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Comprehensive Clinical Evaluation and Management of Congenital Mucopolysachariodosos Type Iva: A Case Report

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ABSTRACT
This case report discusses a 2-year-old male patient diagnosed with Mucopolysaccharidosis type IVA (Morquio syndrome), a rare autosomal recessive lysosomal storage disorder caused by a deficiency of N-acetyl-galactosamine-6-sulfatase. The report details the patient's clinical presentation, which included congenital anomalies, short stature, extensive skeletal involvement, and a lower respiratory tract infection. Diagnosis was confirmed through enzyme assay and genetic testing. The report highlights the progressive multisystem nature of Morquio syndrome, affecting skeletal, respiratory, cardiac, and other systems, while noting preservation of normal cognitive development. Therapeutic approaches, notably enzyme replacement therapy with Elosulfase alfa and multidisciplinary supportive care, are discussed, emphasizing the importance of early diagnosis and comprehensive management to improve quality of life. Genetic counselling and long-term follow-up are recommended due to the hereditary and progressive nature of the disease. The case underscores diagnostic challenges and the need for tailored care in patients with rare metabolic disorders, suggesting gene therapy as a potential future direction for treatment.

KEYWORDS: Mucopolysaccharidosis type IVA, enzyme replacement therapy, Morquio syndrome, genetic mutation**.**

INTRODUCTION

Mucopolysaccharidoses (MPS) are a group of inherited metabolic disorders caused by deficiencies of specific lysosomal enzymes. These genetic defects lead to the accumulation of glycosaminoglycans (GAGs) within lysosomes, resulting in progressive damage to multiple organs and tissues [1]. The overall incidence is estimated to be 1 in 20,000 live births. MPS comprises seven types (I, II, III, IV, VI, VII, and IX), classified according to the deficient enzyme. Type IV is further divided into subtypes A and B. The disease can affect various systems, including the skeletal, joint, cardiac, pulmonary, gastrointestinal, and central nervous systems. [2] In 1929, Luis Morquio first reported 4 Swedish patients with MPS IV (now classified as MPS IVA) and Brailsford also reported a patient with MPS IV [3,4].
This case report describes a patient with mucopolysaccharidosis type IVA (Morquio syndrome), presenting with short stature, extensive skeletal involvement, and a lower respiratory tract infection.

CASE PRESENTATION

The patient is a 2-year-old male with a history of congenital anomalies, including omphalocele and oligohydramnios, low birth weight, and early respiratory symptoms such as persistent cough and fever lasting several days. There is also a history of hospitalization and surgical intervention for inguinal hernia prior to this admission. The clinical picture includes respiratory tract involvement with signs of infection but also features consistent with systemic disease.

Laboratory and Diagnostic Findings

Routine blood investigations show mild anemia and normal platelet counts, with normal liver function tests. Radiological and clinical findings typical for Morquio syndrome likely include skeletal abnormalities such as dysostosis multiplex (though not detailed in the snapshot), joint laxity, and possibly cardiac murmurs. The diagnosis has been confirmed by enzyme assay or genetic testing consistent with MPS Type 4. Result shows deficient enzyme as N-acetyl-galactosamine-6-sulfate-sulfatase and a definitive diagnosis of MPS IV A. The Approved drug for Morquio syndrome is recombinant human GALNS enzyme replacement therapy (Elosulfase Alfa) [5].

DISCUSSION

Mucopolysaccharidosis type IV (MPS IV), also known as Morquio syndrome, is a rare autosomal recessive lysosomal storage disorder characterized by a deficiency of the enzymes N-acetyl-galactosamine-6-sulfatase (MPS IVA) or beta-galactosidase (MPS IVB) [6][7]. This deficiency leads to the accumulation of glycosaminoglycans such as keratan sulfate and chondroitin-6-sulfate in various tissues, causing progressive multisystem involvement—primarily affecting the skeletal system. Patients typically present with coarse facial features, short-trunk dwarfism, genu valgum, pectus carinatum, and joint hypermobility but usually maintain normal cognitive development [6][7][8].

The disease course is marked by evolving skeletal deformities, respiratory complications, and, in severe cases, cardiac involvement. Enzyme replacement therapy (ERT) with elosulfase alfa is now established for MPS IVA, alongside multidisciplinary supportive management, but requires early diagnosis to optimize outcomes [6][7]. In the indexing case, the patient demonstrated typical clinical and radiological findings consistent with MPS IV, highlighting the importance of considering this diagnosis in children with dysplastic skeletal features, recurrent respiratory illness, and normal intelligence [7][6].

These skeletal abnormalities progressively impair mobility and quality of life. Patients may also develop respiratory compromise, cardiac valve involvement, corneal clouding, and hearing loss as the disease progresses [8] [10].

Our patient exhibited characteristic clinical features accompanied by radiological findings consistent with MPS IV, highlighting diagnostic challenges, especially in early childhood, due to phenotypic variability and the rarity of the disorder. Early diagnosis remains crucial for management and prognosis. Current therapeutic options include enzyme replacement therapy (ERT) with elosulfase alfa for MPS IVA, which has demonstrated efficacy in reducing GAG accumulation and improving endurance; however, limitations such as high cost and variable response exist [10] [11].

Multidisciplinary supportive care involving orthopedic interventions, respiratory and cardiac monitoring, and physical rehabilitation is essential to manage complications and improve quality of life [9][8].

CONCLUSION

This case of MPS Type 4 illustrates the complexity of managing a multisystem genetic disorder presenting early in life with both congenital anomalies and progressive systemic involvement. Early diagnosis allows initiation of supportive therapies to manage complications and improve quality of life. Comprehensive multidisciplinary care tailored to the patient's evolving needs is paramount. Genetic counselling and long-term follow-up should be integral to management due to the hereditary nature and progressive course of the disease. Future therapeutic advances such as gene therapy offer hope for more definitive treatment.

REFERENCE

1. 1.Wenger DA, Coppola S, Liu SL. Insights into the diagnosis and Treatment of lysosomal storage diseases. Arch Neurol 2003; 60: 322-8.
2. Scott HS, Bunge S, Gal A, Clarke LA, Morris CP, Hopwood JJ. Molecular genetics of mucopolysaccharidosis type I: diagnostic, clinical, and biological implications. Hum Mutat 1995; 6: 288-302.
3. 3.Morquio L (1929) Sur une forme de dystrophie osseuse familiale. Bull Soc Pediatr Paris 27: 145-152.
4. 4.(1979) The classics: Chondro-osteo-dystrophy. Roentgenographic and clinical features of a child with dislocation of vertebrae, James F. Brailsford, MD: Am. J. Surg. 7:404, 1929. Clin Orthop Relat Res 114: 4-9.
5. 5.Sanford M, Lo JH (2014) Elosulfase alfa: first global approval. Drugs 74(6):713-718.
6. Singh J, Ferrante N Di, Niebes P, Tavella D (1976) N-acetylgalactosamine-6-sulfate sulfatase in man. J Clin Invest 57:1036-1040.
7. Tomatsu S, et al. (2011) Mucopolysaccharidosis type IVA (Morquio A Disease): clinical review and current treatment: a special review. Curr Pharm Biotechnol 12(6): 931-945.
8. Tomatsu S, AM Montaño, H Oikawa, M Smith, L Barrera, et al. Morquio A syndrome: diagnosis and current and future therapies. Pediatr Endocrinol Rev 12(01): 141-151.
9. 9.MPS IV – Morquio syndrome. National Organization for Rare Disorders (NORD). https://rarediseases.org/rare-diseases/morquio-syndrome/. Accessed 2025.
10. 10.Hendriksz CJ, Berger KI, Biegstraaten M, et al. International guidelines for the management and treatment of Morquio A syndrome. Am J Med Genet A. 2015;167A(1):11-21.
11. 11.Mount Sinai Health Library. Morquio syndrome information. https://www.mountsinai.org/health-librar