



Ehlers-Danlos Syndrome

Ehlers-Danlos syndrome (EDS), also known as cutis hyperelastica, is a group of inherited connective tissue disorders caused by a defect in the synthesis of collagen. Depending on the individual mutation the severity of the syndrome can vary from mild to severe. Several types of EDS have been categorized with type 4 considered the most dreadful considering the high propensity of these individuals to develop life-threatening arterial and digestive complications. EDS type 4 is an autosomic dominant defect (missense mutation in the COL3A1 gene) characterize by a fascial acrogeria appearance (large eyes, small chin, thin nose, lobeless ears), small stature with slim build, and thin pale translucent skin. Children with EDS have poor wound healing, hypermobile joints, clotting anomalies, spontaneous pneumothorax and recurrent hernias. Among the catastrophic events associated with type 4 EDS we can find arterial dissection or tear caused by deterioration of congenital fragile tissue leading to hematoma, false aneurysm and intracavitary bleeding. They are responsible for the majority of deaths. The next set of complications are spontaneous and recurrent perforation of the colon associated with a significant risk of leakage after anastomosis and spontaneous perforation or bleeding of the uterus. There is no cure for EDS and management is supportive.

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MALT Lymphoma

Marginal zone mucosa-associated lymphoid tissue (MALT) lymphomas comprised a group of indolent B-cell non-Hodgkin lymphomas which are rare to find in pediatric patients. The gastrointestinal tract is the predominant site for this type of MALT extranodal non-Hodgkin lymphoma. Almost one-third of the patient with malignant

lymphomas has involvement of the stomach (most commonly), small intestine and large intestine. Other sites include salivary gland, tonsils, lungs, thyroid, conjunctiva and even skin. The main categories for MALT lymphomas are the low- and high-grade B-cell MALTomas with or without a low grade component. Small centrocyte-like cells, lymphoepithelial lesions, and reactive lymphoid follicles are the main specific histopathologic features of low-grade MALTomas. On the contrary, large high-grade cells, which usually infiltrate in sheets and between glands without forming lymphoepithelial lesions characterize the high-grade B-cell MALToma. The high-grade MALTomas have shown a worse prognosis than low-grade and mixed types MALTomas. Acquired MALTomas may develop as a reaction to autoimmune disease and infection. Helicobacter pylori infection predisposes to development of MALTomas in the stomach. Management of MALTomas includes surgical resection where anatomically feasible along with adjuvant chemotherapy.

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Central Precocious Puberty

Central precocious puberty (CPP) occurs with premature activation of the hypothalamic-pituitary-gonadal axis. CPP is defined as the onset of secondary characteristics associated with increased linear growth velocity and accelerated bone maturation occurring before the age of seven to 8 years in girls, and nine years in boys. CPP is more common in girls than boys. If left untreated precocious puberty results in early epiphyseal closure and short final stature. The most common cause of CPP is idiopathic caused by early onset of luteinizing hormone, follicle stimulating hormone and estradiol secretion. The goal of therapy is to restore a prepubertal state attenuating the deleterious effect of early sex steroid exposure on physical development, skeletal maturation and ultimate adult height. This can be achieved with parenteral administration of long-acting gonadotropin releasing hormone agonists (GnRHa) which has been found to be effective in retarding progression of secondary sexual characteristics, preventing menses, slowing bone-age maturation and improving final height. Since GnRHa monthly injections are painful, a subcutaneous microporous

hydrogel implant that release GnRHa on a daily basis has been developed and tested efficaciously in suppressing clinical and laboratory parameters of puberty for one year. The implant is placed in the inner aspect of the arm under local or general anesthesia as a minor procedure.

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