A Rare Grotesque Skeletal Deformity: Munchmeyer’s Disease

Debashish Mishra, Aadhaar Dhoooria

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Debashish Mishra1, Aadhaar Dhoooria2

1Department of Internal Medicine, Rheumatology and Clinical Immunology, Post Graduate Institute of Medical Education and Research, Chandigarh, India, 2Consultant Rheumatologist, Department of Rheumatology, Santokba Durlabhji Memorial Hospital, Jaipur, India

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Fibrodysplasia ossificans progressiva (FOP), or Munchmeyer’s disease, is a rare deforming skeletal anomaly caused by intramuscular ossification. Here, we present the clinical images of a patient with this rare grotesque deforming disease.

A 20-year-old gentleman presented to our clinic with inability to open his mouth and difficulty in eating. Progressive deformities had been noticed starting at the age of two and a half months, following intramuscular tetanus immunisation. At present, patient was unable to abduct his arms or stand straight. Examination revealed a hard sheet-like structure binding his arms to the chest and kyphoscoliosis (Figure 1A) and shortened great toes (Figure 1B). Hip movements were limited. A hard submandibular swelling was noted, grossly limiting jaw movements. Radiographs revealed ossification of latisimius dorsi bilaterally (Figure 1C) and monophalangic great toes, confirming a diagnosis of FOP. Patient and family was counselled about the disease, aggravating factors, and avoidance of intramuscular injections.

FOP is a rare disorder (1 in 2,000,000), caused by an autosomal dominant mutation in ACVR1 gene on chromosome 2q 23-24, encoding bone morphogenetic protein (BMP) type1 receptor.1 There is no proven treatment yet, except for several trials with nitrogenous bisphosphonates,2 and few ongoing ones with drugs like palovarotene (MOVE), garetosmab, (LUMINA-1), rapamycin, and saracatinib (STOPFOP group). Avoidance of any sort of trauma is primary to prevent disease progression.

CONFLICT OF INTEREST
The authors declare no conflict of interest.

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