

Teen with Duchenne Muscular Dystrophy (DMD) taking Eteplisren returns to ambulation after complex proximal hip fracture: A Case Report.



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Case Diagnosis

Left minimally comminuted, mildly displaced intertrochanteric fracture.

Case Description

15 year-old male with Duchenne Muscular Dystrophy with genetic testing was amenable to Eteplisren treatment which was started in 2014 at the age of nine. In 2020, patient was ambulating household distances without assistive devices. Patient suffered a ground level fall resulting in a left intertrochanteric fracture requiring operative fixation with an intramedullary. On discharge from IRF patient was walking with supervision and a walker. On follow up 2 months postoperatively, patient had near complete return to baseline.

Assessment / Results

This is the first documented case of a return to ambulation following hip fracture in a teen with Duchenne Muscular Dystrophy.



Discussion

Eteplisren is indicated for patients with Duchenne who have a confirmed mutation in the *DMD* gene that is amenable to exon 51 skipping. Clinical studies have shown increases in the dystrophin protein from .16% to .93% after a 180-week trial, but there is little to no clinical evidence of benefit yet to these patients. This outcome was likely attributed to infusions of Eteplisren from the age of nine and aggressive inpatient rehabilitation.

Conclusion

The patient is in the first generation of patient's receiving Eteplisren and clinical outcomes are only now showing results. Aggregation of outcomes from these patient's will be crucial to understand how gene modifying agents such as Eteplisren are altering the morbidity and mortality of these patients.