



type III encodes by COL3A1. Autosomal dominant inheritance.

Kyphoscoliosis Type (formerly type VI)

Generalized joint laxity and severe muscle hypotonia at birth are seen in this type of EDS. Muscular hypotonia can be very pronounced and leads to delayed gross motor development. Individuals present with scoliosis at birth that is progressive. The phenotype is most often severe, frequently resulting in the loss of ambulation in the second or third decade. Scleral fragility may lead to rupture of the ocular globe after minor trauma. Tissue fragility including atrophic scars and easy bruising may be seen. Spontaneous arterial rupture can easily occur. Other findings may include: marfanoid habitus; microcornea; and radiologically considerable osteopenia.

Kyphoscoliosis Type EDS is the result of a deficient lysyl hydroxylase (PLOD). Autosomal recessive inheritance.

with recurrent subluxations; skin hyperextensibility with easy bruising, tissue fragility including atrophic scars; muscle hypotonia; kyphoscoliosis and radiologically mild osteopenia.

Arthrochalasia Type EDS is caused by mutations leading to deficient processing of the amino-terminal end of pro α 1(I) [type A] or pro α 2(I) [type B] chains of collagen type I. Autosomal dominant inheritance.

Dermatoparaxis Type (formerly type VIIIc)

Individuals demonstrate severe skin fragility bruising. Wound healing is not impaired and the scars are not atrophic, skin texture is soft and doughy. Sagging, redundant skin is evident. The redundancy of facial skin results in an appearance resembling cutis laxa. Large hernias (umbilical, inguinal) may also be seen.

Dermatoparaxis Type EDS is caused by a deficiency of procollagen I N-terminal peptidase. Autosomal recessive inheritance.

Types of EDS

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Ehlers-Danlos Syndrome (EDS) is a heterogeneous group of heritable connective tissue disorders characterized by articular hypermobility, skin extensibility and tissue fragility. There are six major types of EDS. The different types of EDS are classified according to distinct features.

Classical Type (formerly Types I and II)

Marked skin hyperextensibility with widened atrophic scars and joint hypermobility are found. The skin manifestations range in severity from mild to severe expression. The skin is smooth and velvety with the evidence of tissue fragility including; hiatal hernia, anal prolapse in childhood and cervical insufficiency. Hernias may be a post-operative complication. Also evident are molluscoid pseudotumors frequently found over pressure points and subcutaneous spheroids which are mobile and palpable on the forearms and shins.

Complications of joint hypermobility include sprains, dislocations/subluxations and pes planus. Recurrent subluxations are common in the shoulder, patella and temporomandibular joints. Muscle hypotonia, delayed gross motor development may be evident.

Abnormal electrophoretic mobility of the pro α 1(V) or pro α 2(V) chains of collagen type V has been detected. Autosomal dominant inheritance.

Hypermobility Type (formerly type III)

The skin involvement (hyperextensible and/or smooth, velvety skin) as well as bruising tendencies are both variable. Joint hypermobility is the dominant clinical manifestation.

Generalized joint hypermobility that affects large and small joints is evident in Hypermobility Type EDS. Recurring joint dislocations are common occurrences. Certain joints, such as the shoulder, patella, and temporomandibular joint dislocate frequently.

Chronic joint and limb pain is a common complaint amongst individuals with Hypermobility Type EDS. Skeletal X-rays are normal. Musculoskeletal pain is early onset, chronic and may be debilitating. The anatomical distribution is wide, tender points are often elicited.

To date, researchers have identified no distinctive biochemical collagen finding. Autosomal dominant inheritance.

Vascular Type (formerly type IV)

Thin translucent skin reveals the subcutaneous venous pattern, and is particularly apparent over the chest and abdomen. Facial appearance is characteristic in some affected individuals. A decrease in subcutaneous tissue, particularly in the face and extremities is evident. Minor trauma can lead to extensive bruising.

Arterial/intestinal/uterine fragility or rupture commonly arise in this type of EDS.

Spontaneous arterial rupture has a peak incidence in the third or fourth decade of life, but may occur earlier. Midsized arteries are commonly involved. Arterial rupture is the most common cause of sudden death. Life expectancy is shortened with a majority of individuals. Joint hypermobility is usually limited to the digits. Tendon and muscles rupture can occur. Talipes equinovarus is frequently seen at birth. Other manifestations that may be found include: acrogeria; early onset varicose veins; arteriovenous, carotid-cavernous fistula;

pneumothorax/pneumohemothorax; gingival recession and complications during and after surgery.

Vascular Type EDS is caused by structural defects in the pro α 1 (III) chain of collagen