BILATERAL TIBIA FRACTURE IN A CASE OF OSTEOPETROSIS

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- A 22 year-old male patient reported with the chief complaint of pain and swelling in bilateral lower limbs with deformity since last 3 days after an incidence of fall
- Patient has a past history of fracture of b/l tibia 3 years back and was treated conservatively .

General examination revealed a short stature, thin built with normal skin

On extraoral examination hypertelorism, depressed nasal bridge ,broad face,frontal bossing were noted

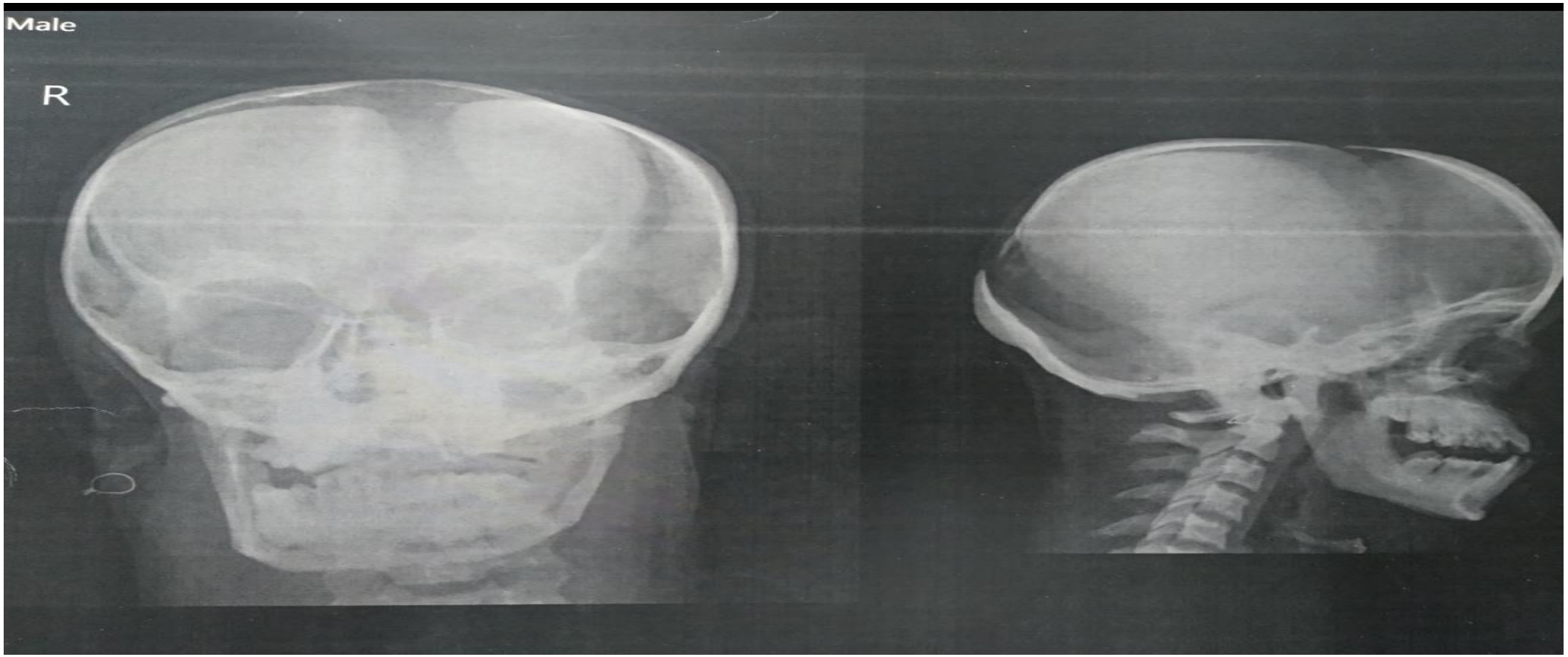
On local examination there was anterior bowing of b/l tibia

Loss of medullary cavity was noted in femur and tibia full length ap lateral views

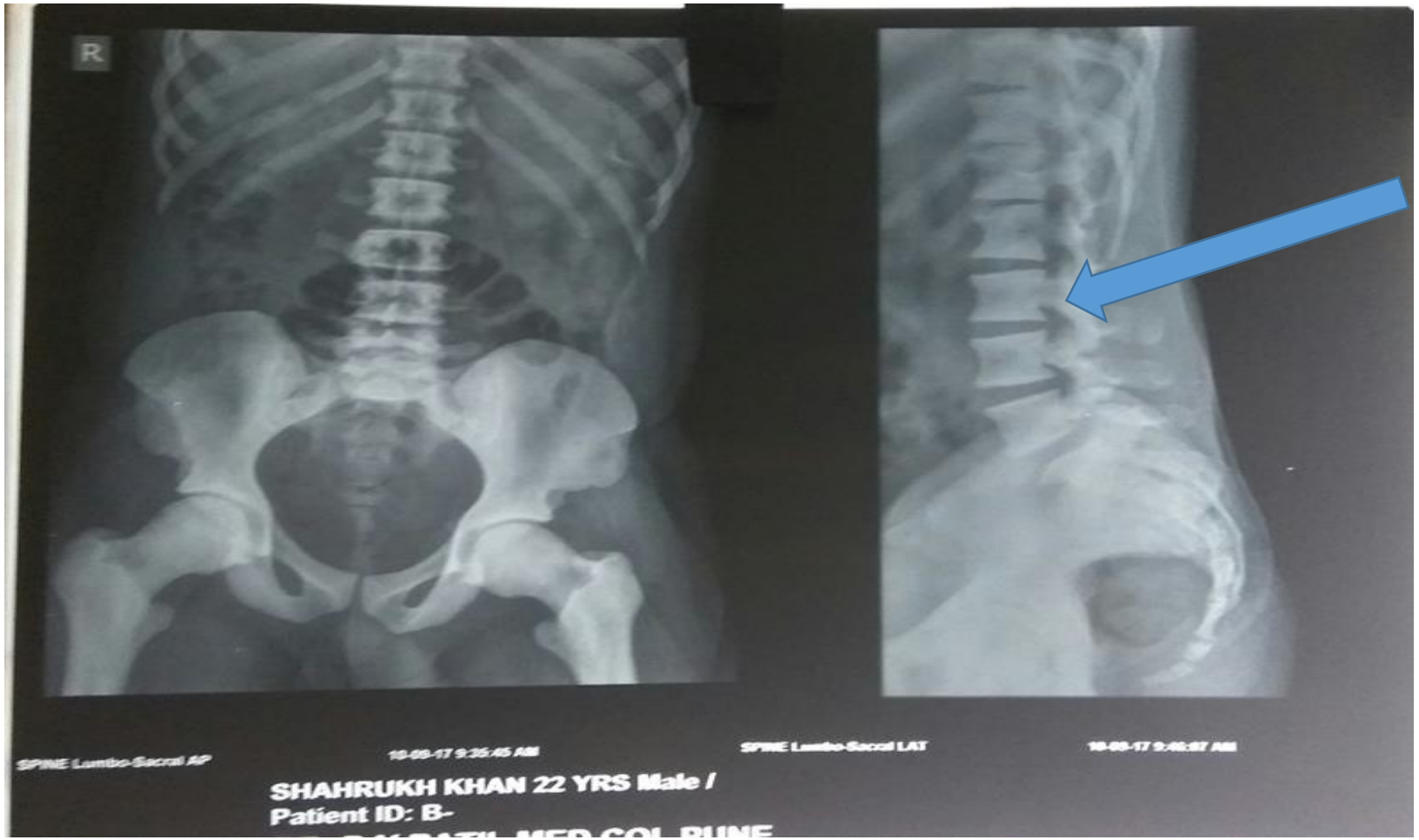
Serum calcium was 8.9mg/dL, and serum phosphorus was 3.0mg/Dl VITAMIN D level was 18.45 ng/ml.

CLINICAL PHOTOS

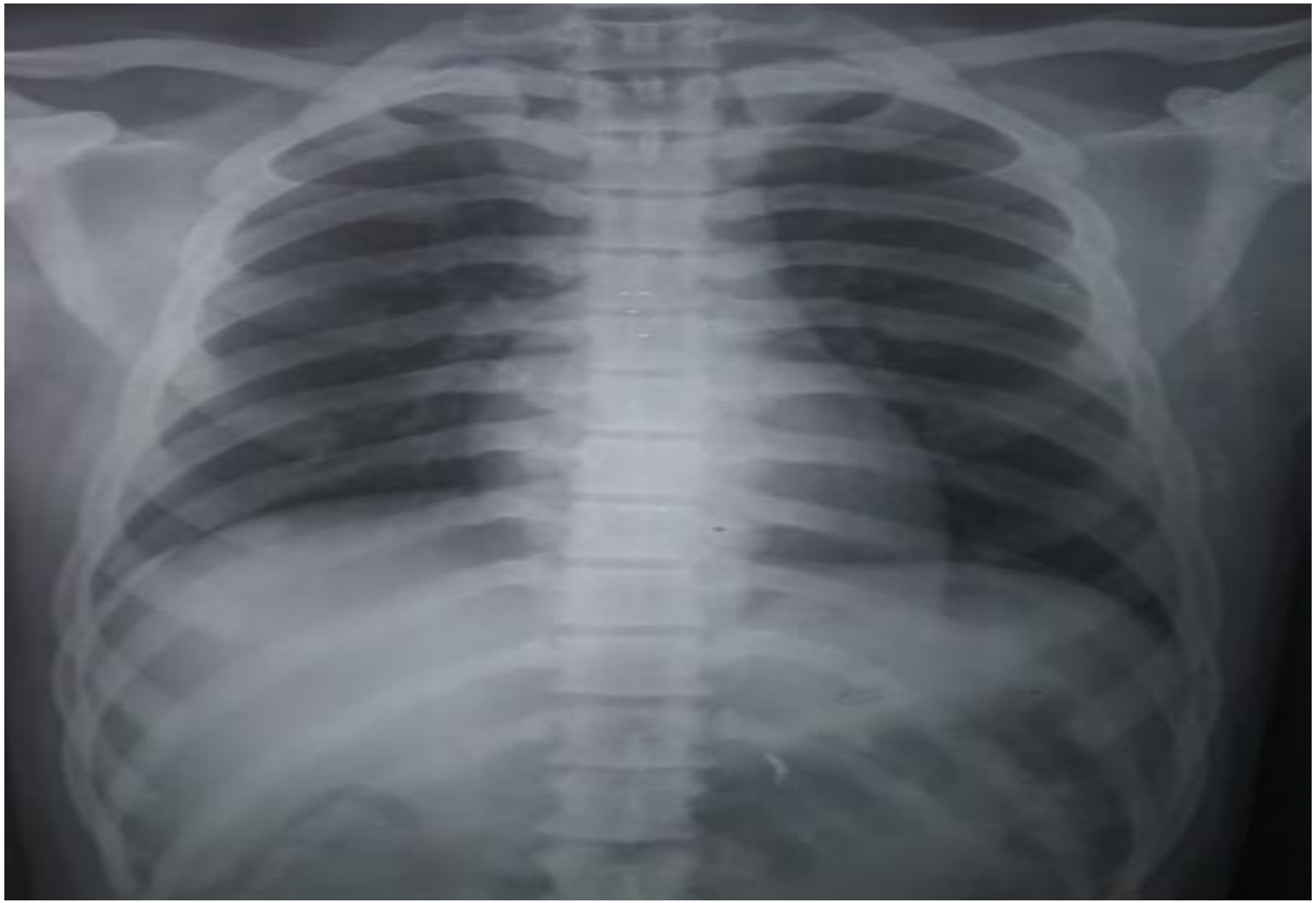
CRANIOSYNOSTOSIS



Anteroposterior view lumbar spine showing rugger jersey spine



CHEST XRAY SHOWING EXCESSIVE CALCIFICATION OF RIBS



Loss of medullary cavity in long bone

PRE OP XRAY

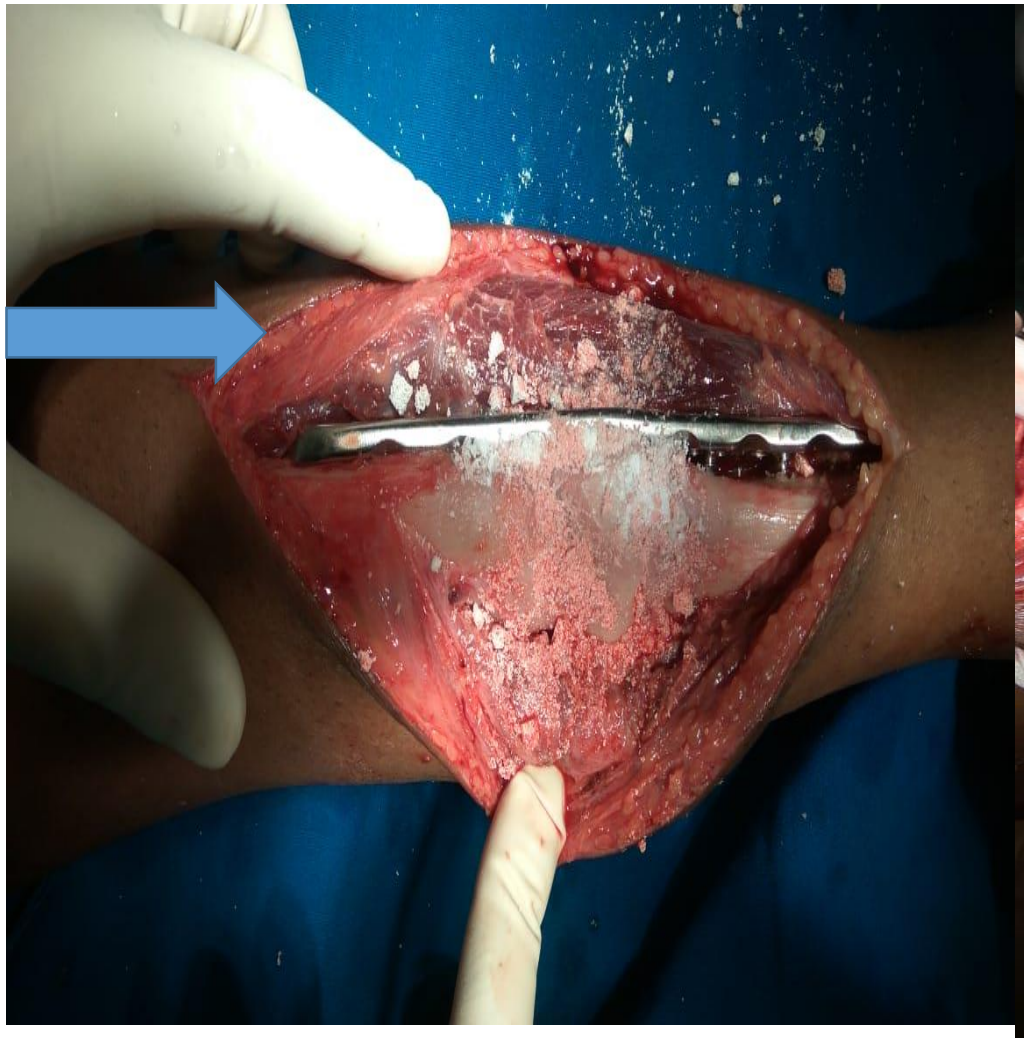
PLAN OF SURGERY:-CORRECTIVE OSTEOTOMY FOR DEFORMITY CORRECTION WITH PLATING

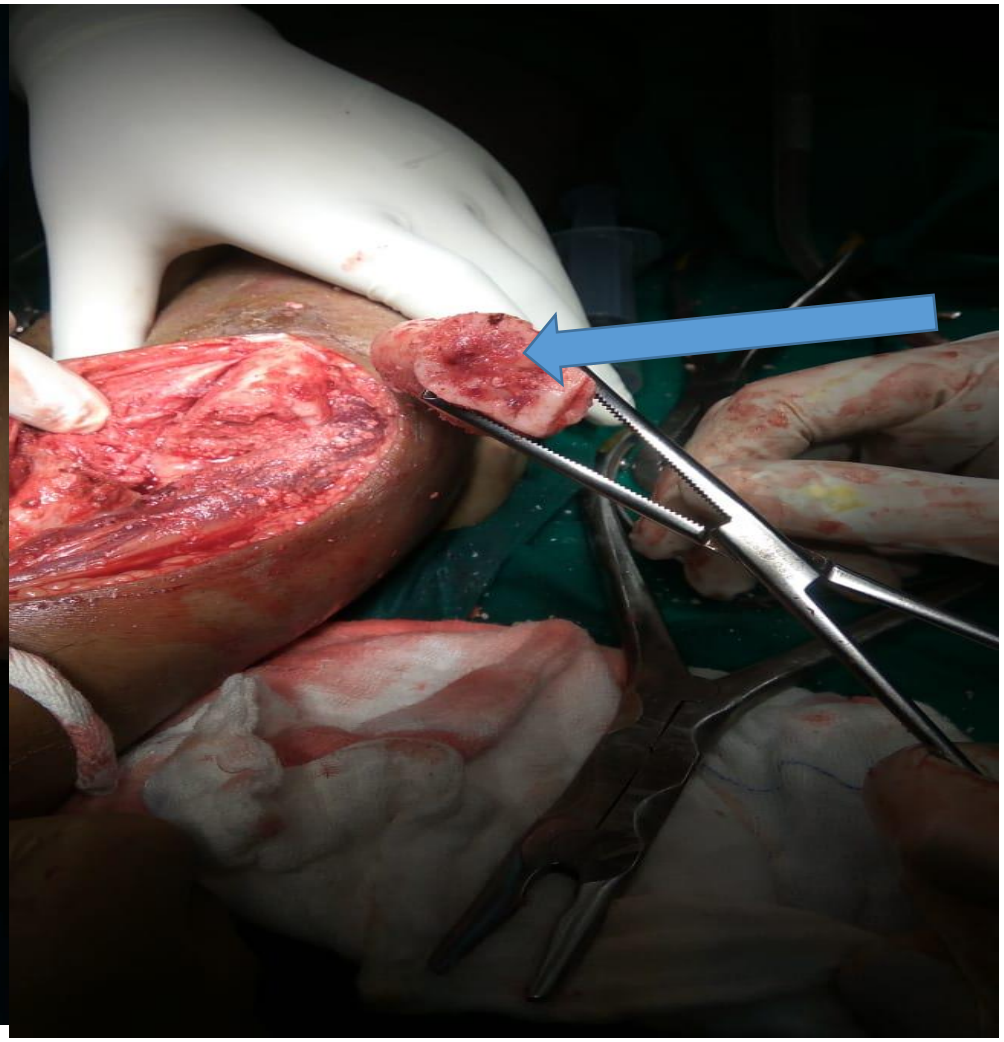
INTRA OP PICTURES

Marble bone appearance

Osteotomy of tibia

Deformity correction with plating was done





Marrow in medullary cavity with lacunae surrounded
By lamellae

AGE:-22 SEX:M

IPD NUMBER:- 54620 OPD NUMBER:- 45155

HISTORY HPE OF BONE

GROSS EXAMINATION:-

SPECIMEN OF SIZE :-3*2.2*1.6 CM

GREYISH WHITE

FIRM TO HARD IN CONSISTENCY

MICROSPCOPIC EXAMINATION:-

SECTIONS SHOWS LAMELLAR BONE WITH CONCENTRIC LAMELLAE

YELLOW MARROW IS SEEN AT PLACES IN MEDULLARY CAVITY

OSTEOBLAST ARE DEFICIENT

NO CARTILAGENOUS AREA SEEN

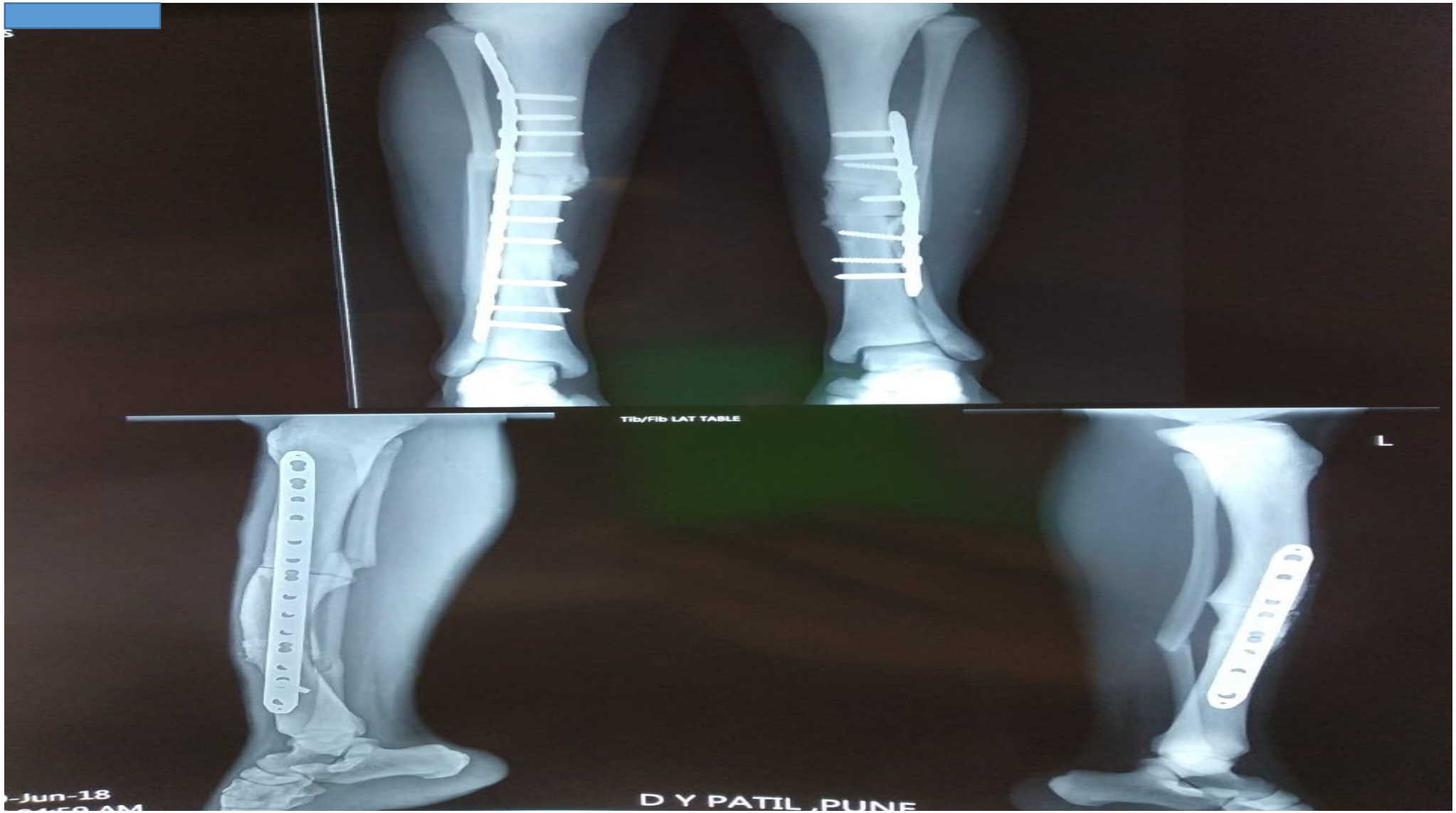
POST OP XRAYS

1st MONTH POST OP

2nd MONTH POST OP OF LEFT SIDE

6th MONTH POST OP OF LEFT SIDE AND 4TH MONTH POST OP OF RIGHT SIDE

9th MONTH POST OP



Physiotherapy

- Partial weight bearing was started at 1 month on bed as the pt couldn’t be started to stand up and walk due to bilateral condition
- The patient was made to lie down on bed and was allowed to put his body weight against a weight machine on a wall with increasing weight by 10 kgs per 7-10 days gradually
- Weight bearing walking with support was started at 8^{th} month post operative
- Full weight bearing was started at 9^{th} month and currently the pt is walking without support

Follow up

- Pt is to be called for regular follow up to see for bone growth and healing and pathological fractures

CHALLENGES IN SURGERY

- Intra medullary nailing was difficult because of absent medullary cavity
- Difficulty to penetrate bone due to hardness of bone
- Care was taken to prevent bit breakage while drilling and inserting implant

Post operative challenges

- Refracture
- Delayed union
- Non union
- High incidence of implant removal failure
- Secondary fractures during implant removal
- Delayed post operative wound healing

DISCUSSION

- The term osteopetrosis is derived from the Greek word “osteo” meaning bone and “petros” meaning stone
- Osteopetrosis is referred to as “marble bone disease” and “Albers Schonberg disease”
- Osteopetrosis has hallmark of increased bone density on radiographs
- The increase in bone density results from abnormalities in osteoclast differentiation and function

- Defects in different genes have been described that lead to a phenotype and mutations in atleast 10 genes have been identified
- These defects include mutations in the gene encoding carbonic anhydrase II, the proton pump gene, and the chloride channel gene

Osteopetrosis has been reported in three clinical forms:

- (1) malignant infantile form with poor prognosis and autosomal recessive inheritance, incidence rate of 1 in 250,000 births
- (2) benign/adult osteopetrosis with autosomal dominant inheritance and with fewer symptoms, incidence rate of 1 in 20,000 live births.
- (3)autosomal recessive intermediate form with clinical manifestations similar to malignant form and lowest incidence rate

Presenting features

- Stunted growth , deformity, and increased likelihood of fractures
- Anemia, recurrent infections, and hepatosplenomegaly due to bone expansion leading to bone marrow narrowing and extramedullary hematopoiesis
- Blindness, facial paralysis, and deafness, due to the increased pressure put on the nerves by the extra bone
- Abnormal cortical bone morphology
- Abnormal form of the vertebral bodies
- Abnormality of temperature regulation
- Abnormality of the ribs
- Abnormality of vertebral epiphysis morphology
- Bone pain
- Cranial nerve paralysis
- Craniosynostosis
- Hearing impairment
- Hypocalcemia

Treatment

- Hematopoietic stem cell transplantation which help in providing cells from which osteoclasts can develop.
- Vitamin D to stimulate dormant osteoclasts, which stimulates bone resorption
- Gamma interferon it improves white blood cell function (leading to fewer infections), decreases bone volume, and increases bone marrow volume.
- Erythropoietin can be used for anemia
- Corticosteroids can be used to stimulate bone resorption.

THANK YOU