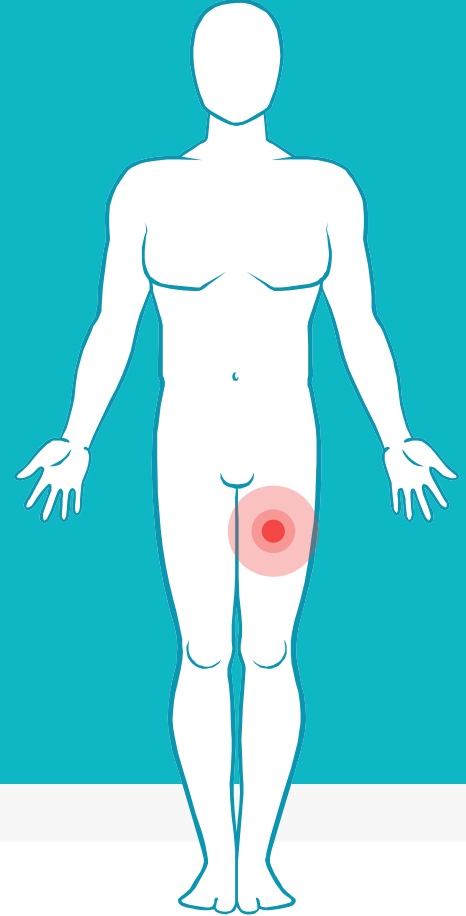


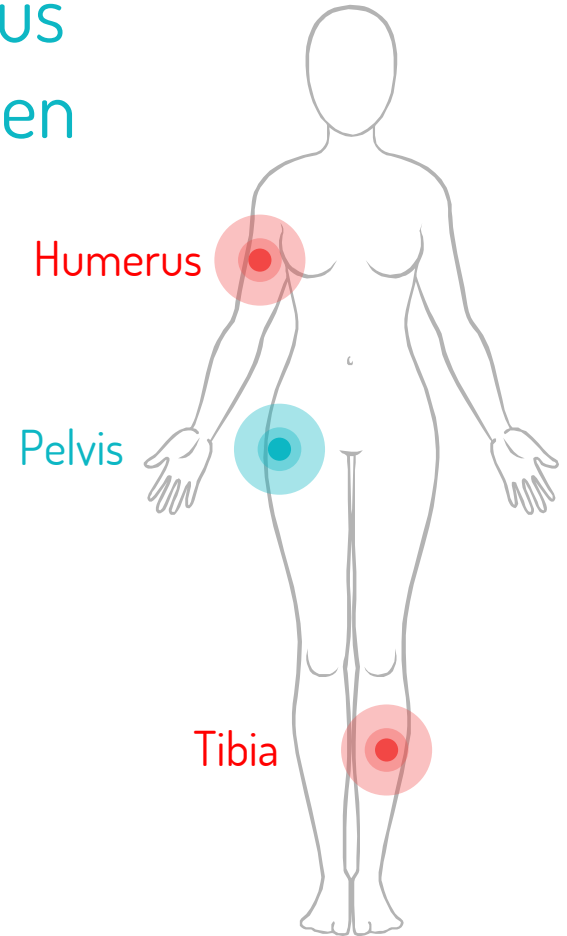
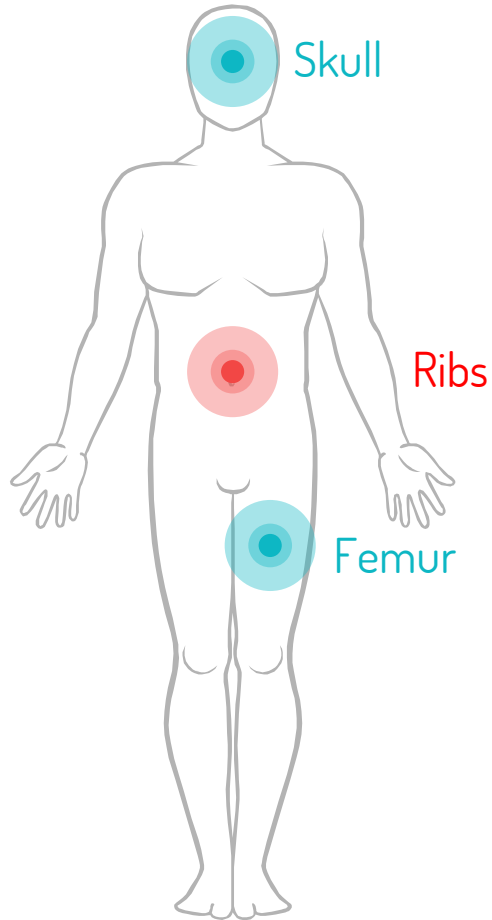
Fibrous Dysplasia



What is Fibrous Dysplasia?

- ▶ A benign bone condition in which abnormal fibrous(connective) tissue develops in place of normal bone
- ▶ Over time, as these areas of fibrous tissue grow, they can weaken the bone, leading to fractures and deformation
- ▶ Linked to a disorder in the bone where bone cells make an abnormal type of fibrous bone
- ▶ Despite this abnormal tissue growing since birth, it is often only discovered until adolescence or even adulthood
- ▶ Fibrous Dysplasia can affect any bone in the body but there are certain areas where it is most common

Where does Fibrous Dysplasia most often occur?



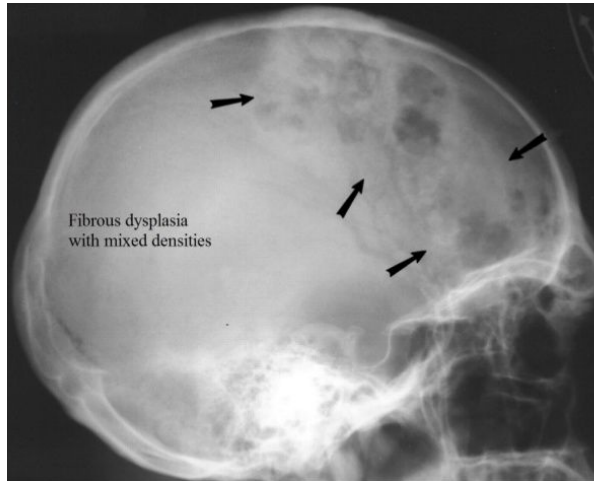
Fibrous Dysplasia Radiology

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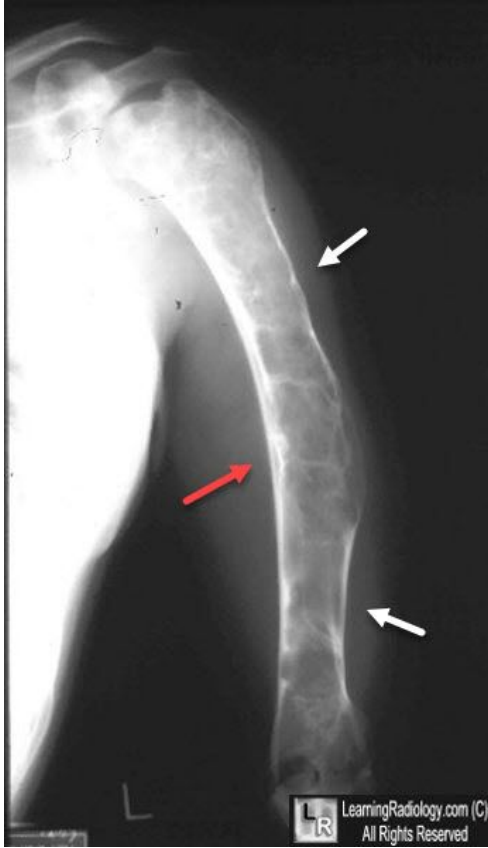


Pelvis/ Proximal Femur

Skull



Distal Femur



Humerus

More X-Ray Images



Tibia



Cherubism

A Glance



Cherubism



What is Cherubism?

- Painless condition
- Maxilla and Mandible bones replaced with less dense fibrous tissue
- More observed in males than females
- Issues:
 - Loss or misplacement of teeth
 - Affected eyesight
 - Difficulty moving jaw



How Does Cherubism Happen?

- Osteoclasts more active than osteoblasts
- Osteoblasts use large cells to replace, resulting in spongy material



Treatment

- Can regress naturally
- Treatment includes
 - Surgery to remove the fibrous tissue
 - Orthodontic treatment to fix teeth
 - **No** radiation!

McCune-Albright Syndrome (MAS)

What is MAS?

Defined as having two or more of the following:

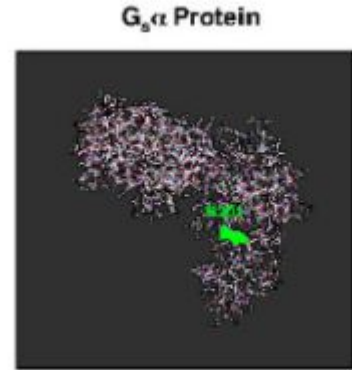
- 1) FD
- 2) Typical cafe-au-lait skin spots
- 3) Hyperfunctioning endocrine disease (e.g. hyperthyroidism, growth hormone excess, precocious puberty)



What causes MAS?

Mutation in a protein known as $G_s\alpha$,
encoded by the gene GNAS

- A genetic disease since the mutation occurs while in early development
- Not hereditary



MAS and PP

- The initial presentation of MAS usually includes signs and symptoms of either precocious puberty or FD
- Children may experience premature sexual developments and quick growths above the normal growth curve



Sample growth chart of a girl with precocious puberty. This early growth spurt results in the child's final short stature.

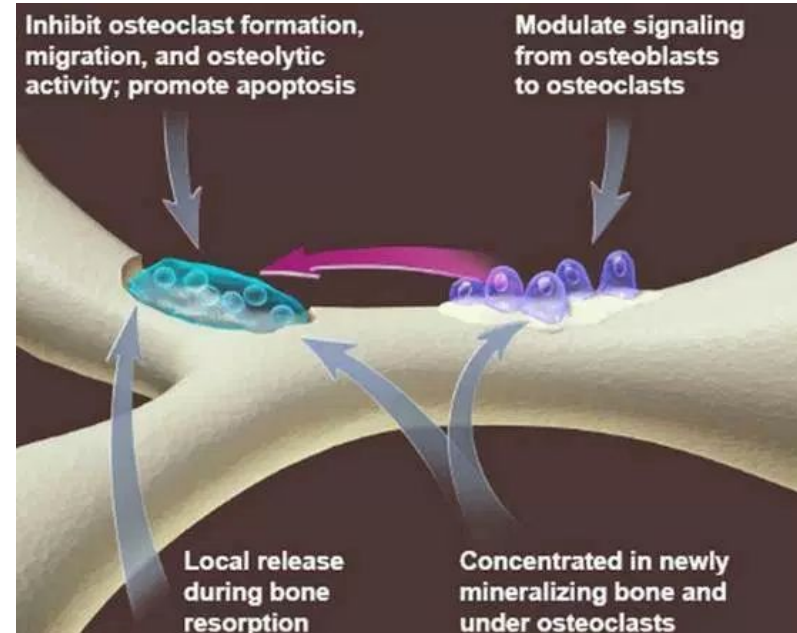
Treatment of FD/MAS

Diagnosed via X-ray, MRI, CT,
DNA biopsy of bone sample

No cure (genetic)

Treatment

- ▶ Surgery to correct mis-shapen bones or fractures
- ▶ Bisphosphonates to relieve pain (but little evidence)
- ▶ Physical activity
- ▶ Sufficient calcium, phosphorus, vitamin D intake



Endocrine Follow-Ups

- ▶ **Pituitary:** excess of GH + PRL common in MAS and CF disease
 - ▶ Oral glucose tolerance test for non-suppressible GH
 - ▶ Non-suppressible GH + elevated IGF-1 treatment available
- ▶ **Thyroid:** hyperthyroidism common
 - ▶ Thyroid function tests
 - ▶ Anti-thyroidal treatments (methimazole, PTU)
 - ▶ Definitive treatment is surgery, not radioiodine
 - ▶ Annual ultrasound
- ▶ **Parathyroid:** primary rare, secondary (Vit.D deficiency) common
 - ▶ Check ionized/total calcium, PTH levels annually

Endocrine Follow-Ups

- ▶ **Adrenal:** neonatal Cushings that rarely last past 1st year
 - ▶ Most resolve spontaneously
- ▶ **Renal:** phosphate wasting is common
 - ▶ Check serum/renal handling of phosphate
 - ▶ Treat with low amounts of TMP/GFR
- ▶ **Gonads:** precocious puberty common, Leydig masses in males
 - ▶ Girls: treat with anti-aromatase + tamoxifen
 - ▶ Boys: treat with anti-aromatase + anti-androgen
 - ▶ Treat secondary PP with long-acting GnRH agonist
 - ▶ Check Leydig masses via ultrasound for cancer

General Follow-Ups

▶ **Craniofacial**

- ▶ Avoid surgeries if vision/hearing is not impaired or if there is no severe pain/disfigurement
- ▶ Annual vision tests and CT (skull/mandible)
- ▶ Screen and treat endocrinopathies that affect bone
- ▶ Bone scans

▶ **Axial and Appendicular Skeleton**

- ▶ Bracing
- ▶ Screen and treat endocrinopathies that affect bone
- ▶ Bone scans
- ▶ Bisphosphonates for pain
- ▶ Strength maintenance (swimming is good!)

Skeletal and Drug Studies

Skeletal Study

- Study in Australia of 5 children with MAS
- Treatment: combination of drugs and surgery
- Bisphosphonates and implantation of in of intramedullary rods in the femur and tibia
- Successful in all patients
- Effect: increased quality of life, decreased pain, decreased fracture rate, improved walking ability



Skeletal Study

- In 43 patients with upper extremity FD
 - Satisfactory results with achieved with non-operative treatment
- Age was a factor for lower extremities
 - Over 18 success in 8/9
 - Under 18 unsatisfactory results

Drug Study

- Nine patients with symptomatic and serious fibrous dysplasia
- Treatment: intravenous pamidronate 60 mg/day for 3 days every 6th month
- Effect: decreased bone pain
- Complete Remission in 12 out of 14 sites