FIBRODYSPLASIA OSSIFICANS PROGRESSIVA- A CASE REPORT

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CLINICAL HISTORY

- A 9-year-old boy comes to pediatric outdoor with HO swelling over the back, which have been present for last 3-4 years, and are gradually increasing in size.
- O/E - Bony prominences were noted on the back
- Prominent 2nd digit of both feet

Fig (A) The photograph shows multiple prominences over the back.

Fig (B) The photograph shows short first toe and long second toe of both feet.

X RAY B/L FEET AP VIEW

X-RAY shows –

- There is microdactyly of first toe of bilateral feet.
- The 2nd toes of bilateral feet show elongated proximal phalange.
- There is Hallux valgus deformity of both feet.
X RAY CHEST PA VIEW

X ray chest and upper abdomen PA view shows columns of bone traversing the right axillary region and left lower chest (probably involving the intercostal muscles, as marked by the arrows in blue.

X-RAY LS SPINE & PELVIS

X-Ray LS spine with pelvis shows bony columns over left lower chest wall with broad femoral neck bilaterally

X-RAY SKULL & HANDS

Fig A

Fig B

Fig (A) X Ray skull shows no apparent bony abnormality.
Fig (B) X Ray BIL hands show micrdactyly of BIL thumbs due to phalangeal shortening
DISCUSSION:

- Fibrodysplasia ossificans progressiva (FOP) is a rare and disabling syndrome, which is characterized by heterotopic ossifications and skeletal deformities.
- So far, around 200 patients with FOP have been reported in the world literature.
- The disorder tends to inherit as an autosomal dominant trait, but the majority of cases are sporadic mutations.
- The age of onset is mostly in the first two decades of life, and no gender preferences have been described.
- C4F-The first clinical feature includes a localized and rapidly progressive swelling, mainly in muscle bundles of the neck, shoulder, and upper arms. The lesions may become warm, erythematous, and tender.

Radiographic Features

After months and even years, other regions, such as pelvis, face, jaws, back, extremities, abdominal wall, and chest will be involved and torticollis and kyphosis may occur. Gradually, muscles are replaced by ossified or calcified tissues, which may be located across joints. This leads to restricted range of motion and even ankylosis in joints, so that the patient would be called a “stone man.” Although the diagnosis of FOP is clinical, it is confirmed by imaging. The imaging features are:

- hallux valgus
- monophalangic first toe
- shortened metacarpals
- pseudoexostoses (ossification of ligamentous insertions)
- Microdactyly of the first metacarpal/metatarsal

DIFFERENTIAL DIAGNOSIS:

- Scleroderma & CREST syndrome
- Juvenile fibromatosis
- Dermatomyositis

REFERENCES:

1. Essentials of skeletal radiology-Yochum & Rowe,vol-2.chapter-8 skeletal dysplasias,P-737
2. Orthopedic Imaging-A practical approach, by Adam Greenspan part VII-congenital and developmental anomalies