Iranian Journal of Orthopaedic Surgery
Vol 16, No 4 (Serial No 63), Autumn 2018, p 332-334

Ehlers-Danlos Syndrome (TYPEI) with Mental Retardation -an Unusual Association
(Reports on Two Brothers)

Abstract
Ehlers Danlos syndrome (EDS) is an inherited connective tissue disease due to impaired collagen metabolism. Joint hypermobility and skin hyper extensibility are the major findings. Six types of EDS are recognized. Type I or Gravis type is characterized by skin hyperextensibility, joint hypermobility, skin splitting autosoml dominancy inheritance, preterm premature rupture of membrane (PPROM) and varicose vein. Mental retardation has not been reported in the literature. Two cases of unusual type 1 EDS with joint deformity and mental retardation will be reported in this article.
Keyword: Ehlers-Danlos Syndrome, Intellectual Disability, Hypermobility

Received: 1 year and 6 months before printing; Accepted: 20 days before printing

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Introduction

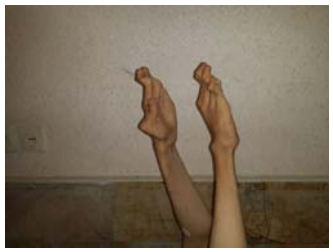
Ehlers Danlos syndrome includes a group of heterogenous disorders of abnormal collagen metabolism, which lead to joint hyperlaxity, skin hyper elasticity and abnormality in some other organs. Meeckeren reported the first case in 1682 as unusual dilatability in skin. Ehlers Danlos syndrome was described in 1901by a Danish dermatologist, Edvard Ehler, as cutis laxa. Hemorragic tendency, and loose- jointedness was described by him [1, 2]. Seven years later Henry-Alexandre Danlos, a French dermatologist, reported one patient with unusual characteristics like mulloscoid pseudotumor due to chronic contusion of soft tissue. “Ehlers-Danlos syndrome” was the name given to it in 1930is. Classification evolusion was progressed up to 1997 when collaborators revised the classification criteria in to six types. The classification is not, however, rigid and sometimes overlap in different types is seen. More than 13 types have been reported by now but some characteristics are shared between the different types [3,4]. Mental retardation which is impaired intellectual and adaptive functioning and is defined by an IQ below 70 has not been reported in correlation with EDS in the literature.

Case Report

First patient was an 18-year-old boy with Mental Retardation (IQ=50) who was preterm. He was floppy baby at birth due to hypotonia. Skin easily became ruptured by simple trauma and easily bruised, but after 10 years old became better. He walked in fourth year of live, and as he became older, swan neck deformity with joint dislocation developed in his hand (fig1-a).

Knee locking was evident during walking. Shoulder dislocation with spontaneous reduction happened in his sleep. He could simply dislocate patella and interphalangeal joint, but with severe pain. PPROM and floppy baby syndrome was present at birth.

He had one brother who discussed later. Parents were first cousin. Their uncle had similar manifestation but he refused to participate in this article.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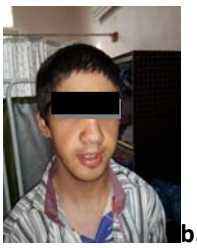
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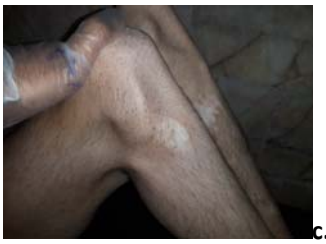
c.

Figure 1. Fig 1a: shows swan neck deformity dislE153-on
Fig 1b: shows cigaret paper scar in for head.
Fig 1c: shows patella alta.

Clinical findings

First patient

He was short stature, severe deformity in chest with right costochodral junction angulation (pectus carinatum) anteriorly, and thoracic asymmetria.

Facies was not typic for type one with cigaret paper scar in forehead (fig1-b).

Epicatic fold was present but teeth were normal. Skin was hyper extensile.

In his buttocks mulloscoid spheroid was seen, Swans neck Deformity in digit cubitus valgus in arm extention and subcutaneous spheroid in elbow also was present. Patella alta (fig1-c) and in knee, positive adams test, hammer toe in foot was seen. Rocker bottom feet (fig 2) and cigaret paper in anterior of the calf was seen. Bed sore was also present. Neurologic examination was normal. No sign of cerebral palsy and no focal neurological deficit was present.

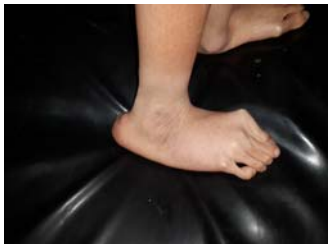

Fig 2: shows rocker bottom feet.

Patient 2

The other brother was ten years old boy with Mental Retardation (IQ=60), like his brother PPROM and floppy baby was present at birth. He underwent two operations due to intoeing gait. He only has had recurrent dislocation in his shoulder not in knee.

Short stature was present. He never walked in his life due to severe flexion contracture and genu valgum. Thoracic cage deformity was similar to his brother but with more severity. Joint deformity and hyperextensibility was less severe than his brother.

Cigaret paper was seen in medial of right foot

Proximal interphalangeal subluxation was present. Cubitus varus severe hyperextesibility was also present. Hallux valgus was present. The patient had severe mental retardation with no focal neurological deficit.

Discussion

Ehlers Danlos syndrome is an inherited connective tissue disease due to impaired collagen metabolism. Joint hypermobility and skin hyperextensibility are the major characteristics.

Ehlers Danlos syndrome nosology first time was described in the Berlin nosology in 1986 [5]. In 1998 the nosology was revised, in this revision major and minor criteria established for each type. Our diagnostic assessment was 1998 nosology and both patients fulfilled the criteria for type 1[3].

Six type of EDS are classified: type I or Gravis type is characterized by skin hyperextensibility, joint hypermobility, skin spility,[3, 4] Autosomal Dominant inheritance, preterm premature rupture of membrane (PPROM) and varicose vein, mental retardation was not reported in literature, three case of unusual EDS scour joint deformity type I with mental retardation and will be discussed in our article.

Classic form of EDS type is a rare syndrome which is inherited autosomal dominate, present by skin hyperlaxity, atrophic scar and joint hapermobility as major criteria and mulluscoid pseudotumor, subcutaneous spheroid delay coarse motor movement and hypothesis.

In 1960, EDS was categorized on its various clinical symptoms, first into three types in 1967,[6] one year later to five types,[7] and finally in 1972 into seven types[8].

In mid-1980 EDS was categorized in Berlin, then revised in Vill efranch in France in 1992. They categorized it into six types to simplify clinical diagnosis[1].

Mental Retardation is characterized by impaired intellectual and adaptive functioning. It is defined by IQ under 70. 2-3 opercent of general population are affected and has different etiologies such as genetic disorder, toxin exposure like mercury, iodine deficiency, and absence of the arcuate fasiculus and so on. Mental retardation was not reported in correlation with EDS in literature [3]. These two brothers with similar clinical presentation and no reason for mental retardation, and severe EDS are unique.

There is no specific treatment in patients with EDS. Immobilization is the method of choice in joint subluxation treatment, but recurrences are expected which is difficult to treat[9]. Probably bone realignment procedures whenever possible in recurrence is better to stabilize a joint. Wound problems are common and surgeons should have a meticulous wound closure technique, and patient should be warned about possibility of unusual scar formation. In anterior spinal surgery sever bleeding have been reported, therefore some authors managed kyphoscoliosis with posterior surgery and reported good result[10,11]. Visceral and vascular rupture management discussion is beyond this case report. Restrictions in this case report were: the patients and their family did not agree with genetic study and their uncle who had similar characteristic for EDS did not participate.

Conclusion

Mental retardation accompanied by joint deformity are not reported characteristics of EDS type I. This can be categorized as a new type with new mutation. More study should be done to recognize genetic abnormality in these patients.

The authors declares that there is no conflict of interest regarding the publication of this paper. Both patients had informed consent.

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