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## Missed Diagnosis

# Ehlers Danlos syndrome – masquerading as primary muscle disease

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**Summary: A 9 year old Libyan boy presented with a history of delayed walking and abnormal gait. The presence of marked muscle under-development with hypotonia led to the initial diagnosis of primary muscle disease; later, he was found to have hyperelastic, fragile skin and hypermobile joints – the cardinal features of Ehlers Danlos syndrome. In this instance the disease seems to have been inherited in an autosomal recessive manner.**

### Introduction

Ehlers Danlos syndrome is a rare heritable disorder of collagen. Generally, the cutaneous and articular involvement provide fairly characteristic findings. Occasionally the patient may have additional features, such as progressive periodontitis, congenital hypotonia with or without muscular under-development, where the focus of attention may be diverted away from the primary diagnosis. Recently, we encountered a boy in whom the diagnosis had been missed because of a marked muscle under-development.

### Case report

A 9 year old Libyan boy presented with the complaints of delayed walking and abnormal gait. He had also recurrent episodes of cough for the past 7 months. There were no symptoms referable to the ocular, cardiovascular, genito-urinary or central nervous systems. His other developmental milestones and intelligence were normal. He was born to a fourth gravida mother after a full-term normal delivery. His parents were first degree cousins. One of his female cousins also had a similar illness.

The child had been admitted to this hopsital for evaluation of similar complaints two years previously. At that time he was thought to have congenital myopathy but electromyographic studies were normal. Muscle biopsy was advised, but was refused by the parents. He was discharged with this diagnosis.

On physical examination he was found to be thinly built. Anthropometric examination revealed his height to be 135 cm (75th percentile), weight 20.5 kg (3rd percentile), upper/lower segment ratio 0.93 ($\pm$1.0 s.d.), middle finger/hand ratio 0.4 cm (20th percentile), canthal index 0.4 and inter-pupillary distance 5.5 cm (60th percentile). The skin had multiple, thin, papery scars mainly on the forehead, knees and both shins, some of which were hyperpigmented. There was a well-defined, non-scarred, irregular area of pigmentation on the skin over the left cubital fossa. Small and large peripheral joints showed marked hypermobility. Arches of the feet were poorly formed. There was marked generalized reduction in the muscle mass with near normal muscle tone and power. Deep tendon reflexes were normal. Ocular examination did not reveal corneal anomalies, blue sclera or ectopia lentis. The oral cavity was normal. There was no evidence of aortic or mitral regurgitation. Parents

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and siblings were found to be normal on clinical examination.

Laboratory investigations and intravenous pyelogram were normal. His karyotype was 46, XY

## Discussion

The patient presented above had the cardinal features of Ehlers Danos syndrome (EDS), i.e. hyperelasticity, fragility and bruisability of skin with marked hypermobility of large and small joints of the body. The diagnoses of Marfan syndrome, Marfanoid hypermobility and Noonan syndrome, which might resemble Ehlers Danlos syndrome were excluded on clinical examination. Phenotypically our patient resembled the gravis variety (type I) of EDS. Patients with type I EDS usually inherit the disease in an autosomal dominant fashion while our patient inherited it in an autosomal recessive fashion.

To the best of our knowledge, autosomal recessive inheritance has not been reported in EDS type I. Ecchymotic type (type IV), ocular type (type VI) and arthrochalasis multiplex congenita (type VIII) may be inherited as autosomal recessive, but these types of EDS are distinct phenotypically from that of our patient.

The presence of marked under-development of muscles with mild hypotonia in patients with Ehlers Danlos syndrome may mislead the physicians to the erroneous diagnosis of primary muscle disease.[2] Muscular under-development and hypotonia have been described in other disorders of connective tissue, i.e., Marfan syndrome, osteogenesis imperfecta and Ehlers Danlos syndrome type III and type VII. Therefore muscle under-development in association with the manifestations of connective tissue disorders like extreme hypermobility of the joints, hyperelastic, fragile or bruisable skin should alert the physicians to a diagnosis other than primary muscle disease.

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