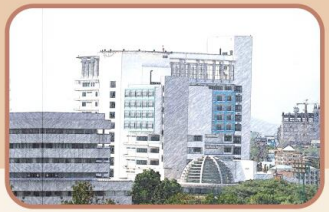


Interhospital conference

ผู้ป่วยหญิงไทยโสด อายุ 31 ปี ไม่ได้ประกอบอาชีพ
ภูมิลำเนา จ.นครศรีธรรมราช

Chief complaint : ปวดต้นขาซ้าย 4 วัน ก่อนมาร.พ.

Present illness:

4 months “ปวดตามเส้น” บริเวณต้นขาซ้าย

4 days ปวดต้นขาซ้ายมากขึ้น ให้หมอบ้านบีบนวด จากนั้นเจ็บต้นขาซ้ายมากขึ้นทันที เดินไม่ไหว จึงมาร.พ.

- ไม่ดื่มเหล้า ไม่สูบบุหรี่
- ปฏิเสธยาที่ใช้ประจำ
- เคยผ่าตัดบริเวณกรามด้านซ้ายเนื่องจากก้อนที่ทำให้หน้าผิดรูปเมื่อ อายุ 20 ปี
- เคยแขนซ้ายหักจากอุบัติเหตุทางจราจร อายุน้อยกว่า 5 ปี
- ประวัติอื่นๆ unremarkable

Physical Examination

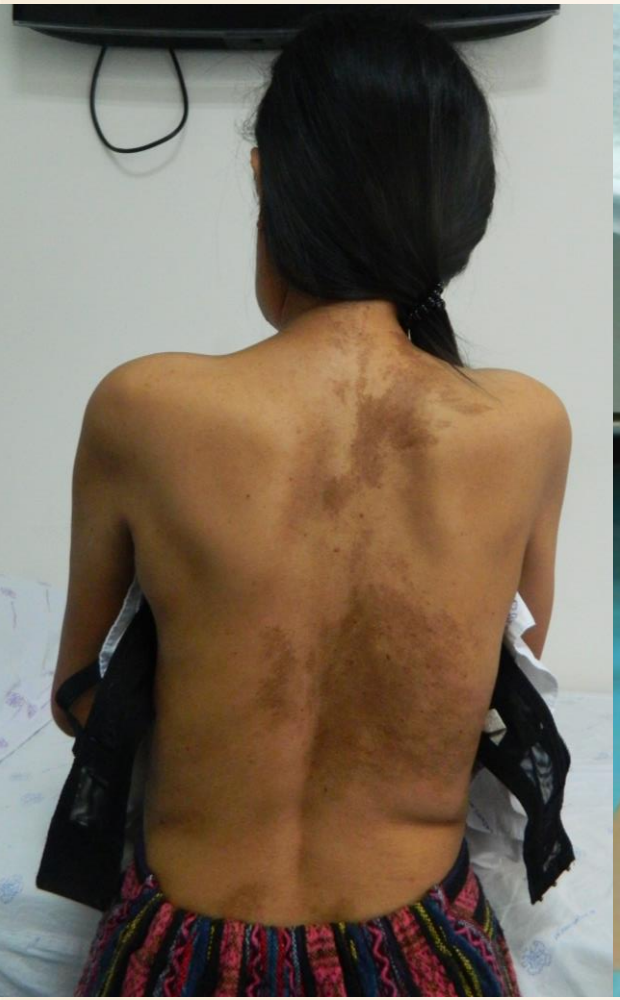
- Vital signs :BT 36.5 C BP 120/75 mmHg HR 110 bpm RR 24/min
- Weight and height can’t be measured due to left leg pain
- Good consciousness
- HEENT : not pale, no jaundice, abnormal facial feature [as pictures]

- Thyroid : not enlarged, size 15 g
- Heart : normal S1S2, no murmur
- Lungs : normal breath sound both lungs, no adventitious sound
- Abdomen : soft, not tender, no mass,no hepatosplenomegaly
- Skeleton
  - Kyphoscoliosis
  - Lt humerus : mild swelling, not tender
  - Extremities : Lt thigh : mild swelling, markedly tender at mid thigh, shortening with internal rotation of left leg

Problem list

Would you like more information ???

Skin : as pictures



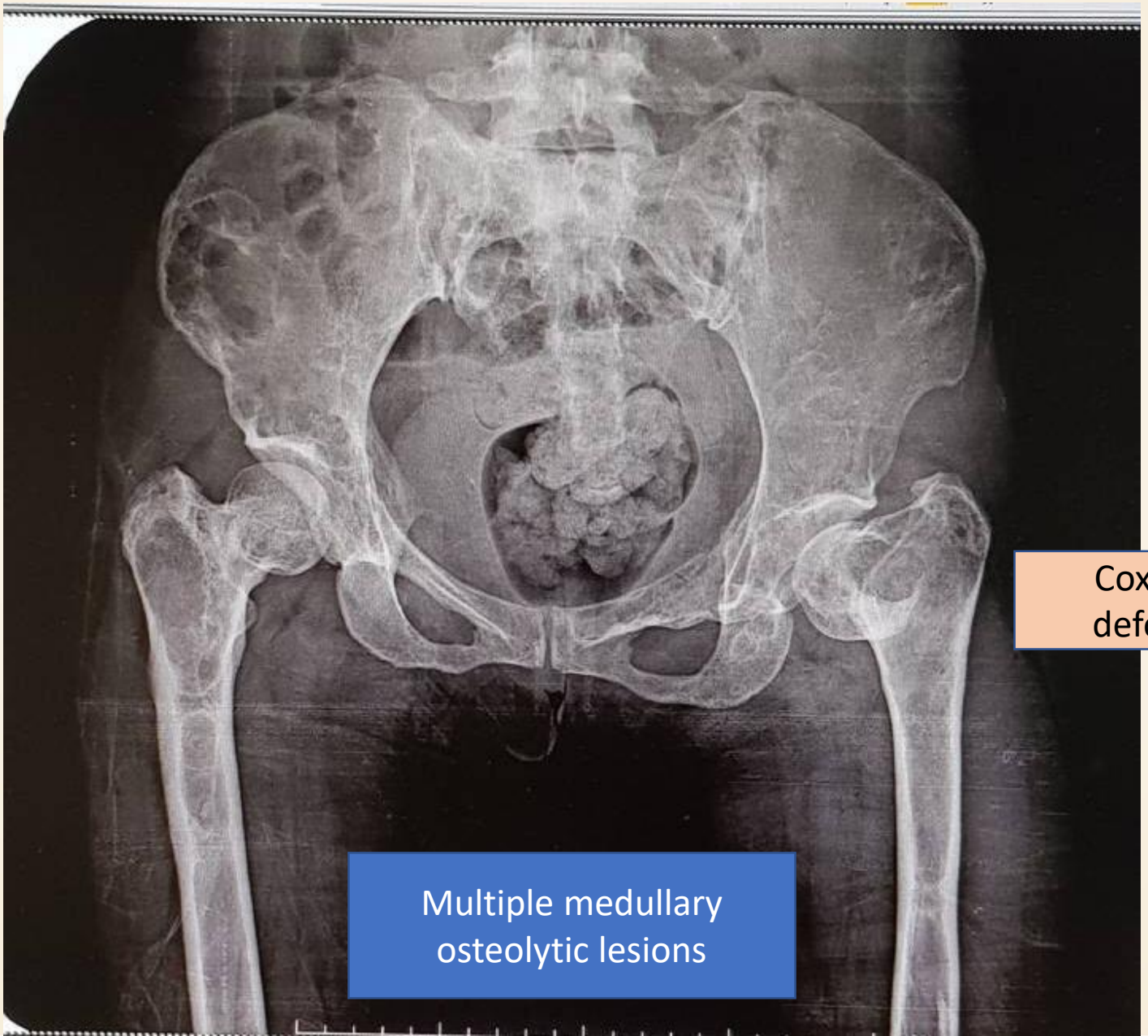
Problem list

1. Limping and shortening of left leg
2. Mass at left mandible
3. Skin lesions

Investigation

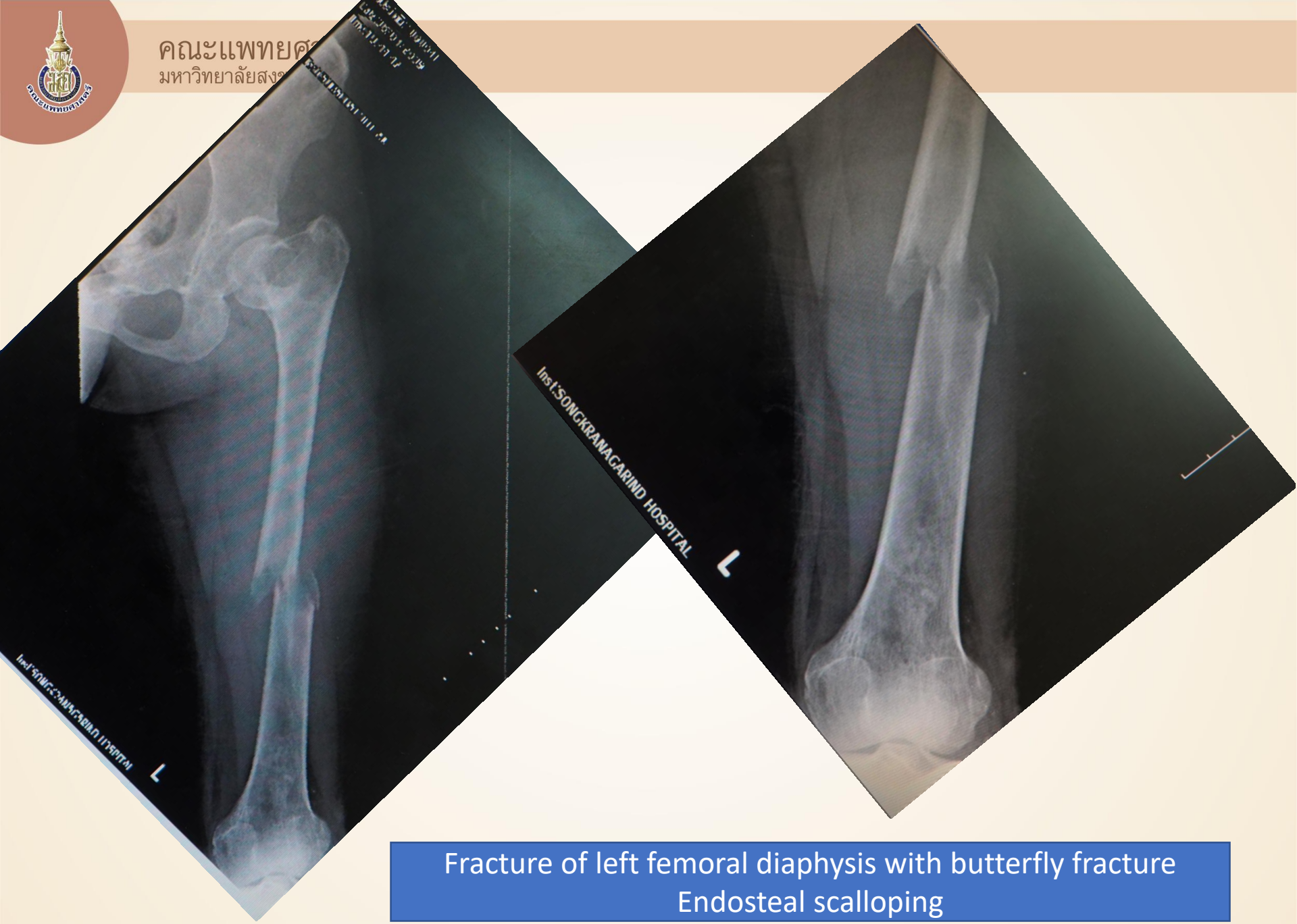
- CBC : Hct 41.4% Hb 14.2 MCV 86.4
  WBC 10260 N 65.9% plt 221000
- BUN 9.3 Cr 0.39
- Na 134.9 K 3.52 Cl 103.8 HCO3 22.7
- Ca 9.1 mg/dl ,PO4 3.2 mg/dl
- ALP 601 [39-117 U/L]
- UA [20/4/52] : Sp.gr 1.025 pH 6.5 glucose: neg ketone 1+ blood : neg , protein : neg , no cell

- FT4 1.56 [0.7-1.75 ng/dl]
- TSH 0.026 - 1.32 [0.25-4 mIU/L]
- Morning Cortisol 17.28 mcg/dl
- Prolactin 15.5 ng/ml
- PTH 36.79 [11-62 pg/ml]



Coxa vara deformity

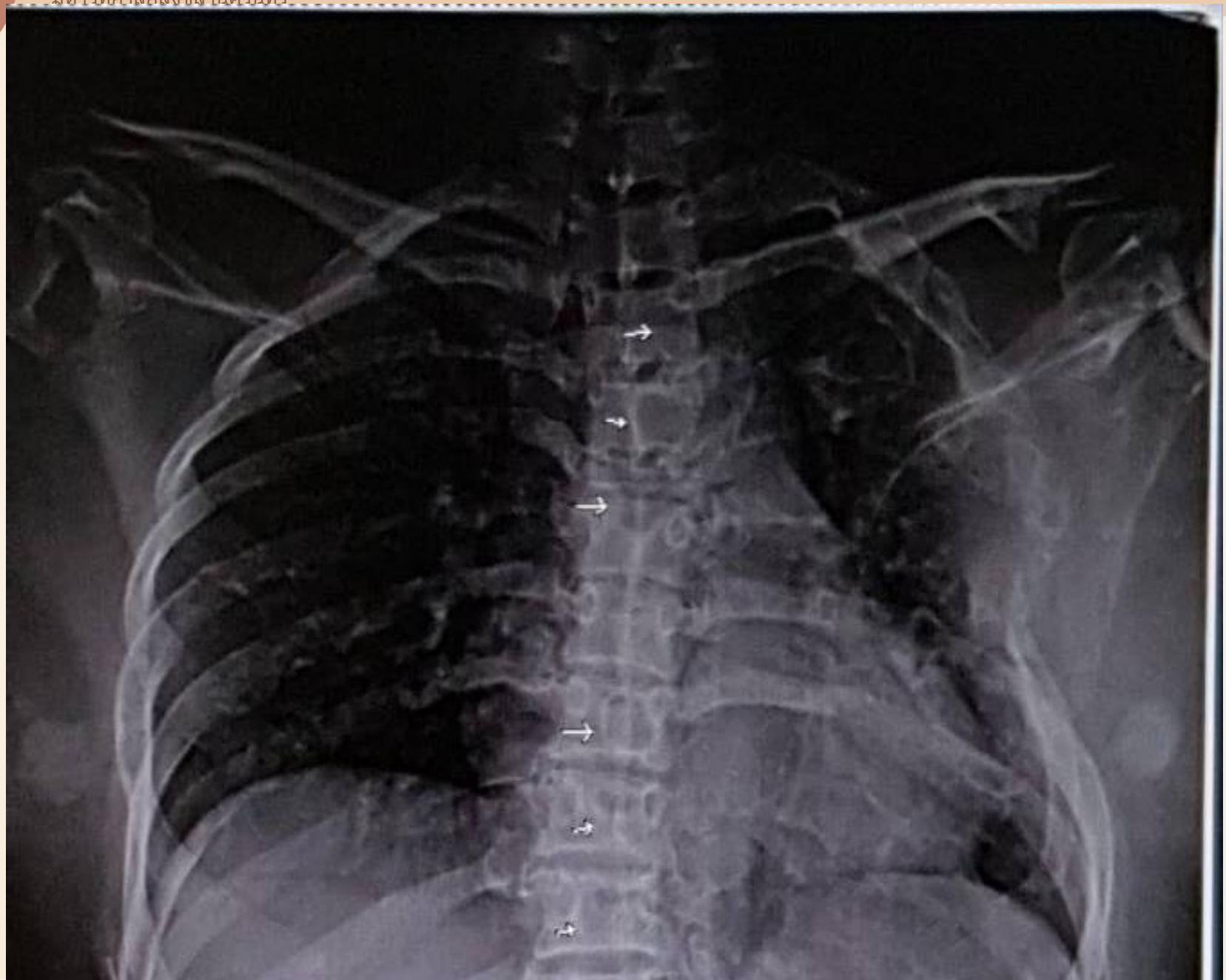
Endosteal scalloping



Fracture of left femoral diaphysis with butterfly fracture
Endosteal scalloping


R


R
cm



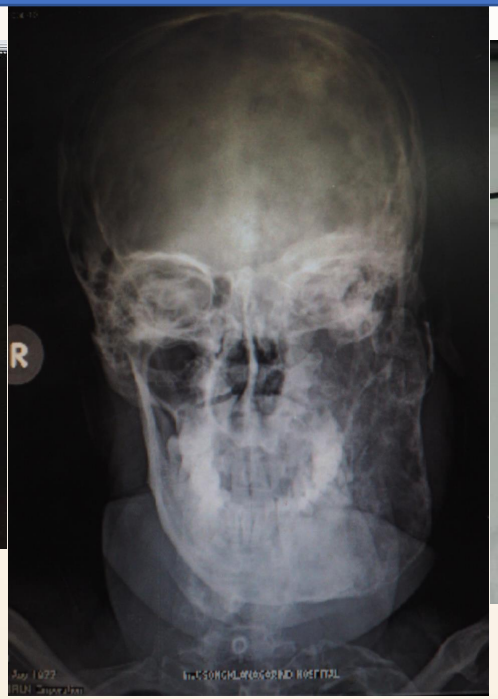
Front

Back

Problem list

1. Limping and shortening of left leg : fracture of left femur
2. Mass at left mandible with multiple osteolytic lesions
3. Skin lesions : café -au- lait

McCune-Albright syndrome (MAS)



Differential diagnosis of slow progressive mass-like bone lesion

- Fibrous dysplasia
- Hyperparathyroidism [brown tumor or Jaw tumor]

Hyperparathyroidism
brown tumor
(osteitis fibrous cystica)

Fibrous dysplasia

McCune-Albright syndrome (MAS)

- estimated prevalence : 1/ 100,000 - 1/1,000,000
- The triad
  - polyostotic fibrous dysplasia of bone(FD)
  - café-au-lait skin pigmentation
  - Endocrinopathies : hyperthyroidism, growth hormone (GH) excess, precocious puberty (PP), Cushing syndrome
- renal phosphate wasting

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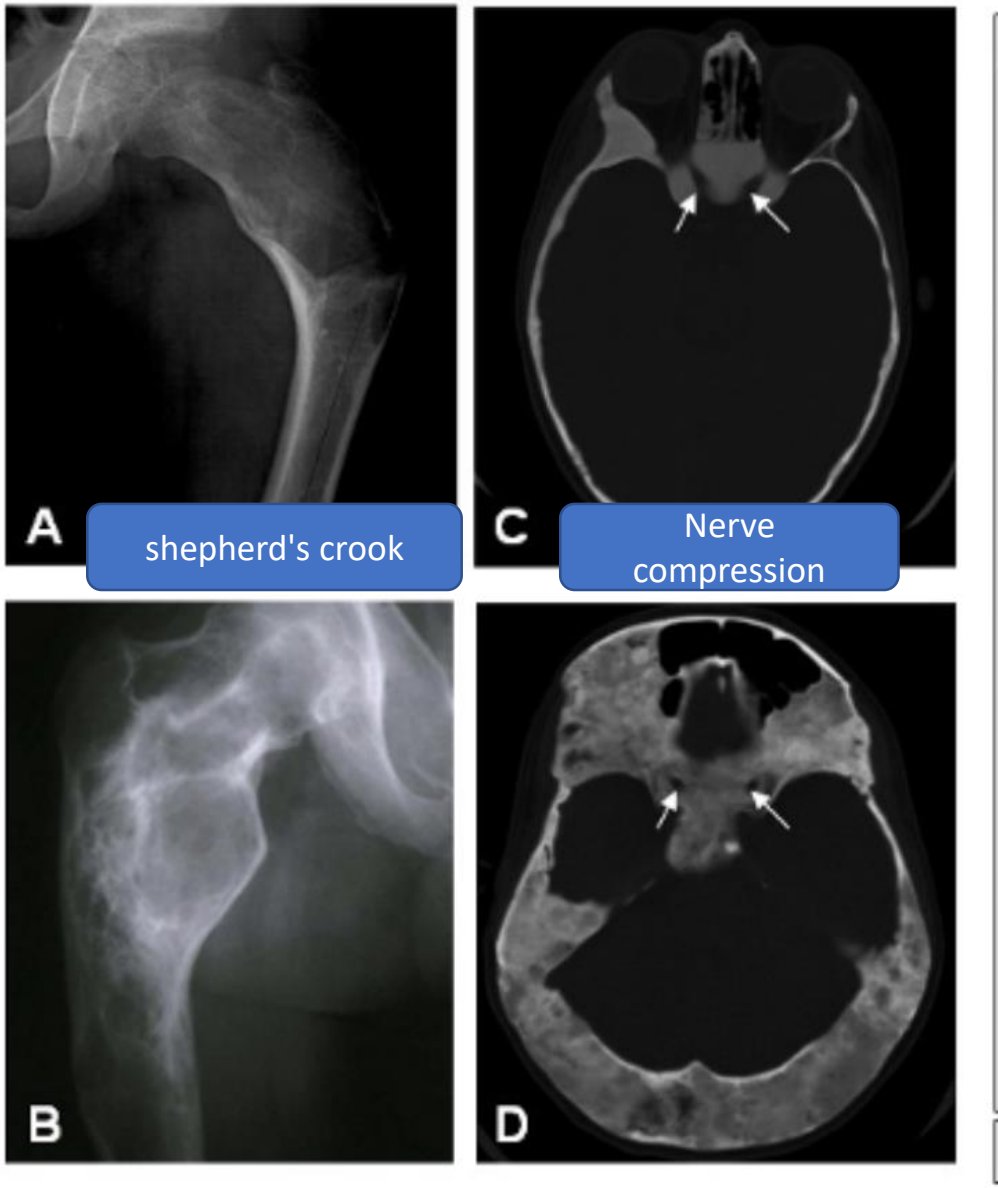
- Fibrous dysplasia[FD]
  - limb deformity and/or pain
  - pathologic fracture
  - craniofacial bones : painless "lump" or facial asymmetry
  - shepherd's crook (coxa vara deformity)
  - Nerve compression
  - most common : proximal femur and skull base



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Café-au-lait spots

McCune Albright Syndrome [MAS]
Coast of Maine : irregular border

Neurofibromatosis
Coast of California : smooth/regular border

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Endocrinopathies

Pituitary	GH and prolactin excess (21%)
Thyroid	Hyperthyroidism : common (38%)
Adrenal	Cushing syndrome can occur in the neonatal period
Gonads	Central precoccious puberty

Renal phosphate wasting

- FGF23 (by FD tissue)

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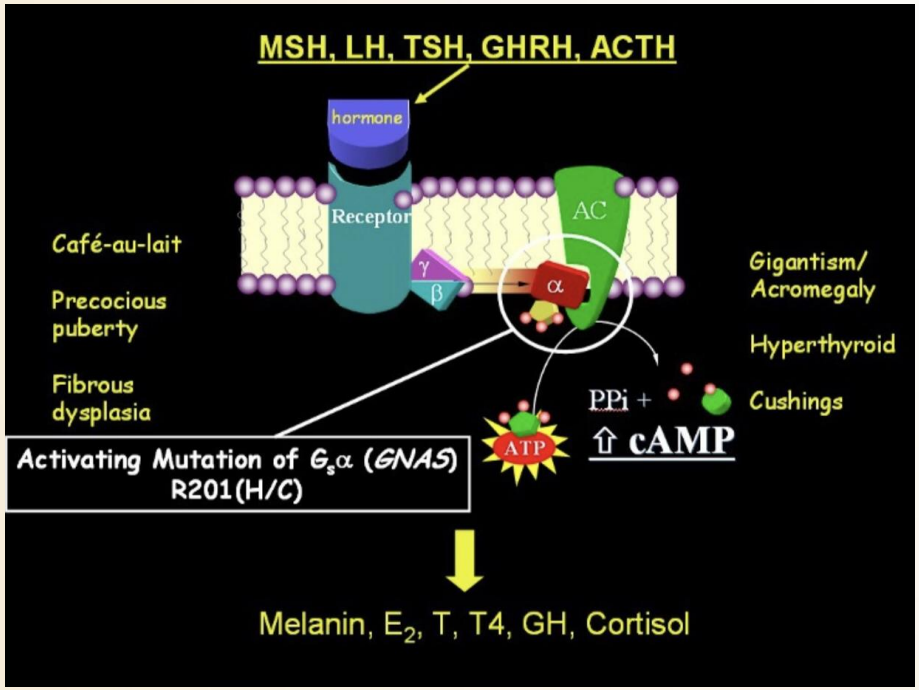
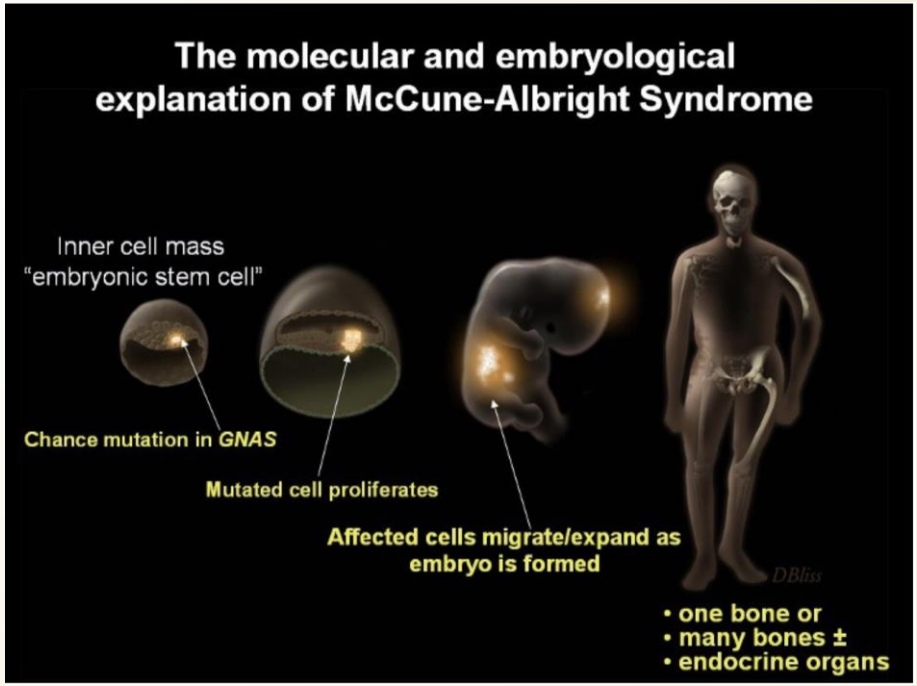
Etiology

- Postzygotic mutations in the Gsα protein (encoded by the GNAS gene)
- Long (q) arm of chromosome 20 at position 13.3





GNAS = stimulatory alpha subunit, guanine nucleotide-binding protein (G protein)

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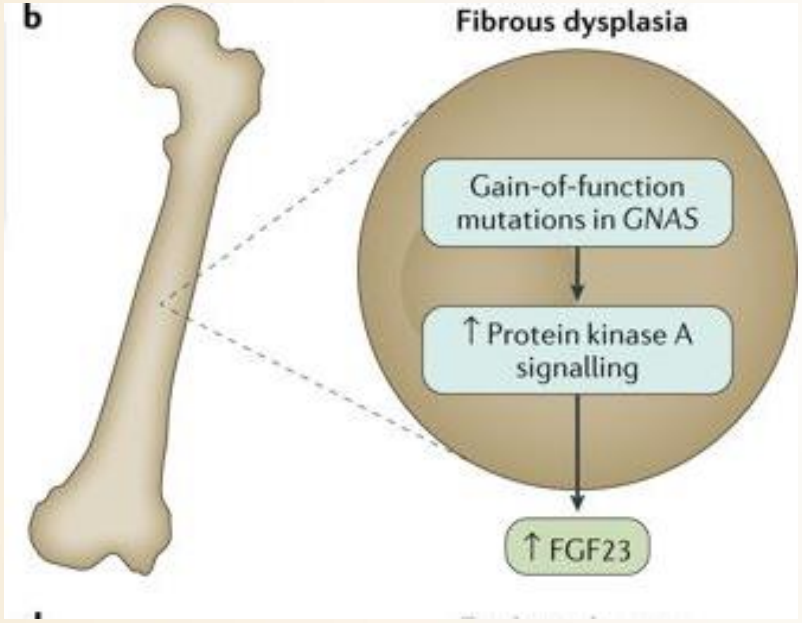

Hypophosphatemia

-overproduction of “**FGF23**” by abnormal skeletal progenitor cells

*Nature Reviews Disease Primers***volume3**, Article number: 17101 (2017)

Management

Genetic counseling : no vertical transmission of the disease

Craniofacial FD
Observation - annual vision and hearing testing - imaging => CT of skull and/or mandible (MRI :the optic nerves)
Indications for surgery : -cranial nerve compression -Severe disfigurement
Bisphosphonates may relieve pain

Axial and appendicular skeleton
The surgical management : Intramedullary device [preferable]
Bisphosphonates :relieve bone pain
strengthening exercises and strength maintenance (Swimming and cycling)

Condition	Management
GH excess (associated with vision and hearing loss, and macrocephaly) (20%)	-Treatment options : medications, surgery, and radiation -long-acting somatostatin analogues -GH receptor antagonist(pegvisomant)
GH secreting tumors co-secrete prolactin	dopamine agonists (cabergoline, etc.)
Hyperthyroidism	Anti-thyroidal drugs
Precocious puberty	-long-acting GnRH analogues -Aromatase inhibitors -Ultrasound screen and follow testicular masses
Cushing syndrome (rarest)	Adrenalectomy Ketoconazole, Metyrapone
Renal phosphate wasting(50%)	high dose of oral phosphate with high dose of calcitriol
Secondary hyperparathyroidism due to vitamin D deficiency	Vitamin D

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Bisphosphonate treatment

- Pain= primary indication for treatment and the clinical end point
- minimum dose and longest interval [pain control]
- Dose as treatment in Paget's disease

Treatment of hypophosphatemia

Goal: Serum phosphorus = low normal level

Phosphate and vitamin D treatment

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Conclusion

- Present with Pain, pathologic fracture
- Look for skin lesion ,endocrinopathies
- Skull involvement => nerve compression
- follow up Endocrinopathies (occur later)
- Bisphosphanate for relieving bone pain(clinical symptom)