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Ehlers Danlos Syndrome - A Review

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Abstract:

The study aims in reviewing the causes, symptoms, prevalence and diagnosis of Ehlers Danlos syndrome.Ehlers– Danlos syndrome (EDS) is an inherited connective tissue disorder.It is caused by a defect in the structure, production, or processing of collagen or proteins that interact with collagen, such as mutations in the COL5A or COL3A genes.The extra oral manifestations of EDS are the presence of scarring on the chin and forehead, a history of repeated luxations of the TMJ, epicanthus, hypertelorism, a narrow curved nose, sparse hair and hyper-elasticity of the skin.As fragile as the skin, the mucosa tears easily when touched by instruments. Periodontal tissues lead to the premature loss of deciduous and permanent teeth.

Keywords :Ehlers Danlos Syndrome, Hypermobility joints, Collagen fibres, hyperelastic skin.

INTRODUCTION :

Ehlers - Danlos syndrome (EDS) belongs to a heterogeneous group of disorders of collagen characterised by hyper extensibility and fragility of the skin with easy bruisability and hyper mobility of the joints [1]. Following the identification of specific mutations in the genes encoding collagen types I, III, and V, as well as several collagen processing enzymes, the EDS classification scheme was collapsed into six distinct clinical syndromes (2).People with EDA are often misdiagnosed with hypochondriasis,depression,chronic fatigue syndrome,and other conditions [3] because there is generally poor knowledge about EDS among practitioners [4,5,6]. Signs vary widely based on which type of EDS the patient has. In each case,however,the signs are ultimately due to faulty or reduced amounts of collagen.EDS typically affects the joints,skin,and blood vessels[7].The classification system for EDS included 10 specific types and also acknowledged that other extremely rare types existed. At this time, the classification system underwent an overhaul and was reduced to 6 major types using descriptive titles [8]. These six variants are:

a) classical - Skin and joint hyper mobility,Atrophic scars,Easy bruising
b) Hyper mobility- Joint hyper mobility,Pain,Spontaneous dislocations
c) Vascular- Thin skin,Arterial or uterine rupture,Bruising,Small joint hyper-extensibility
d) Kyphoscoliosis- Hypotonia,Joint laxity,Congenital scoliosis,Ocular fragility
e) Arthrochalasia- Severe joint,Hyper-mobility,Skin mild with bruising,Scoliosis
f) Dermatosparaxsis- Severe skin fragility,Cutis laxa,Bruising[12]

DIAGNOSIS / CLINICAL SYMPTOMS:

EDS individuals may have one or more of the following symptoms/conditions

- Hyperextensibility of the joints: Extended, flexible joints, circus-like contortions, vulnerability to sprains, and spontaneous dislocations of joints [13].
- Hyperextensibility of the skin: Hyperextensibility of the skin, gaping wounds even in relatively minor trauma, poor wound healing and the development of severe scar tissue with excessive pigmentation known as keloids [14].
- Vascular changes: Arteries lose tensile strength leading to aneurysms (ballooning of the blood vessels) [13].
- Pulmonary alterations: The development of thick, sticky mucus secretions within the lung tissue (cystic lung disease), fluid accumulation and weakening of the lung adherence to the chest wall (subpleural blebs) and fluid-filled blisters at the top portions of the lungs (apical bullae), which can lead to collapse of the lung unrelated to pressure- or physical-induced trauma (spontaneous pneumothorax) [15].
- Dental abnormalities: Partial absence of the teeth (hypodontia), delayed eruption of permanent teeth, and malformation of the dentin (the chief material of the tooth which is harder and denser than bone that surrounds the pulp) [13].

Other congenital cardiac anomalies associated with EDS include atrial and ventricular septal defects, coarctation of the aorta (formation of a shelf-like blockage within the descending thoracic aorta), bicuspid aortic valve and complex conditions such as Tetralogy of Fallot (the most common cyanotic heart lesion comprised of four cardiac anomalies: narrowing of the pulmonic valve, a large hole between the two ventricles, thickening of the right ventricle, and misalignment of the aorta) [12].

CASE DISCUSSION:

The first comprehensive description of a syndrome displaying laxity and fragility of the skin associated with hyper- mobility of the large joints was published in 1892 when Dr Tschernogobow presented two patients at the Moscow Venereology and Dermatology Society. The first patient was a 17-year-old male with epilepsy who suffered from ‘fragility and hyperelasticity of the skin, and a failure to hold sutures. He also had hypermobility and luxation of joints, and molluscoid pseudo tumours of the knees, elbows, and other areas’, the combination of all the above features are suggestive of EDS.[9]. In an other study, a 41 year old Hindu female attended the medical ,out-patient department with the complaints of gradually increasing looseness of the skin all over the body, generalized weakness, and laxity of skin and joints for the last 8 years. A history of bruising of the skin with even very trivial trauma could be easily ascertained but no episode of a

major haemorrhagic accident had ever occurred in the past. The patient was an outcome of a full term normal delivery with no neonatal or perinatal morbidity. The family history revealed that she has 8 issues and none of them has any such complaint of skin and joints. Her mother died at the age of about 50 years and had similar hyperelastic skin and loose jointedness as did also her brother who has recently died of a cerebra-vascuular accident, under our care, at the age of 35 years.[10].The exact etiology of the syndrome is not yet known. Deficiency of certain enzymes has been detected of which procollagen lysilhydroxylase and procollagen protease are the important ones [11].This is supposed to result into defective cross linking of various hydroxyproline and hydroxylysine units. The collagen meshwork thus formed is loose and is responsible for the symptomatology [10].

CONCLUSION:

Knowledge of the anatomy and clinical significance of Ehlers Danlos syndrome may help the surgeons to make accurate diagnosis and provide appropriate treatment.

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