EJPMR, 2026, 13(3), 203-206 **Case Study** **Impact Factor: 8.158**

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

ISSN (O): 2394-3211
ISSN (P): 3051-2573
Coden USA: EJPMAG

A CASE OF MUCOPOLYSACCHARIDOSIS PRESENTING AS SHORT STATURE

[1]*Dr. Vinayak Moorthy, [2]Dr. Rathnakumar MD, [3]Dr. Marchwin Kingston MD, [4]Dr. Arjunan MD

[1]Post Graduate Student, Tirunelveli Medical College Hospital.
[2]Professor and HOD of Medicine, Tirunelveli Medical College Hospital.
[3]Professor of Medicine, Tirunelveli Medical College Hospital.
[4]Assistant Professor of Medicine, Tirunelveli Medical College Hospital.

***Corresponding Author: Dr. Vinayak Moorthy**

Post Graduate Student, Tirunelveli Medical College Hospital.

DOI: https://doi.org/10.5281/zenodo.18796528

How to cite this Article: [1]*Dr. Vinayak Moorthy, [2]Dr. Rathnakumar MD, [3]Dr. Marchwin Kingston MD, [4]Dr. Arjunan MD. (2026). A Case of Mucopolysaccharidosis Presenting As Short Stature. European Journal of Biomedical and Pharmaceutical Sciences, 13(3), 203–206.

Article Received on 28/01/2026 **Article Revised on 18/02/2026** **Article Published on 01/03/2026**

ABSTRACT

The diagnostic process of short stature requires a disciplined, stepwise approach that moves beyond simple height velocity. It involves the scrutiny of body proportions, skeletal maturation, and subtle dysmorphic features. In many cases it is merely the most visible marker of a systemic disorder affecting connective tissue, neurological function, and visceral organ integrity.[1] This report focuses on the diagnostic journey of a 14-year-old male presenting with short stature and a constellation of multisystemic symptoms. By integrating clinical history, examination findings and imaging evidence we present a compelling of Mucopolysaccharidosis.

INTRODUCTION

Short stature in the pediatric population, defined as a height more than two standard deviations (SD) below the mean for age and sex, is a frequent reason for referral to pediatric endocrinology and genetics services. While the majority of these referrals represent normal variants such as familial short stature (FSS) or constitutional delay of growth and puberty (CDGP), a significant subset harbors underlying pathological conditions ranging from endocrine deficiencies to complex skeletal dysplasias and inborn errors of metabolism. The diagnostic challenge lies in differentiating the immense heterogeneity of these etiologies and the subtle, often overlapping phenotypic presentations that characterize the early stages of disease.[1]

The Mucopolysaccharidoses represent a group of rare, inherited metabolic diseases caused by the deficiency of specific lysosomal enzymes required to degrade glycosaminoglycans (GAGs), formerly known as mucopolysaccharides. The accumulation of undegraded glycosaminoglycans within the lysosomes results in progressive cellular damage and a distinct skeletal phenotype characterized by short stature, joint abnormalities, systemic involvement and coarse facial features.[1]

A critical component of this analysis is the differentiation of MPS from other skeletal dysplasias.[3],[4] The overlap in radiological findings, necessitates a nuanced understanding of specific radiographic signs. Markers such as the "bullet-shaped" metacarpal and the "oar-shaped" rib serve as vital discriminatory tools in the radiologist's arsenal. Furthermore, the life-threatening implications of craniovertebral junction (CVJ) instability in these patients underscore the necessity for early and precise identification to prevent catastrophic neurological sequelae.

CASE DETAILS

History

A 14 year old male presented with chief complaints of insidious, progressive breathlessness for past 2 weeks and a 2-day history of left-sided chest pain aggravated by inspiration and coughing. The past medical history revealed that the patient underwent surgery for inguinoscrotal swelling (hernia) at 4 months and again at 10 months of age. He was evaluated at age 10 for unilateral hearing loss and was advised hearing aids. Developmental history showed significant gross motor delays in form of head holding at 6 months, sitting with support at 1 year, walking at 1.5 years, and climbing stairs (one feet per step) by age 2. Speech was also

delayed (1-2 words by age 2 and half years). Antenatal/Perinatal history revelaed that patient was born via normal delivery (3 kg) from second degree consanguinity.

Examination

The general examination of the patient showed the following features - coarse facies, frontal bossing, flat nasal bridge, low set ears and a low posterior hairline, protruded tongue and macroglossia, short stature, short neck, umbilical hernia and an equinus deformity.

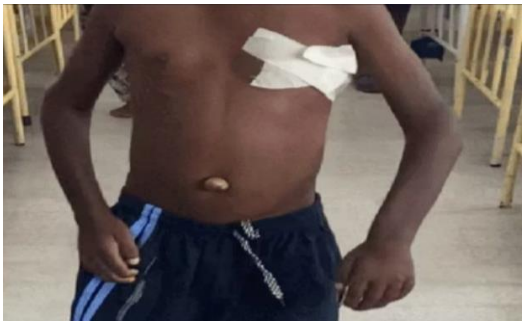

Figure 1: Cubitus valgus, umbilical hernia.

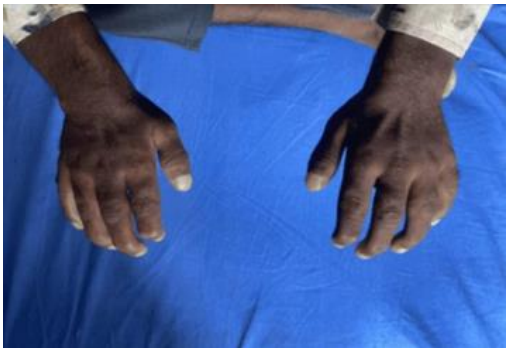

Figure 2: Hands of The Patient.

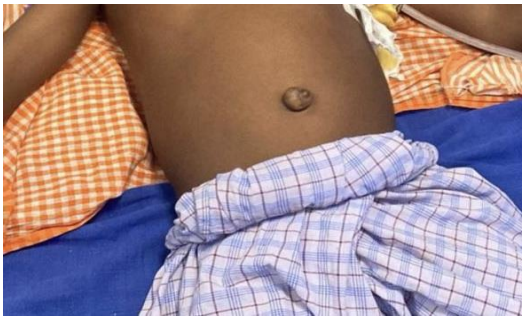

Figure 3: Umbilical Hernia.

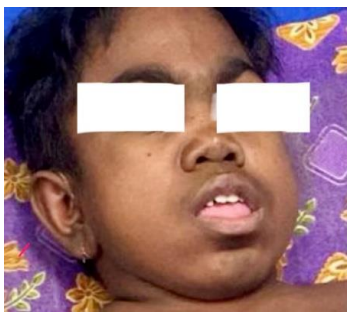

Figure 4: Facial dysmorphism.

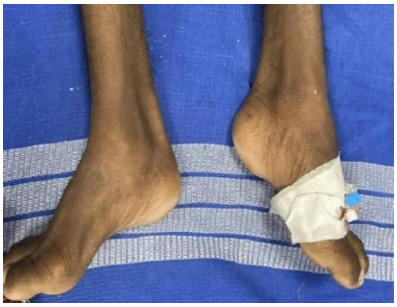

Figure 5: Equinus deformity.

INVESTIGATIONS

The complete blood counts, differential counts, renal and liver function tests, coagulation profile were in the normal range and inflammatory markers were elevated. Other basic investigations like LDH, uric acid, CKMB was normal. Blood cultures and urine culture was negative. Thyroid function tests were within normal limits. Skeletal analysis showed features suggestive of dysostosis multiplex.

X ray imaging showed the following features

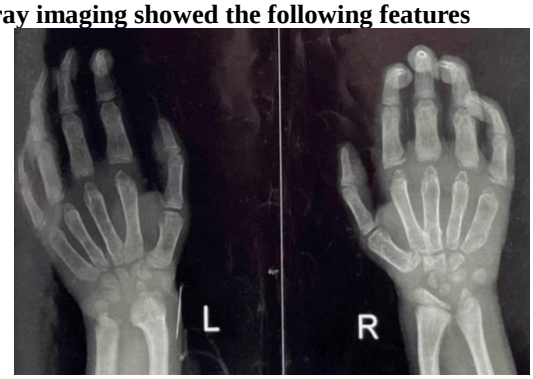

Figure 6: Metacarpalchanges.

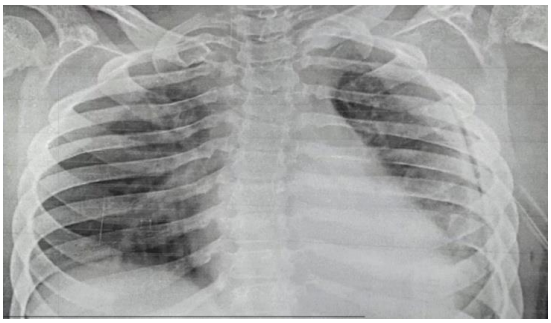

Figure 7: Changes in ribs.

The ultrasonogram of the abdomen showed hepatosplenomegaly and umbilical hernia.

Brain MRI revealed features of basilar invagination, platybasia, J shaped sella with a NORMAL PITUITARY. Other findings included increased AP diameter, widened CSF spaces anterior to the temporal lobes, pachygyria, a thinned out corpus callosum, ventriculomegaly and diffuse cortical atrophic changes with prominent sulcal spaces. These features were suggestive of mucopolysaccharidoses with communicating hydrocephalus.

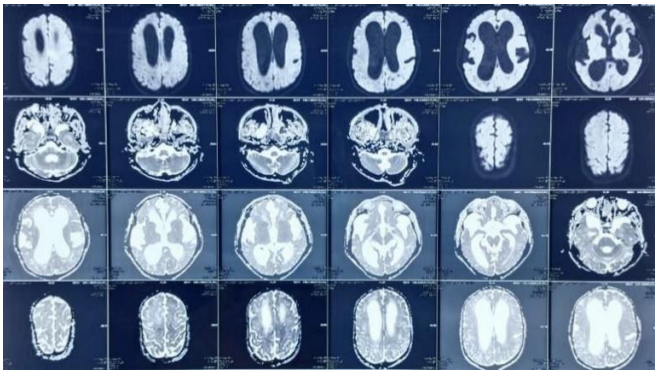

Figure 8: MRI - ventriculomegaly.

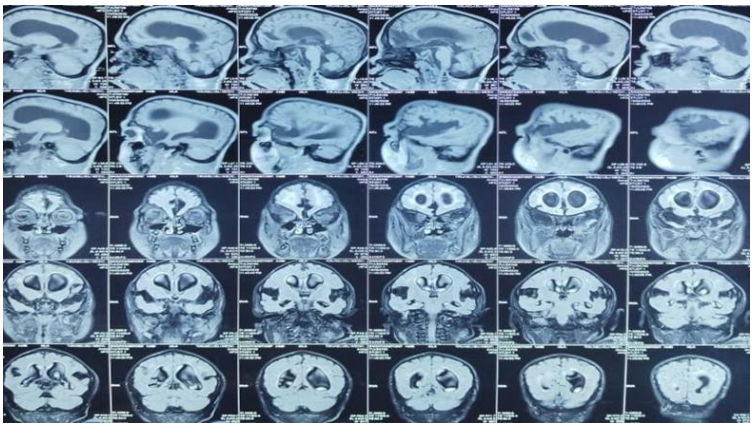

Figure 9: Normal Pituitary.

DISCUSSION

The evaluation of this case follows a structured diagnostic paradigm designed to categorize short stature and isolate the underlying pathology. The initial step involves establishing the presence of short stature. Clinically, this is defined as a height less than the 3rd percentile or 2 standard deviation scores (SDS) below the mean for the patient's age and sex.[1] However, a static height measurement is insufficient for diagnosis. The evaluation must assess *growth velocity*, as a velocity below the 25th percentile is a potent indicator of active pathology rather than a variant of normal growth like FSS or CDGP. In this case the history is notable for a growth delay noticed by the mother within the first year of life as she said that she had noticed a difference in the rate of growth compared to children of her relatives (though not scientifically quantified or evaluated at the initial stages). This early onset of growth failure, persisting into adolescence (current age 14), signals a congenital or early-onset genetic etiology rather than an acquired endocrine deficit like late-onset hypothyroidism.

ASSESSMENT OF BODY PROPORTIONS- Once short stature is confirmed, the second step involves the critical differentiation of body proportions. The Upper Segment to Lower Segment (US:LS) ratio is the primary metric used to classify the defect. The patient's height was 140 cm and the US:LS ratio was 0.97. Proportionate Short Stature is suggested by a US:LS ratio appropriate for age (e.g., ~1.0 at age 10, dropping to ~0.9 post-puberty). Common causes include constitutional delay, familial short stature, malnutrition, systemic chronic illness, or Growth Hormone (GH) deficiency. Disproportionate Short Stature is indicated by an abnormal ratio, which subdivides the differential diagnosis into two distinct categories: Short Limbs (Increased US:LS Ratio): Conditions such as Achondroplasia and Hypochondroplasia typically present with rhizomelic (proximal) shortening, leading to a relatively long trunk compared to the limbs. Short Trunk (Decreased or Paradoxically Normal US:LS Ratio): Conditions such as Spondyloepiphyseal Dysplasia (SED) and Mucopolysaccharidoses (MPS) primarily affect the spine (platyspondyly), leading to a shortened trunk height. In MPS, the trunk is often shortened due to flattened vertebrae, but the limbs may also show shortening as there is skeletal dysplasias affecting the long bones and the knees, thereby creating a picture where there is reduction of both upper and lower segment dimensions and when it occurs more or less proportionately it can produce an **apparently normal US:LS ratio.** In such cases the arm span is evaluated. In this case the ratio (arm span to height) was less than 0.89. In the setting of a suspicion of an apparently normal US:LS ratio this is an important differentiator as normally in this age the arm span is almost equal to or more than the height and the abnormality here helps to narrow down the etiology significantly as in endocrine causes we do not expect such a deviation of the arm span to height. The third step involves Bone Age assessment to determine growth potential and refine the differential.

Bone Age - Typically assessed using the left wrist in younger children. For older adolescents, assessment may require evaluation of the hip and elbow ossification centers. In MPS, bone age is often discordant; that is there will be a lack of synchrony in the appearance and maturation of the ossification centres. A delayed bone age guides the etiology towards – CDGP, GH deficiency, hypothyroidism, chronic systemic illness and malnutrition. Whereas an advanced bone age occurs in conditions such as precocious puberty, hyperthyroidism etc. Normal bone age can occur in cases of familial short stature and certain genetic syndromes. Normally the femur lower end and the upper ends of tibia and fibula (and also the phalanges) – appears at the age of 14 and fuses at the age of 17, distal end of radius and ulna appear at 14 and fuse by 18 with the carpal bones appearing by 9 and fusing by 12. In this case the expected findings include asynchronous maturation, abnormal morphology and malformed epiphysis due to the accumulation of the glycosaminoglycans. In our case the bone age assessment did not provide diagnostic information.

The next step involved the phenotypic assessment - In this case the findings included – coarse facies, frontal bossing, flat nasal bridge, low set ears and a low posterior hairline, protruded tongue and macroglossia, short neck, umbilical hernia and an equinus deformity. Although this phenotype is not specific to the diagnosis this combination of skeletal with radiological determinants did give a strong direction to the diagnostic algorithm as the skeletal assessment – showed skeletal dysplasia (dysostosis multiplex) and basilar invagination.

The diagnosis of Mucopolysaccharidosis is established not by a single finding, but by the irrefutable convergence of multiorgan pathology. The presence of

CLUES FROM HISTORY

1. **Inguinal hernias** in infancy requiring repeated surgery is a classic sign of the connective tissue integrity failure seen in MPS.
2. Combined with **sensorineural/mixed hearing loss** in childhood, the clinical backdrop is highly suggestive of a storage disorder.

CLINICAL EXAMINATION CLUES

3. Facial dysmorphism
4. Developmental delays and organomegaly
5. Clues derived phenotypic assessment of body proportions and skeletal analysis - The identification of Dysostosis Multiplex, specifically the bullet-shaped metacarpals.[5]

CLUES FROM INVESTIGATIONS

6. The CT findings of CV junction anomaly.[7]
7. The MRI findings that confirm the CV junction anomalies and also gave insight to the presence of communicating hydrocephalus and other CNS structural changes like - **tip of the dens was lying 3.9mm above the chamberlin line and 9mm above the McGregor line indicating basilar invagination** with a NORMAL pituitary – suggestive of mucopolysaccharidosis.

CONCLUSION

This case is presented for highlighting the significance of evaluation of short stature and offering possible therapies for the reversible causes like (hypothyroidism, growth hormone deficiencies) and also identifying risk factors (like a CV junction anomaly in this case, The MRI findings of basilar invagination (dens >3.9mm above Chamberlain's line) highlighted the life-threatening CVJ instability, a critical "silent" killer in these patients.[6],[7] and providing adequate care to preventing complications of the same and thereby improving morbidity and mortality as far as possible. It also signifies the fact that a stepwise clinical detailed evaluation can provide a constellation of clues which when put together systematically can aid in the identification etiology. The work up of this case also throws light into the diagnostic approach to short stature and how the basic clues can aid in further decision making and therapeutics.

The patient was further planned for genetic studies with the clinical diagnosis of hunters syndrome keeping in mind

1. The X linked inheritance pattern,
2. Sparing of the cornea
3. Significant past history of the patient.

But the patient lost to follow up after discharge from the hospital.

REFERENCES

1. Kliegman RM, St. Geme JW, editors. Nelson Textbook of Pediatrics. 21st ed. Philadelphia: Elsevier, 2020.
2. Spranger JW. Mucopolysaccharidoses. In: Kliegman RM, St. Geme JW, Blum NJ, Shah SS, Tasker RC, Wilson KM, editors. Nelson Textbook of Pediatrics. 21st ed. Philadelphia: Elsevier, 2020; 107.
3. Lachman RS. Taybi and Lachman's Radiology of Syndromes, Metabolic Disorders and Skeletal Dysplasias. 5th ed. Philadelphia: Mosby, 2007.
4. Greulich WW, Pyle SI. Radiographic Atlas of Skeletal Development of the Hand and Wrist. 2nd ed. Stanford: Stanford University Press, 1959.
5. Husler J. Dysostosis multiplex mit Schwachsinn: Typus Hurler. Z Kinderheilkd, 1924; 3: 651.
6. Ghanem IB, Elhage S, Rachkidi R, Kharrat K. Pediatric cervical spine instability. J Child Orthop, 2008; 2(2): 71-84.
7. Talukdar R, Yalawar RS, Kumar M. Imaging in craniovertebral junction (CVJ) abnormalities. IOSR J Dent Med Sci., Dec. 2015; 14(12): 33-49.