Abstract: The Ehlers-Danlos syndromes (EDS) form a clinically and genetically heterogeneous group of inherited connective-tissue disorders characterized by joint hypermobility, tissue fragility and skin abnormalities. Six subtypes have been well characterized based on clinical features and molecular genetic abnormalities. The arthrochalasia type EDS (formerly type VIIa and VIIb) is an extremely rare condition characterized by severe generalized joint hypermobility with multiple dislocations including congenital bilateral dislocation of the hips, muscular hypotonia and distinct dysmorphic features. We report a 7 year old girl who presented with joint hyper mobility and bilateral dislocation of hips. This was a case of EDS- ARTHROCHALASIS type. This case is being reported for its rarity. Till date only 30 cases have been reported in literature worldwide.

Keyword: EDS, ARTHROCHALASIS, HYPER MOBILITY.

INTRODUCTION: The Ehlers-Danlos syndromes (EDS) are a clinically and genetically heterogeneous group of inherited connective-tissue disorders characterized by joint hypermobility, tissue fragility and skin abnormalities. Other organ involvement, depending on the type of EDS, include blood vessel, skeletal, gastrointestinal, dental, brain, genitourinary and other system. The most recent nosology has simplified the classification of EDS into 6 subtypes based on their clinical features and molecular abnormalities, with a seventh group of miscellaneous forms of EDS. The prevalence of all forms of EDS is estimated to be 1:5000. Of these, the classic and hypermobility types (EDS I-III) account for ~90%, and the vascular type (EDS IV) for ~5%. All other subtypes are extremely rare.

CASE REPORT: A 7 month old girl child 2nd born out of non consanguineous marriage presented with h/o fever, cough, fast breathing of 3 days duration. There was h/o previous similar episode which was treated as bronchiolitis. At 3 month of age she was diagnosed as congenital dislocation of left hip joint and hip-spica was applied, surgery was planned at 1 year of age. Antenatal, natal, family history was uneventful. On examination she was alert, tachypneic with normal vitals. Head to foot examination revealed hyper extensibility of skin, hyper mobility of joints, micrognathia and criss cross pattern of skin creases in palms and soles. The nine point BEIGHTON hyper mobility score was 8/9. (>5 indicates hyper mobility ). Systemic examination was normal except for hypotonia of limbs.

X-ray pelvis showed dislocation of left hip. X-rays of Cranium, cervical and dorsolumbar spine were normal. Cardiac and ophthalmological evaluation were normal. Skin biopsy done showed loosely dispersed collagen under light microscopy. However the confirmatory investigation, which is the gene
Ehlers-Danlos syndrome (EDS) is a group of genetically heterogeneous connective tissue disorders. Affected children appear normal at birth, but skin hyperelasticity, fragility of the skin and blood vessels, delayed wound healing, and joint hypermobility develop later. The essential defect is a quantitative deficiency of fibrillar collagen.

It is characterized by generalized joint hypermobility with recurrent dislocations of other joints including congenital bilateral hip dislocation, muscular hypotonia and mild dysmorphic features. Motor development is significantly delayed in the neonatal and postnatal period due to muscular hypotonia and recurrent joint luxations, but cognitive development is usually normal. Characteristic for the arthrochalasia type is bilateral congenital hip dislocation that is difficult to treat. There is significant skin redundancy, causing one of the key features of arthrochalasia type EDS i.e. a criss-cross pattern that is seen on the palms and soles. Patients have dystrophic features which include mild hypertelorism, bilateral epicanthic folds, large fontanels (mainly the anterior fontanel), and especially pronounced micrognathia. Confirmation of diagnosis is by skin biopsy and genemutational analysis. Classic (COL5A1, COL5A2, COL1A1 Genes; Previously EDS Type I—Gravis, EDS Type II—MITIS) This autosomal dominant disorder is characterized by premature birth caused by rupture of membranes, skin hyperelasticity and fragility, easy bruising, generalized and severe joint hypermobility, scoliosis, mitral valve prolapse, recurrent hernias, muscular hypotonia.

Hypermobile (COL3A1 Gene; Previously EDS Type III): This disorder has autosomal dominant inheritance and manifests as generalized severe joint hypermobility and minimal skin manifestations. Musculoskeletal pain is common, and osteoarthritis may develop prematurely.

Vascular (COL3A1 Gene; Previously EDS Type IV—Arterial Echymotic) This autosomal dominant disorder shows the most pronounced dermal thinning of all.

The skin is thin, translucent and minimal hyperextensibility, and the joints are not hypermobile, except perhaps during childhood. Premature birth, extensive ecchymoses from trauma, a high incidence of keloids, rupture of the bowel (especially the colon), uterine rupture during pregnancy, rupture of the great vessels, dissecting aortic aneurysm, and stroke all contribute to the increased morbidity and shortened life span. Kyphoscoliosis (Lysyl Hydroxylase [PLOD Gene] Deficiency; Previously EDS Type VI) Patients with this autosomal recessive type have joint hyperextensibility, hypotonia, kyphoscoliosis, fragile ocular globe, keratoconus, skin hyperelasticity, and fragile bones. They are at risk of rupture of medium sized arteries and respiratory compromise if kyphoscoliosis is severe. Dermatosparaxis (Type 1 Collagen N-Phenylas; Previously EDS Type VII) This autosomal recessive condition that includes premature rupture of membranes, delayed closure of fontanels, skin fragility and laxity, easy bruisingability, growth retardation, short limbs, umbilical hernia, and characteristic facies with micrognathia, and prominent, puffy eyelids. DIFFERENTIAL DIAGNOSIS:

Joint hypermobility is seen in Marfan syndrome, EDS, Osteogen sis imperfecta. Absence of marfanoid features, bone changes ruled out Marfans and Osteogenesis imperfecta. MANAGEMENT:

Investigations include electron microscopic examination of skin biopsy specimen, electrophoretic analysis of collagen proteins in cultured fibroblasts, gene mutational analysis. There is no specific treatment available. Supplementation of ascorbic acid (Vitamin C), a cofactor for crosslinking of collagen fibrils, ameliorates the tendency toward bruising in some patients. The use of special braces may help to stabilize affected joints. Precautions should be taken to prevent injuries and trauma, such as may occur during contact sports. Appropriate precautions and careful monitoring are essential before, during, and after dental or surgical procedures. Females with EDS vascular type should be counseled concerning the increased risk of certain complications during pregnancy and delivery and the need for meticulous obstetric care.

PROGNOSIS:

Ehlers-Danlos syndrome type IV is a severe form, and patients with this disease often have a shortened lifespan. Arterial aneurysms, valvular prolapse, and spontaneous pneumothorax are common complications. The prognosis with this type is poor. Sudden death can occur after visceral perforation or after the rupture of a large vessel, most commonly an abdominal and splenic vessel. The other types usually are not as dangerous and affected individuals can live healthy with restrictions.

FOLLOW UP:

In ARTHROCHALASIS type patients are followed up for recurrent dislocations, developmental evaluation and orthopedic evaluation regarding braces. Genetic counseling should be offered regarding the disease. Each child of an individual with EDS has a 50% chance of inheriting the mutation. Type IV Ehlers-Danlos syndrome patients should be carefully monitored because they are at high risk for spontaneous rupture of a large artery (eg, splenic artery, aorta) or perforation of internal organs. These patients should be educated that surgery can pose life threatening risks.

REFERENCES:
