Hypermobile Ehlers-Danlos syndrome: a video demonstration of Beighton score

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DESCRIPTION

An adolescent boy was brought by his parents with an 8-year history of increased elasticity of skin and flexibility of multiple joints. He also had associated bilateral knee joint pain for the last 6 months. The child's grandfather also had similar abnormal joint hypermobility. The medical history revealed a malunion of supracondylar fracture of the right humerus, which led to a gun-stock deformity. Cutaneous examination showed hyperelastic skin with a congenital melanocytic nevus over the right forearm. The joints were hypermobile with a Beighton score (BS) of 8 (figure 1, video 1). Hyperextension of the right elbow was not performed due to the gunstock deformity. There was no scoliosis or joint dislocation. Routine haematological and urine investigations did not reveal any abnormality. Neurological, cardiac and ophthalmological evaluation was found to be normal. A diagnosis of hypermobile Ehlers-Danlos Syndrome (hEDS) was established as the child satisfied the 2017 International Classification of the Ehlers-Danlos Syndromes criteria. Oral analgesics were prescribed for chronic knee joint pain along with knee stabilising braces. Health education regarding proper posture maintenance and mild strengthening exercise was provided to the patient.

EDS is a rare autosomal dominant connective tissue disorder affecting the skin and joints. There is a defect in the collagen metabolism that leads to the deposition of disordered collagen. Depending on the type of collagen affected, it has been classified into various phenotypes. Hypermobile EDS is one of the subtypes characterised by generalised hypermobility of joints, recurrent joint dislocations, chronic joint pain and osteoarthritic changes. Skin is usually soft, mildly hyperextensible with easy

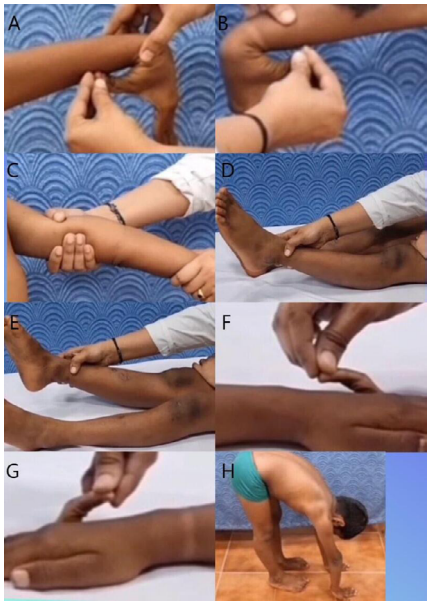
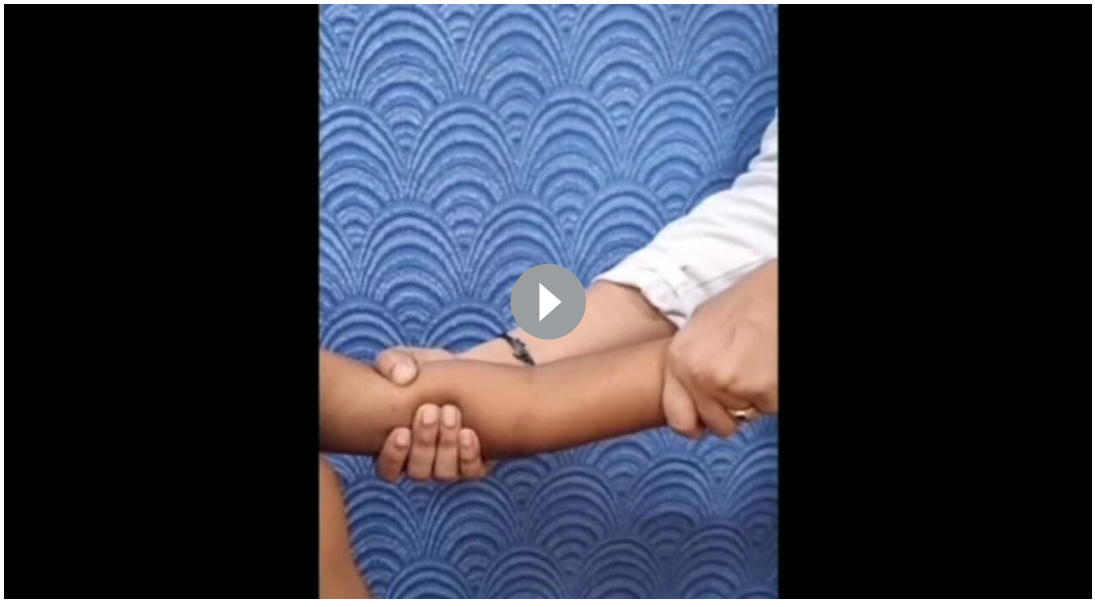


Figure 1 (A–H) performance of various manoeuvres of Beighton scoring system in our patient.



Video 1 A video demonstration of Beighton score in a hypermobile Ehlers-Danlos syndrome patient.

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Table 1 Beighton score for the assessment of generalised joint hypermobility

No.	Instructions given	Components	Left	Right
1	Keep your arms straight and wrist flexed, I will bend your thumb to the front of your forearm	Flexion of the thumb towards the flexor aspect of the forearm—passive	1	1
2	I shall extend your elbow backward	Hyperextension of elbow beyond 10°—passive	1	1
3	I shall extend your knee backward	Hyperextension of knee beyond 10°—passive	1	1
4	Keep your forearm on the flat surface, I will bend your little finger to the back of your hand	Dorsiflexion and hyperextension of the little finger beyond 90°—passive	1	1
5	Can you bend and touch the floor with your palms, keeping your knees straight	Forward flexion of the trunk with palms touching the floor, with knees straight—active	1	
Total score 9				

bruisability. Patients may also have autonomic dysfunction with orthostatic intolerance and gastrointestinal symptoms.[1]

BS is an internationally recognised screening tool introduced in 1973 for the assessment of generalised joint hypermobility (GJH). This nine-point scoring system requires the performance of five manoeuvres, which include four bilateral passive and one active performance (table 1). According to Beighton, the original cut-off value for GJH was ≥5,[1–3] However, since the mobility of joints decreases with age, the International Consortium on the Ehlers-Danlos Syndrome Committee proposed the following criterion of BS for GJH in hEDS as (1) BS ≥6 for adolescents and prepubertal children, (2) ≥5 for men and women from puberty to 50 years of age and (3) ≥4 above 50 years of age. The limitations of this scoring system are as follows: (1) Examination of only a few joints and (2) 'All or none' nature, that is, cannot assess the degree of joint hypermobility. This case reiterates the importance of the BS in the diagnosis of joint hypermobility disorders.[1]

Learning points

- Beighton score is used to assess generalised joint hypermobility in heritable connective tissue disorders.
- A score of 5 and above out of 9 is considered generalised joint hypermobility.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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