Marfanoid Hypermobility Syndrome: Reminscising a Forgotten Entity…

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Sir,

There are several hereditary connective tissue disorders (CTDs), which occur due to mutation of specific genes. Common CTDs, which may present to a dermatologist, include Marfan syndrome (MFS) and Ehlers–Danlos syndrome (EDS); hence, we need to become acquainted with the diagnostic clinical features of these conditions. However, on rare occasions, patients may present with overlapping features of both these conditions, when the entity is called Marfanoid hypermobility syndrome. We describe here a rare case of Marfanoid hypermobility syndrome to highlight the perplexing clinical presentation.

A 34-year-old male presented to us with complaints of generalized lax skin and hypermobility of several joints. Skin laxity was present all over the body since birth; although asymptomatic in nature. The joints turned hypermobile in a gradually progressive fashion. No other cutaneous abnormality was reported. There was no family history of abnormal skin, hypermobile joints, eye abnormality, and cardiovascular involvement.

General examination revealed a height of 164 cm and arm span 172 cm with the upper segment to lower segment ratio 0.90 (normal range 0.93 + 0.8). He weighed 45 kg; vitals were stable. His fingers were thin and elongated with the presence of high arched palate. Cutaneous examination revealed enhanced laxity of skin with reduced elasticity [Figure 1]; however, the consistency of skin was normal. No striae were noted in any part of the body. There was sparse subcutaneous fat with poorly developed muscles. The joints of hand and feet showed hyperextension on manipulation. His arm-span to height ratio was found to be 1.05 [Figure 2]. Both Steinberg’s thumb sign [Figure 3] and Walker-Murdoch wrist sign were positive.

Routine blood biochemistry including complete hemogram, blood urea, serum creatinine, and liver function tests was within normal limits. Urine examination, chest X-ray, electrocardiogram, and echocardiography were unremarkable. Skin biopsy from the lax skin revealed thinner collagen fibers in the dermis with respect to normal skin [Figure 4]. A diagnosis of Marfanoid hypermobility syndrome was made. The patient has been counseled and advised regular follow-up.

Figure 1: (a) Face, (b) arm - demonstrating enhanced laxity of skin

Figure 2: Tall patient with increased arm-span to height ratio

Figure 3: Positive Steinberg’s thumb sign indicative of Marfanoid syndrome
Marfanoid hypermobility syndrome is a genetically distinct generalized heritable connective tissue disease with features of both MFS and EDS.

EDS is a heterogeneous group of inherited CTDs. The hallmarks of EDS are fragility of the skin and blood vessels, hyperextensibility of the skin and joint hypermobility.\(^1\)

MFS encompasses skeletal, ocular, and cardiovascular defects. The patient is often, but not invariably, exceptionally tall, and the skeletal proportions are abnormal. The limbs are long, the excess being greatest distally, giving rise to arachnodactyly, and the length of the hallux is often particularly conspicuous. The skull is dolichocephalic, the paranasal sinuses are large and the palate high and arched. Lax capsules result in unstable or hyperextensible joints, kyphoscoliosis, pectus excavatum, and flat foot. Muscles may be underdeveloped and hypotonic, and subcutaneous fat is sparse.

Simple screening tests that may be helpful include the thumb sign (positive, if the thumb when completely opposed in the clenched hand, projects beyond the ulnar border), the wrist sign (positive if the thumb and little finger overlap when wrapped around the opposite wrist) and the ratio of the lower segment (pubic ramus to floor) to the upper segment (height minus lower segment), but this ratio varies with age and sex. Some tall people have high, arched palates, and some degree of arachnodactyly. This is probably of no consequence in many cases, although a Marfanoid habitus in women may be associated with mitral valve prolapse.\(^2,3\)

Our patient had some features of MFS (tall height, arm span > height, arachnodactyly, high arched palate, hypermobility of joints) with some features of EDS (hyperextensibility of skin and joints) suggesting that this patient had Marfanoid hypermobility syndrome, a heritable connective tissue disease with features of both MFS and EDS. Relatively, few patients have been described with concomitant EDS and MFS; and our case happens to be the first report from this part of the world. Currently, research is underway to identify whether this condition is an overlap of EDS and MFS or a genetically distinct syndrome. Several reports have associated this syndrome with a variety of cardiovascular anomalies (coarctation of aorta, flail mitral valve, etc.).\(^4-8\) However, no cardiovascular anomaly was present in our patient. Terada and Wanibuchi\(^9\) suggested surgery as one of the treatment modalities of Marfanoid hypermobility syndrome. However, our patient disagreed to go under the surgeon’s knife.

This case has been reported to highlight a rare condition from a dermatologist’s perspective. To the best of our knowledge, this is the first such case being reported from India.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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