

FOP: THE RARE BUT UNMISSABLE DISABLER

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BIO

Dr. Lawrence Owino Okong'o is a paediatrician and paediatric rheumatologist, and a lecturer in the Department of Paediatrics and Child Health, University of Nairobi, Kenya. He is the immediate past National Chairman of the Kenya Paediatrics Association. He acquired his Master of Medicine in Paediatrics and Child Health (2007) from the University of Nairobi, and Master of Philosophy in Paediatric rheumatology (2015) from the University of Cape Town, as well as Fellowship Certificate in Paediatric Rheumatology from the CMSA (2015).



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INTRODUCTION 1: FOP

- Fibrodysplasia ossificans progressiva (FOP) is an **autosomal dominant disorder of bone formation**.
- The disease is caused by an **activating mutation in the ACVR1**, a bone morphogenetic protein type I receptor, which promotes osteogenic differentiation of connective tissue progenitor cells.
- Most cases of FOP arise from spontaneous new mutation.
- A few cases are genetically transmitted, in an autosomal dominant pattern (can be inherited from either mothers or fathers).

EPIDEMIOLOGY

- The prevalence of FOP is **1 in 2 million individuals**.
- There is **no ethnic, racial, gender, or geographic predisposition**.

INTRODUCTION 2: Clinical features

- Malformations of the great toes evident at birth (even in utero)
 - Episodic flares and
 - progressive heterotopic endochondral ossification (HEO).
- Heterotopic bone replace skeletal muscles and connective tissues leading to an armament-like encasement of bone and permanent immobility.



INTRODUCTION 3: Triggers of flares

- **Minor trauma such as**
 - intramuscular immunizations,
 - mandibular blocks for dental work,
 - muscle fatigue,
 - blunt muscle trauma from bumps, bruises, falls, or
 - influenza-like viral illnesses can trigger painful new flare-ups of FOP leading to progressive heterotopic ossification
- **Attempts to surgically remove the heterotopic bone risk** provoking explosive and painful new bone growth.



CASE PRESENTATION

INTRODUCTION

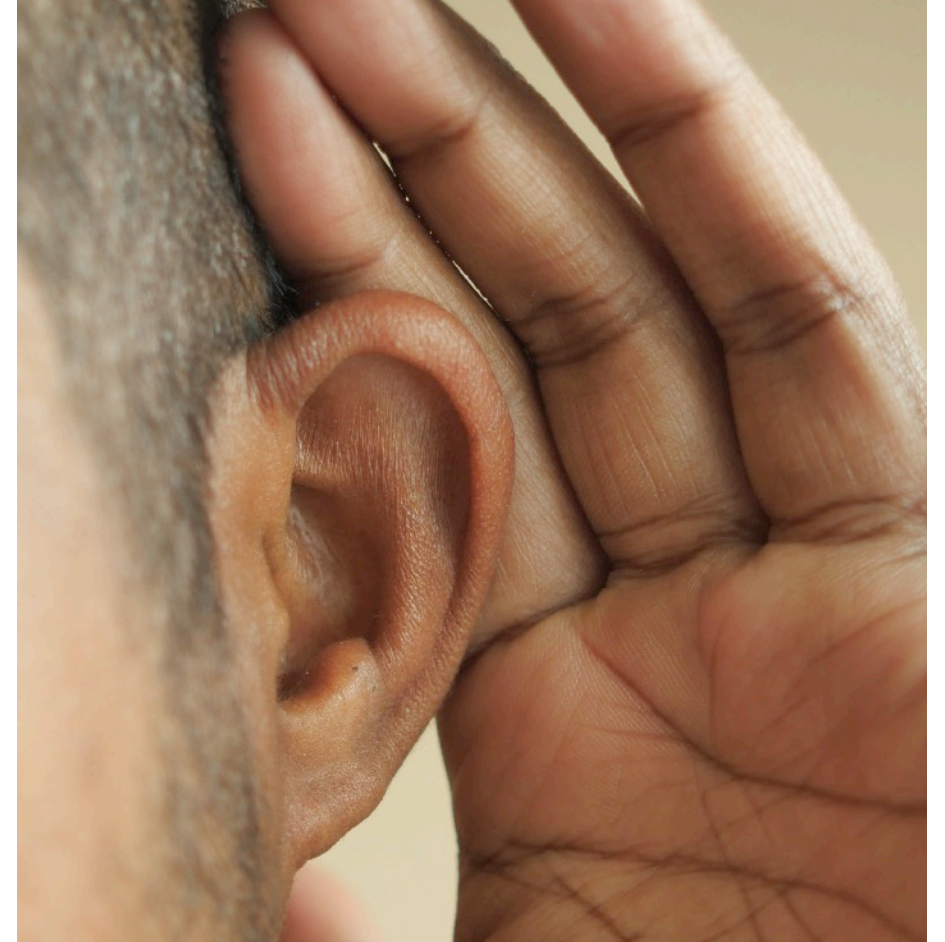
AG, 26 years old.

- History of firm **swellings on the back** and at sites of trauma from early childhood.
- Family noted **abnormal large toes** from birth but didn't know the significance of the same.
- Similarly, noted recurrent firm swellings at trauma sites and assumed these were just normal swellings from trauma associated with play especially since this was a boy.
- The **swellings**, however, **did not go away** as expected though it never occurred to the family that these were signifying a serious chronic disease.



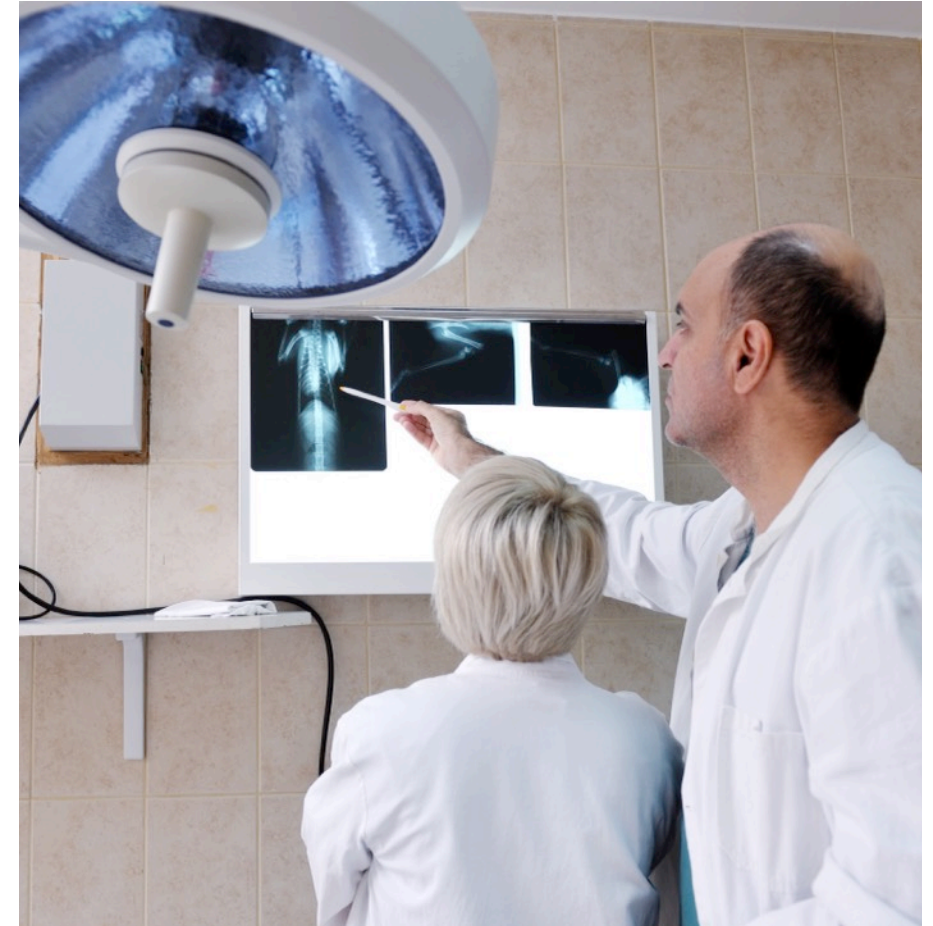
SCHOOL

- He enrolled in a **normal school** but from the age of 6 years, was noted to have challenges with hearing.
- **Hearing impairment worsened** and he was transferred to a school for the deaf in class 7 and 8 (age 12-14 years).
- Subsequently, he attended high school in a school for the hearing impaired completing his high school education in 2016.
- He learnt sign language as part of the subjects for his high school.
- His final exam performance was below average.



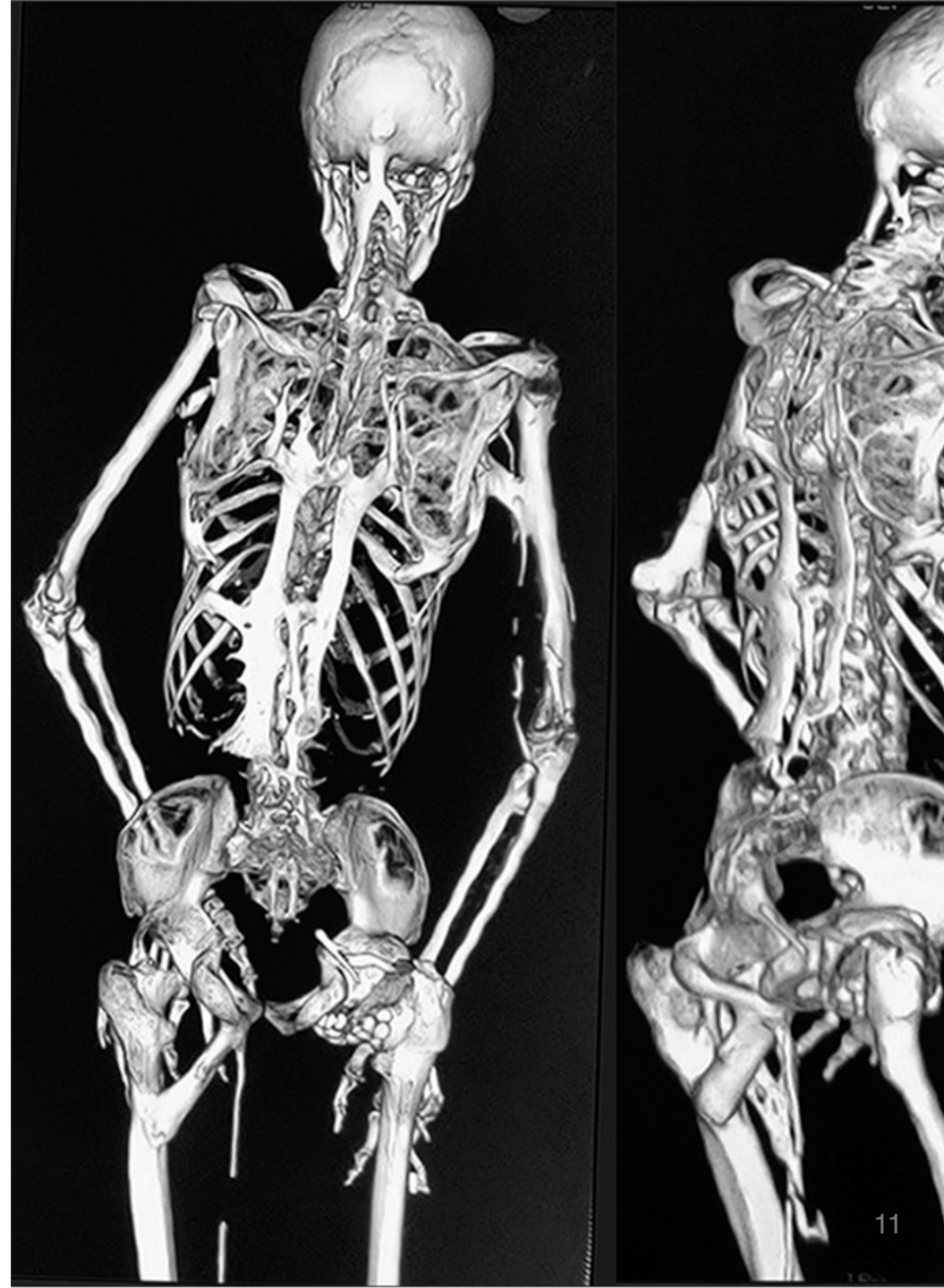
THE FANTASTIC RADIOGRAPHER

- While at the school that he was referred to a mission hospital in Kenya that is known for its orthopedic service for assessment for
 - abnormal bony growths as well as for
 - the possible cause of his hearing impairment.
- It was during this that a keen radiologist came up with a diagnosis of FOP.



THE ALIEN DISEASE OR DISEASE OF ALIENS

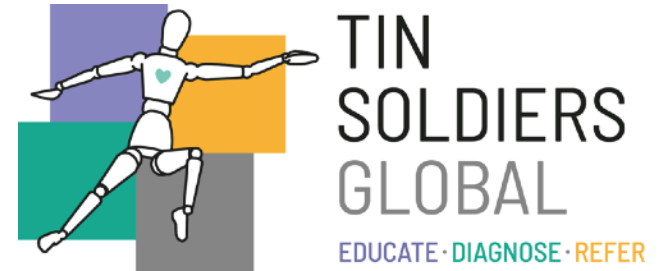
- The FOP champion teacher narrated the shock and disbelief of the radiologist who is reported to have quipped thus:
- “had it not been for the fact that the patient was right there on the Xray table in front of me, I would have thought he was an alien”.
- This was way back in 2015, and the patient was approximately 16 years of age.



COMMUNITY LINKAGE:

Joining the army

- After this, a teacher at the school took interest in the case and reached out to the FOP community in 2015.
- We lost contact, and it was difficult to trace the patient since he had completed high school and went back home, and no one was sure where exactly home was.



LOST AND FOUND:

The march to find missing tin soldiers

- In 2024, we were supported by Tin soldiers to search for the lost patients in Kenya;
- With the help of the teacher FOP champion, we were eventually able trace the patient from their home in the remote arid northern part of Kenya, some 550KM from Nairobi.
- The patient is under the care of the elder sister who has been linked to global FOP community.



THE UNFORTUNATE OCCURRENCE:

The good but harmful intention?



- Unfortunately, in an attempt to improve mobility and reduce pain in the left hip that had bothered the patient, he underwent hip replacement surgery in June 2023 before we got to him.



Needless to say, this has not improved mobility and instead triggered more profuse heterotopic bone formation at the surgery site.

CURRENT CONDITION 1

Activities of daily living

- He walks with a limp and some difficulty
- He is unable to sit on a low chair.
- Ever wondered if “**pooping**” would be a big deal?
- The family have made for him a high stool where he is able sit while “almost standing”.

CURRENT CONDITION 3

When hearing and inability to enjoy your favourite dish become issues



He is unable to **open the mouth wide** enough to eat and has to select the foods that are easy to bite and chew.



He has **hearing impairment** and would like assistance with hearing aids.

CURRENT CONDITION 3

Wounded soldier

- In June 2024, the patient developed painful ulcer around the medial malleoli of the legs.
- The ulcer has been recalcitrant, not responding to the care that the patient is able to access locally including antibiotics.
- Sometimes, the ulcer heals then recurs.
- Currently, has an ulcer on the right and measures close to 5cm in diameter.



CURRENT CONDITION 3

The recurring ulcer



TRAUMA? Once beaten twice shy.

- The patient reached out for help.
- Due to the chronicity of the ulcers, a diagnosis of **malignancy** (squamous cell carcinoma) was entertained.
- There was **suggestion for biopsy**, but this possibility was put aside due to the ulcer characteristics that were not suggestive of malignancy and so biopsy was avoided.
- We recommended **treatment with boric water 3%** in compresses twice per day followed collagenase cream with antibiotic.
- The patient however was not able to get this, partly due to availability at their remote location and partly due to cost.
- The ulcers remain.



QUESTIONS

SUMMARY

- In summary, we present the case of AG, now 26 years old diagnosed with FOP by a radiologist and linked to the FOP community.
- Due to remoteness of his location, has not had physical assessment someone with some knowledge of FOP.
- He has hearing impairment, trouble with activities of daily living, inability to sit, impaired locomotion, impaired jaw opening and bothersome chronic recurrent leg ulcer.
- He is one of four “soldiers” known to exist in Kenya.



THANK YOU



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