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Photo Clinic

A Rare Disorder of Bleeding Tendency: Ehlers-Danlos Syndrome

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A 8-year-old girl was referred to hematology clinic because of easy bruising and several ecchymotic lesions mostly over his lower extremities. She had been symptomatic since early childhood. The parents were consanguineous. They also complained of loose skin, twisting ankles and recurrent falling in their child causing old scars. On physical examination, she was in good general condition. The facies was noticeable with epicanthal fold, mild prominent eyes and fixed lip borders. The most remarkable signs were dispersed ecchymoses throughout lower extremities, ulcerated hematomas on his knees, stretchy loose skin visible over elbows and hypermobility in small joints of the hands (Figure 1-4). Laboratory workup and coagulation screening tests including BT, PT and PTT were normal. According to the clinical manifestations, a provisional diagnosis of Ehlers-Danlos syndrome was made for the child.

Ehlers-Danlos syndromes (EDS) are a group of clinically and genetically heterogeneous disorders of connective tissue characterized by generalized joint hypermobility, skin hyperextensibility, and tissue fragility resulting in easy bruising, delayed wound healing and atrophic scarring. In March 2017, an updated International Classification of EDS and related disorders identified 13 variants with mutations in 19 different genes.[1]

The bleeding tendency is most prominent in EDS. Most EDS-subtypes are caused by mutations in genes encoding fibrillar collagens type I, III or V, or genes encoding enzymes involved in the posttranslational modification of collagens.[2] Easy bruising is, to a variable degree, present in all subtypes of EDS, and is because of fragility of the capillaries and the perivascular connective tissues. However, collagen proteins are also involved in platelet activation, adhesion, and aggregation. Vascular fragility affecting medium-sized and large arteries and veins is typically observed in the vascular subtype of EDS, caused by a molecular defect in collagen type III, an important constituent of blood vessel walls. A variable bleeding tendency, manifesting as gum bleeding, menometrorraghia, postnatal or perioperative hemorrhage is observed in many EDS-patients of varying EDS subtypes.[3]

The diagnosis of classic EDS is established in a patient with the minimal clinical diagnostic criteria (skin hyperextensibility and atrophic scarring and either joint hypermobility or ≥3 minor clinical criteria) and identification of a heterozygous pathogenic variant in



Figure 1: Ulcers and ecchymoses on lower extremities

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Figure 2: Hypermobility of the wrist and metocarpophalangeal joints



Figure 3: Hypermobility of the wrist and metocarpophalangeal joints



Figure 4: Loose skin over elbow

COL5A1, COL5A2, or (less commonly) COL1A1 on molecular genetic testing.[4]

Although, EDS is considered primarily as a connective tissue disorder with symptoms involving musculoskeletal system, the pediatric hematologists should keep in mind this rare group of disorders when encountering children with easy bruising, widespread ecchymoses and dysmorphic scarring and hence an evaluation of joint mobility should be included in every initial visit of such children particularly when coagulation tests are within normal range.

Conflict of Interest: None declared.

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