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

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Introduction

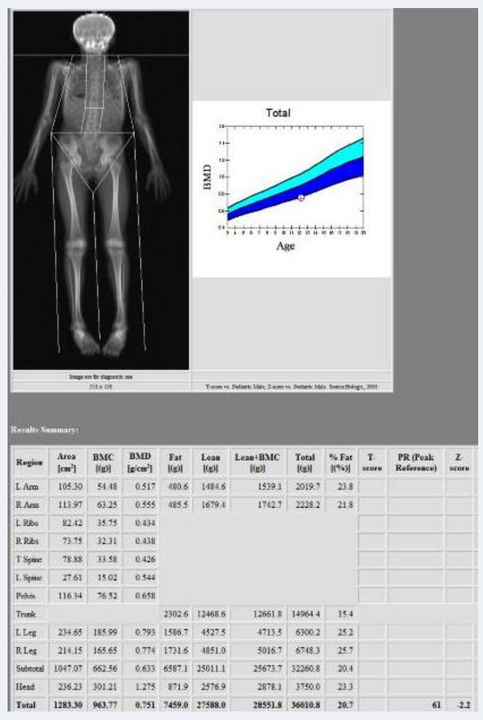
- Osteoporosis in children and young people can be primary due to Osteogenesis Imperfecta (OI) or secondary to chronic disease.
- We report 2 patients with Idiopathic Juvenile Osteoporosis (IJO), a rare primary osteoporotic disorder

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 anthropometry [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/COL1A*2

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 SDS) and signs of early puberty.
- MRI spine showed loss of height of T7 to L1 vertebrae. DEXA scan showed a lumbar spine BMD Z-score of -2.8.
- Genetic testing was negative for common mutations in *COL1A1/COL1A*2.
- 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.



Results Summary:

Region	Area [cm²]	BMC [g]	BMD [g/cm²]	Fat [g]	Lean [g]	Lean+BMC [g]	Total [g]	% Fat [(%)]	T score	PR (Peak Reference)	Z score
L Arm	105.30	54.48	0.517	480.6	1484.6	1539.1	2019.7	23.8			
R Arm	113.97	63.25	0.555	485.5	1679.4	1742.7	2228.2	21.8			
L Ribs	82.42	35.75	0.434								
R Ribs	73.75	32.31	0.438								
T Spine	78.88	33.58	0.426								
L Spine	27.61	15.02	0.544								
Pelvis	116.34	76.52	0.658								
Trunk				2302.6	12468.6	12661.8	14964.4	15.4			
L Leg	234.65	185.99	0.793	1586.7	4527.5	4713.5	6300.2	25.2			
R Leg	214.15	165.65	0.774	1731.6	4851.0	5016.7	6748.3	25.7			
Subtotal	1047.07	662.56	0.633	6587.1	25011.1	25673.7	32260.8	20.4			
Head	236.23	301.21	1.275	871.9	2576.9	2878.1	3750.0	23.3			
Total	**1283.30**	**963.77**	**0.751**	**7459.0**	**27588.0**	**28551.8**	**36010.8**	**20.7**		**61**	**-2.2**

Fig 1: Total body BMD Z-score -2.2

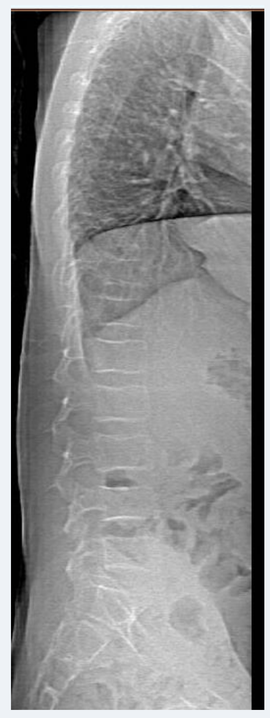


Fig 2: VFA
Loss of vertebral body height at levels T6-T10

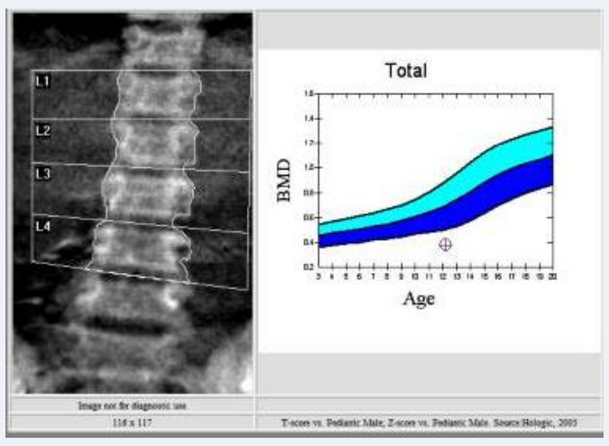


Fig 3: Lumbar Spine BMD Z-score -3.2

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.