CASE REPORT

Becker Muscular Dystrophy with Exon 27-43 Deletion: Clinical and Genetic Insights in a Pediatric Case: A Case Report

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Article History:
Received date: December 1, 2025
Revised date: June 9, 2026
Accepted date: June 30, 2026
Published date: August 1, 2026

Online Access

DOI: 10.64772/mjapfn.2.2.36

Abstract

Introduction: Becker muscular dystrophy is an X-linked disorder of dystrophin that leads to gradually progressive muscle weakness, and cardiac abnormalities may appear regardless of the degree of skeletal muscle involvement.

Case Presentation: An eight-year-old boy, born to non-consanguineous parents with an unremarkable birth and family history, began showing progressive lower-limb weakness at five years of age with troubles in activities of daily living. Examination revealed proximal weakness, hypotonia, and diminished reflexes, with no other systemic abnormalities with positive Gower's sign, and bilateral calf hypertrophy. Creatine kinase was significantly raised. Genetic testing identified an in-frame deletion of exons 27-43 in the DMD gene, confirming Becker Muscular Dystrophy. The patient was managed with physiotherapy and corticosteroid therapy.

Conclusions: The case emphasizes importance of early recognition, genetic diagnosis, and comprehensive long-term care in patients with Beckers muscular dystrophy.

Keywords: *frameshift mutation; genetic testing; muscular dystrophies.*

Introduction

Becker muscular dystrophy (BMD) is an X-linked recessive dystrophinopathy caused by pathogenic mutations in the DMD gene.[1] It represents the milder phenotype of dystrophin deficiency compared with Duchenne muscular dystrophy, due to the production of partially functional dystrophin.[2] BMD typically presents in late childhood or adolescence with progressive muscle weakness and calf hypertrophy.[3] The estimated prevalence of BMD is 1 in 18,000–30,000 live male births, although rates vary by population.[4] Cardiac involvement, particularly dilated cardiomyopathy, is a major contributor to morbidity and may occur independent of skeletal muscle severity involvement.[5] Diagnosis is based on clinical features, elevated creatine kinase, and confirmed further with genetic testing.[4] Management focuses on cardiac surveillance, physiotherapy, and emerging gene-targeted therapies.[4]

This case illustrates a rare Becker muscular dystrophy genotype characterized by a deletion of exons 27–43 in the DMD gene. The patient exhibited a relatively mild clinical phenotype, with preserved muscle strength and no cardiac involvement, consistent with in-frame deletions. This case underscores the importance of identifying Becker muscular dystrophy early through clinical features, elevated creatinine

How to cite (Vancouver Style)
K.C. SR, Chand B, Poudel D, Shah PM, Karki R. Becker Muscular Dystrophy with Exon 27-43 Deletion: Clinical and Genetic Insights in a Pediatric Case: A Case Report. Med J APF Nepal. 2026;2(2):89-91.

phospho-kinase, and genetic testing. Documenting such cases enhances understanding of genotype–phenotype relationships and informs timely management and routine cardiac surveillance.

Case Report

An 8-year-old boy, born to non-consanguineous parents, presented with progressive lower-limb weakness first observed at five years of age. His mother reported that he was slower than peers while running and was hesitant to climb or jump. He currently experienced difficulty rising from the floor but could perform activities such as combing his hair and dressing independently. There was no history of cognitive delay, scholastic difficulties, constipation, rash, chronic medication use, or exertional pain. Family, antenatal, and birth histories were unremarkable. No history of similar illness or unexplained death among maternal lineage.

On examination, he was conscious, oriented, and thin built, with no dysmorphic features and normal vital signs. His weight was at the 3rd percentile and height was at the 25th percentile (WHO growth charts). Early motor milestones were achieved on time, and speech and cognitive development were normal. Central nervous system examination revealed normal higher mental functions and intact cranial nerves. Motor examination demonstrated calf hypertrophy (Figure 1), wasting of extensor muscle groups, hypotonia, and reduced deep tendon reflexes predominantly in the lower limbs. Muscle strength was assessed using the Medical Research Council (MRC) scale, showing mild weakness (4/5) in the hip extensors and knees extensors bilaterally. Gower's sign was positive (Figure 2). Upper-limb tone, reflexes, and power were preserved. Sensory examination was normal. There were no neurocutaneous manifestations. The examination of the spine and other systems were unremarkable.

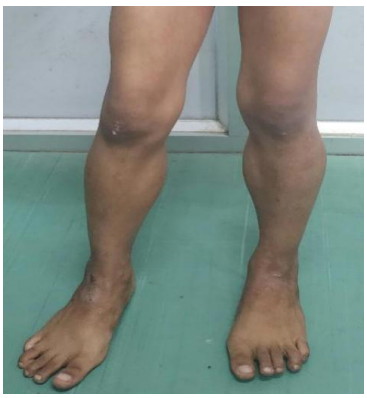

Figure 1: Bilateral calf hypertrophy as seen from front.

Figure 2: Demonstration of positive Gower's sign.

With provisional diagnosis of muscular dystrophy, baseline investigations including complete blood counts, renal and hepatic function test were normal. Serum Creatine Phosphokinase – N-Acetylcysteine (CPK-NAC) was markedly elevated at 3011 U/L. Genetic testing via Next Generation Sequencing (NGS) revealed a deletion of exons 27-43 in the DMD gene, classified as likely pathogenic, confirming the diagnosis of BMD. Echocardiography and electrocardiogram were normal.

After the diagnosis, the child was managed with low impact strengthening physiotherapy and corticosteroid therapy (Deflazacort 0.9 mg/kg). Genetic counselling was done and the patient was kept on regular follow up.

Discussion

This 8-year-old boy's presentation is consistent with Becker muscular dystrophy, a dystrophinopathy with later onset and slower progression than Duchenne muscular dystrophy. Symptoms beginning at five years, including difficulty running, climbing stairs, and rising from the floor with a positive Gower's sign, indicate early involvement of the pelvic girdle muscles.[6] The presence of calf hypertrophy and normal early developmental milestones further supports the diagnosis, as cognitive function and early motor development are typically preserved in BMD.[7]

Proximal lower-limb weakness, hypotonia, and reduced reflexes reflect selective involvement of pelvic and thigh muscles, while upper-limb strength is generally maintained in early stages.[8] The absence of sensory deficits, systemic complaints, or skin changes helps exclude other neuromuscular or metabolic disorders.[9] Although this child currently has no cardiac manifestations, dilated cardiomyopathy may develop independently of skeletal muscle involvement, emphasizing the need for early and ongoing cardiac monitoring.[10] Genetic testing remains the definitive method for diagnosing BMD, distinguishing it from other dystrophies, and guiding family counseling as well as access to emerging targeted therapies.[11] Early identification allows

timely physiotherapy and proactive management to optimize long-term function.[12]

Genotype–phenotype studies show that multi-exon deletions in BMD commonly occur in two "hotspot" regions of the DMD gene: a proximal region (exons 3–20) and a mid-to-distal region (exons 45–55).[13] In a study of 163 patients, 78% had in-frame deletions, most frequently in exons 45–47, with 92% of all deletions located within exon 45–55.[14] Another large series of 750 patients confirmed this distal hotspot, with occasional large deletions but none specifically highlighting exon 27–43.[15]

Our patient carries the rare exon 27-43 deletion, outside the typical hotspot and spanning a larger contiguous segment than usually observed. The limited number of reported cases with this specific deletion, particularly with detailed clinical and follow-up information, underscores the uniqueness and significance of this report.

Conclusions

Early genetic testing in suspected dystrophinopathies helps confirm the diagnosis, improves understanding of genotype–phenotype relationships in Becker muscular dystrophy, provides useful prognostic information, and supports timely genetic counselling and appropriate supportive care for better patient management.

Source of Funding: None

Acknowledgement: None

Conflict of Interest:

The authors declare no competing interests. Dr. Drishti Poudel is currently serving as an Editor of Medical Journal of Armed Police Force Nepal (MJAPFN). She was not involved in the editorial decisions, peer-review processing, and publication-related activities concerning this manuscript.

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