Case Report

Melnick- Needles Osteodysplasty Presenting With Quadriparesis

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Abstract

Melnick-Needles syndrome or osteodysplasty, a monogenic heritable bone dysplasia, is characterized by a typical facies and characteristic radiological findings. Less than 70 well-documented cases have been reported in literature; most of them were sporadic. We report the first case from Eastern India in an adolescent male, who had cranio-vertebral junction anomalies and presented with spastic quadriparesis at the age of 13 years. ©

Introduction

A case of rare Melnick-Needles osteodysplasty producing cervico-medullary compression due to complex craniovertebral (CV) junction anomalies is reported.

Case Report

A 13 years male presented with progressive weakness of all four limbs for two weeks. The weakness first started in the left lower limb distally and progressed rapidly over next one to two days to involve the other limbs. There was no history of loss of consciousness, convulsion, headache, sensory abnormalities, abnormal movements, sphincter dysfunction, involvement of facial muscles, dysphagia, respiratory distress, oscillopsia, or similar history in the past. The only child of non-consanguineous parents, he was born with several bony deformities, had delayed developmental and motor milestones and was able to walk only at the age of five years. His vision and hearing were normally developed but he could not utter meaningful words. Family history was non-contributory.

On examination, facial features were remarkable with broadened forehead, exophthalmos, widely set eyes with anti-mongoloid slant, depressed temples, fleshy nose, large ears, prominent upper jaw, micrognathia, and dental malalignment (Fig. 1). Neck: height ratio was 1: 16.8. Skeletal examination revealed scoliosis to the right, pectus excavatum, flexion deformity of the elbow, metacarpo-phalangeal and interphalangeal joints, long slender fingers, genu valgus, hallux valgus, and flexion contracture of the toes. A clinical diagnosis of Melnick-Needles syndrome was made. Neurological examination revealed diminished power, hypertonia, bilateral extensor plantar responses, and exaggerated deep reflexes. Cranial nerves were not affected. Examination of other systems was unrevealing.

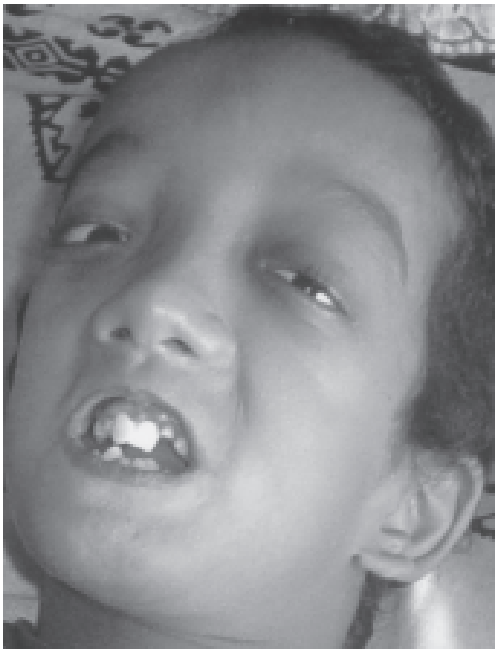

Fig. 1 : Photograph showing typical facial appearance.

Routine hemogram, renal and hepatic biochemical profile was normal. Skeletal survey showed generalized osteosclerosis, bony sclerosis of base of the skull structures, abnormal dental alignment, platybasia, fusion of upper cervical vertebrae (C_2- C_4), sclerosis and irregularity of clavicles and ribs, scoliosis of thoracic vertebrae (T_4-T_8), cortical irregularity of tubular bones

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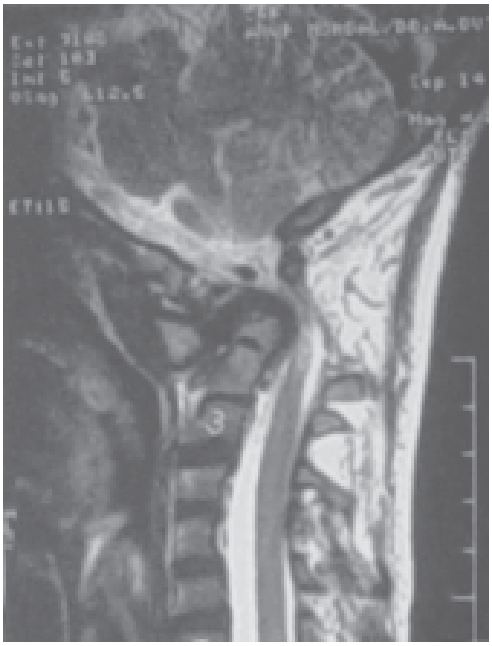

Fig. 2 : T_2- weighted MRI of skull and upper cervical spine (sagittal view) showing platybasia, partial arthrosis (C_2-C_3), anterior atlanto-axial subluxation and posterior angulation of the odontoid tip.

with metaphyseal flaring and diaphyseal bowing, and broadening of iliac crest and vertebral bones. Abdominal ultrasound, electrocardiogram, echocardiogram, and radiography of lung fields were unremarkable. On multiplanar magnetic resonance imaging (MRI), atlanto-occipital assimilation was seen with almost fusion of posterior arch of atlas with occipital bone. There was platybasia, partial arthrosis between C_2-C_3 vertebrae, mild anterior atlanto-axial subluxation along with posterior angulation of the odontoid tip. The odontoid tip was well below McGregor's line. Narrowing of the C V junction was seen producing indentations on the lower brain stem and at cervico-medullary junction with hyperintense intramedullary signal changes suggestive of cord edema / myelomalacia (Fig. 2). No significant disc bulge was seen. A final diagnosis of Melnick-Needles osteodysplasty with multiple C V junction anomalies was made. Screening of the family members was negative.

Discussion

Melnick-Needles syndrome or osteodysplasty is a highly characteristic syndrome of the skeletal system, the affected individual having typical facial appearance.[1] Main clinical findings are relatively large cranium with high prominent forehead and generalized delay in closure of fontanelles, small facial part, exophthalmos, hypertelorism, fleshy nose, full cheeks, relatively large ears, micrognathia, malalignment of teeth and malocclusion.[2] Variable deformities of the arms, legs, fingers and toes are usually present along with characteristic radiological changes (especially cortical irregularities and narrowing of diaphyses) involving long bones and axial skeleton.[3] Apart from abnormal facies, patients usually attract attention because of an abnormal gait and bowed limbs.[2] Affected individuals attain normal adult height, and mental development is not affected.[2,3]

This monogenic heritable diseases is transmitted either as X-linked dominant or autosomal dominant, associated with lethality in males and normal life expectancy in females, and till date less than 70 cases have been reported in literature.[4] Affected children may have initial failure to thrive, and increased susceptibility to infection of upper respiratory tract and middle ear.[3] Pelvic deformity may cause difficult child bearing in adult, and narrow thorax may give rise to impairment of respiration. Premature arthritis can develop.[2] Prenatal recognition of skeletal changes is possible. Some superficial similarities to conditions like pyknodysostosis, craniometaphyseal dysplasia or Engelmann Caurati syndrome can be easily ruled out by appropriate radiological studies.[1] Partially expressed forms may be recognized only by chance. Several reports of 'serpentine fibula-polycystic kidney syndrome' with marked similarities to Melnick-Needles syndrome appear independently in literature.[5]

Though a definite genetic diagnosis was not possible, the typical facial features (particularly the dental malalignment and antimongoloid slant of eyes) and widespread skeletal deformities in this case were closely similar to those described for Melnick-Needles syndrome in literature. The patient presented with quadriparesis at 13 years of age as a result of cervico-medullary cord compression due to associated cranio-vertebral junction anomalies. There is no report of such a presentation in the literature. In absence of any family history, the patient was considered to have first phenotypic expression of a new mutation in his family. This may also be the reason for the course being not as lethal as reported in literature for the affected males. At present, patient is awaiting a spinal fixation procedure through the anterior approach considering the complexity and extent of his spinal deformities.

References

1. Greco F, Mauceri L, Finocchiaro M, *et al.* Melnick-Needles syndrome. Report of a case. *Pediatr Med Chir* 1998;20: 149-52.
2. van der Lely H, Robben SG, Meradji M, *et al.* Melnick-Needles syndrome (osteodysplasty) in an older male—report of a case and a review of the literature. *Br J Radiol* 1991;64:852-4.
3. Albano LM, Kim CA, Lee VK, *et al.* Clinical and radiological aspects in Melnick-Needles syndrome. *Rev Hosp Clin Fac Med Sao Paulo* 1999;54:69-72.
4. Donnenfeld AE, Conard KA, Roberts NS, *et al.* Melnick-Needles syndrome in males: a lethal multiple congenital anomalies syndrome. *Am J Med Genet* 1987;27:159-73.
5. Majewski F Enders H, Ranke MB, *et al.* 'Serpentine fibula-polycystic kidney syndrome' and Melnick-Needles syndrome are different disorders. *Eur J Pediatr* 1993;152:916-21.