# Report of a case – Progeroid Ehlers Danlos syndrome

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## Introduction

Ehlers Danlos Syndrome (EDS) is a group of inherited disorders affecting connective tissues and characterized by hyper extensive skin and joints, fragility of blood vessels and poor wound healing[1]. Progeroid type is a rare subgroup present in children and adolescence with autosomal recessive inheritance[1]. They have unique clinical features such as sparse hair and eyebrows, aged appearance with loose facial skin in addition to key clinical features of classic EDS. Despite their aged appearance they have neither premature aging nor decreased lifespan[2].

## Case history

A 1 year and 4 month old boy presented to a pediatric unit with failure to thrive. He was a product of a non-consanguineous marriage, a normal antenatal period, and birth. However perinatal period was complicated with poor sucking and shallow breathing. Child failed to gain weight from three months onwards. He was exclusively breast fed till 6 months then weaned with satisfactory food intake. No history of recurrent infections noted. Despite adequate calorie intake his height and weight was persistently below the 3rd centile – so admitted for further evaluation.

## On examination

He was small for age, looked old and had unusual facial appearance with lack of subcutaneous fat .He had hyper extensible skin and joints. Poor head control was noted in addition to muscle hypotonia with that he had inability to sit up without support. Palmer grasp was present but mature pincer grasp was not developed. He was able to communicate with single words and recognized strangers. Vision and hearing were normal.

**Figure 1.** *The child look older for the age. He had poor head control and inability to sit without support, compatible with grossmortor developmental age of less than 3 months.*

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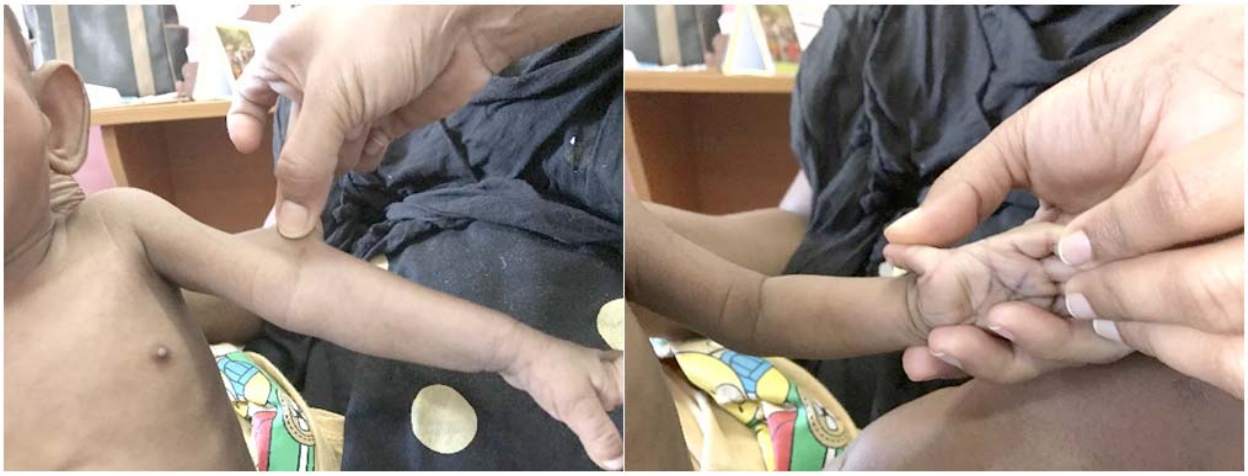

**Figure 2.** *Hyperextensible skin and joints.*

## Investigations and management

Investigations including full blood count, liver and renal functions, urine full report, blood and urine cultures, retroviral and intrauterine infection screening were normal in this patient as well as the imaging studies (CXR, 2d echo, USS abdomen, CT-brain.)

Depending on the characteristic clinical features progeroid type of EDS and associated predominantly gross-motor developmental delay was diagnosed, however genetic confirmation of the clinical diagnosis was not performed due to financial and technical constrains.

Patient was managed with supportive care as there was no specific therapy, parents were educated regarding importance of longitudinal evaluation for possible complications and possibility of genetic transmission.

## Discussion

EDS is a group of inherited disorders of connective tissues that support the skin, bone, blood vessels and many other organs. even though it is characterized by fragile, hyper elastic skin and hypermobile joints – EDS is known to have multiorgan involvement[1]. Progeroid type is a rare form of EDS and there are just eight reported patients with the progeroid variant of EDS in the medical literature, only three of which have supporting genetic diagnoses[2]. This syndrome is due to a mutation in B4GALT7 gene located on chromosome 5 – which encodes for the enzyme xylosyl protein 4-beta-galactosyl transferase which is involved in biosynthesis of dermatan sulfate proteoglycan[2]. Progeroid EDS has autosomal recessive inheritance pattern and commonly present in childhood and adolescence. Typical clinical presentation includes aged appearance, loose skin folds of the face, sparse hair and eye brows, developmental delay, fragile bones, muscle hypotonia mild intellectual disability, failure to thrive and short stature in addition to the key clinical features of classic EDS[1]. Clinical diagnosis can be confirmed by genetic testing for B4GALT7 mutation. The management of these patients consists of genetic counselling and supportive measures as there is no definitive treatment.

## References

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