

# Role of Imaging in A Case of Fibrodysplasia Ossificans Progressiva

RACHIT HARJAI, SUSHIL. G. KACHEWAR

## ABSTRACT

Fibrodysplasia ossificans progressiva is one of the rarest and one of the most debilitating and disabling disorders known to affect the mankind. Short big toes and unrestricted progressive ossification of soft tissues, are its hallmarks. Failure of early diagnosis and erroneous interpretation of

histology has resulted in patients being mistreated with surgical excision and several cycles of radiotherapy. This case report describes a rare case of this bizarre entity which presented to our institute. Typical imaging findings are highlighted that clinch the diagnosis and thus enable one to avoid biopsy which can further hasten this process.

**Keywords:** Big toes, Calcifications; 'Munchmeyer's disease', Paraspinal ossification, Synphalangism

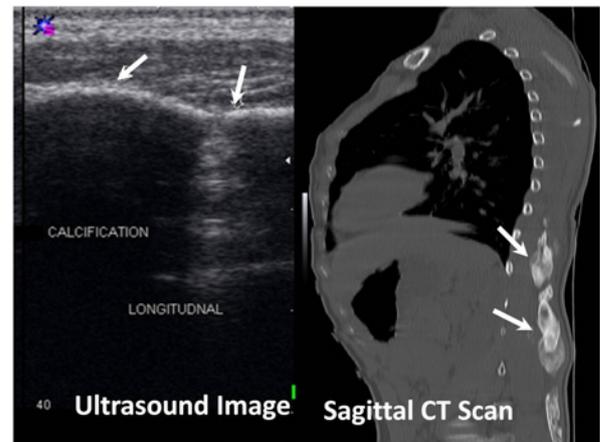
## CASE REPORT

A 15-year-old, presented with multiple swellings over the back which she has been noticing for the past three years. The swellings over the back were insidious in onset, progressive in nature, painless. She had a bowed posture as her spinal movements were restricted. There was no history of any significant trauma. Laboratory investigations were essentially normal. On examination, there were multiple, non-tender, hard (bone like) swellings varying in size from 2 cm to over 15 cm. The swellings had indistinct edges and there were no signs of active inflammation. What was unique in this patient was that the great toes of both feet very significantly and symmetrically short.



**[Table/Fig-1]:** Clinical photograph showing multiple swellings over the back. Plain Radiograph of D-L spine showing ossification in bilateral paravertebral soft tissues

As shown in [Table/Fig-1], clinical photographs revealed multiple swellings over the back. Plain Radiograph of D-L spine showed ossification in bilateral paravertebral soft tissues.



**[Table/Fig-2]:** Ultrasound of the swellings showing hyperechoic foci within it associated with posterior acoustic shadowing suggestive of dense calcification / ossification inside it. A sagittal reformatted CT image better depicts the location and extent of these ossifications

Ultrasound of the swellings [Table/Fig-2], showing hyperechoic foci within it associated with posterior acoustic shadowing suggestive of dense calcification / ossification inside it. These were confirmed in CT scan images. A sagittal reformatted CT image better depicted the location and extent.

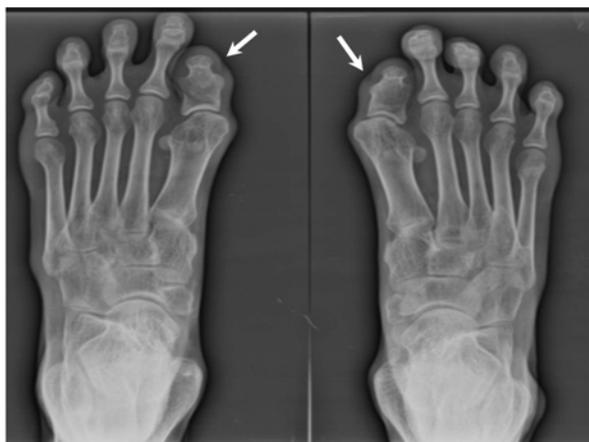
Clinical photograph showed short great toes of both feet [Table/Fig-3] and Plain Radiograph of both feet [Table/Fig-4]

showed the characteristic short great toes of both feet.

As the clinical and imaging features were typical of this entity, the final diagnosis of Fibrodysplasia Ossificans Progressiva (FOP) was made. The patient and her parents were counseled about the nature of the disease. Symptomatic treatment in the form of analgesics only when required was provided.



**[Table/Fig-3]:** Clinical photograph showing short great toes of both feet



**[Table/Fig-4]:** Plain Radiograph of both feet shows the characteristic short great toes of both feet

## DISCUSSION

It has been mentioned that the condition was first described by Guy Patin in 1692 when he wrote about the woman who turned to wood [1]. Von Dusch in 1868 first used the name myositis ossificans progressiva, which means “muscle turns progressively to bone.” Munchmeyer (1869) first gave a comprehensive description of the disease with an account of 12 cases-hence the eponym ‘Munchmeyer’s disease’. The name was officially modified to fibrodysplasia ossificans progressiva in 1970 by Dr. Victor McKusick of Johns Hopkins University, following Bauer and Bode (1940), in order to acknowledge that other soft (or fibrous) tissues in addition to

muscle (for example tendons and ligaments) are replaced by bone [2].

The diagnosis is firmly established based on clinical and radiological findings, which are very distinctive and as a result there are only few differentials. Clinical criteria include male or female, usually presenting in the first decade with acute episodes of swelling (flare ups) usually after insignificant / minor trauma or surgery. The swellings universally start in upper paraspinal muscles and involve the sternocleidomastoid, the shoulder girdle and the pelvic girdle. Sites of primary involvement include the neck (50%), dorsal paraspinal region (30%), head (10%), or limbs (10%) [3,4].

Malformed great toe resulting from shortened first metatarsal and proximal phalanx is a hallmark that can be noticed since birth [5].

As a result of endochondral bone formation causing ankylosed temporo-mandibular joint patient may die of starvation. Similarly when rib cage is affected, the patient dies of respiratory distress [5]. Periodic remissions and exacerbations have been reported.

Typical of the heterotopic bone formation in this entity is its progression scheduled on lines of normal embryonic skeletal formation. Interestingly, muscles of heart, diaphragm, larynx, tongue, sphincter and eyes are spared. Laboratory and haematological work-up is essentially normal.

Microdactyly of the great toe associated with synphalangism and progressive heterotopic ossification are the diagnostic imaging characteristics [6,7]. Sometimes, synostosis, exostoses, broad neck of femur and malformed cervical vertebrae may also be encountered [8].

All patients with classic clinical features of FOP (great toe malformations and progressive heterotopic ossification) are reported to have heterozygous mutation (c.617G4A; p.R206H) in the Glycine and Serine residue (GS) activation domain of activin A type I receptor/activin-like kinase 2 (ACVR1/ALK2), a bone morphogenetic protein (BMP) type I receptor.

Till today, specific genetic or biological markers of this entity have remain to be identified.

With progression, FOP restricts eating and breathing by the patient and is thus fatal. Invasive procedures like muscle biopsy or removal of new bone that are formed, can further hasten the disease process and are therefore contraindicated [9,10]. Definitive treatment for this entity is still elusive, although short term steroids may have some role. Trials of Radiotherapy can have disastrous effects too as they can further hasten the disease process. Hence, counselling plays the most important role because there is autosomal dominant inheritance pattern.

## CONCLUSION

FOP is a rare bizarre entity. Proper knowledge, timely diagnosis, “Non invasive treatment” and counselling play an important role in its management.

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### AUTHOR(S):

- 1. Dr. Rachit Harjai
- 2. Dr. Sushil. G. Kachewar

### PARTICULARS OF CONTRIBUTORS:

- 1. Senior Registrar, Department of Radiodiagnosis, Rural Medical College (RMC), PIMS, Loni, India.
- 2. Professor, Department of Radiodiagnosis, PDVVPF’s Medical College, Viladghat, Ahmednagar, India.

### NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Sushil Kachewar,  
 Professor, Department of Radio-diagnosis, PDVVPF’s  
 Medical College, Viladghat, Ahmednagar, India.  
 E-mail: sushilkachewar@hotmail.com

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