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Ehlers-Danlos Syndrome

Pathology DEN2322

Ehlers-Danlos Syndrome (EDS) is a group of genetically inherited disorders that affect the connective tissue. There are six major types of EDS. EDS occurs in about 1 in 5000 births worldwide. Experts believe that EDS is under-diagnosed due to the wide range of severities in the disorder. Most cases have overlapping symptoms which makes it difficult to categorize. Although there are six types, the prevalence within these types varies dramatically. The most common types are the hyper mobility and classical types. EDS does not have prevalence amongst a particular gender, race or age.

EDS is caused by a defect in the synthesis of collagen. Collagen helps tissues resist deformation and gives tissue strength and elasticity. Collagen is the main component of connective tissue which accounts for approximately 45% of total body weight. This makes EDS affect a wide range of tissue, from the skin, joints, and musles to blood vessels and organs. The following chart is short summary of the different types of EDS and the involvement on the body:

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| Hypermobility | Mainly involves joints. Frequent joint dislocation and subluxation. Pain is common. |
| Classical | Similar symptoms to Hypermobility but involves the skin |
| Vascular | Life threatening. Involves blood vessels and organs. Prone to tearing and rupture. Easily bruised, translucent skin. |
| Kyphoscoliosis | Very rare. Scoliosis, fragile eyes, severe muscle weakness. |
| Arthrochalasia | Very rare. Very loose joins and dislocations involving the hips. |
| Dermatosparaxis | Very rare. Extremely fragile and sagging skin. |

Clinically, many people with EDS have velvety skin that is very elastic and fragile. As stated in the chart, the Vascular type of EDS bruise easily and have translucent skin. Classic type EDS patients may have abnormal shallow scarring. Dermatosparxis EDS is characterized by saggy and wrinkly skin. A diagnosis can be made by clinical observations but further tests such as collagen gene mutation testing and biopsy is needed to further categorize which type of EDS a person has. The symptoms of EDS can vary from mild to life threatening. Some symptoms can severely restrict a person's daily life. Chronic musculoskeletal pain, instability in joints, and scoliosis can limit a person's mobility. Autonomic dysfunction and cardiovascular problems are secondary conditions that a person with EDS may develop. There is no cure for EDS, but surgical intervention as well as palliative treatments, such as casts and braces, can be used to stabilize joints. Physical therapy can also be used to assist in strengthening muscles.

The symptoms of EDS can be easily mistaken for other disorders such as Marfan syndrome. In Marfan syndrome, there is instability in the joints and patients have similar cardiovascular problems. Clinical findings of those who have Marfan Syndrome are tall with long arms and legs but clinical findings for EDS are short in stature. Another disease that EDS is mistaken for is Cutis laxa, where the skin is loose, saggy and wrinkled. However, people with EDS have elastic skin, when pulled, it will return to its original state. Patients are sometimes misdiagnosed with fibromyalgia and other bleeding disorders due to the similarities of weak blood vessels of those who have EDS.

In the article *Oral Phenotype and Scoring of Vascular Ehlers-Danlos Syndrom: a Case-Control Study,* concentrates on the specific vascular type of EDS and its connection to gingival recession. This study was done on 17 patients with vEDS. A thorough dental assessment was completed on these patients, the results were then compared to a set of age and sex matched control patients who did not have vEDS. Contrary to the assumption that patients with vEDS will have gingival recession, the results of this study showed little evidence of that. It did, however, show other oral deformities that were prevalent in vEDS patients.

Over 80% of the patients with vEDS had TMJ disorders as oppose to only 24% of the control. Radiographs showed many dentin deformities prevalent in vEDS patients such as decreased pulp volume, thistle-shaped pulp chamber, increased length of mandibular molar roots and root fusion. vEDS patients also showed more gingival thinness and bleeding upon probing compared to the control group. Although this study showed interesting results, the study group was too small, with only 17 participants who had vEDS. The sex ratio was unbalanced and there were no children or teenager participants which limits the data collected.

*Oral Health in Prevalent Types of Ehlers-Danlos Syndomres,* is another article that studied the oral health of EDS patients. Instead of concentrating on one specific type of EDS, this study was executed on the three more common types of EDS - hypermobility, classical and vascular. All of the EDS patients had TMD, caries rate was high and their overall oral hygiene was poor compared to the control group. Patients had increased mucosal fragility; over half of the patients had lesions from prolonged and poor tissue healing. Like the other study, root deformities were found in EDS patients as well as abnormal pulp shape, and obliteration of the pulp chamber.

These studies future shows the importance of dental assessment and a clear medical history. As a hygienist I need to pay close attention to the details the patient is giving me and the data I collect form the assessment. Everything I see and hear will aid me in helping my patients to achieve optimum oral health. Due to this disease's symptoms that may hinder a person's movement, oral health may be severely compromised. It will be my responsibility to introduce different types of methods and dental aids to help them achieve good oral hygiene. Another issue that arises with EDS patients is TMJ problems. Due to unstable joints in EDS patients, their TMJ can be easily dislocate making it difficult for patients to keep their mouth open. Therefore it is important to communicate and be cautious when dealing with EDS patients.