International Journal of Contemporary Pediatrics
Anitha GFS et al. Int J Contemp Pediatr. 2017 Jan;4(1):261-263
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20164614

Case Report

A rare case of Ehler Danlos syndrome - Progeroid type: a case report

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Received: 19 November 2016
Revised: 01 December 2016
Accepted: 03 December 2016

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ABSTRACT

Ehler Danlos syndrome (EDS), is a group of genetically heterogenous connective tissue disorder. A very rare type of this syndrome is the Progeroid type which is included in the NIH group of rare diseases list. The prevalence is < 1 / 1000000. Along with the usual clinical features, patients with Ehler Danlos syndrome-Progeroid type have old age appearance. Here we report one such rare case of this syndrome diagnosed in a 8 yr old child with characteristic clinical features and supportive genetic confirmation.

Keywords: Ehler Danlos syndrome-progeroid type

INTRODUCTION

Ehler Danlos syndrome is a group of inherited disorders that affect the connective tissues primarily of skin, blood vessels, joints etc. Progeroid type is a rare form of EDS. It usually presents by childhood or adolescence. Individuals with progeroid type have unique features of sparse scalp hair, eyebrows, old man look, loose elastic skin on face, along with the usual features of EDS such as hypermobility of joints, fragile elastic skin and easy bruises.[1] The mode of inheritance is autosomal recessive.[2] The name itself is a misnomer, as individuals with this progeroid form may have old age appearance, but they neither have premature ageing nor a decreased life span.

CASE REPORT

An eight year old female child third born of a third degree consanguineous marriage, presented to the paediatric OPD of a government medical college hospital with complaints of swelling in the left side of the neck for a duration of 1 month. There was no history of fever or associated constitutional symptoms. The child was immunised for age and there was no TB contact.

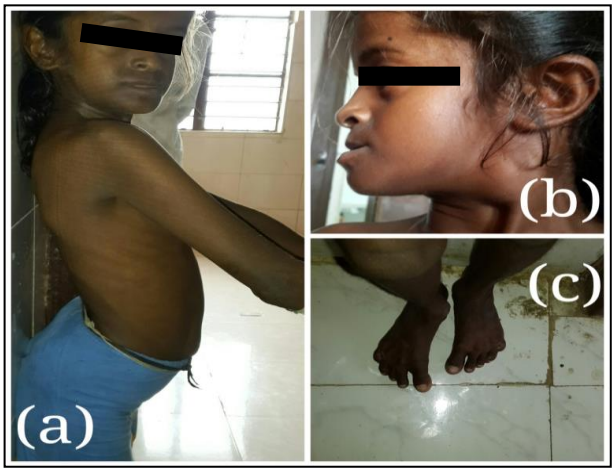

Figure 1: (a) Lumbar lordosis with progeroid features; (b) Tuberculous lymphadenitis of the neck; (c) Flat feet.

The antenatal period was uneventful but the child required neonatal admission of 1 week for preterm, low birth weight care- birth weight 1.5 kg. The child had a delay in attaining age appropriate motor milestones.

On examination, child was afebrile, malnourished with hypertelorism, prognathism, and had an old age appearance. There were loose skin folds over the extremities, laxity and hypermobility of joints, lumbar lordosis, and small muscle wasting of both hands and flat feet. The swelling in the left side of the neck was of size 4 x 3cm, with firm consistency, smooth surface and no local warmth or tenderness (Figure 1).

She had a positive 'Gorlin sign' which is the ability to touch the tip of the nose with the tongue. Her anthropometry was suggestive of grade 3 malnutrition (IAP), short stature (Mclaren) and microcephaly (WHO Z scores). Her vitals were stable and systemic examination was clinically normal. Based on the above history and clinical picture, a genetic syndrome was considered and investigations proceeded.

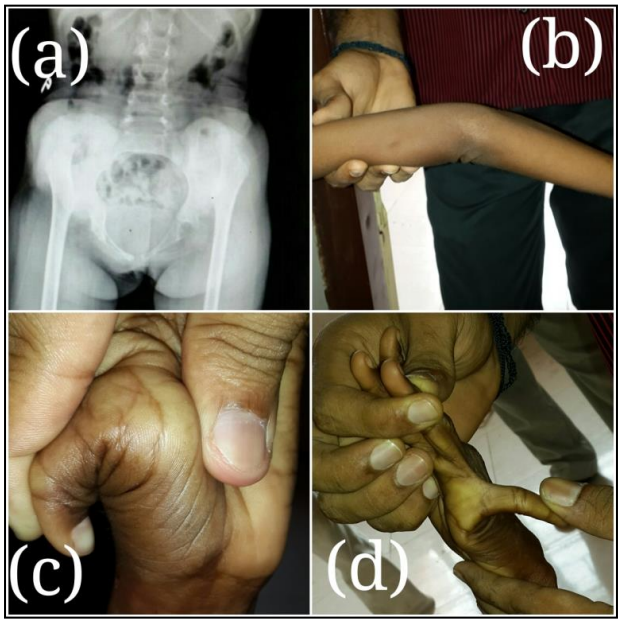

Figure 2: (a) Dislocation of hip-high acetabular type; (b) (c) and (d) Hyperextensibility of skin and joints.

The basic blood investigations, thyroid profile, chest X-ray, ECHO and USG abdomen were normal. Retroviral screening was negative. The ophthalmic and ENT assessment was normal. FNAC of the neck swelling was suggestive of tuberculosis and she was started on category 1 ATT. Orthopedic consultation with radiological imaging revealed dislocation of hip-high acetabular type and spina bifida of L3,4,5. With a background of third degree consanguinity, hyperextensibility of joints, loose skin folds etc, a connective tissue disorder was suspected and skin biopsy was done which confirmed Ehler Danlos syndrome (Figure 2). Genetic studies revealed it to be progeroid type of Ehler Danlos syndrome.

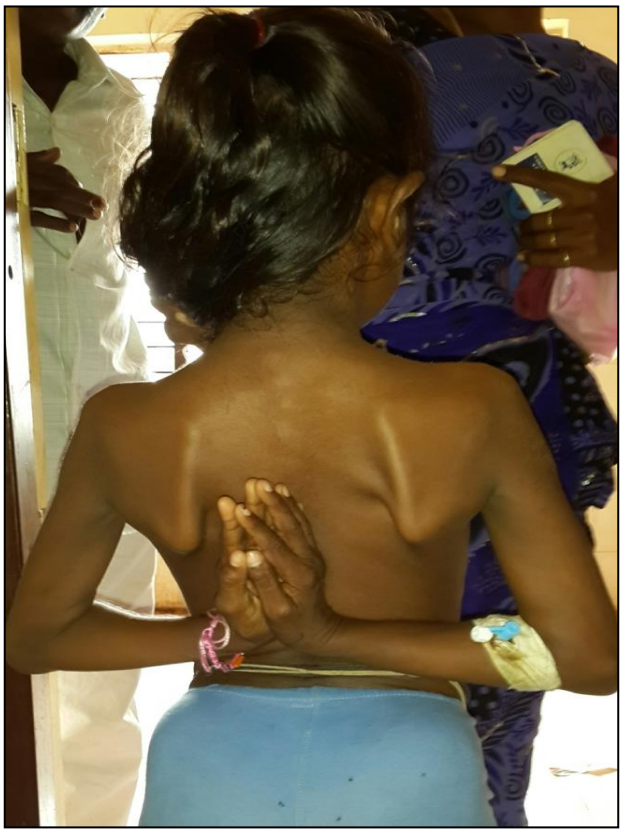

Figure 3: Reverse Namaskaar sign.

DISCUSSION

Ehler Danlos syndrome is a group of inherited disorders that affect the connective tissues that support the skin, bone, blood vessels and many other organs. It is characterised by "kneadable" skin, hypermobile joints, delayed wound healing etc. The progeroid type is a rare form of EDS. In addition to the above features, individuals with progeroid type have aged appearance, loose skin folds over face, sparse eyebrows, developmental delay, thin curly hair, fragile bone weakness, weak muscle tone, mild intellectual disability, delayed growth, short stature etc.[3] It is caused by mutation in B4GALT7 gene located on chromosome 5.[4,5]

This gene provides instructions for making an enzyme that is involved in the production of collagen. When the enzyme is deficient, collagen is not formed correctly in connective tissue. The deficient enzyme identified is xylosyl protein 4-beta-galactosyl transferase, with defective biosynthesis of dermatan sulfate proteoglycan. As it has an autosomal recessive pattern of inheritance, there is a 25% chance of recurrence in each pregnancy.

The patient described here was a product of third degree consanguineous marriage, with old age appearance, hypermobile joints and skin laxity. She had a positive

Gorlin sign and reverse Namaskar sign (Figure 3). Although Gorlin sign can be present in 10% of the normal population, it is a characteristic finding in 50% of patients with EDS.

Namaskar is the typical Indian way of greeting people, where the forearms are folded in front of the chest and the palms are opposed together.

Reverse Namaskar is the ability to fold the forearms at the back and to oppose the palms facing each other to say 'Namaskar'. The sign demonstrates the hypermobility of joints. Although 'progeroid' means 'appearance similar to old age' individuals with the progeroid type of EDS, do not actually have premature ageing and are not expected to have a shortened life span. Her skin biopsy and genetic studies were also supportive for the diagnosis.

CONCLUSION

There is no specific treatment and patients are managed with supportive care. Physiotherapy improves muscle strength and coordination. Lifestyle changes and precautions during exercise and intense physical activity should be advised to reduce the chance of accidents to skin and bone.

This case is reported for the rare presentation of the progeroid type of Ehler Danlos syndrome which was incidentally diagnosed in a child who presented with TB lymphadenitis.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Kresse H, Rosthoj S, Quentin E, Hollmann J, Glossl J, Okada S, et al. Glycosaminoglycan-free small proteoglycan core protein is secreted by fibroblasts from a patient with a syndrome resembling progeroid. Am J Hum Genet. 1987;41:436-53.
2. Wenstrup R, Paepe AD. Ehlers-Danlos syndrome, classic type. Gene Reviews, 2010; Available at http://www.ncbi.nlm.nih.gov/books/NBK1244/#eds.Differential_Diagnosis. Accessed on 07 November 2011.
3. Hernandez A, Aguirre-Negrete MG, González-Flores S, Reynoso-Luna MC, Fragoso R, Nazará Z, et al. Ehlers-Danlos features with progeroid facies and mild mental retardation: further delineation of the syndrome. Clinical Genetics. 1986;30:456-61.
4. Guo MH, Stoler J, Lui J, Nilsson O, Bianchi DW, Hirschhorn JN, et al. Redefining the progeroid form of Ehlers–Danlos syndrome: Report of the fourth patient with B4GALT7 deficiency and review of the literature. Am J Med Genet Part A. 2013;161A:2519-27.
5. Faiyaz HM, Zaidi SHE, Ali M, Mureikhi MS, Kennedy S, Thani G, et al. A novel missense mutation in the galactosyl transferase-I (B4GALT7) gene in a family exhibiting facioskeletal anomalies and Ehlers–Danlos syndrome resembling the progeroid type. Am J Med Genet. 2004;128:39-45.

Cite this article as: Anitha GFS, Shanmugam VK, Rajendran VV. A rare case of Ehler Danlos syndrome - progeroid type: a case report. Int J Contemp Pediatr 2017;4:261-3.