CASE REPORT

Weismann-Netter-Stuhl syndrome: report of two cases and treatment

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SUMMARY

Weismann-Netter-Stuhl syndrome is a rarely reported cause of bowed legs; hence a thorough clinical and radiological examination is needed for its diagnosis. In view of the paucity of reports guiding the treatment of the deformity, we propose a one-level/two-level osteotomy with intramedullary nail fixation as a treatment modality for the tibial bowing.

BACKGROUND

Weismann-Netter-Stuhl syndrome (WNS), first described by Weismann-Netter and Stuhl (France) in 1954, is a rare diaphyseal dysplasia associated with anterior tibial bowing and posterior cortical thickening of the tibia and fibula.¹ With most cases having bilateral involvement,²⁻⁴ short stature and tibialisation of fibula are also commonly seen.^{5, 6} Majority of the literature on the syndrome exists in the form of case reports and series,^{1, 2, 4, 5} with most of them failing to address the appropriate treatment guidelines in correcting the deformity. We present two cases with this syndrome who responded well to fixation using an intramedullary tibial nail after one-level/two-level osteotomy of the tibia and fibula along with bone grafting.

CASE PRESENTATION

Two siblings, a 152 cm tall 22-year-old man and a 148 cm tall 28-year-old woman presented to us with bowing of both legs. A detailed family history revealed two more siblings (all 4 of the progeny) and their father being affected in a similar manner with none of their progeny or the previous generation suffering from the same affliction (figure 1). Clinical evaluation of both cases showed anterior bowing of bilateral tibia with deformity in the upper limbs. Neck movements, facial features,

spine examination and biochemical studies including serum calcium, phosphate and alkaline phosphatase levels, and screening CT of craniovertebral junction were normal. Psychiatric evaluation showed normal intelligence.

INVESTIGATIONS

Radiographs of lower limbs (figure 2) showed bowing of bilateral tibia and fibula with broadening of fibula and thickening of posterior cortices of the tibia and fibula.

DIFFERENTIAL DIAGNOSIS

Differentials include persistent physiological varus, vitamin D deficiency rickets, renal osteodystrophy, proximal tibial metaphyseal focal fibrocartilaginous defects, blount disease, type IX Ehlers-Danlos syndrome, fibrous dysplasia, hypophosphatasia and osteogenesis imperfecta.⁷

Biochemical markers and routine laboratory investigations, such as serum calcium, serum phosphate and alkaline phosphate levels, are used in children to differentiate from congenital syphilis and rickets which are in addition usually associated with a larger and more generalised involvement of the skeleton.⁵ The radiographic findings and clinical history of these diseases are also quite different. While rickets involves mainly the metaphyseal region of the bone, the bowing in congenital syphilis usually shows bowing at mid-tibial level with the thickening of the cortex having a uniform symmetry.⁵ Patients with rickets also usually have a history of vitamin D deficiency in childhood, with resulting thoracic and epiphyseal lesions.² Blount disease clearly lacks a history of illness, generalised involvement of the skeleton, multiple deformities and terminal short stature in comparison with the other differentials.⁷

Some of the other differential diagnosis can be differentiated from this syndrome by lack of association with neurofibromatosis type 1 as in fibrous dysplasia.⁵

In comparison, patients with WNS usually have persistently raised serum alkaline phosphatase levels with a history of increased bowing all through puberty with greater incidence of short terminal stature. They may also have abnormal pelvic radiographic findings.⁸

TREATMENT

Both patients underwent corrective osteotomies of bilateral tibia and fibula and fixation using intramedullary interlocking tibial nail and autologous bone grafting (figure 3). Three of the four procedures

Figure 1 Pedigree chart, filled objects denote individuals affected with Weismann-Netter-Stuhl syndrome; and 1 and 2 denote the index cases who underwent treatment.

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Figure 2 Preoperative radiographs of bilateral legs of both patients.

required a two-level osteotomy to correct the deformity while one procedure required just a single-level osteotomy of the tibia.

OUTCOME AND FOLLOW-UP

At the end of the 6-month follow-up period, all the osteotomies had united and the patients were satisfied at the end of 3 years of follow-up.

DISCUSSION

Bowing of legs is frequently labelled as vitamin D deficiency rickets and a significant percentage of affected children are misdiagnosed and mismanaged. A careful examination and a high index of suspicion will enable the clinician to look for other causes of bowing of legs including WNS syndrome. It is genetically associated with mutation of an unknown gene. Most case series reported are from a single family and many constitute siblings from unaffected parents. Most of the reported patients are in European literature^{1 5 6 9} and a major bulk of them are French adults. Diagnosis in children is delayed because of the rarity of the entity in this age group^{6 10} and more prevalence of other common causes of deformation of bone, which is of profound importance in a developing country like India. Another reason for delay of diagnosis is unfamiliarity of the clinician and radiologist with the entity and unclear presenting symptoms and radiological appearance.

Bowing of tibia and fibula is typically seen at the junction of the middle and distal thirds of the bones in radiographs as in our cases. Bilateral symmetrical tibialisation of fibula, which is apparent as increased diameter, enlargement and thickening of fibula, is the pathognomonic feature for the diagnosis of this

syndrome.^{5 6} Although unilateral involvement has been reported, it is quite rare.^{1 5 6} Tibial exostosis,^{5 6} involvement of upper limbs and femur have also been reported.^{1 5} Defect in longitudinal growth of the bone is another feature of the syndrome resulting in significantly short stature, the reason for which is still obscure. The reported heights of adults in literature are within the range 135–155 cm and the majority range from 145 to 155 cm⁶ and is true of both the patients in our report. Two cases with associated craniovertebral malformations have been reported by Kaisi *et al*,⁹ who also recommended a routine CT scan to rule out such a malformation due to its life-threatening implications though none of our patients had this malformation. Family history, though uncommonly reported, suggests that the syndrome is genetically transmitted in an autosomal-dominant pattern with incomplete penetrance.^{5 6}

Patients (siblings) in our report were diagnosed on the basis of clinicoradiological correlation of the findings. Patients were less than third centile for their heights. Their radiographs showed bowing of the tibia and fibula along with features of tibialisation of fibula. The posterior cortices of the bones were thickened. Metabolic investigations were within normal limits. Involvement of other family members was a reliable clue which aided in the diagnosis. Alp *et al*⁶ reported a case series which involved a father and two other family members of the paternal tree in a pedigree analysis.

Learning points

- ▶ Weismann-Netter-Stuhl syndrome is a rare condition which is often difficult to diagnose especially in children of developing countries where bowing of legs due to nutritional causes is more prevalent.
- ▶ A positive family history and awareness of this rare condition are very important in making a diagnosis of this syndrome. Delayed detection and misdiagnosis are common because of the lack of awareness.
- ▶ Biochemical markers and serology differentiate it from other commonly prevalent differential diagnosis.
- ▶ We recommend a one-level/two-level osteotomy of the tibia and the fibula with fixation using intramedullary tibial nail along with bone grafting as a treatment modality in this condition on the basis of the successful outcome of the same in our report.

Figure 3 Postoperative radiographs of bilateral legs of both patients.

Contributors PG, RM, SM and VS were involved in the preparation of the manuscript, patient care and approval of the final manuscript.

Competing interests None.

Patient consent Obtained.

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