

Monostotic fibrous dysplasia with Raynaud's phenomenon

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Abstract

Fibrous dysplasia (FD) is a benign bone disorder characterized by alteration in bone morphology. Monostotic FD is the commonest variant and affects the craniofacial bones. Raynaud's phenomenon is recurrent vasospasm of the fingers and toes due to cold exposure. The disease is usually idiopathic or secondary to connective tissue disorders. Raynaud's phenomenon is not described previously with FD. We recently encountered two interesting patients of craniofacial monostotic FD with Raynaud's phenomenon and report the same in this report.

Key words: Bisphosphonates, craniofacial dysplasia, monostotic fibrous dysplasia, Raynaud's phenomenon

INTRODUCTION

Fibrous dysplasia (FD) of bone is a benign disorder characterized by replacement of medullary bone with the fibro-osseous tissue. The overgrowth of the affected bone leads to distortion, poor strength leading to characteristic deformities.^[1] The FD is divided into three types, based on the clinical presentation and bone involvement. They are monostotic (single bone involved), polyostotic (multiple bones) and McCune Albright Syndrome (FD along with endocrinopathies and skin macules).^[2] Craniofacial form of FD presents in young adults with no specific sex predilection. The lesion is more common in maxilla than mandible. The disease may be self-limiting by third decade, and both sexes are equally affected. The disease usually presents with deformity, bone pains and recurrent fractures.

Raynaud's phenomenon is a vascular disorder characterized by discoloration of the fingers and toes.^[3] This is classically described with decreased blood supply triggered by cold exposure. The phenomenon is considered primary (Raynaud's disease) with no apparent underlying etiology and secondary due to underlying systemic disorders like connective tissue disorders. Raynaud's phenomenon is not commonly described with bone disorders and literature search revealed one such report in hyperparathyroidism.^[4] We recently encountered two interesting patients of monostotic FD with Raynaud's phenomenon. We report these cases for the unusual

association and to highlight the relevant review of the literature.

CASE REPORTS

Case 1

A 30-year-old man presented with painless, progressively increasing swelling over the right mandibular region with the onset 4 years prior to the visit. He denied difficulty in mouth opening, food chewing, facial trauma or temporomandibular joint clicking. He gives a history of bluish discoloration of the fingers on exposure to cold for past 2 years with no residual disability. He had no history of recurrent fractures, joint pains, skin rash, hair loss or oral ulcers and denied similar complaints in the family members. Examination revealed normal vital parameters with asymmetry of the face [Figure 1b] due to expansion of the right half of mandible. Exposure to cold revealed classical Raynaud's phenomenon in different stages [Figure 1c and d]. The rest of the systemic and cutaneous examination was normal. Hormonal profile revealed normal thyroid, adrenal and gonadal axes evaluation. A skeletal survey revealed an expansible osteolytic lesion over right mandible and no features of metabolic bone disorders. Other laboratory work-up showed elevated alkaline phosphatase (347 U/L) with normal calcium, phosphorus, parathyroid hormone and 25 hydroxy Vitamin D levels. Immunological profile revealed normal C-reactive protein, antinuclear antibody and Rheumatoid factor titer. Further work-up including

the imaging of the chest and abdomen, drug screen and peripheral Doppler studies were normal. He was diagnosed finally to be a case of monostotic FD with Raynaud's disease. He was treated with zoledronic acid 5 mg as an intravenous infusion, once every 6 months, calcium, Vitamin D supplements and Nifedipine. The patient was reviewed every 3 months for the last 1-year during which, he remained asymptomatic with no recurrence of Raynaud's phenomenon and fall in the level of alkaline phosphatase (103 U/L).

Case 2

A 26-year-old female presented with painless swelling over the left mandibular region with no definite duration of the onset. The swelling was noticed by a photographer and she sought consultation for the same. She denied difficulty in mouth opening, food chewing, facial trauma or any other local symptoms. She had no history of recurrent fractures, joint pains, skin rash, hair loss or oral ulcers. Past medical history revealed that she had two episodes of painful bluish discoloration of the fingers on exposure to the extreme cold climate. The episodes were diagnosed as chilblains and treated symptomatically. She was not prescribed any regular medicine for the same and denied any residual disability. Examination revealed normal vital parameters with asymmetry of the face [Figure 1a] and expansion of the left mandible body. She had mild discoloration of fingers on exposure to cold water. The rest of the systemic and cutaneous examination was normal. Hormonal profile revealed normal thyroid, adrenal and gonadal axes evaluation. A skeletal survey revealed an expansible osteolytic lesion involving the body and condyle of the left side of the mandible [Figure 2a-d]. Other laboratory work-up showed mild elevation of alkaline phosphatase (215 U/L) with normal calcium,

phosphorus, parathyroid hormone and 25 hydroxy Vitamin D levels. Her immunological and biochemical profile were normal. She was treated with zoledronic acid 5 mg as an intravenous infusion, once every 6 months along with conservative measures for the Raynaud's phenomenon. During last review, 6 months after first injection she remained asymptomatic without any further worsening of facial asymmetry and normal serum biochemical parameters.

DISCUSSION

We present two cases of craniofacial FD with Raynaud's phenomenon. Extensive literature search did not reveal any previous report with this association. Etiological work up in both the patients did not reveal any known condition for Raynaud's phenomenon. The suggested mechanism in these patients could be due to release of an unidentified chemokine substance of the dysplastic bone triggering the vascular phenomenon. Raynaud's phenomenon was associated with connective tissue disorders such as systemic sclerosis, osteopoikilosis and fibromuscular dysplasia.^[5] Increased collagen breakdown products triggering the cutaneous reactions were reported in systemic sclerosis.^[5]

Raynaud's phenomenon is divided into primary where the disease is idiopathic, and secondary when it is due to a systemic disorder. The conditions associated with Raynaud's symptoms are connective tissue disorders, obstructive vascular disorders and drugs like beta blockers and ergotamine. Metabolic bone disorders are reported to have Raynaud's phenomenon rarely.^[4] Progressive diaphyseal dysplasia is the only dysplastic condition of the bone with Raynaud's phenomenon.

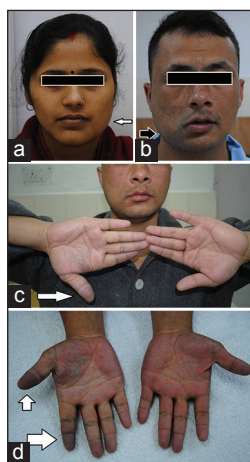


Figure 1: (a and b) Two patients showing left and right mandibular enlargement of craniofacial dysplasia (c and d) Raynaud's phenomenon in 1st patient during different phases showing acrocyanosis (arrows)

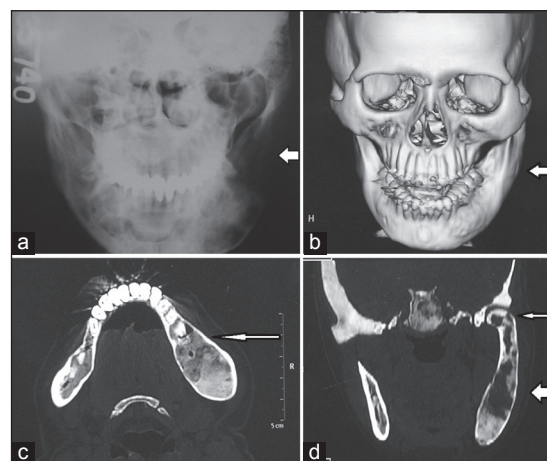


Figure 2: Imaging findings of 2nd patient showing radiolucent area (a) on X-ray the expansile lesion is seen on three-dimensional image of computed tomography scan (b) and involvement of left mandible body and condyle on axial (c) and coronal sections (d)

Craniofacial dysplasias are most common site of involvement in monostotic FD. Skeletal survey did not reveal involvement of other bones in both the patients. The use of bisphosphonates has transformed the treatment of FDs. They reduce the pain and swelling, fracture rates and the progression of deformities. Bisphosphonates are given as intravenous or oral preparation and act by suppressing the osteoclast activation.^[6] The treatment protocols for the use of bisphosphonates in FD are not standardized.^[7] Pamidronate was used in the majority of the patients and recent reports suggest the beneficial effects with Zoledronic acid. We used the later drug for its more potent actions and the flexibility of using twice yearly dosing schedule.^[8]

To conclude, we present two interesting cases of craniofacial FD with Raynaud's phenomenon. This unusual combination was not reported earlier in the world literature.

REFERENCES

- Jayaraman M, Karikumar K, Verma A, Modi KD. Alendronate therapy in polyostotic fibrous dysplasia presenting with pathologic fracture. *Am J Orthop (Belle Mead NJ)* 2011;40:E48-50.
- Kumar KH, Dhull P, Bisht Y. Hansen's disease with McCune-Albright syndrome. *J Glob Infect Dis* 2012;4:215-7.
- Goundry B, Bell L, Langtree M, Moorthy A. Diagnosis and management of Raynaud's phenomenon. *BMJ* 2012;344:e289.
- Schwartz J, Rosenfeld V, Stelian J, Wohl Y, Schindler D, Leibovitz A, *et al.* Hyperparathyroidism associated with Raynaud's phenomenon — A case history. *Angiology* 1993;44:731-3.
- Istok R, Czirják L, Lukác J, Stancíková M, Rovenský J. Increased urinary pyridinoline cross-link compounds of collagen in patients with systemic sclerosis and Raynaud's phenomenon. *Rheumatology (Oxford)* 2001;40:140-6.
- Lee JS, FitzGibbon EJ, Chen YR, Kim HJ, Lustig LR, Akintoye SO, *et al.* Clinical guidelines for the management of craniofacial fibrous dysplasia. *Orphanet J Rare Dis* 2012;7 Suppl 1:S2.
- Amit M, Collins MT, FitzGibbon EJ, Butman JA, Fliss DM, Gil Z. Surgery versus watchful waiting in patients with craniofacial fibrous dysplasia — A meta-analysis. *PLoS One* 2011;6:e25179.
- Raj N, Rajan S, Krishnan V. Treatment of fibrous dysplasia with zoledronic acid infusion case report, review of literature and future prospectives. *Webmedcentral Orthop* 2010;1:WMC001206.

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