Idiopathic Juvenile Osteoporosis (IJO): 
Common symptoms in an uncommon condition

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Introduction

- Osteoporosis in children and young people can be primary due to Osteogenesis Imperfecta (OI) or secondary to chronic disease.
- We report 2 patients with Idiopathic Juvenile Osteoporosis (IJO), a rare primary osteoporotic disorder

Patient 1

- A 12 year old boy presented with 12 months of lower back pain and stiffness, against a background of chronic pain in knees, wrist and ankles.
- There was no history of fractures or systemic disease. Examination revealed normal anthropology [height 154.1cm (+0.42SDS), weight 38.3kg (+0.09SDS)] and he was pre-pubertal.
- He had mild tenderness over the thoracolumbar spine, knee and ankle joints but there was no limitation of movement. He had no skeletal/extra skeletal manifestations of OI.
- X-ray and MRI of the spine revealed multiple thoracic vertebral compression fractures [T6-T10]. DEXA scan revealed a low lumbar spine bone mineral density (BMD) Z-score of -3.2.
- Iliac crest bone biopsy showed high turnover osteopenia, increased osteoid surface with no definite mineralization defect and was predictive of responsiveness to bisphosphonate therapy.
- He was commenced on 6 monthly intravenous zoledronic acid therapy with good recovery.
- Genetic testing was negative for common mutations in COL1A1/COL1A2.

Patient 2

- A 14 year old boy was referred with severe back pain of subacute onset.
- He had several childhood fractures (wrist, ribs and navicular bone) and limb pains for 6 years prior to presentation.
- Examination revealed normal height [158 cm, +0.3SDS], excessive weight [100kg, +2.9 SD] and signs of early puberty.
- MRI spine showed loss of height of T7 to L1 vertabrae. DEXA scan showed a lumbar spine BMD Z-score of -2.8.
- Genetic testing was negative for common mutations in COL1A1/COL1A2.
- Bone biopsy showed adynamic bone with reduction in thickness of the cortices, osteoblast and osteoclasts.
- He received intravenous pamidronate therapy with symptomatic relief. Repeat DEXA scan after 1 and 3 years showed improvement in BMD Z-scores to -1.1 and +0.4 respectively. He is on maintenance oral risedronate therapy.

Conclusions

- IJO is a diagnosis of exclusion based on clinical and histological findings. Non-specific symptoms can lead to delay in the diagnosis.
- IJO should always be considered in the differential diagnosis of pre- and peri-pubertal adolescents with chronic back/bone pain in the absence of other causes.
- Bone biopsy is an important part of the diagnostic workup, however histology can be variable.
- Early diagnosis and appropriate therapy with bisphosphonates can promote bone remodeling thereby alleviating chronic pain and morbidity.