

pancreatic circulation, which led to ischemia, autodigestion and chronic pancreatitis.

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Melnick Needles Syndrome

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Melnick Needles Syndrome (MNS) is a hereditary skeletal dysplasia involving most of the bones, first reported in 1963. A primary biochemical defect in collagen synthesis is hypothesized to be at fault(1). Clinical features, including a

typical facies, combined with characteristic radiographic features make its diagnosis relatively secure. We report the first case of MN syndrome of Indian origin.

Case Report

A 6-year-old female child presented with complaints of cough with expectoration for 15 days. There was no history of bone pains or poliomyelitis. Clinical examination demonstrated a thin built child of normal height and intelligence, with a slightly abnormal posture. She had very prominent supra-orbital ridges, normal eyes and a small chin. There was no evidence of hemi-hypertrophy or *cafe-au-lait* spots. Respiratory and cardiovascular systems were normal. No neurological deficit was found and spine was normal. Serum calcium, phosphorous, alkaline phosphatase and

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BRIEF REPORTS

blood urea levels were normal.

A chest skiagram done revealed a right middle lobe consolidation and a peculiar appearance of the thoracic cage. The thoracic cage was small with a markedly diminished anteroposterior diameter. There was a distorted 'ribbon-like' appearance of ribs bilaterally with cortical irregularity; and bone resorption at the lateral end of right clavicle (*Fig. 1*). Skull skiagram exhibited dolicocephaly and a wavy inferior margin of the mandible on the left side. Lamina dura was normal. There was scalloping of the posterior margin of vertebral bodies in the dorsolumbar spine with widening of the spinal canal

(*Fig. 2*). Concavity was also seen along the anterior margins of L₁ and L₂ vertebral bodies. Pelvis demonstrated flared, angular iliac blades with tapering of their caudal halves. The ischium and pubis were small with narrow, flattened obturator foramen. Also, protrusio acetabuli and marked coxa valga was present bilaterally (*Fig. 3*). Meta-physeal ends of long tubular bones were expanded. Angulation of upper ends of both radii was observed. Both tibia revealed a S-shaped bowing in the frontal plane only (*Fig. 4*).

On the basis of above clinical and radiological features a diagnosis of Melnick Needles Syndrome was made.

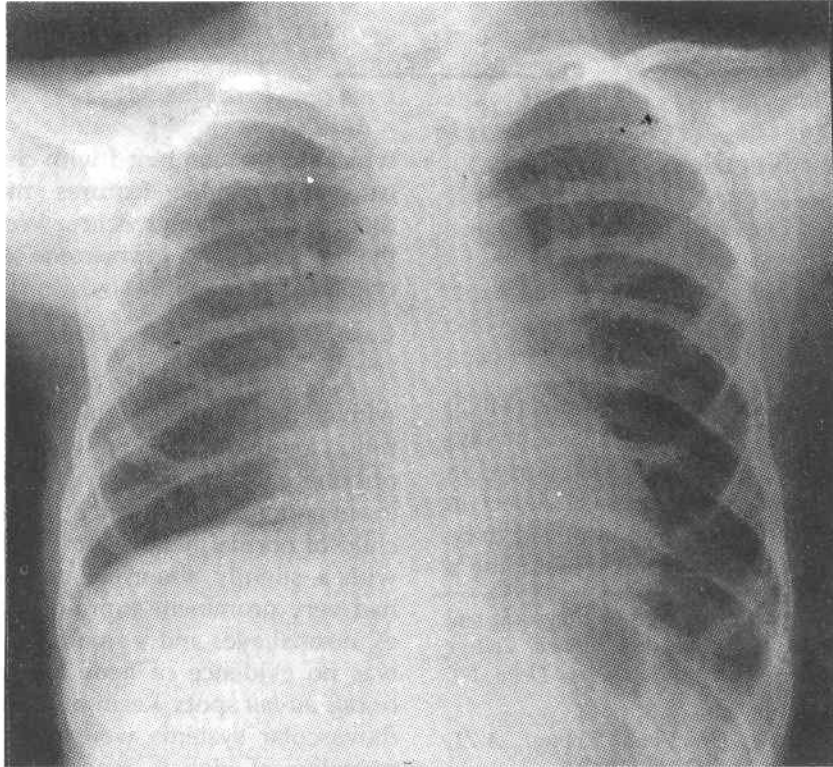


Fig. 1. Ribbon ribs with marked contour irregularity. Deficient ossification at lateral end of right clavicle and middle lobe consolidation is also seen.

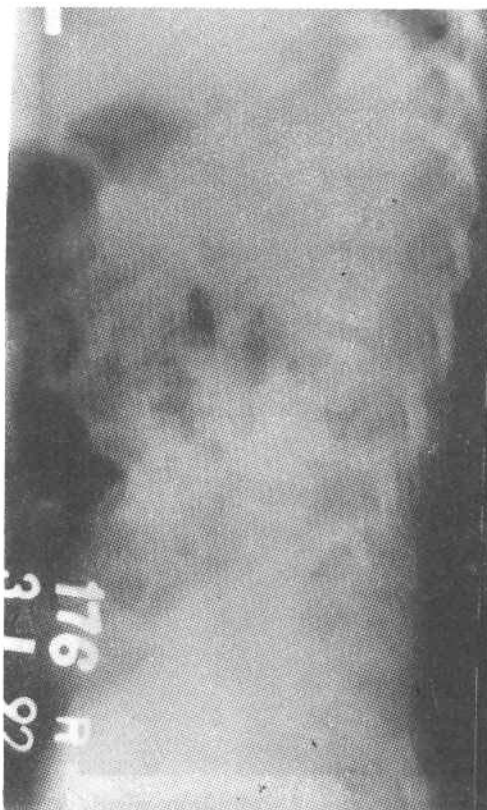


Fig. 2. Expansion of lumbosacral canal with posterior scalloping of vertebral bodies. Anterior concavity of L1 and L2 vertebral bodies is also evident.

Discussion

MN Syndrome or Osteodysplasty is a rare connective tissue disorder producing somatic abnormalities with characteristic radiographic features. Inheritance is thought to be X-linked/autosomal dominant(1-3), lethal in the male. There are less than 40 documented cases reported to date, of which only one was an Asian female thought to be of Japanese ancestry(1). We present the first patient of Melnick Needles Dysplasia seen in the Indian subcontinent.

Somatic abnormalities of this dysplasia include a small facies with high forehead, full cheeks, micrognathia, tooth malalignment and proptosis(4,5). Stature is mildly reduced while intelligence is normal. Patients are frail and have narrow shoulders and a small chest. They suffer from recurrent pneumonia and ear infections. The child may occasionally present with growth failure and delayed motor milestones. Pulmonary hypertension may occur, presumably due to small thoracic cage, pectus excavatum and repeated bouts of pneumonia and upper respiratory infection(6).

Skull shows dolicocephaly, large posterior fossa, mild basilar sclerosis, poorly developed mastoids, elongated posterior clinoids and thickened supraorbital ridges. Mandible appears hypoplastic with malaligned teeth and radiolucencies near the angle of mandible(2).

Thoracic cage is narrow and ribs shows striking cortical irregularity and appreciable focal constrictions which may mimic the 'twisted ribbon ribs' seen in neurofibromatosis. Clavicles may have cortical irregularity and deficient ossification at their distal ends(4), as seen in our case (*Fig. 1*). Spinal abnormalities include slight scoliosis, thoracolumbar gibbus or increased lumbar lordosis. Vertebrae are increased in height relative to their width and lumbar disc spaces are narrowed. Excessive concavity of anterior vertebral bodies produces a 'double-beak' appearance in upper dorsal and lumbar spine(1,3,4,7,8). Expansion of lumbosacral canal was thought to represent an expanding intraspinal lesion rather than

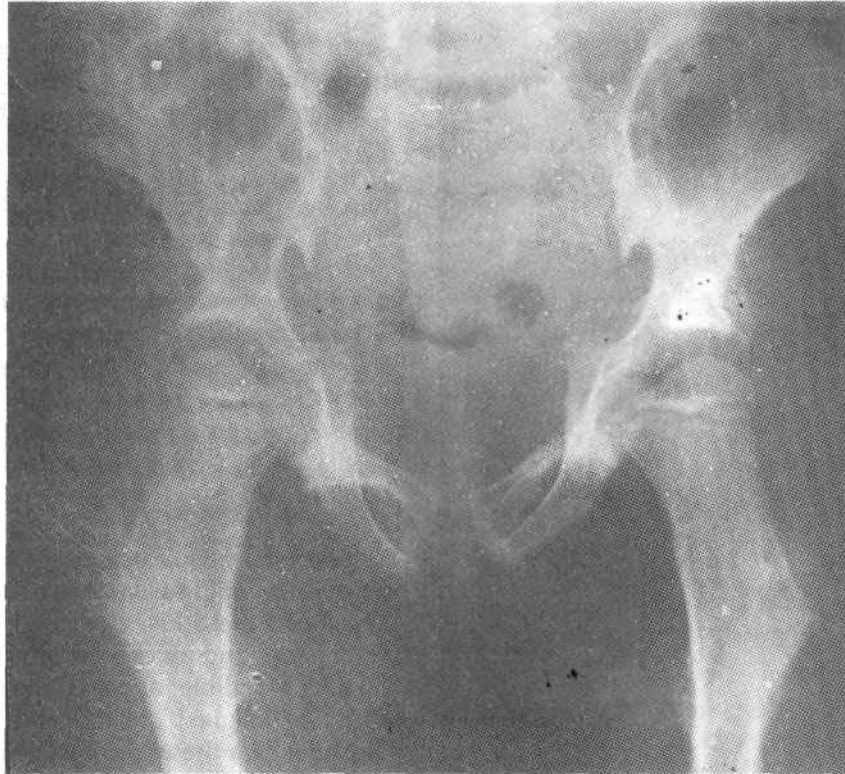


Fig. 3. Flared iliac blades with constriction of their bases. Hypoplastic ischii, pubic bones and obturator foramen with bilateral coxa valga.

a primary bony abnormality(6). Increase in both interpedicular and AP diameters of D-L spine was observed in our patient too, alongwith posterior scalloping of the vertebral bodies, which has not been described earlier. However, myelography was deferred due to absence of any neurological symptoms. In our opinion posterior scalloping seen in MNS without any neurological deficit, is likely to be due to a primary bony abnormality rather than an intraspinal tumor.

Pelvis shows a bizarre modelling in MNS, with a narrowed and triangular

superior pelvic opening, flared iliac crests with constriction of base of ilia. Ischii and public bones are hypoplastic. Obturator foramen are distorted. Protrusio acetabuli may be seen. Marked coxa valga and genu valgum may be observed. Limb bones show an alteration in the usual smooth contour of their cortices. S-shaped tibial and radial bowing is the hallmark of this syndrome, as observed in the present case.

Biochemical findings, *e.g.*, serum alkaline phosphatase, calcium and phosphorus levels and blood urea are normal(7).

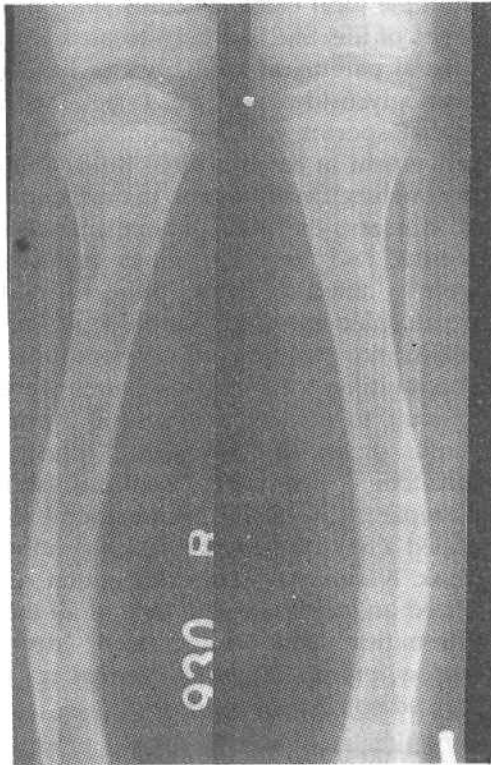


Fig. 4. Hallmark of MNS—S-shaped tibial bowing in the frontal plane.

In conclusion, we would like to stress that recognition of MN Syndrome is important because susceptibility to recurrent pneumonias, due to a small thoracic cage, may be responsible for early death of these patients.

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