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Camurati- Engelmann Syndrome: A Case Report

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Summary:

A case of Camurati- Engelmann's syndrome, a form of progressive diaphyseal dysplasia of unknown aetiology in a boy of 14 months of age is reported. The boy presented with delayed motor mile stones, weakness, anterior bowing of tibia, and firm and prominent musculature. Diagnosis was made on the basis of characterstic radiological findings. This is the first case reported so far in Bangladesh.

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Introduction:

Prograssive diaphyseal dysplasia or Camurati-Engelmann syndrome or Engelmann's disease (as designated in earlier literature) is a rare autosomal disease, primarily affecting long bones and musculature. Cockaine in 1920 for the first time reported the disease in a 9.5 year old boy who presented with sclerosis of long bones and base of the skull[1]. The disease historically known as Camurati-Engelmann's disease was re-designated as progressive diaphyseal dysplasia in 1948 to stress the involvement of diaphysis and progressive nature of the disorder[2]. In course of time it was revealed that membranous bones, and in advanced stage even vertebral columns are also affected[2,3]. There is also involvement of muscles, resulting in weakness, waddling gait and pain[3]. Camurati reported this disease as a case of hereditary symmetrical osteitis of the lower limbs in a seven year old boy in 1923. The characterstic roentgenographic description came from Engelmann in 1929. Hence the name Engelmann's disease or Camurati-Engelmann's disease[1]. About 100 cases are so far been reported in different literature[4].

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Diversity of presentations was found in different literature according to the site and extent of involvement, and it has been suggested that the disease could be a systemic disease with predominant muscle and bone manifestations[4,5].

This case is reported with characteristic clinical and radiological presentation, only difference with others is that the child had increased muscle bulk.

Case report:

A 14 month old boy was brought to the hospital by his father with the complaints of anterior bowing of both legs with delayed mile stones. On inquiry, it was revealed that the child was lagging behind in motor abilities only. Other mile stones were normal. His birth history was uneventful. His nutrition seemed to be adequate. He was properly immunized. There was no history of other sibs or any relatives of the child being affected. The child had muscle weakness. He could stand up with difficulty.

He walked with waddling gait. He also had difficulty to keep on standing with a tendency to fall when attempted to walk. The father said that his son was clumsy in walking. This, he compared with motor development of other children. This is however the age by which a child can stand independently and walk steadily without fall. He did not have any pain in his limbs. His father's main concern was the anterior bowing of the legs. His muscles looked prominent and firm on feeling. His leg muscles were more prominent and pelvic girdle muscles were weaker than others.

Initial presentation aroused suspicion of myopathy. Other systemic examination revealed no abnormality. There was no neurological abnormality either.

His biochemical markers were normal (Table-I) and excluded the possibility of myopathy. X-ray of the legs (Fig.-1) showed diaphyseal sclerosis with sub-periosteal lamellar bone formation. Muscle biopsy could not be done. Bone biopsy also could not be done.

Table-I

Biochemical markers of the patient

CPK	30 U/L(Range: 5-195 U/L)
LDH	245 U/L(Range: 30-460 U/L)
Serum calcium	09 mg/dl(Range: 8.1- 10.4 mg/dl)
Serum phosphate	04.1 mg/dI(Range: 2.4-4.5 mg/d l)
Serum alkaline phosphatase	285 U/L(Range: 98-279 U/L)*

*Higher level may be considered normal for a growing child

So, it was diagnosed to be a case of Engelmann's disease. The child is in follow-up for one year. He is improving gradually in terms of muscle weakness and gait abnormality.

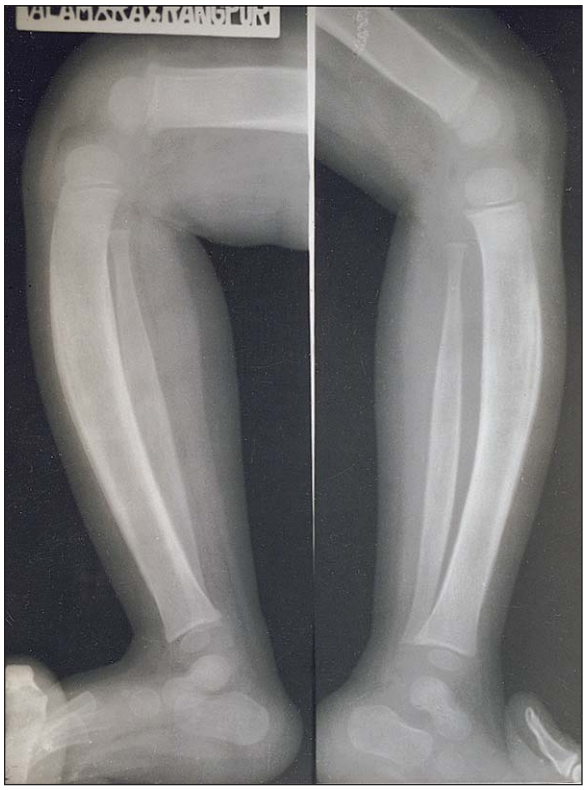

Figure-1 : *Radiological finding of tibia and fibula of the case*

Discussion:

Camurati-Engelmann syndrome is a combination of muscular dystrophy and dysplasia of bone. Often it has been described as progressive diaphyseal dysplasia of bone. But it has been reported by many authors that almost all bones of the body including long bones, axial bones and skull bones may be affected[1-8]. In this patient only the long bones of lower extremity were involved.

Since Camurati and Engelmann reported the syndrome, cases are being reported from different parts of the world with diverse clinical presentations with the key presentations like muscle weakness, gait disturbance (characteristically waddling or woobling gait) and sclerotic change in the bone remaining the constant features[1,4].

These were the features in this case also. In addition, the boy had anterior bowing of legs.

Applegate and colleagues reported cranial neuropathy due to stenosis of foramina of skull bones resulting in impaired hearing, difficulty in talking and chewing [4]. However, no neurological deficit was detected in this boy. About 100 cases have been reported up till now[4].

Engelmann's disease is yet to be studied as regards its cause and pathogenesis. Extensive literature review reveals that this rare disease presents with gait disturbance, muscular weakness, pain in the leg, thin and small muscle mass, anterior bowing of tibia, shiny skin over anterior surface of tibia, expression less face, tight skin of maxilla, enlarged jaw and occasional involvement of cranial nerves[1,4]. This patient presented with many of these features. Patients are biochemically normal, as was the reported case here. Bone biopsy shows thickening of diaphyseal cortices and endosteally and subperiosteally formed new bones. However, bone biopsy could not be done in this case.

Patients sometimes present with the features of muscular dystrophy, particularly of pelvic girdle type. Radiology of long bones confirms the diagnosis. Among the atypical features, delayed mile stone and systemic manifestation like fatigue, poor appetite, lamellated periosteal reaction, joint involvement with contracture and crippling pain are also noticed[5,7-10]. In this case all the features except joint involvement and pain were noticed.

There are reports of sporadic cases but familial propensity is marked. Sparks and Graham have suggested autosomal dominant mode of inheritance with variable expressivity of the gene[8]. No familial tendency was found in this case.

Treatment of Engelmann's disease is uncertain. Corticosteroid has been tried with limited success. Reports of alleviation of symptoms of pain and other manifestation to some degree has been reported but its mode of action could not be explained[9,10]. Surgical intervention in crippling disease like joint involvement and contracture has been suggested but not strongly recommended. Analgesics failed to show any response[8]. No treatment was given to this boy. But on follow up he has shown some noticeable improvement in gait and muscle strength.

The case reported here simulates with the manifestations of Engelmann's disease in many respect. The typical presentations, muscle weakness, gait disturbance and anterior bowing of the legs were clinically very obvious in this case. Absence of biochemical abnormality and radiological evidence of diaphyseal sclerotic change with subperiosteal lamellar bone formation suggests Engelmann's disease. Noticeable deviation here is improvement in gait and weakness of the patient. However, improvement has been rported in earlier literature also.

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