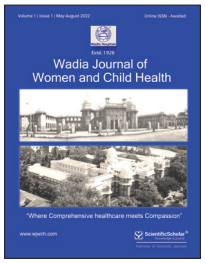
www.wjwch.com

Wadia Journal of Women and Child Health



Clinical Image

Arthrogryposis multiplex congenita with multiple pituitary hormone deficiency

Atharv Jayant Tungatkar[1], Kavya DR[2], Snehith Reddy Kasu[1], Rajesh Joshi[2], Sudha Rao[2]

Departments of [1]Pediatrics and [2]Pediatric Endocrinology, Bai Jerbai Wadia Hospital for Children, Mumbai, Maharashtra, India.

***Corresponding author:**
Atharv Jayant Tungatkar,
Department of Pediatrics,
Bai Jerbai Wadia Hospital
for Children, Mumbai,
Maharashtra, India.

ajtungatkar@gmail.com

Received: 02 April 2023
Accepted: 04 November 2023
Published: 01 February 2024

DOI
10.25259/WJWCH_17_2023

Arthrogryposis multiplex congenita with multiple pituitary hormone deficiency (MPHD) can present unique diagnostic challenges. A 4-year and 5-month-old girl, born of non-consanguineous marriage, first of twin, presented with failure to thrive, bilateral club foot, joint contractures since birth and gross motor delay. The second twin died *in utero* in the second trimester. An antenatal

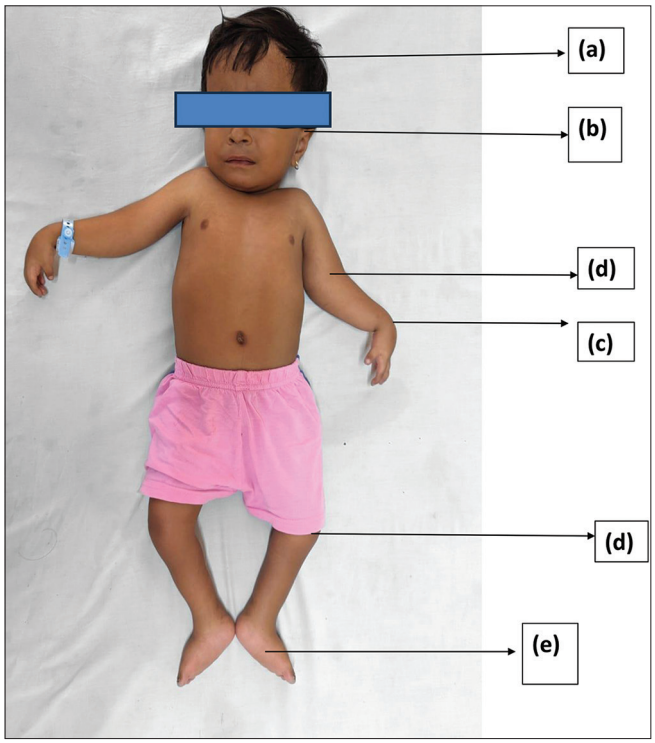


Figure 1: Physical features: (a) Recession of anterior hairline, (b) Low set ears, (c) Flexion deformity of wrist, (d) Large joint contractures of elbow and knee, (e) Flat foot.

How to cite this article: Tungatkar AJ, Kavya DR, Kasu S, Joshi R, Rao S. Arthrogryposis multiplex congenita with multiple pituitary hormone deficiency. Wadia J Women Child Health. 2023;2(3):144-5. doi: 10.25259/WJWCH_17_2023

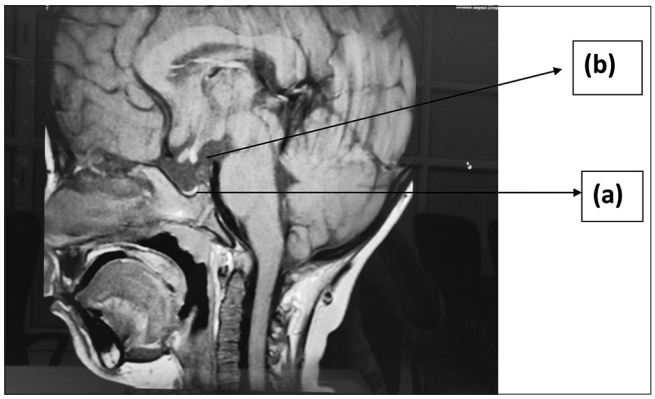


Figure 2: Magnetic resonance imaging pituitary showing: (a) Small anterior pituitary, (b) Ectopic posterior pituitary bright spot.

scan revealed bicornuate uterus, oligohydramnios, fetal limb abnormalities, and breech presentation. The mother had perceived decreased fetal movements.

On examination, she was short (length - 63.5 cm) and weighed 6.5 kg with a weight for height of −1 standard deviation, suggestive of short stature. She has multiple contractures and short stature [Figure 1] suggestive of arthrogryposis multiplex congenita with short stature.

The evaluation was suggestive of hypothyroidism (low free thyroxine: 0.77 ng/dL [normal range: 1.0–1.7 ng/dL]; mildly elevated thyroid-stimulating hormone: 6.16 uIU/mL [normal range: 0.4–4.8 uIU/mL]) and growth hormone (GH) deficiency (peak GH of 1.94 ng/mL on clonidine stimulation test). Magnetic resonance imaging pituitary was suggestive of pituitary stalk interruption syndrome [Figure 2].[1]

Very few cases of arthrogryposis multiplex congenita with MPHD have been reported, some of which have a genetic basis.[2] However, we did not find any pathogenic variant on mutation analysis. At present, the child is on thyroxine and GH replacement, orthopedic intervention, and physical rehabilitation measures.

Ethical approval

The research/study complied with the Helsinki Declaration of 1964.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

Dr. Sudha Rao is on the Editorial Board of the Journal.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

REFERENCES

1. Narayanan VK, Kirk J, Hogler W. Multiple pituitary hormone deficiencies in two patients with arthrogryposis multiplex congenita. Endocr Abstr 2013;33:68.
2. Gregory LC, Shah P, Sanner JR, Arancibia M, Hurst J, Jones WD, *et al*. Mutations in MAGEL2 and L1CAM are associated with congenital hypopituitarism and arthrogryposis. J Clin Endocrinol Metab 2019;104:5737-50.