# Pyle Metaphyseal Dysplasia

NEERJA GUPTA
MADHULIKA KABRA
CHANDAN J DAS*
ARUN K GUPTA *

**ABSTRACT**

*Pyle type metaphyseal dysplasia is a rare autosomal recessive disease that primarily affects metaphyses. We report a 12 year old boy with Pyle's disease. He had mild facial dysmorphism, genu valgum and wasting of legs. Skeletal radiology revealed the characteristic Erlenmeyer flask sign at distal femoral and proximal tibial metaphyses along with platyspondyly.*

**Key words**: *Erlenmeyer flask deformity, Pyle's disease.*

## INTRODUCTION

Pyle's disease is a rare familial metaphyseal dysplasia disorder with few reports worldwide(1-7). This paper presents a case report, review of literature, possible treatment options for Pyle's disease and discusses the differential diagnosis for Erlenmeyer flask deformity.

## CASE REPORT

A 12-year-old male child born to a third-degree consanguineous couple was referred to our genetic clinic as a case of suspected Gaucher's disease. He presented with increasing wasting of lower limbs for 6 months. There was history of fracture of right forearm after trivial trauma. There was no history of bone or joint pains, jaundice, blood transfusion or developmental delay. There was no family history of similar problem. His weight was 28 Kg (at 10th centile NCHS) and height was 138 cm (between 10th and 25th centile NCHS). He had hypotelorism, round face, misaligned teeth and small chin. He also had widening of bones of the forearm along with genu valgum and wasting of legs. There was no scoliosis, anemia, or hepatosplenomegaly. Examination of the parents was normal. Investigations revealed normal hematological, renal and hepatic parameters. Skeletal survey showed expansion of cortical marrow in bilateral distal femora (***Fig.* 1**) and proximal tibiae (***Fig.* 2**) suggestive of Erlenmeyer flask deformity. There was mild expansion of lower end of ulna. Multiple growth arrest lines, diffuse osteopenia and bowing of the bilateral tibiae was also seen (***Fig.* 2**). *X*-ray of the dorsolumbar spine revealed platyspondyly. *X*-ray skull and chest were normal. Bone densitometry was suggestive of osteoporosis (*z* score-5.3). Based on clinical features and radiological findings, we made a diagnosis of Pyle metaphyseal dysplasia.

## DISCUSSION

Pyle metaphyseal dysplasia has peculiar radiological findings with relatively unremarkable phenotypic features. It is characterized by defect in metaphyseal remodeling that leads to grossly widened metaphyses of long bones with marked cortical thinning and osteoporosis (Erlenmeyer-flask deformity) especially in the distal end of femur and proximal tibia. Proximal two-thirds of humerus and distal two-thirds of radius and ulna may show similar changes. Similar but less striking changes are seen in other distal long bones, distal metacarpals and proximal phalanges. Spinal involvement varies from moderate platyspondyly to biconcave lens appearance of the vertebral bodies(3,4). Jaw involvement has also been documented(5). Skeletal survey of obligate heterozygotes may show minor skeletal changes. Along with these roentgenographic changes, there

*From the Genetic Subdivision, Department of Pediatrics and *Department of Radiodiagnosis, All India Institute of Medical Sciences, New Delhi 110 029, India.*

*Correspondence to: Madhulika Kabra, Additional Professor, Genetics Subdivision, Department of Pediatrics, All India Institute of Medical Sciences, New Delhi 110 029, India.*
*E-mail: madhulikakabra@hotmail.com*

*Manuscript received: March 21, 2007;*
*Initial review completed: July 12, 2007;*
*Revision accepted: October 23, 2007.*

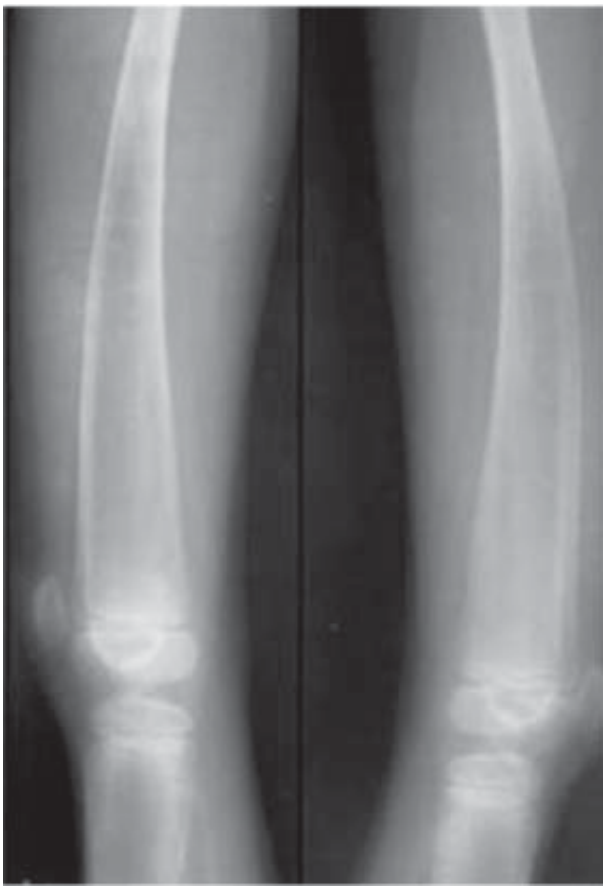

FIG. 1. *X-ray of lower thigh lateral view showing Erlenmeyer flask deformity in distal femora. Note diffuse osteopenia.*

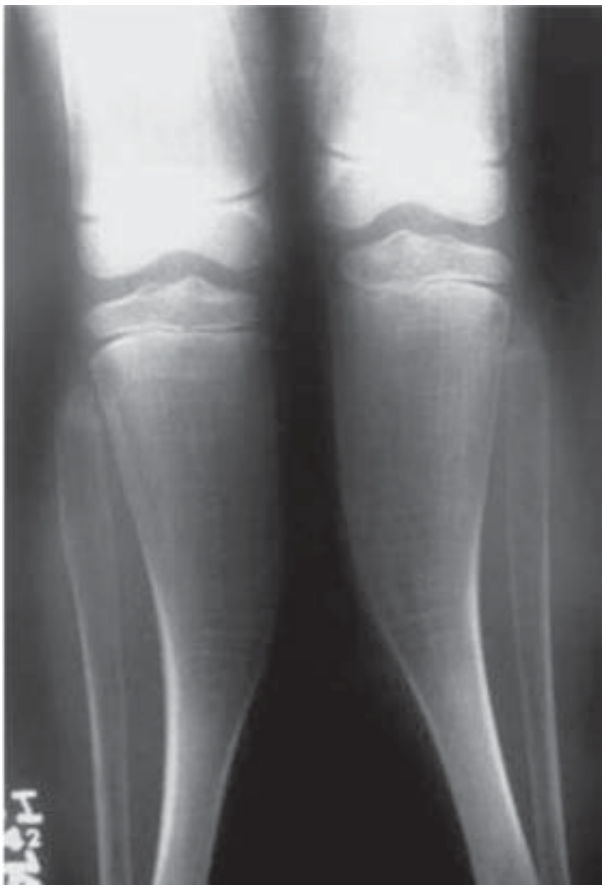

FIG. 2. *X-ray of upper leg AP view showing Erlenmeyer flask deformity in bilateral proximal tibia. Note multiple growth arrest lines and bowing of the tibia.*

may be few clinical signs and symptoms like muscle weakness, joint pain, genu valgum, scoliosis, and limited extension of elbow. Temporary asymmetrical tibial epiphysiodesis or bilateral osteotomies can correct the progressive genu valgum not only by provoking the momentary arrest of growth in the growth cartilage on the internal tibial aspect, but also by allowing for bone remodeling in the tibial metaphysis(6,7).

Other differential diagnoses of Erlenmeyer flask deformity are craniotubular dysplasias including craniometaphyseal and craniodiaphyseal dysplasia, Gaucher disease, osteopetrosis, thalassemia and Niemann Pick disease. Absence of cranial involvement excludes first three diagnoses in our patient. Moreover, our patient did not have anemia, jaundice, hepatosplenomegaly, increased bone density or any history of blood transfusion ruling out the latter four possibilities also.

*Contributors:* NG, MK: Preparing and reviewing the literature; and CJD, AKG-Radiological description.

*Funding:* None.

*Competing interests:* None stated.

## REFERENCES

1. Percin EF, Percin S, Koptagel E, Demirel H. A case with Pyle type metaphyseal dysplasia: clinical, radiological and histological evaluation. Genet Couns 2003; 14: 387-393.
2. Raad MS, Beighton P. Autosomal recessive inheritance of metaphyseal dysplasia (Pyle disease). Clin Genet 1978; 14: 251-256.
3. Heselson NG, Raad MS, Hamersma H, Cremin BJ, Beighton P. The radiological manifestations of metaphyseal dysplasia (Pyle disease). Br J Radiol 1979; 52: 431-440.
4. Turra S, Gigante C, Pavanini G, Bardi C. Spinal involvement in Pyle's disease. Pediatr Radiol 2000; 30: 25-27.

5. Narayananan VS, Ashok L, Mamatha GP, Rajeshwari A, Prasad SS. Pyle's disease: an incidental finding in a routine dental patient. Dentomaxillofac Radiol 2006; 35: 50-54.

6. Lindberg EJ, Watts HG. Postosteotomy healing in Pyle's disease (familial metaphyseal dysplasia). A case report. Clin Orthop Relat Res 1997; 341: 215-217.

7. Ferrari D, Magnani M, Donzelli O. Pyle's disease. A description of two clinical cases and a review of the literature. Chir Organi Mov 2005; 90: 303-307.

# Atypical Teratoid/Rhabdoid Tumor Mimicking Tuberculous Meningitis

**NILGUN SELCUK**
**MURAT ELEVLI**
**DICLE INANC**
**HUSEYIN ARSLAN**

## ABSTRACT

*Atypical teratoid/rhabdoid tumor of the central nervous system is a highly malignant neoplasm in infants and young children. We report a 6 year-old girl with atypical teratoid/rhabdoid tumor. Based on cerebrospinal fluid examination MRI scan and family history of tuberculosis; we diagnosed tuberculous meningitis. There was inadequate response to the antituberculosis therapy; so we performed stereotactic brain biopsy. Pathologic result revealed high grade atypical teratoid/rhabdoid tumor.*

**Key words:** *Atypical teratoid/rhabdoid tumor, Tuberculous meningitis.*

## INTRODUCTION

Primary atypical teratoid/rhabdoid tumors (AT/RT) are extremely rare malignant intracranial neoplasms. To date, approximately 200 cases of atypical teratoid/rhabdoid tumor (AT/RT) of the central nervous system have been described in the literature(1). It is a highly aggressive neoplasm, often has central nervous system dissemination, does not respond to therapy and typically is fatal within 1 year(2).

*From the Department of Pediatry, Haseki Education and Research Hospital, Istanbul, Turkey.*

*Correspondence to: Dr. Nilgun Selcuk, Sef Yardimcisi, Haseki Egitim ve Arastirma Hastanesi, Cocuk Klinigi, 34300, Fatih – Istanbul, Turkey.*
*E-mail: nilgunduru@yahoo.com.*

*Manuscript received: October 19, 2006;*
*Initial review completed: December 5, 2006;*
*Revision accepted: November 1, 2007.*

## CASE REPORT

A 6-year-old girl was admitted to our clinic with confusion, headache, vomiting, aphasia, and right hemiparesis for last two months. She had family history of tuberculosis. On admission, the patient was lethargic. She was not alert and oriented. On clinical examination right hemiparesis was detected. Cerebrospinal fluid (CSF) examination revealed 350 lymphocytes/$mm^3$, protein 40.8 mg/dL and glucose 36 mg/dL. At the same time blood glucose was 136 mg/dL. MRI brain showed marked leptomeningeal involvement and basal meningitis. The initial working diagnoses included tuberculous meningitis and malignant infiltration. Spinal MRI was normal. Cerebrospinal fluid was negative for viral serology and PCR for *Mycobacterium tuberculosis*. No malignant cell were demonstrated in cerebrospinal fluid. Chest radiography did not reveal any abnormality. The tuberculin test was negative. Antitubercular and antiedema treatment was initiated. Post-treatment course showed an improve-ment in neurological status, but two weeks later the child developed ptosis and loss of vision. Cranial CT scan showed meningial enhancement in the extracerebral sub-arachnoid spaces. There was minimal dilatation of third and lateral ventricles, and homogeneous contrast enhancement on both temporal lobes, right insular cortex, ambient cistern, and left sigmoid sinus. These findings were concluded as suggestive of tuberculous meningitis. Lumbar puncture revealed 120 lymphocytes/$mm^3$ with protein 129 mg/dL and sugar 19 mg/dL.