Late diagnosis of Osteogenesis Imperfecta: A Case Report

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INTRODUCTION

Osteogenesis imperfecta (OI) is a genetically inherited disorder that occurs in 1 in 15,000 to 20,0000 births.[1] This condition is primarily characterized by bone fragility, blue or gray sclera, hearing loss, short stature, multiple deformities, positive family history, and tooth abnormalities called dentinogenesis imperfecta. Diagnosis of mild OI is challenging due to variability in mutations affecting the two genes (COL1A1 & COL1A2) and variable phenotypic expression. Sillence types (Type I-IV) comprise most cases with a mutation on collagen type I (COL1A1 and/or COL1A2). [2,3,4] The purpose is to present a rare case of a late onset diagnosis of OI in a 12-year-old patient which is unsuitable for the Sillence Classification System (Type I-IV).

CASE DESCRIPTION

- A 12 y/o boy presented to the clinic with spontaneous fractures (fx.) with no history of trauma
- PE was unremarkable except for bluish sclera **(Figure 1)**
- Upper and lower extremities were atraumatic in appearance and without deformities.
- No family history, both parents appear with normal bones anatomy and no history of frequent bone fractures.
- In physical examination:
  - Height: 4' 11"
  - Weight: 130 lbs.
  - Body Mass Index: 26.3
  - Blood Pressure: 130/80 mmHg
- Serum calcium: 10.4
- Serum vitamin D: 23 ng/mL
- Creatinine: 0.46 mg/dL
- BUN: 10 mg/dL
- Serum phosphate levels: 5.1 mg/dL
- Serum PTH levels: 27.80 pg/mL
- Alkaline Phosphate: 259 U/L
- Genetic testing was performed and showed that patient was positive for COL1A1 and COL1A2.

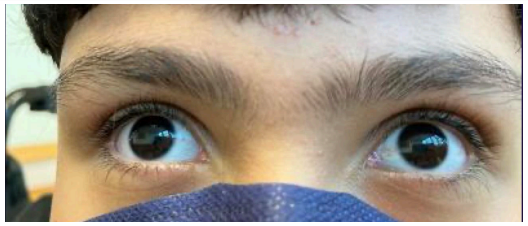

Figure 1. Bluish Sclera presentation on a 12-year-old boy

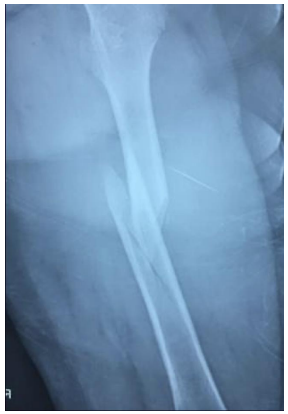

Figure 2. Right femur fx.

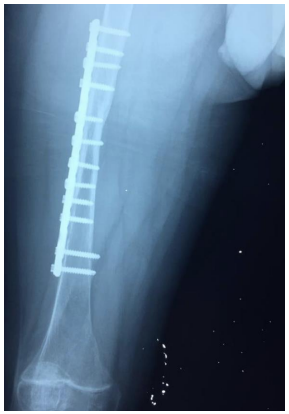

Figure 3. Post-op femur ORIF

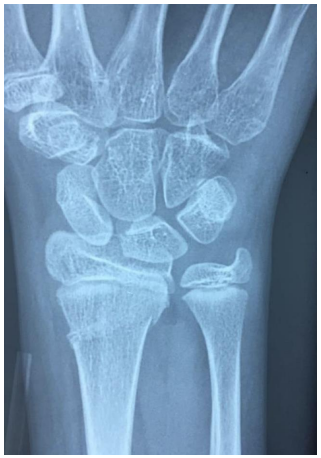

Figure 4. Right Distal Radius Fx.

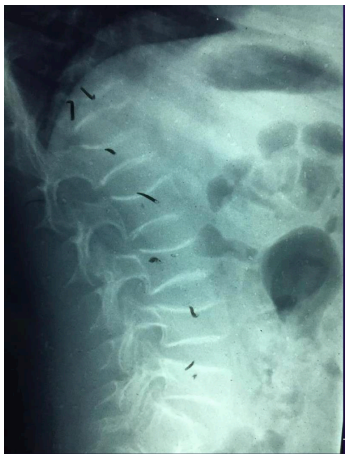

Figure 5. Codfish vertebrae

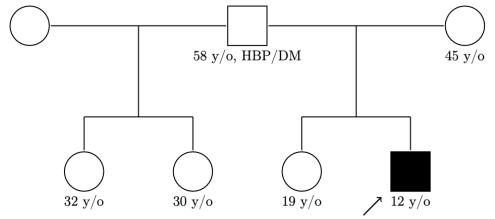

Figure 6. Family Pedigree

DIAGNOSIS

The patient was diagnosed with de novo OI, confirmed with positive COL1A1 and COL1A2 gene tests; first manifestation was at the age of 12 with two spontaneous fractures, which indicates a late diagnosis of OI.

Patient was sent home and started on bisphosphonate infusions.

DISCUSSION

Osteogenesis Imperfecta is commonly diagnosed in early childhood due to the common clinical findings during physical examination, such as bluish sclera, decreased bone mass, dentinogenesis imperfecta and hearing loss [1]. The presented case is particular in that it showed bilateral bluish sclera and spontaneous fractures at 12 years of age, which suggests a late onset and eventually diagnosis of OI. Imaging exhibited codfish vertebrae but no other musculoskeletal anomalies which increased the necessity of in-depth analysis and comparison. OI has a broad clinical spectrum which appears to be variable in phenotypic expression. [5]. Further work include analyzing in depth family genetics in order to establish a detailed inheritance pattern to be able to classify this case (**Figure 6**).

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