Classical Ehlers-Danlos Syndrome (EDS)

Within the system of Classical Ehlers-Danlos Syndrome (EDS), there were initially 6 subtypes. However, with the identification of additional genetic and biochemical markers, in 2017 the classification was revised to include 13 types. We previously discussed vascular EDS in **the last blog**. In this blog we will discuss classical EDS.

With classical EDS, the major diagnostic criteria includes skin hyper extensibility, widened atrophic scars and join hyper mobility. It should be noted that the severity varies even in family members.

Certain proteins called collagen, which provides strength and structure to the extracellular matrix of tissues and organs. The genes that are associated with classical EDS are passed on with autosomal dominant inheritance, which means you only need one parent to get the gene.

The major criteria include:
1. Skin hyper extensibility and atrophic scaring.
2. General hyper mobility (a Beighton score of 5 or more)

Minor criteria include:
1. Easy bruising
2. Soft doughy skin
3. Skin Fragility or traumatic splitting
4. Mulluscoid pseudotumors, which are fleshy lesions associated with scars at pressure points.
5. Subcutaneous spheroids, which are small round hard bodies that are mobile and commonly located on the forearms and chin.
6. Hernia or history of hernia
7. Epicanthal folds
8. Complications of joint hyper mobility such as sprains, subluxation, pain or flexible flat foot.
9. First degree relatives who meet clinical criteria.

Minimal criteria suggestive of classical EDS include: skin hyper extensibility and atrophic scaring plus generalized joint hyper mobility and 3 or more minor criteria.

Genetic conformation is required for definitive diagnosis. More than 90% of classical EDS patients labor a mutation of one of the genes that encode for type V collagen. A reduction in type V collagen is central to the pathogenesis of classical EDS.

While musculoskeletal joint hyper-mobility is present in classical EDS, the skin is the key to establishing the diagnosis of classical EDS. The skin is hyper extensible and soft with severe atrophic scarring and hemosiderin deposits, or brown areas over the shin and extensor surfaces, due to easy bruising. Poor wound healing is often seen in classical EDS.

A characteristic facial feature has been described. These are epicanthic folds, excess skin on the eyelids, a prematurely aged appearance and scars on the forehead and chin. Absence of striae or stretch marks has also been noted in classical EDS patients.

Other problems are gastrointestinal problems, most commonly nausea, vomiting and gastroesophageal reflux, followed by chronic constipation. Abnormalities in the cornea are also found in classical EDS with thin and steep and transparent corneas.