Ehlers-Danlos Syndrome, Type III

Alternative Names
Ehlers-Danlos Syndrome, Hypermobility Type
EDS III
EDS3
Benign Hypermobility Syndrome

Record Category
Disease phenotype

WHO-ICD
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations and deformations of the musculoskeletal system

Incidence per 100,000 Live Births
6-10

OMIM Number
130020

Mode of Inheritance
Autosomal dominant

Gene Map Locus
6p21.3, 2q31

Description
Ehlers- Danlos syndrome (EDS) is a family of disorders that cause abnormalities in the synthesis and metabolism of the connective tissue components in skin, bones, blood vessels, joints, and other organs. As a result, the tensile strength and integrity of the affected organs will decrease. EDS is classified into many types according to the symptoms and mode of inheritance. The hypermobility (unusual range of joint movement) type, or type III, is the most common, but the least severe form of EDS that is characterized by musculoskeletal complications, degenerative joint disease, chronic pain, and soft skin. Type III EDS is found to affect 1 in 10,000 to 15,000 people worldwide. Diagnosis of this type of EDS depends mainly on clinical manifestations and family history.

Molecular Genetics
Ehlers-Danlos syndrome (EDS) is inherited as an autosomal dominant pattern; therefore, family history is the most important diagnostic tool. Some people with EDS, hypermobility type have mutations in the Tenascin X (TNXB) gene. The normal product of the TNXB gene is a protein known as tenascin that is important for organizing and maintaining the supportive body tissues. Scientists believe that low tenascin-X levels in the body cause changes in collagen deposition as well as disruption the elastic fiber network in joint ligaments and tendons. A mutation of the "collagen, type III, alpha 1" (COL3A1) gene was reported in a single family with hypermobility EDS.

Epidemiology in the Arab World
Saudi Arabia
Banjar (2003) reported a 10-year-old male patient with a comorbid association of type III Ehlers-Danlos syndrome (EDS) and cystic fibrosis. The patient had a history of recurrent chest infection since one month of age and multiple admissions for pneumonia. Pulmonary function tests revealed severe obstructive lung disease and air-trapping. He had pectus carinatum, severed clubbing, hypermobility of all joints, and elastic skin. His mother and two paternal aunts had hypermobile joints. The patient presented features of cystic fibrosis and he died from respiratory failure. Since EDS caused the abnormal elasticity of the lung, the patient suffered form a progressive cystic fibrosis course.

References
Banjar H. Simultaneous occurrence of cystic fibrosis and Ehlers- Danlos Syndrome. Emirates Med J. 2003; 21(1):105.

Related CTGA Records
Cystic Fibrosis

External Links
http://ghr.nlm.nih.gov/condition=ehlersdanlossyndrome
http://www.emedicine.com/ped/topic654.htm

Contributors

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