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Ehlers-Danlos Syndrome

Ehlers-Danlos Syndrome is a genetic defect which encompasses a spectrum of conditions and clinical manifestations. Described in 1901 by Danish dermatologist Edward Ehlers and respectively in 1908 by French dermatologist Henri-Alexandre Danlos, the co-named Ehlers-Danlos Syndrome is a multi-systemic disorder that primarily affects skin, ligaments, joints, blood vessels, and internal organs. This heritable connective tissue disease generally results either from mutations in genes that encode fibrillar collagens or the enzymes involved in collagen synthesis. Fibrillar collagens consist of collagen types I, II, III, V, and XI, and provide the strength and structure to the extracellular matrix of all the tissues and organs of the body.

Ehlers-Danlos Syndrome is classified into 6 subtypes, differentiated by the defective collagen type involved and the pattern of inheritance: Classical, Hypermobility, Vascular, Kyphoscoliotic, Arthrochalasia, and Dermatosparaxis. Each subtype is accompanied by a set of major and minor diagnostic criteria with the classical, hypermobility, and vascular types being the most common and the latter three occurring very rarely. The hereditary pattern of EDS will either be autosomal dominant, autosomal recessive, or dependent on the X chromosome.

Although each subtype of EDS has a different clinical presentation, there are some general characteristics which present in each in varying degrees. These features include: smooth velvety hyperextensible skin, (or) thin transparent fragile skin, joint hypermobility, muscle hypotonia, and easy bruising. The patients with the thin fragile skin type will often display widened atrophic scars from minor trauma, and joint hypermobility often leads to subluxations, sprains, and chronic joint pain.

The incidence of EDS is 1 in 20,000-100,000 (depending on the subtype). This syndrome is not age, gender, or race specific. An afflicted individual is born with the defect and depending on the severity of the EDS subtype may or may not be diagnosed in childhood. Some patients show a very severe phenotype that is recognizable at birth, while other diagnoses are delayed due to a milder condition. In conjunction with the clinical manifestations, various imaging studies are used in diagnosing EDS including CT scanning, MRI scanning, and ultrasonography. However to confirm a suspected diagnosis of EDS, ultrastructural examination of the collagen fibrils is completed alongside direct DNA analysis. The different subtypes of EDS can be confused with multiple other conditions. The differential diagnosis should include fibromyalgia, Aarskog-Scott syndrome, fragile X syndrome, congenital flaccidity of the skin (cutis laxa), osteogenesis imperfecta, achondroplasia, and other diseases of the muscles and nervous system. Genetic testing can be used to locate or rule out a specific molecular defect.

For patients with Ehlers-Danlos Syndrome a multidisciplinary approach is needed for treatment. Patients will require a cardiologist, physiotherapy, pain management, and psychological support. The EDS subtype with the worst prognosis is the vascular type where hollow organs and arteries are likely to rupture at a young age. Vascular fragility is of main importance in managing these patients; arterial rupture and sudden death often result in their 3rd and 4th decades. Surgical interventions are discouraged, and manipulation of tissues should be done with extreme care. Conservative therapy is recommended and pregnancy is not advised. Studies show that desmopressin may be effective in bleeding associated with EDS, and high doses of vitamin C can be used adjunctively to treat the kyphoscoliotic subtype. Even in patients with a "milder" form of EDS, the recurrent joint dislocations and chronic joint pain accompanying physical activity often leads to social isolation and depression requiring counseling and sometimes medication.

As previously stated, the prognosis for patients with vascular type EDS is poor with a median age of death being 33, usually by arterial rupture. With vessel rupture, vascular interventions are performed with a survival rate of just over half. With all EDS subtypes there is high risk of uterine rupture and premature birth associated with pregnancy, and a pregnancy related mortality rate ranging from 12-25%. Regardless of treatment, patients often have excessive scarring, wrinkles and overly-aged appearance, tooth-loss, crippling joint pain, and generalized weakness. Joint hypermobility worsens in severity over time and becomes disabling with repetitive dislocations.

As dental hygienists, health care professionals working on the "front lines" of the public health system, we encounter patients with a multitude of conditions and need to be able to recognize and effectively treat these individuals. Patients with Ehlers-Danlos Syndrome are prone to developing periodontitis, as EDS is a defect in collagen; they often lose periodontal tissues including the PDL and alveolar bone. Patients with EDS have a poor healing response and bleed easily, manipulation of the oral tissues should be kept to a minimum and very conservative measures should be taken. Patients with EDS bleed so readily, most report abnormal bleeding with tooth brushing. The focus of patient treatment with EDS is prevention and homecare education, along with appropriate pain management and maintaining patient comfort. Despite the preventative measures taken, these patients still often lose their teeth very early. Additionally, without the proprioceptive effects of collagen they are prone to self-injuries, and often have difficulty brushing because of weakness and inability to manipulate the tooth brush. These patients also suffer from serious TMJ problems which make chewing and speaking painful. Careful effort must be made to avoid subluxation of the jaw during treatment.

SOURCES

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