

RECESSIVE MULTIPLE EPIPHYSEAL DYSPLASIA: A CASE REPORT

Saeed Bin Ayaz^{✉1}, Saima Rauf¹, Fatima Ayaz²

ABSTRACT

Multiple epiphyseal dysplasia (MED) is characterized by abnormal bone and cartilage development particularly affecting epiphysis of long bones. Individuals with MED present during childhood presents with hip pain, knee pain and waddling gait. The autosomal recessive MED is differentiated from the dominant type by the presence of hands, knees and feet malformations with scoliosis. We present here, a case of autosomal recessive MED, an eleven and half years old boy who presented with pain in knees, difficulty walking, abnormal fingers and abnormal toes. The radiographs of hands, knees and pelvis were suggestive of an abnormal epiphyseal development. After diagnosis, patient was counseled and he was advised analgesics and different exercises.

KEY WORDS: Epiphyseal Dysplasia (MeSH); Knee Pain (MeSH); Waddling Gait (MeSH); Clinodactyly (Non-MeSH).

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1. Department of Rehabilitation Medicine, Combined Military Hospital Quetta and Quetta Institute of Medical Sciences, Quetta 87300, Balochistan, Pakistan.

Email ✉: saeedbinayaz@gmail.com

2. Registrar Internal Medicine, Holy Family Hospital, Rawalpindi, Pakistan

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Radiographs of the pelvis (Figure 2B) showed irregular iliac blades with cortical flaring, fused sacroiliac joints, marked irregularity of the acetabular margins with loss of concavity, sclerosis, irregularity of the femoral epiphysis with metaphysis bilaterally and irregularity of inferior pubic rami with pubic symphysis. The radiographs of the lumbosacral spine (Figure 3; A&B) revealed irregularity of end plates with central depression and no evidence of kyphoscoliosis. The knee radiographs (Figure 3; C&D) showed osteopenia and mild flaring of metaphysis of both tibias towards lateral half.

All essential laboratory investigations were carried out. The thyroid profile, erythrocyte sedimentation rate and renal function tests were within normal limits. The bone mineral density measured at the spine was 0.424g/cm² with a Z-score of -1.5 which is lower than the normal limits for his age and sex. The bone-age of the boy was 10 years which is a year and half less than his actual age. Thus, he was diagnosed to have rMED.

He was advised to take oral diclofenac sodium 50 mg twice daily, strengthening exercises of quadriceps with glutei and stretching exercises for the hamstrings. He was also counselled about the prognosis, potential complications and other treatment options for the future.

DISCUSSION

The term MED was introduced by Thomas Fairbank in 1947.³ The frequency of MED per million is 11.2 in index patients only while 16.3 if affected relatives included.⁴ Autosomal MED was described in 1999 by Superti-Furga which is mutations in DTDST

INTRODUCTION

Multiple epiphyseal dysplasia (MED) is characterized by abnormal bone and cartilage development particularly affecting the epiphysis of the long bones of arms and legs. MED is of two types (autosomal dominant and autosomal recessive) that differ in their inheritance pattern and some phenotypical characteristics.¹ Both subtypes are diagnosed clinically during childhood in bulk of cases and affected individuals present with hip pain, knee pain and waddling gait.¹ The recessive MED is differentiated from the dominant type by the presence of hands, knees and feet malformations with scoliosis.^{1,2} This is a case of a patient who was suffering from autosomal recessive MED (rMED).

CASE REPORT

An eleven and half years old boy presented with pain knees and difficulty in walking for about two and half years, his hands, feet and stature were shorter relative to his peers since birth. The pain

was rapid in onset and developed during a playing activity though there was no history of trauma. The boy had minimal relief with oral paracetamol.

On examination, he had a short stature when analyzed on growth charts. His fingers and toes were short and stubby (Figure 1).

The fingers showed an unusual curving (clinodactyly). The neurological examination was normal and he had a normal intelligence. He did not have obvious spinal deformity and his oral examination was normal. The radiographs of both hands revealed short stubby metacarpals with cupping of distal metaphysis at 2nd, 3rd, 4th and 5th metacarpals (Figure 2A).

The distal metaphysis of metacarpals showed sclerosis and flaring, while the proximal metaphysis had cupping, flaring and sclerosis in all the digits. The ulnar styloid process was not visible however; the medial ulnar metaphysis showed flaring with an adjacent radiolucent area. The radius and the carpal bones appeared normal.

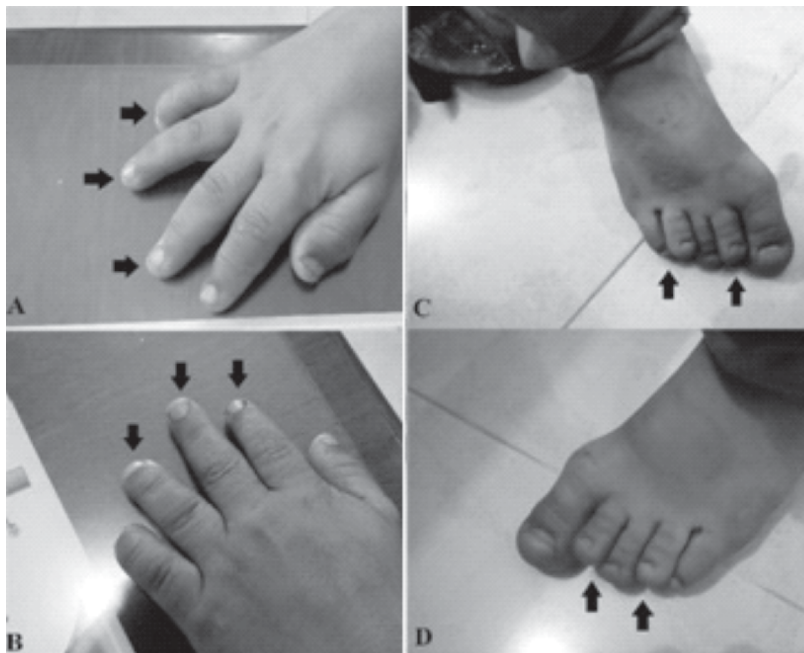


Figure 1: Short and stubby fingers (A and B) and toes (C and D)



Figure 2: (A) Short stubby metacarpals with cupping of distal metaphysis of 2nd, 3rd, 4th, and 5th metacarpals. The distal metaphysis of metacarpals showed sclerosis and flaring, while the proximal metaphysis had cupping, flaring, and sclerosis in all digits. The ulnar styloid process is not visible; however, the medial ulnar metaphysis shows flaring with an adjacent radiolucent area. (B) Irregular iliac blades with cortical flaring, fused sacroiliac joints, marked irregularity of the acetabular margins with loss of concavity and sclerosis, irregularity of femoral epiphysis, metaphysis, and apophysis bilaterally, and irregularity of inferior pubic rami and pubic symphysis.

(SLC26A2) gene and are attributed to the development of rMED.⁵ The disease generally manifests itself in late childhood with pain in hip and /or knee joints, difficulty walking, waddling gait, easy fatigability, contractures at the elbow joints and angular deformities in the lower extremity e.g. coxa vara, varus or valgus deformities of the knees and valgus deformity at the distal tibia.^{1,2} Chest wall deformities (funnel-shaped or pigeon-shaped chest) may also occur.² In about 50% children, there is at least one abnormality noticeable at birth i.e. cleft palate, clubfoot, clinodactyly or cystic ear swelling.^{1,2}

The differential diagnosis of rMED includes Legg-Calvé-Perthes disease, pseudoachondroplasia, spondyloepiphyseal dysplasia, congenital hypothyroidism, mucopolysaccharidoses, and other disorders that are genetically related.¹ Pseudoachondroplasia despite the radio-clinical similarities with rMED is more severe and presents earlier.⁶ Spondyloepiphyseal dysplasia demonstrates significant scoliosis that is not a prominent feature of rMED. Mucopolysaccharidoses and congenital hypothyroidism can be segregated using urinalysis and thyroid function tests.¹

The goals of treatment in rMED are pain relief and halting the joint destruction.¹ The conservative management includes weight reduction, physical modalities, specific muscle strengthening, stretching exercises and analgesic medications.^{1,2} Occupational or recreational activities including sports causes joint overload should be avoided.² Orthopedic surgery is sometimes used to relieve pain, adjust deformities and improve joint contractures.¹ Treatment options include realignment procedures and arthroplasty to deal with advanced osteoarthritis. The affected individuals also need psychological, social and vocational support to tackle with problems of short stature, disability and unemployment.

Though rare, the patient of rMED must be thoroughly investigated at the time of very first presentation to reduce future morbidity associated with the disease.

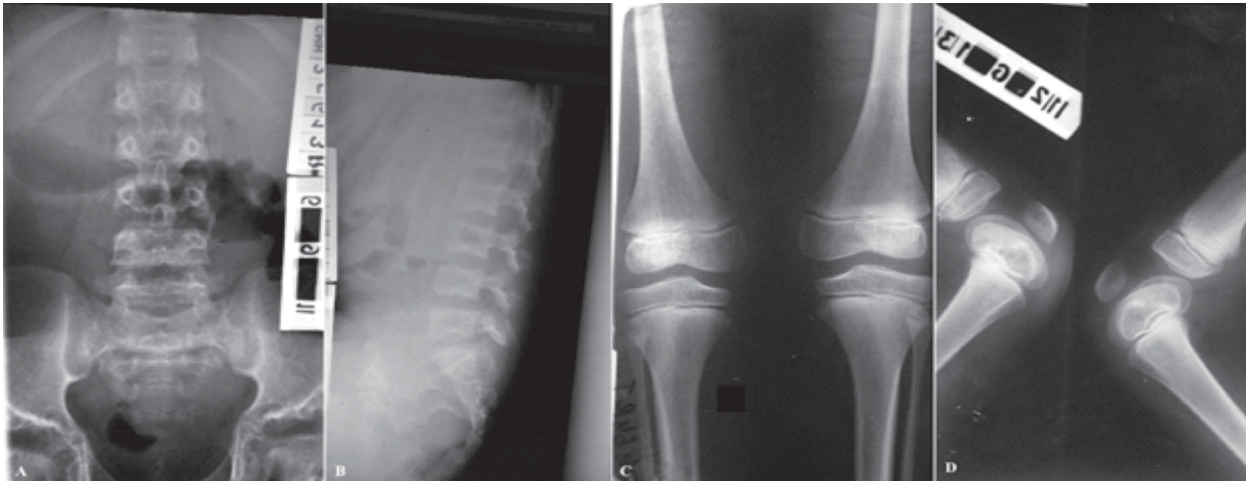


Figure 3: (A and B): Irregularity of vertebral end plates with central depression but no evidence of kyphoscoliosis; (C and D): Osteopenia and mild flaring of metaphysis of both tibia towards lateral half.

REFERENCES

1. Anthony S, Munk R, Skakun W, Masini M. Multiple epiphyseal dysplasia. J Am Acad Orthop Surg 2015;23(3):164-72. DOI: 10.5435/JAAOS-D-13-00173.
2. Santra G, Banerjee S, Sarkar RN. Multiple epiphyseal dysplasia. J Assoc Physicians India 2013;61(6):408-9.
3. Fairbank T. Dysplasia epiphysialis multiplex. Br J Surg 1947 Jan;34(135): 225-32.
4. Wynne-Davies R, Gormley J. The prevalence of skeletal dysplasias. An estimate of their minimum frequency and the number of patients requiring orthopaedic care. Bone Joint J 1985;67(1):133-7. DOI: 10.1302/0301-620X.67B1.3155744.
5. Superti-Furga A, Neumann L, Riebel T, Eich G, Steinmann B, Spranger J, et al. Recessively inherited multiple epiphyseal dysplasia with normal stature, club foot, and double layered patella caused by a DTDST mutation. J Med Genet 1999;36(8):621-4. DOI: 10.1136/jmg.36.8.621.
6. Kumar KJ, Nayana, Manjunath VG, Shetty C. Pseudoachondroplasia: A Rare Cause of Short Limbed Dwarfism. J Compr Ped 2017; 8(1):e39867. DOI: 10.5812/compreped.39867.

AUTHORS' CONTRIBUTIONS

Following authors have made substantial contributions to the manuscript as under:

SBA & SR: Identification & diagnoses of the case, management of the case, manuscript witting, final approval of the version to be published

FA: manuscript witting, final approval of the version to be published

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

CONFLICT OF INTEREST

Authors declared no conflict of interest

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KMUJ web address: www.kmu.edu.pk

Email address: kmu@kmu.edu.pk