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Morquio Syndrome Type A in Adulthood: A Rare Case Report with Multisystem Involvement

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Abstract

Introduction: Morquio syndrome type A (MPS IV) is an infrequent disorder of recessive autosomal pattern lysosomal storage disorder due to an inadequacy of N-acetylgalactosamine-6-sulfatase. It typically presents in early childhood with progressive skeletal dysplasia but may remain undiagnosed until adulthood in milder forms. Adult presentations are rare and often complicated by multisystem involvement.

Keywords: Morquio syndrome, MPS IV A, lysosomal storage disorder, adult presentation, skeletal dysplasia, enzyme deficiency, pneumonia, urinary tract infection.

Introduction

Mucopolysaccharidosis type IV (MPS IV), is an infrequent recessive autosomal lysosomal storage disorder characterised by defective degradation of glycosaminoglycans (GAGs), primarily keratan sulphate and chondroitin-6-sulphate [1,2]. MPS IV is subclassified into type A and type B, with type A resulting due to a deficiency of the enzyme N-acetylgalactosamine-6-sulfatase (GALNS). This enzymatic defect results in progressive buildup of GAGs in various tissues, resulting in multisystem involvement, notably skeletal dysplasia, respiratory complications, cardiac anomalies, and ocular changes.

Unlike other mucopolysaccharidoses, intelligence in MPS IV A is typically preserved[1,3]. Clinical manifestations usually begin in early childhood, with patients presenting with short stature, kyphoscoliosis, pectus carinatum, joint hypermobility, and gait abnormalities. However, adult presentation is extremely rare and often under recognized due to phenotypic

variability and overlap with other skeletal dysplasias. Additionally, late diagnosis may lead to delayed management of serious complications such as respiratory failure, spinal instability, and cardiovascular compromise [3,4].

This case is unique because it represents the survival and presentation of an undiagnosed 28-year-old female with Morquio syndrome type A, who displayed acutely with bilateral pneumonia, urinary tract infection, and characteristic skeletal features.

Case presentation

A 28-year-old female, born of non-consanguineous marriage, presented to the emergency department with complaints of low-grade intermittent fever and productive cough for the past 3 to 4 days, followed by progressive breathlessness over 2 days. The breathlessness was insidious in onset and gradually worsened, prompting hospital evaluation. According to her relatives, she had an abnormal posture and a progressively curved spine noted since early childhood.

Her birth and perinatal history were uneventful, and early developmental milestones were initially normal but later delayed. At around 4 years of age, she began to show signs of progressive kyphoscoliosis, waddling gait, and frequent falls. Despite multiple healthcare visits during childhood, no definitive diagnosis was established. There was no history of seizures or cognitive delay. Family history was non-contributory, with no known genetic disorders or similar skeletal abnormalities in siblings or extended family.

On examination, the patient was conscious, orientated, and cooperative. She was notably short-statured with a short neck and disproportionate limb length. Respiratory examination revealed RR- 32/min and Spo2- 84% on room air. Ocular examination showed corneal clouding. Other significant findings included pectus carinatum, kyphoscoliosis (Figure 2), valgum genu and coxa valga (Figure 1), joint excessive movement(Figure 3), and an atypical waddling stance. Neurological examination did not reveal any focal deficits.

Figure 1:

Radiological evaluation showed bullet-shaped vertebrae with central beaking, odontoid hypoplasia, and metaphyseal irregularities in the long bones. Chest imaging revealed consolidation in the basal segments of the RL lobe and the superior segment of the LL lobe along with moderate cardiomegaly.

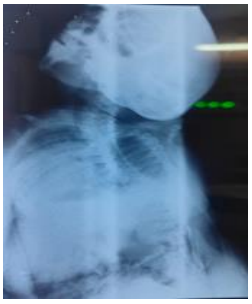

Figure 2:

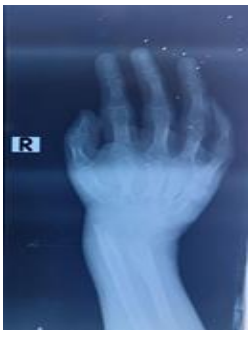

Figure 3:

Laboratory investigations revealed Elevated urinary glycosaminoglycan (GAG) levels 5.5 mg/mM Creatinine (biological reference interval 1.5-5.1) was also noted. Enzyme assay confirmed markedly reduced action of N-acetylgalactosamine-6-sulfatase 0.4 (biological reference interval 23-283), establishing the diagnosis of Morquio syndrome type A (MPS IV A).

The patient was treated with intensive care, including intravenous fluids, antibiotics for pneumonia and urinary tract infection, and respiratory support. She showed gradual clinical improvement and was discharged with advice for regular follow-up.

Discussion

Morquio syndrome is a rare lysosomal storage disorder resulting from a inadequacy of specific enzymes involved in the decay of glycosaminoglycans (GAGs), primarily keratan sulphate and chondroitin-6-sulphate. MPS IV is subclassified into two forms: type A, caused by a defect in the GALNS gene leading to deficient N-acetyl-galactosamine-6-sulphate sulfatase, and type B, due to an alteration in the GLB1 gene affecting beta-galactosidase activity[3]. Unlike other mucopolysaccharidoses, Morquio syndrome spares the degradation of heparan and dermatan sulphate, which accounts for the preservation of normal cognitive function in affected individuals [3].

Our patient, a 28-year-old female, presented with classical phenotypic and radiological features of Morquio syndrome type A, including short stature, kyphoscoliosis, genu valgum, joint laxity, and odontoid hypoplasia. She exhibited a waddling gait, short neck, and pectus carinatum, all of which are hallmarks of the condition and typically manifest in early childhood [3, 4]. Notably, the patient had survived into adulthood without enzyme replacement therapy, highlighting a milder phenotype. Although she appeared healthy at birth and initially met developmental milestones, progressive musculoskeletal deformities became evident by the age of four—a common pattern in MPS IV A [3].

Radiological findings in our case were consistent with established descriptions of Morquio syndrome, including bullet-shaped vertebrae with central beaking, platyspondyly, and odontoid hypoplasia, which may predispose to cervical spine instability and neurological complications [5]. Similar to the observations by Langer and Carry [5], our patient also exhibited a long pelvis with flared iliac bones and acetabular changes. These radiographic features play a critical role in distinguishing MPS IV from other skeletal dysplasias.

Additional systemic involvement in our case included bilateral pneumonia, moderate cardiomegaly, and urinary tract infection—likely secondary to structural deformities of the thoracic cage and reduced respiratory capacity. The presence of minimal interbowel free fluid and moving echoes in the urinary bladder suggested cystitis, an association not commonly highlighted but important in the context of impaired mobility and bladder dysfunction. These complications underscore the multisystem nature of Morquio syndrome, particularly in untreated adults. Screening tests for MPS, such as spot urine assays for mucopolysaccharides, can yield false positives or negatives. Hence, confirmation requires specific enzyme assays or fibroblast studies. In our patient, the diagnosis was established through marked inadequacy of GALNS enzyme action, in concordance with the definitive diagnostic approach recommended in the literature [6-8].The management of Morquio syndrome remains largely supportive. Our patient was treated with antibiotics, intravenous fluids, and electrolyte correction during the acute illness. Long-term

management involves orthopedic surveillance, respiratory care, and cardiac monitoring. Currently, enzyme replacement therapy (ERT) with elosulfase alfa is available in some settings, although it was not accessible in this case. Potential future therapies include gene therapy and hematopoietic stem cell transplantation, which may offer disease-modifying benefits [3].

Although the average life expectancy in Morquio syndrome may extend into the 3 or 4 decade, quality of life is often compromised by progressive skeletal and cardiorespiratory complications [4]. Early diagnosis, even in adulthood, can facilitate timely multidisciplinary interventions aimed at mitigating morbidity.

This case key points the importance of considering MPS IVA in adults with unexplained skeletal dysplasia, short stature, and preserved intelligence. It confirms the need for awareness among clinicians to recognise such phenotypes beyond childhood and to pursue enzyme testing where appropriate. Given the rarity of adult presentations, every such case adds to the growing body of evidence regarding the natural history and spectrum of Morquio syndrome.

Conclusion

Morquio syndrome type A is a rare disorder with predominant skeletal involvement and potential multisystem complications. Although usually diagnosed in childhood, delayed recognition can occur in milder cases. This report of a 28-year-old female with classical skeletal features, pneumonia, and urinary tract involvement highlights the importance of considering MPS IV A in adults with unexplained skeletal dysplasia. Early diagnosis and multidisciplinary care are essential to enhance outcomes and quality of life.

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