

## RETT SYNDROME: A RARE DISEASE WITH LOW ENERGY FEMUR FRACTURE

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**Introduction:** Rett syndrome is a rare genetic neurological and developmental disorder that affects the way the brain develops, causing a progressive loss of motor skills and speech. Epilepsy affects the majority of patients in a specific clinical stage of the disease. Generalized tonic-clonic (GTC) seizure is one of the most frequent seizure type and its association with supracondylar femur fracture is less reported in the literatures.

**Discussion:** We reported a case of 10 years old girl with underlying Rett syndrome stage 3 in its clinical form, presented to our center with pain and swelling of distal left thigh after episodes of GTC seizure at home. There was no history of fall during the seizure episodes. Clinically, her distal left thigh was swollen, tender with limited range of motion due to pain. Plain radiograph showed minimally displaced supracondylar fracture left femur with anterior angulation. Cast was applied and satisfactory reduction obtained post application of cast. She was discharged home and advised for wheelchair ambulation.

**Conclusion:** Children with severe disability experience functional limitations and often develop comorbidities including osteoporosis, and those with limited ambulation are at higher risk of osteoporosis and subsequent fracture than those who can walk independently<sup>1</sup>. As the clinical stage of the disease progress, most people who have Rett syndrome experience seizures at some point during their lives. GTC type of seizure may cause fracture despite its low energy trauma because of osteopenic bone (disuse osteopenia). Literature regarding treatment for this condition is still lacking but most of the time conservative treatment was favored. The complication of malunion has been reported in approximately one third of children treated in this manner<sup>2</sup>.