collagen, enzymes, and glycoproteins, that provide structural and biochemical support of surrounding cells.

These tissues form a framework, or matrix, for the body, and are composed of two major structural protein molecules: collagen and elastin. There are many different types of collagen protein in each of the body's tissues.

Connective tissue diseases can have strong or weak inheritance risks, and can also be caused by environmental factors.

Collagen is the main structural protein in the extracellular space in the various connective tissues in the body. It is mostly found in fibrous tissues such as tendons, ligaments and skin.

Depending upon the degree of mineralization, collagen tissues may be rigid (bone), compliant (tendon), or have a gradient from rigid to compliant (cartilage). It is also abundant in corneas, blood vessels, the gut, intervertebral discs and the dentin in teeth.

A group of genetic disorders cause Ehlers-Danlos syndrome, which results in a defect in collagen production.

In most cases EDS is an inherited condition. The minority of cases are not inherited. This means that they occur via spontaneous gene mutations. Defects in the genes weaken the process and formation of collagen.

All of the genes listed below provide instructions on how to assemble collagen, except for ADAMTS2. That gene provides instructions for making the proteins that work with collagen. The genes that can cause EDS, while not a complete list, include:

- ADAMTS2
- COL1A1
- COL1A2
- COL3A1
- COL5A1
- COL6A2
- PLOD1
- TNXB

People with EDS may initially be misdiagnosed with hypochondriasis, depression, PoTS (postural tachycardia syndrome) or chronic fatigue syndrome.

What are the symptoms of EDS?

•**Fragile tissue**:

Skin tearing,
small blood vessels bursting,
easy bruising,
osteopenia (weaker than normal bones)

•**Pain**:

Joints and muscles affected,
loss of elasticity,
disrupted signalling
(e.g. proprioceptive dysfunction (difficulties in sensing where body parts actually are, clumsiness),
dysautonomia (malfunction of the autonomic nervous system, which is responsible for maintaining a constant internal temperature, regulating breathing patterns, keeping blood pressure steady, and moderating the heart rate. It is also involved in pupil dilation, sexual arousal, and excretion),
and dystonia (a movement disorder where muscles contract uncontrollably. The contraction causes the affected body part to twist involuntarily, resulting in repetitive movements or abnormal postures.))

•**Other major symptoms**:

Fatigue,
haemorrhaging (bleeding),
constipation,
gastro-oesophagal reflux,
dyspnea (shortness of breath/ 'air hunger') ,
respiratory and sleeping disturbances,
random bradypnea (abnormally slow breathing rate),
tachycardia (heart rate exceeding normal rate)
bradycardia (heart rate slower than normal rate),
unstable blood pressure,
cognitive difficulties (eg brain fog)

Collagen holds the body together, so it's no wonder EDS patients' symptoms can be so diverse. With numerous medical specialities, it's easy to miss a collagen defect presenting in various ways within potentially every bodily system.

Not all EDS patients suffer from frequent dislocations.
A subluxation is an incomplete or partial dislocation of a joint or organ.
People with EDS may be constantly subluxing joints, and not be aware of it because to them it is normal. Subluxations in EDS involve the joints easily sliding in and out of position.

Treatment and management of EDS

There is no known cure. Treatment is supportive in nature. Physical therapy and bracing may help strengthen muscles and support joints.

While some forms of EDS result in a normal life expectancy, those that affect blood vessels generally decrease life expectancy.

The genetically non-typical collagen causes no degeneration in the body.
It is the damage that accumulates from the body compensating for the non-typical collagen which results in degeneration.

Collagen is stretchy-gluey stuff so a rubber band analogy may aid understanding.
Most of the time when you stretch a rubber band, it goes back to its original shape and position. However, as the rubber band ages the time taken for it to return to that original shape and position lengthens. Then eventually either the rubber band stretches and stays stretched or it breaks.

Collagen is affected by hormones, so its natural strength waxes and wanes. This means someone with EDS will have good days, bad days, and truly horrible days. Collagen is definitely impacted in a woman by her menstrual cycle, strongest at ovulation, weakest at pre-menstruation.

If you have seen a puppet or marionette on a set of strings, it is a useful image for describing EDS. Because the usual connective tissue is weak (tendons, ligaments etc), the muscles have to do extra duty to keep the body in alignment. With weak connective tissue the default is for the body to slump, so it takes extra effort from the muscles (and consequently extra energy) to stop the body from slumping.

Someone with EDS has to tread a fine line between not doing too little and not doing too much. Keeping still isn't an option because that reduces muscle tone. At the same time doing anything that over-stretches the muscles risks a subluxation, dislocation or muscle damage. This is why getting into and out of a vehicle can be very troublesome and why carrying anything remotely heavy is risky.

For the same reason keeping away from crowds, where the risk of being bumped out of alignment is high, is a good thing. Anything that can be done to prevent a person with EDS from being pushed or pulled beyond what they know his/her body can take is important. They know what pace is within their limits, so any request for them to hurry is ultimately counter-productive.

Soft flexible braces to help the muscles keep the body in alignment and prevent major subluxations and dislocations are good, but can often be beyond the person's ability to put the braces on by themselves.

Mobility aids can be helpful on bad days, or when traversing distances beyond what energy levels can tolerate. However, if by using the walking stick or activating the wheels on the wheelchair too much pressure goes on the various parts of the upper limbs, any benefit can be negated. It is all a very fine balancing act to get the optimum mix of aids and effort.

The pain levels a person with EDS experiences are similar to or worse than rheumatoid arthritis.

Non-typical collagen has a big impact on the digestive system because collagen is a major component of the valves that open and shut in the various parts of the digestive tract, and in connecting the muscles that push what is inside the digestive system along. So if the valve between oesophagus and stomach isn't working well, gastric reflux happens (heartburn); if the stomach doesn't empty properly then bloating happens; if the contents move too slowly then pooling happens, the contents get harder and constipation results. With this kind of scenario for constipation you can also get the situation where there are spurts of watery leakage around hardened stools.

The jaw is also subject to subluxation and dislocation which can impact upon chewing and speech.

Surgery for someone with EDS is very risky due to how fragile and prone to tearing his/her connective tissue is. Recovery times are also longer than average post-surgery. In particular, orthopaedic surgeons need to be reminded that their EDS patients have non-typical muscle function.

Extreme caution should be exercised in helping get someone who has EDS up from a fallen position. Anyone likely to face such a task (eg family) should study and get training in how to help someone with EDS up from a fall correctly and/or be familiar with how to call for an ambulance and inform them how prone to subluxation and dislocation the patient is.

Getting in to and out of chairs and beds and vehicles is always something a person with EDS is cautious in doing. Should they begin to experience sudden changes in blood pressure when doing these activities, then they will need someone to watch over them carefully when they attempt these activities for the purpose of preventing falls.

If at all possible, exposure to infectious people should be reduced. The last thing a person with EDS wants is the extra worry of getting neck and jaw out of alignment due to sneezing or coughing.

Advancing stages of degeneration may see trouble with swallowing, voicing and speaking. To suddenly not be able to call for help is a frightening thing, so plans should be put in place to minimise this eg. Small bell to ring; ICE button on mobile phone set up, someone else within line of sight, one of the various Vital Call or Medical Alert systems in place.

When swallowing becomes troublesome, dental care becomes crucial due to food parts remaining in the mouth that didn't get swallowed. Mouth washes and similar may be needed. Dentists need to be informed why a patient cannot open mouth as wide as others and why their dentin is more fragile.

Physiotherapy with someone who understands the need for gentle exercise to maintain muscle tone, and can give guidance as to how to exercise gently at home, will be beneficial and help maintain quality of life.

Anything that can be done to help a person with EDS to keep their body in good posture is beneficial.

Even a regular person tires of holding a phone to the ear for too long. Some kind of blue-tooth ear piece for phone calls would make receiving anything longer than a short phone call more comfortable for a person with EDS.

Sometimes a person with EDS will experience bouts of 'brain fog' and disorientation. It is something they have no control over.

Rearranging a bathroom so that it is possible to shower while sitting down, can reduce levels of muscle fatigue in people with EDS.

Headaches and migraines are rather common, and sensitivity to light (photophobia) caused by dry eyes is also frequently reported by people with EDS.

Advancing stages of degeneration will see people with EDS deciding to no longer drive vehicles. This is because the fast and sudden requirements to put on the brakes or to swivel the head to check for danger become more difficult, and the increasing risk of having a dislocation that takes time to fix (which produces the fear 'Will I be able to get home?')