

Ribbing Disease, An Unusual Cause of Leg Pain in a Young Woman

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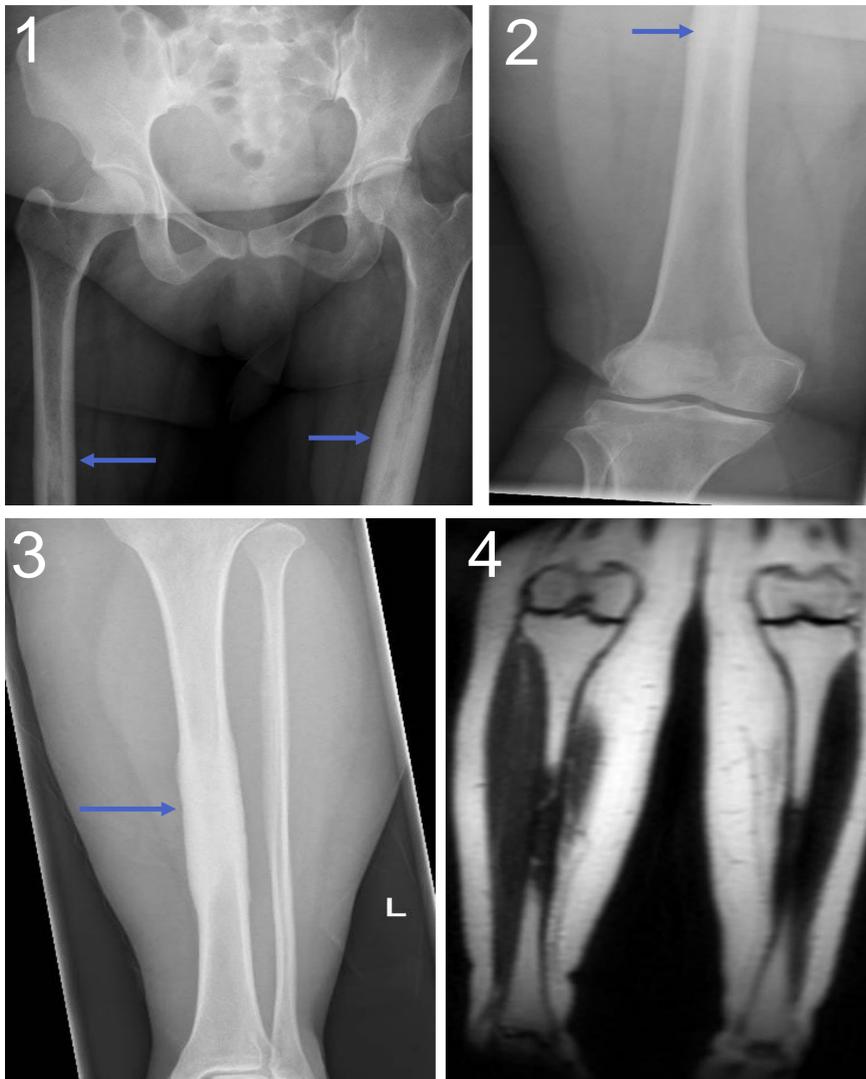
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Introduction

- Ribbing Disease (also known as Multiple Diaphyseal Sclerosis) is a rare form of sclerosing dysplasia characterised by benign endosteal and periosteal bone growth confined to the diaphysis of long bones.
- It was first described in 1949 by Ribbing. Most commonly the disease presents with leg pain in young women, classically involving the femora and tibiae.

Case description

- We present the case of a 32 year old woman, presenting to her GP with right lower leg pain. This pain was worse at night, but responded to simple analgesia. She was initially diagnosed with arthritis but the leg pain got worse and became bilateral. She had no other significant past medical history with no history of fevers, trauma, fractures or dental problems. She had no relevant family, social or medication history.
- On examination, there was no tenderness on bone palpation, or active and passive adjacent joint movements. There were no overlying skin changes or joint swellings.
- Investigations confirmed normal biochemistry including alkaline phosphatase (51iu/L (NR 30-130)) and inflammatory markers. Plain radiographs revealed bilateral focal segmental cortical sclerosis involving the mid-diaphyseal region of the tibiae and femora, with resultant narrowing of the medullary cavity. MRI of the lower legs demonstrated associated marrow oedema and bone scanning showed increased tracer uptake in these mid-diaphyseal areas. Bone densitometry however was entirely normal. Based on the clinical and radiological features she was diagnosed with Ribbing Disease.
- Images 1-3 showing sclerosis with cortical thickening of diaphysis of long bone with sparing of epiphyses on plain radiographs.
- Image 4 shows T1-weighted coronal image demonstrating cortical thickening of tibial diaphysis with hypointense signal in mid-diaphysis.



Discussion

- The cause of the Ribbing Disease is currently unknown, although some cases appear to be genetic and inherited in an autosomal recessive fashion. Recent studies showed a *TGFβ1* mutation in some cases of Ribbing disease.
- It is important to exclude other causes of bone pain including osteomyelitis, fractures, osteosarcoma, osteopetrosis, Erdheim-Chester, Van Buchem and Camurati-Engelmann Disease. In Ribbing disease the markers of bone formation and resorption are normal.
- MRI demonstrates a significant bone marrow oedema and cortical thickening that can explain significant pain associated with the disease. Radiographs and tomographic studies show benign-appearing endosteal and periosteal cortical thickening. Intense uptake of radionuclide tracer is confined to the shaft of all involved bones. All pathologic specimens reveal nonspecific changes that include a slow increase in the mass of cortical and endosteal bone.
- Treatment is mainly supportive with analgesia but can include steroids, NSAIDs, bisphosphonates and orthopaedic intervention. If conservative treatment fails then intramedullary reaming or fenestration is performed to relieve the severe pain. It can be a self limiting disease but may progress in some patients. It can be associated with fatigue, anaemia, waddling gait and muscle weakness.

Cardiovascular & Renal complications

- Patients with Ribbing's disease may have significant alterations in their left ventricular systolic and diastolic function associated with supraventricular and ventricular arrhythmias. Many patients with Ribbing disease develop severe ventricular hypertrophy with hypertension. There is increased incidence of ischaemic heart disease in these patients.
- Rarely renal failure related to arteriosclerotic changes of the renal arteries can happen.

Conclusion

- This case demonstrates a rare cause of a common symptom presenting to the Metabolic Bone Clinic and highlights the importance of the history, examination and investigation pathway as it is often a diagnosis of exclusion in a young woman.
- It may be initially diagnosed as low-grade osteomyelitis, but it may also be confused with other causes of increased bone density on radiological investigations (sclerosing dysplasia) like Camurati-Engelmann disease, Van Buchem disease, Erdheim-Chester disease.
- The clinical presentation and imaging findings of patients with Ribbing disease are becoming more apparent. However, there is lack of evidence on the natural disease progression and effectiveness of treatment modalities.

Key references

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