**Ehler-Danlos Syndrome Type VII C/ Dermatosparaxis**
By Ita Glick
Oral Pathology 2018
Section: Thursday AM

 **Overview**
Ehlers-Danlos Syndrome Dermatosparaxis type 7C is a heterogenous inherited connective tissue disorder, characterized by skin texture abnormalities, vascular and soft tissue fragility and joint hypermobility. The underlying molecular defect can also manifest itself within organs in a more severe way.

**Etiology**
Ehlers-Danlos Syndrome Dermatosparaxis type is a genetically autosomal recessivly inherited connective tissue disorder, caused by a deficiency in the ADAMTS-2 enzyme activity. This enzyme splits the amino terminal propertide domain of type I, II and III procollagen. The ADAMTS-2 gene helps process the procollagen molecules type I, II, and III which are the precursors to the collagen protein. The collagen provides the structure and strength for the connective tissue. Patients with this gene mutation, have a defect in this protein and the connective tissue is weakened. This causes all kinds of skin abnormalities such as really fragile, almost dough-like skin that bruises easily or joint hypermobility and instability.

**Clinical Presentation**
There are many different clinical presentations that patients with Ehlers-Danlos Syndrome may display. There are major criteria and well as minor criteria. The major criteria for those to be diagnosed with the Dermatosparaxis subtype include severely fragile skin, sagging and redundant skin at the wrists and ankles, and skin tearing either at bith or within the first few years of life. Craniofacial features that are indicative of this disease, are congenital or postnatal and progressive. Examples of these are swelling of the periorbital soft tissue with prominent protuberant eyes and puffy edematous eyelids, blueish sclera, down-slanting palperbral fissures, delayed frontal closure, epicanthic folds, micrognathia, wide cranial sutures, and/or hypoplastic chin. Often it will present with severe bruisablility and other risks of subcutaneous hemmorages and hematomas. Short limbs, severe palmer wrinkling, umbilical hernia, postnatal growth retardation and perinatal complications are also inherent to this disease. Some of the complications due to the fragile connective tissue include skull fractures, intracerebral hemorrhage, friable umbilical cord, congenital skin tears and neonatal pneumothorax. Minor criteria include soft doughy skin texture, osteopenia, joint hyperextensivity, premature rupture of fetal membranes resulting in preterm birth, atrophic scars, rupture of the bladder or diaphragm, myopia, and delayed motor development. There have also been some periodontal manifestations seen clinically in patients with dEDS such as severe gingival hyperplasia in both jaws, nodular, fragile, and inflamed gingival enlargement, mobility of the TMJ, hypodontia, microdontia, abnormal morphology of molars, and generalized opalesence tooth discoloration. In order to be diagnosed with dEDS someone must present with two major criteria: 1) extreme skin fragility, and 2) the craniofacial features listed above. Patients must also have three of the minor criteria indicated earlier, and/or one other major criteria.

**Demographic**
The demographic of people with dEDS is extremely rare and there are only 15 diagnosed cases known to date as of September 2016. The ratio is more male dominant with a total of 11:4. Because this is a connective tissue disease that has an inheritance pattern that is autosomal recessive it is something that a patient is born with and clinical features are recognized at birth or during early childhood. This disease is congenital; the abnormal mutation of ADMTS2 gene cannot be contracted midlife. As of the research we have today, there are no known or specific race popularity for Deramtosparaxis type EDS.

**Biopsy / Histology / Radiographs**
A skin biopsy is taken, and under an electron microscope the histology shows characteristics of hieroglyphic fibrils. The research does not show any data of using radiographs as a means of diagnosing of Ehlers-Danlos Syndrome.

**Differential Diagnosis**

In order to properly diagnose a patient with Dermatosparaxis type Ehlers-Danlos Syndrome a genetic test must be done to find the abnormal gene mutation of ADAMRS2. A skin biopsy is not enough to diagnose someone with this Dermatosparaxis EDS because the hieroglyphic pattern is present with the Arthrochalasia type EDS as well and will look almost identical to dEDS microscopically.

**Treatment**
There is no cure for this syndrome, although there are different treatments that are available to keep the patient with EDS as stable and healthy as possible. Different options can alleviate symptoms such as surgery to remove an umbilical hernia. Using a wheelchair, leg braces or walker can relieve severe joint instability. Physical therapy is often prescribed for joint hypomobility. Padding or bandages can protect skin areas like knees, shins or forehead. Special care should be taken with the patient’s loose skin, to prevent skin tearing and bruising. Rough contact sports or physical activity should be avoided due to patients skin sensitivities.

**Prognosis**
There is no prognosis for patients with Dermatosparaxis Ehlers-Danlos Syndrome. From the research that I have seen this condition is extremely rare. Patients who suffer from dEDS live with extreme discomfort; perhaps with continued research and development in new therapies they can achieve a better quality of life.

**Professional Relevance**
As a hygienst it is important to look at the bigger picture in clinical practice. Firstly, it is imperative to evaluate a patient as they sit in the chair to see if they have any conditions, and to assess its effects on their cognitive abilities. Often people assume that those with facial abnormalities are impaired mentally. This is not the case with EDS patients as it is not for many other diseases. Secondly, if a patient presents in clinical practice with Dermatosparaxis Ehler-Danlos Syndrome the hygienist must be extra careful to be gentle with him/her because of their extremely sensitive skin. The hygienist should also recognize their dental anomalies mentioned earlier as being part of their greater condition. A patient with this connective tissue disorder may also have difficult dexterity because of their doughy, shortened, bruised fingers and joint hypomobility. When using anesthesia to treat a patient it is important to consult with the patient’s EDS specialist first regarding the risks of bleeding, and ineffectiveness of local anesthetic in the past. If the patient has muscle weakness, their neuromuscular blockade should be monitored before emergence of the anesthesia. If the patient is immobile, one should avoid using depolarizing agents (succinylcholine). Adhesive tapes or wound dressings for attaching devices should be easily removable or avoided because of the risk of skin damage. The postoperative care must include carefully sitting the patient up and helping them out of the chair to reduce the risk of joint luxation. Patients should be checked thoroughly for development of bleeding or hemotomas at the operating site.

**Citations**
1)Sobey, Glenda. “Ehlers–Danlos Syndrome: How to Diagnose and When to Perform Genetic Tests.” Archives of Disease in Childhood, vol. 100, no. 1, Mar. 2014, pp. 57–61., doi:10.1136/archdischild-2013-304822.

2)Damme, Tim Van, et al. “Expanding the Clinical and Mutational Spectrum of the Ehlers–Danlos Syndrome, Dermatosparaxis Type.” Genetics in Medicine, vol. 18, no. 9, 2016, pp. 882–891., doi:10.1038/gim.2015.188

3)Wiesmann, Thomas, et al. “Recommendations for Anesthesia and Perioperative Management in Patients with Ehlers-Danlos Syndrome(s).” Orphanet Journal of Rare Diseases, vol. 9, no. 1, 2014, doi:10.1186/s13023-014-0109-5.

4)Kapferer-Seebacher, Ines, et al. “Periodontal Manifestations of Ehlers-Danlos Syndromes: A Systematic Review.” Journal of Clinical Periodontology, vol. 44, no. 11, 2017, pp. 1088–1100., doi:10.1111/jcpe.12807.

5) Brady, Angela F., et al. “The Ehlers-Danlos Syndromes, Rare Types.” American Journal of Medical Genetics Part C: Seminars in Medical Genetics, vol. 175, no. 1, 2017, pp. 70–115., doi:10.1002/ajmg.c.31550.

6) “Dermatosparaxis Ehlers-Danlos Syndrome.” Genetic and Rare Diseases Information Center, U.S. Department of Health and Human Services, rarediseases.info.nih.gov/diseases/2089/dermatosparaxis-ehlers-danlos-syndrome.