

## Case Report

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## A neonate with metatropic dysplasia: A case report

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## Abstract

Metatropic dysplasia is a rare form of skeletal dysplasia in which characteristic clinical and imaging features are found at birth. The short limbs with relatively long trunk seen in the neonatal period change to short trunk in childhood due to progressive kyphoscoliosis. This rare condition should be diagnosed in the neonatal period to avoid diagnostic dilemma in later life. A 2-day-old baby girl was referred for a skeletal survey study to diagnose the underlying skeletal dysplasia. The metatropic dysplasia was diagnosed based on plain radiographic findings. This case report highlights the characteristic radiographic findings of this rare disease entity.

**Keywords:** *Skeletal dysplasia, Metatropic dysplasia, Neonate, dumbbell shape long bones, Case report*

## INTRODUCTION

Metatropic dysplasia (MD) is a rare form of spondyloepimetaphyseal dysplasia [1] and it is an autosomal dominant disorder [2]. There are unique clinical and imaging characteristics, seen at birth in MD [3]. The plain radiographic features of dumbbell-shaped long tubular bones and severe platyspondyly are said to be specific features of MD [4]. In addition to the skeletal anomalies, concomitant association of MD with peripheral neuropathies have been reported [4,5]. Although affected newborns have short limbs with long trunk at birth, they develop progressive kyphoscoliosis leading to short trunk that causes

apparent equalization of trunk and limbs [1,3,6]; hence, the term metatropic is used [1,7]. The term metatropic, which means changing dysplasia, is used to indicate the change in the appearance from neonatal period to childhood. Thus, it is important to identify and diagnose MD at birth, since the imaging features will change in later life causing diagnostic dilemma. Since the presence of short limb rhizomelia seen at birth and associated metaphyseal dysplasia, MD can be misdiagnosed for more common achondroplasia. Similarly, due to severe platyspondyly, MD can be mistaken for spondyloepimetaphyseal dysplasia congenita.



## CASE PRESENTATION

A 2-day-old baby girl with an uncomplicated antenatal history delivered by an uncomplicated normal vaginal delivery was presented to the radiology department for a skeletal survey study. No skeletal dysplasia was diagnosed on her parents and there was no family history of skeletal anomalies. She was having short upper limbs and lower limbs compared to elongated trunk with scoliosis of the thoracic spine. The skull and facies were normal. Her full blood count did not reveal any abnormality. Due to disproportionate short limbs with elongated trunk, a skeletal anomaly was suspected, and a skeletal survey was planned.

## SKELETAL SURVEY

Her plain radiographs of the spine showed flattened dense vertebral bodies with well-developed posterior elements. There was severe platyspondyly. In the limb radiographs the tubular bone diaphysis was short with broad metaphyses resembling dumbbells. The X-ray of the thorax showed narrow and elongated chest with short ribs. There was scoliosis of the thoracic spine. Skull radiographs were unremarkable.

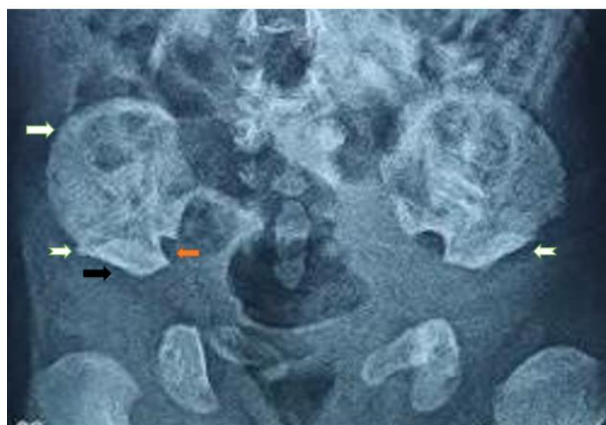


Figure 1: Plain X-ray Pelvis: Hypoplastic ilia (solid white arrow) with narrow sacrosciatic notches (orange arrow), horizontal acetabular roofs (black arrow) with small associated notches superior to their lateral borders (notched white arrow)

The plain radiograph of the pelvis (Figure 1) showed hypoplastic ilia with narrow sacrosciatic notches, horizontal acetabular roofs with small associated notches superior to their lateral borders and prominent flaring of crescentic iliac

wing giving rise to the characteristic halberd appearance.

The thin dense vertebrae with severe platyspondyly were seen in the lateral thoraco-lumbar vertebrae (Figure 2).

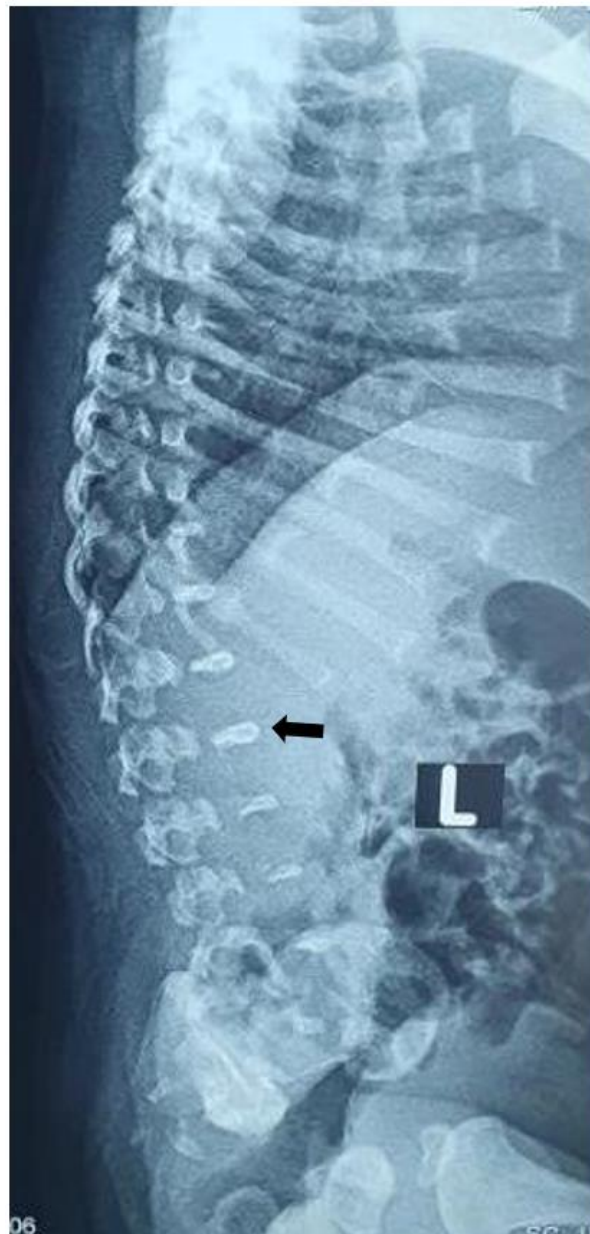


Figure 2: Plain X-ray lateral thoracolumbar spine showing thin dense vertebrae with severe platyspondyly (black arrow)

The lower limb radiograph (Figure 3) showed short diaphyses with widened metaphyses giving rise to dumbbell shape.

The clinical and radiological features lead to the diagnosis of MD and the child was referred to the specialised children hospital for follow-up care.

## DISCUSSION

MD is a rare form of spondyloepimetaphyseal dysplasia [1]. The disease spectrum comprises of a nonlethal type with autosomal recessive transmission, a nonlethal dominant type, and a lethal type with death before or shortly after birth and possibly autosomal recessive inheritance [1]. Recent genetic studies have revealed that mutations in the transient receptor potential vanilloid 4 (TRPV4) that encodes calcium-permeable, non-selective cation channels as the cause for MD [2,8,9]. Further, mutations in TRPV4 is known to be associated with peripheral neuropathies and muscular dystrophies [4,5]. Therefore, coexistence of MD and neuropathy has been observed in some patients [4,5]. The cartilage overgrowth in the perichondral ring and dysfunctional endochondral ossification are thought to be the causes for the skeletal abnormalities seen in MD [2].

The characteristic radiological features of MD seen at birth are described in the literature. These include short diaphysis with metaphyseal flaring leading to dumbbell shaped long bones, hypoplastic ilia with narrow sacrosciatic notches, horizontal acetabular roofs with small associated notches superior to their lateral borders leading to characteristic halberd pelvis, severe platyspondyly with thin dense vertebra and kyphoscoliosis [3,5,6,9]. All these features were present in our patient. The genetic studies were not done in our patient and the diagnosis was based on the characteristic clinical and imaging features. In addition to these features, a coccygeal prominent tail has been stated in the literature, which occurs due to excessive ossification of the coccyx [7].

It is stated that the kyphoscoliosis associated with MD is often resistant to surgical treatment [3]. However, spinal bracing is used as the standard method of treatment for kyphoscoliosis [1,3,9]. In addition, cervical instability due to odontoid hypoplasia, upper respiratory obstruction following laryngo-tracheal dysfunction and severe restrictive lung disease due to thoracic anomalies

are well document complications associated with MD [3]. Thus, it is important to educate the parents and emphasize the importance of attending long term follow-up care.

Since certain clinical and radiological features are shared by MD with other skeletal dysplasia, early identification of the unique radiological features seen in MD is extremely important. Furthermore, in resource limited clinical settings, where the access to specific genetic studies is limited, the imaging findings play a key role in the diagnosis of rare skeletal dysplasia like MD.

## Abbreviations

Metatropic dysplasia - MD

Transient receptor potential vanilloid 4 - TRPV4

### Author declaration

#### Acknowledgement

None

#### Author contribution

MCW, EH and NH were involved in the interpretation of the imaging findings. MCW and NDW prepared the original draft of the manuscript. MCW, NDW, NH and EH read and approved the final version of the manuscript.

#### Conflict of interest

The authors declare that they have no competing interests.

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#### Ethics approval and consent to participate:

Informed verbal consent was obtained from the parents of the child.

**Consent for publication:** Informed verbal consent was obtained from the parents of the child. The case report only contains X-rays and the case report, and the images included do not contain any personally identifiable information.

## REFERENCES

1. Song HR, Sinha S, Song SH, Suh SW. A case of metatropic dysplasia: operative treatment of severe kyphoscoliosis and limb deformities. *Oman Medical Journal*. 2013 Nov;28(6):445. doi: 10.5001/omj.2013.123
2. Hamabe F, Edo H, Yamashita T, Matsumoto H, Tamada S, Sumi K, Shinmoto H. Mild metatropic dysplasia: emphasis on the magnetic resonance imaging of articular cartilage thickening. *BJR| case reports*. 2021 Mar;7(2):20200155. doi: 10.1259/bjrcr.20200155
3. Kannu P, Aftimos S, Mayne V, Donnan L, Savarirayan R. Metatropic dysplasia: clinical and radiographic findings in 11 patients demonstrating long-term natural history. *American Journal of Medical Genetics Part A*. 2007 Nov 1;143(21):2512-22. doi.org/10.1002/ajmg.a.31941
4. Ürel-Demir G, Şimşek-Kiper PÖ, Öncel İ, Utine GE, Haliloğlu G, Boduroğlu K. Natural history of TRPV4-Related disorders: From skeletal dysplasia to neuromuscular phenotype. *European Journal of Paediatric Neurology*. 2021 May 1; 32:46-55.
5. Gucev Z, Kalcev G, Laban N, Bozinovski Z, Popovski N, Saveski A, Daskalov B, Plaseska-Karanfilska D, Tasic V. Characteristic diagnostic clues of metatropic dysplasia: The lumbothoracic humpback with dumbbell appearance of the long bones. *Balkan Journal of Medical Genetics*. 2018 Dec 1;21(2):35-8.
6. Graversen L, Haagerup A, Andersen BN, Petersen KK, Gjørup V, Gudmundsdottir G, Vogel I, Gregersen PA. Novel TRPV4 variant causes a severe form of metatropic dysplasia. *Clinical Case Reports*. 2018 Sep;6(9):1774.
7. Narayanan DL, Bhavani GS, Girisha KM, Phadke SR. Metatropic dysplasia with a novel mutation in TRPV4. *Indian pediatrics*. 2016 Aug;53(8):735-7. doi: 10.1007/s13312-016-0921-1
8. Panda A, Gamanagatti S, Jana M, Gupta AK. Skeletal dysplasias: a radiographic approach and review of common non-lethal skeletal dysplasias. *World journal of radiology*. 2014 Oct 10;6(10):808. doi: 10.4329/wjr.v6.i10.808
9. Tchou Tchoumba MA, Bai Y, Jin R, Yu X, Male M. Metatropic Dysplasia of Nonlethal Variant in a Chinese Child—A Case Report. *Orthopaedic Surgery*. 2020 Feb;12(1):333-6.