A case of osteogenesis imperfecta in a patient diagnosed in adulthood

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Abstract

Background
Osteogenesis imperfecta (OI) is an extremely rare inherited connective tissue disorder, with an incidence of 1 in 20,000 births in the United States. Our aim is to assist other providers in diagnosing and managing patients with OI and concurrent medical morbidities.

Case Presentation
A 21 year old male with a BMI of 34 and a lifetime history of over 10 fractures presented to the Emergency department after a complex seizure at home. The seizure was witnessed by his mother and lasted less than one minute. He had another seizure two weeks prior to presentation. In the ED, the patient had another witnessed seizure lasting 45 seconds with bladder incontinence. He has one cousin with epilepsy. EEG and MRI Brain were unremarkable. CT of the upper extremities showed displaced fracture of the left humeral head, displaced fracture of the right anterior humeral head, and fracture of the posterior rim of the right glenoid fossa.

His seizures were controlled on levetiracetam. The patient's mother detailed a life-long history of fractures with minimal trauma, including breaking his R leg after hitting a sofa at age 5, breaking a knee while playing football at age 14, and breaking a thumb while playing catch at age 18. Upon follow-up with an Endocrinologist, the patient was diagnosed with osteogenesis imperfecta.

Conclusions
Identifying pathologic fractures and taking a thorough history is essential for timely and appropriate diagnosis of OI. OI can be complicated by the comorbidities including epilepsy and obesity. Appropriately addressing these comorbidities can decrease future risk of fracture.