ABSTRACT

Loeys-Dietz syndrome is characterised by vascular aneurysms, hypertelorism, and a bifid uvula. We report on an 11-year-old boy with Loeys-Dietz syndrome who presented with bilateral radial head dislocations and severe osteopaenia with changes of avascular necrosis in both hips causing an out-toeing, wide gait. Considering the poor prognosis for elbow movement and possible radial head dysplasia, surgical reduction of the radial heads was deferred. A subtrochanteric de-rotation osteotomy of the left hip was performed to improve the gait.

Key words: hypertelorism; Loeys-Dietz syndrome; Marfan syndrome; radial heads, posterior dislocation of

INTRODUCTION

Loeys-Dietz syndrome is characterised by vascular aneurysms, hypertelorism, and a bifid uvula, and a variety of skeletal abnormalities. We report an 11-year-old boy with Loeys-Dietz syndrome who presented with bilateral radial head dislocations and severe osteopaenia with changes of avascular necrosis in both hips causing an out-toeing, wide gait.

CASE REPORT

In February 2010, an 11-year-old boy presented with progressively increasing deformities of both elbows and hips since birth. There was no hindrance in activities of daily living. The patient was born of non-consanguineous parents and had no major trauma, fever, early morning stiffness or any systemic complaints. Developmental stages were normal.

The child had hypertelorism with dolicocephalic features, along with a high-arched palate and a bifid uvula (Fig. 1). He also had a mildly flexible dorsolumbar scoliosis with convexity to the left, without any chest wall deformity. Dolicostenomelia was evident, with an arm span to height ratio of 1.1. Both upper limbs had a fixed elbow flexion deformity of 50º with further flexion up to 130º, and...
the forearms were fixed in a mid-prone position (Fig. 1). The 3-point bony relationship was well-maintained. The radial heads were prominent and dislocated posteriorly. There was no distal neurovascular deficit. Arachnodactyly was present, and the Steinberg’s sign1 (the entire distal phalanx of the thumb protruding beyond the ulnar border of the clenched fist) and Walker-Murdoch sign2 (the thumb covering the entire distal phalanx of the little finger when wrapped around the contra lateral wrist) were positive. Movement of the left hip was severely restricted with a flexion range of 20° to 90° and a fixed abduction and an external rotation deformity of 30° and 50°, respectively, causing an out-toeing, wide gait. The right hip was relatively normal except for terminally restricted rotations. The knees appeared normal. Both feet had mild (grade 1) metatarsal adductus with heel valgus (Fig. 1), according to the Bleck classification.3

Radiographs revealed grossly dysplastic elbow joints, and the radial heads were posteriorly dislocated (Fig. 2). The pelvis had severe osteopaenia

---

**Figure 1** (a) A high-arched palate and a bifid uvula, (b) hypertelorism, a flexion deformity of both elbows, and an external rotation deformity of the left hip, (c) restricted range of motion of both elbows with a flexion deformity of 50° with further flexion up to 130°, (d) positive Steinberg sign (the entire distal phalanx of the thumb protruding beyond the ulnar border of the clenched fist), (e) positive Walker-Murdoch sign (the thumb covering the entire distal phalanx of the little finger when wrapped around the contra lateral wrist), and (f) mild bilateral metatarsus adductus.

**Figure 2** (a) Posteriorly dislocated and severely dysplastic radial heads. The dysplastic lower end of the humerus and the posteriorly bowed ulna. (b) Severe osteopaenia with changes of avascular necrosis in both hips.
and changes suggestive of avascular necrosis with arthritis in the left hip and early avascularity in the right hip (Fig. 2). The spine had a mild dorsolumbar scoliotic curve from D8 to L3 with a Cobb’s angle of 20°.

Aneurysmal changes involving the major vessels were ruled out by 2-dimensional echocardiography. Ophthalmic examination was normal and did not reveal any lenticular anomalies. The cardiovascular system was normal with no audible heart murmur.

Surgical reduction of both dislocated radial heads was deferred because of the parental concern of possible loss of range of motion as well as the poor prognosis for elbow movement and possible radial head dysplasia. A subtrochanteric de-rotation osteotomy of the left hip was performed to improve the gait. At the 6-month follow-up, the patient was walking with a relatively normal gait with the hip externally rotated by 10°. His cardiac status was regularly monitored by echocardiography.

DISCUSSION

Loeys-Dietz syndrome is characterised by abnormalities in the cardiovascular, musculoskeletal, and craniofacial systems. It was initially considered a subgroup of Marfan syndrome, with additional features such as hypertelorism, cleft palate, and uvular abnormalities. These features along with cardiovascular abnormalities (vascular tortuosity and widespread aneurysms) distinguish it from the Marfan syndrome. Delayed or missed diagnosis is the leading cause of mortality in patients with Loeys-Dietz syndrome. Our patient had not yet developed aneurysms; the mean age of manifestation of cardiovascular anomalies is in the third decade of life. Previous cases have been reported in Caucasian, Japanese, Korean, and Chinese people.

The differential diagnosis of Loeys-Dietz syndrome includes Ehlers-Danlos syndrome, Marfan syndrome, Shprintzen-Goldberg syndrome, and arthrogryposis complex. Loeys-Dietz syndrome has a number of distinctive features including craniosynostosis, hypertelorism, cleft palate, bifid uvula, extremit contractures, talipes equinovarus, metatarsus adductus, cervical malformations, and instability. Many patients with Loeys-Dietz syndrome have dermatological changes in the form of translucent skin and easy bruising. Prompt cardiovascular evaluation and early treatment with antihypertensive drugs (beta blockers and angiotensin-converting enzyme inhibitors) may prevent vascular catastrophe.

There are 2 types of Loeys-Dietz syndrome. Type I is characterised by the presence of cleft palate, hypertelorism, and craniosynostosis, whereas type II is associated with none of these abnormalities but an isolated bifid uvula. The craniofacial severity index (0–11) has been used to evaluate the syndrome; higher scores indicate more severe abnormalities. A score of 2 is given for marked hypertelorism, 1 for subtle hypertelorism (inter-pupillary distance at or around the 97th percentile), and 0 for no hypertelorism. A score of 0 is given for absence of cleft palate and craniosynostosis, 6 if both are present, and 3 if either is present. A score of 3 is given for bifid uvula, 2 for midline raphe, 1 for a broad uvula, and 0 for a normal uvula. Our patient had type-1 Loeys-Dietz syndrome and a craniofacial severity index of 5, which has a low risk for vascular anomalies.

In the largest retrospective analysis of musculoskeletal anomalies in Loeys-Dietz syndrome, the mean age at presentation was 21 years; 18 of 65 patients were initially diagnosed as having Marfan syndrome or Ehlers-Danlos syndrome. Spine and foot abnormalities were the most common features. Upper cervical spine abnormality was found in 51% and thoracolumbar scoliosis in 55% of the patients, with a mean cobb angle of 30°±18°. Two of the patients also had mild spondylolisthesis. Combined Steinberg and Walker-Murdoch signs were present in 25% of the patients, whereas 23% had talipes equinovarus and 41% had severe hindfoot varus. Although most patients responded well to early serial casting, they tended to have a final hindfoot valgus. A combination of joint contractures with hyper extensible joints was common. Mild acetabular protrusion was present in 33% of the patients. Two of the patients underwent varus de-rotation osteotomy for dysplastic hips. One patient underwent arthroplasty at age 30 years for avascular necrosis of the hip.

For contractures of the upper limb, management has been non-surgical, with physiotherapy and serial stretch casts. All joint contractures reported have been soft-tissue contractures and not bony deformities. Congenital radial head dislocations have been associated with the Cornelia De Lange syndrome, Klinefelter syndrome, and Mieten syndrome, but never in Loeys-Dietz syndrome. Congenital radial head dislocations have a poor prognosis after surgical reduction and are best left alone. Radial head dysplasia may also be present.

DISCLOSURE

No conflicts of interest were declared by the authors.
REFERENCES