

OSSEOUS MANIFESTATIONS IN VON RECKLINGHAUSEN'S DISEASE

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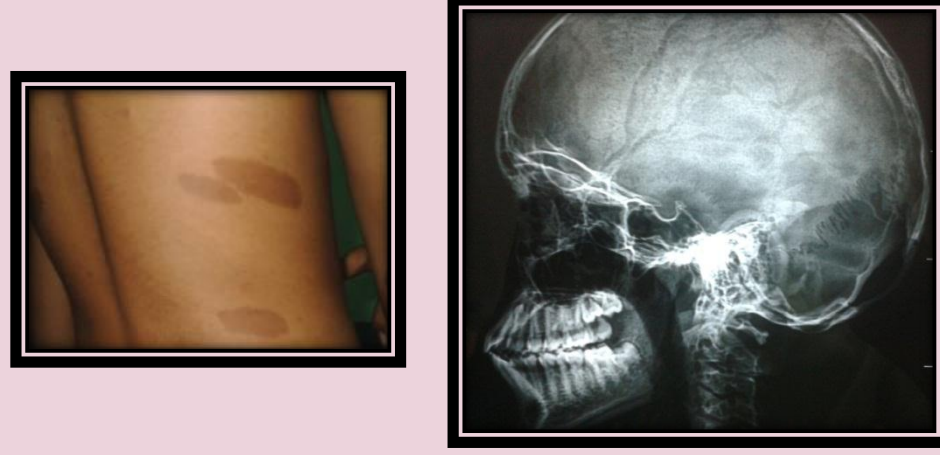
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The Spectacle of Deformity of "The Elephant Man" Joseph Carey Merrick - A TRIBUTE!

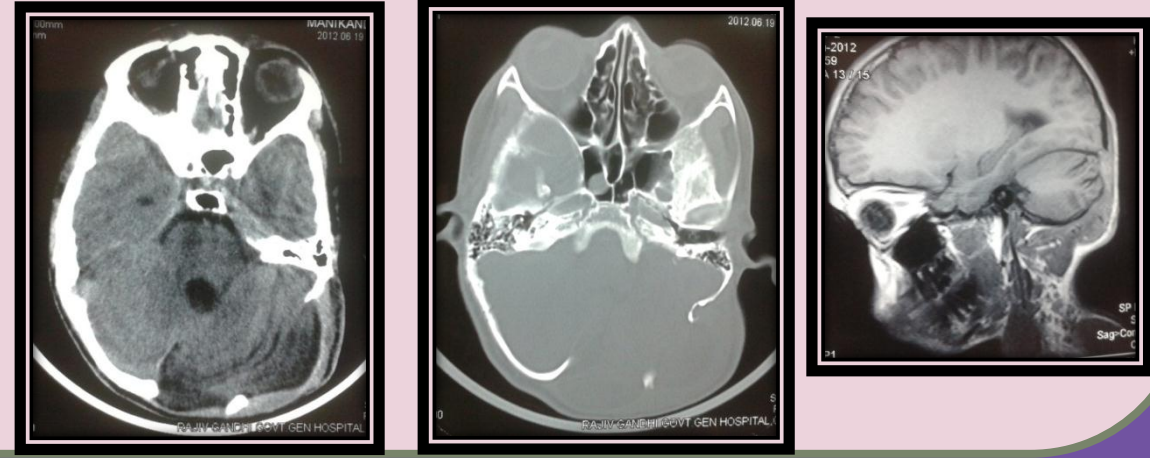
Skull defects

Asterion defect

Radiolucent defect is sometimes seen in the calvaria adjacent to the lambdoidal suture.

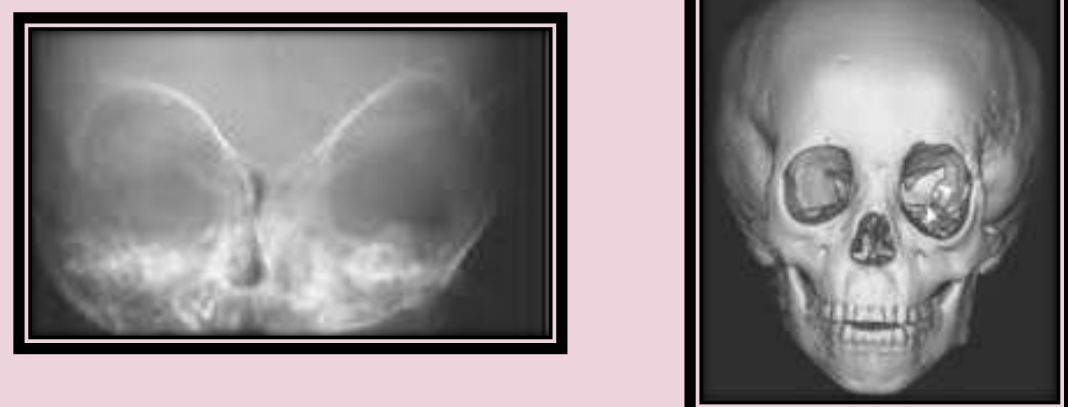


Virtually diagnostic of neurofibromatosis.

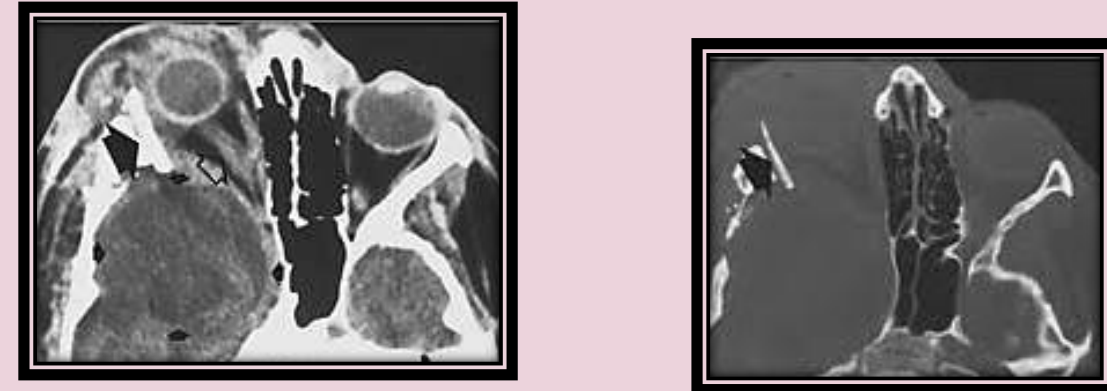


Orbital defects

Agnesis or hypoplasia of the posterior wall of the orbit, wings of the sphenoid, and the orbital plate of the frontal bone.

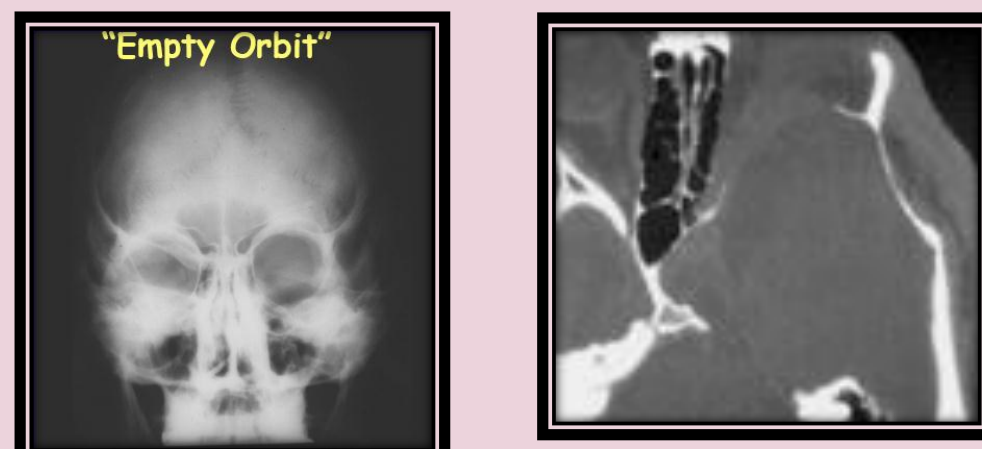


Creates bare orbit. These changes are usually unilateral.



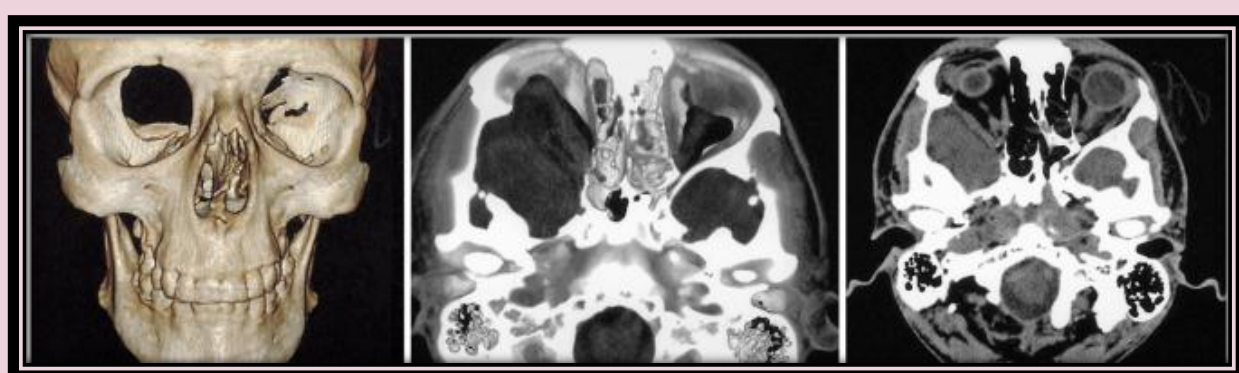
Sphenoid wing agnesis

Rare, most characteristic bony change

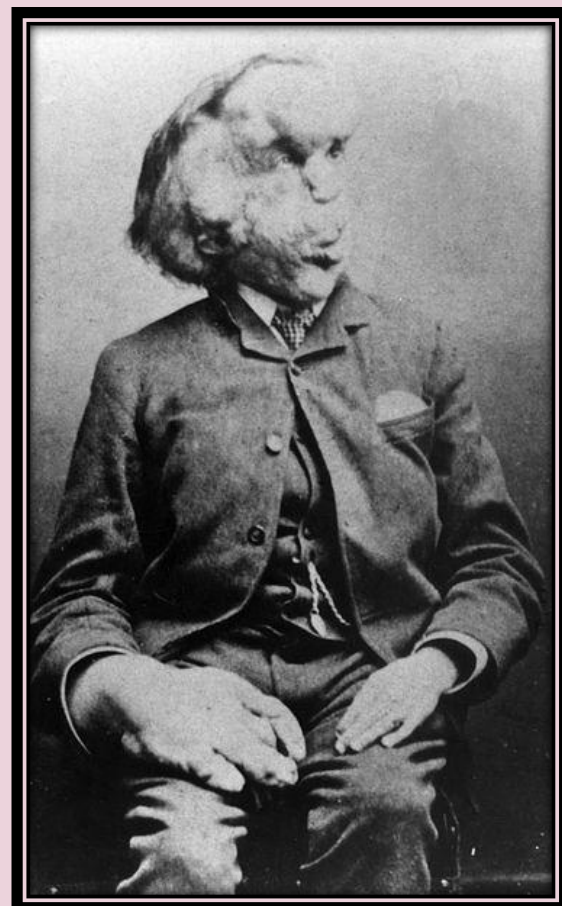


Produces "empty orbit" in plain film

May lead to pulsating exophthalmos.



The Elephant Man's Bones Reveal Mystery



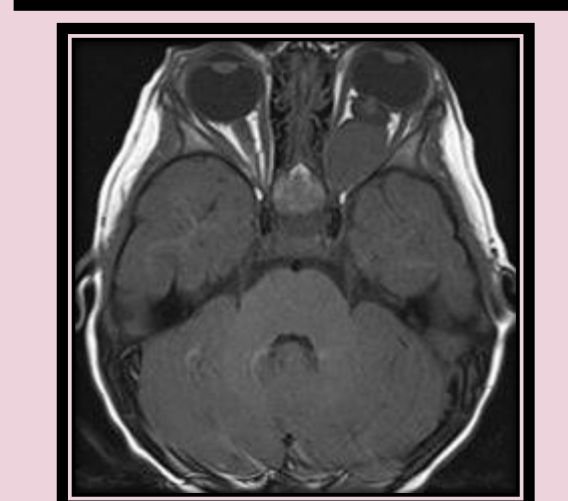
Joseph Merrick, the "Elephant Man"



The skeleton of Joseph Merrick

Optic canal expansion

Neurofibroma affecting the optic nerve may lead to enlargement of the optic foramen



Spine defects

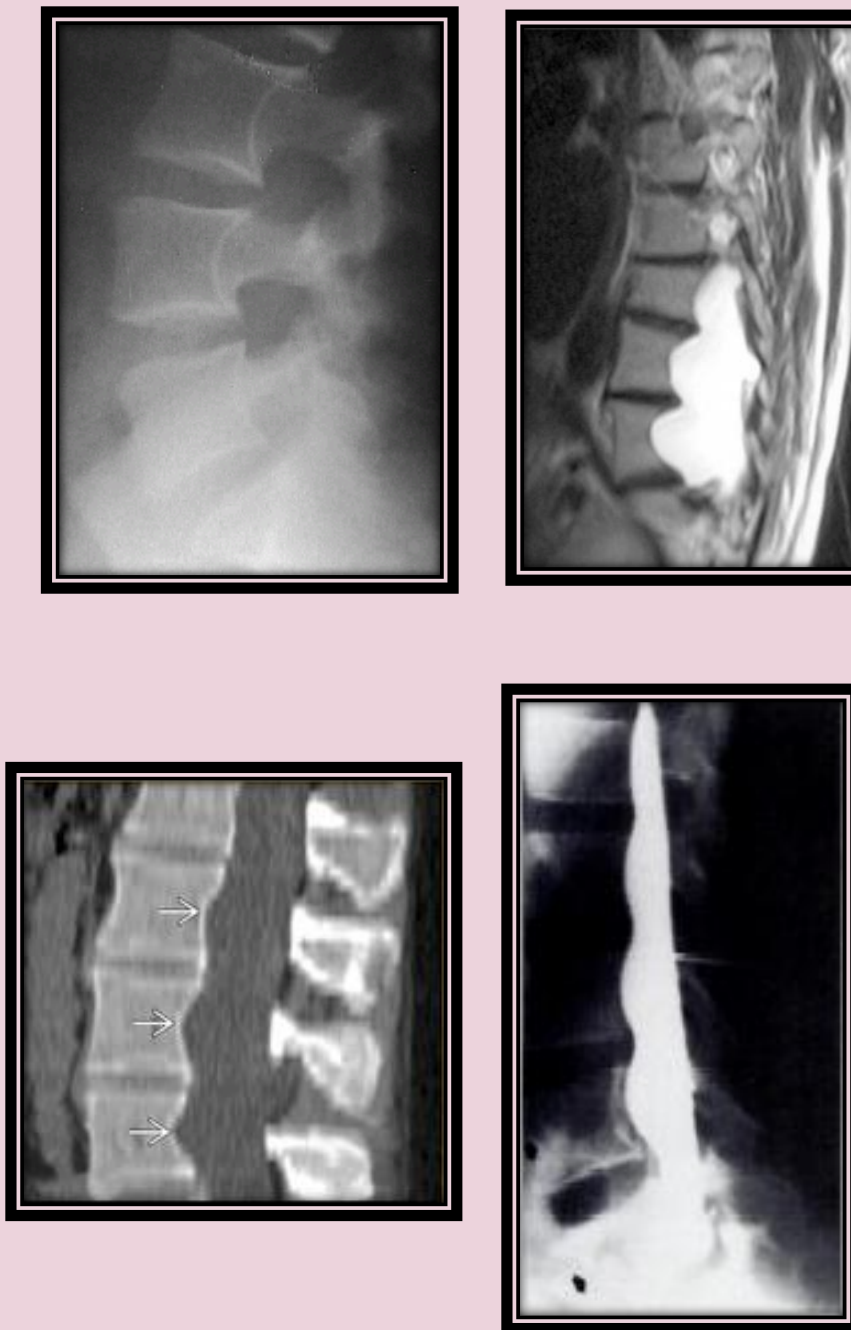
Kyphoscoliosis

Most common skeletal abnormality. Occurs in 50% of NF1. Short segment (5-7 vertebrae) involvement and acutely angular. Lower thoracic spine commonly affected.



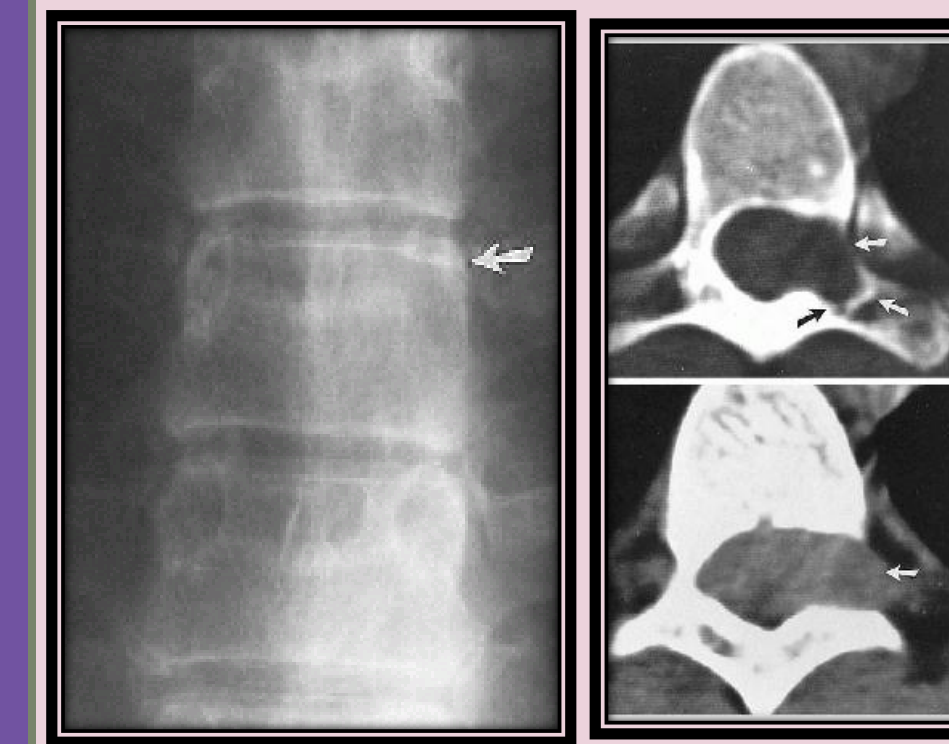
Posterocentral vertebral scalloping

Usually affecting more than one vertebral level. Secondary to dural ectasia. Lack of disc space narrowing differentiates from pott spine.



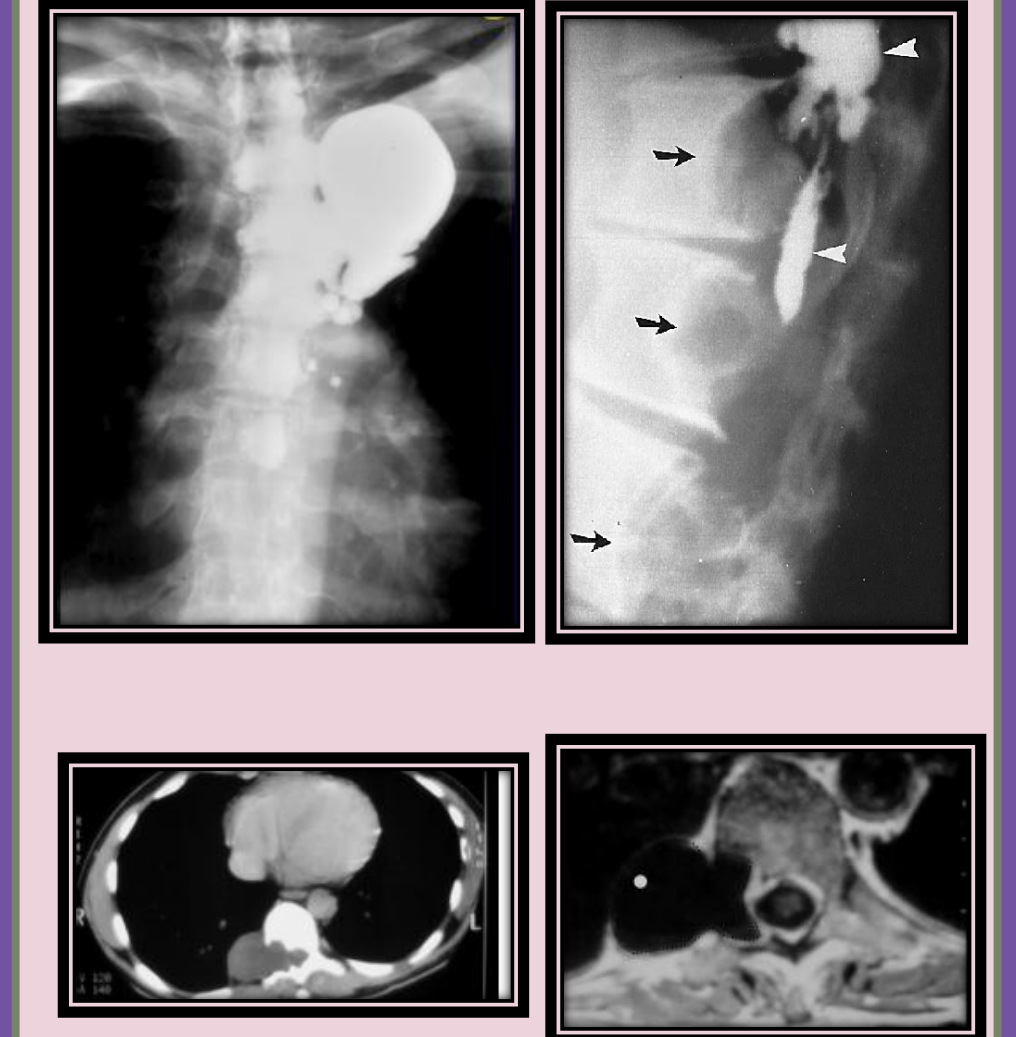
Eccentric unilateral vertebral scalloping

Due to dumbbell neurofibroma arising from a spinal nerve root and protruding through the adjacent intervertebral foramina (IVF) causing erosion of the posterior body and adjacent pedicle.



Intra thoracic meningocele

Occurs in approximately 66% of cases. Common presentation- asymptomatic posterior mediastinal mass and are erroneously interpreted as a neurofibroma. Myelography reveals it.



Neurofibromatosis- type 1

Neurofibromatosis (Recklinghausen's disease) is a congenital and familial disease with abnormalities of skin, nervous system, bones, and soft tissues.

A typical case demonstrates (1) multiple soft, elevated cutaneous tumors (fibroma molluscum), (2) cutaneous pigmentation (café au lait spots), and (3) neurofibromas of peripheral nerves, frequently palpable in the subcutaneous tissue.

Variety of skeletal defects are associated with neurofibromatosis. 50% of NF 1 develop skeletal abnormalities. Spine and skull are commonly affected.

These are of particular interest to the radiologist, since certain types are characteristic of the disease, while others are most suggestive.

Associated skeletal abnormalities include severe scoliosis, defects of the walls of the orbits, asterion defect, erosive defects caused by adjacent neurogenic tumors, bowing and pseudarthrosis of the lower leg, and disorders of growth of bone associated with elephantoid hypertrophy of soft tissue, macrocrania. Knowledge of these findings enables the radiologist to help confirm the clinical diagnosis.

Defects in long bones

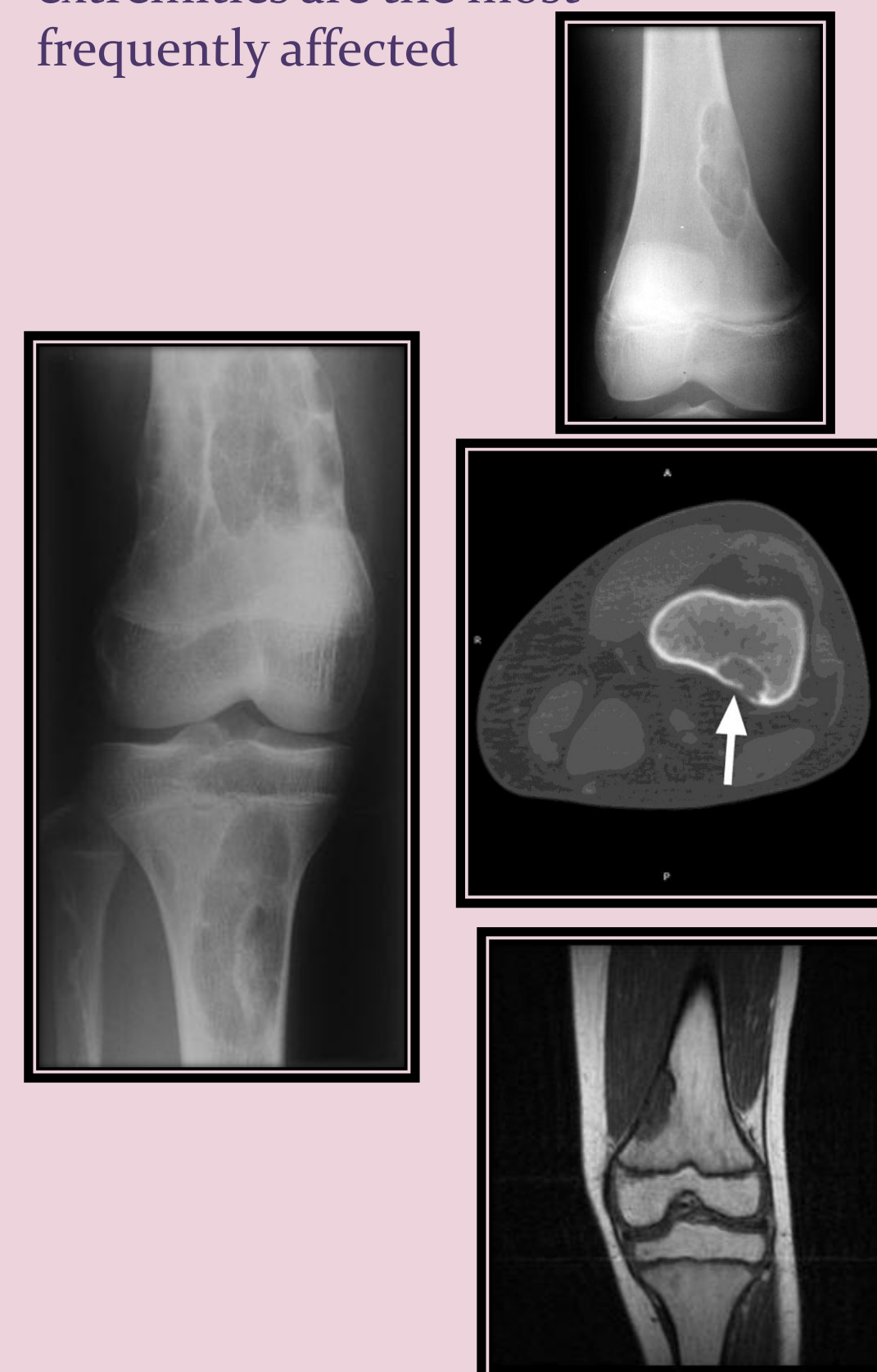
Pseudoarthrosis

Due to pathologic fracture and inability to form normal callus in healing. Deossification and bending of weight-bearing bones may also occur. The tibia in its lower two thirds is the most commonly affected bone.



Non ossifying fibroma

Present with cystic intramedullary lesions in the metaphysis of the long tubular bones. Often multiple, bilateral. The bones of the lower extremities are the most frequently affected



Focal gigantism

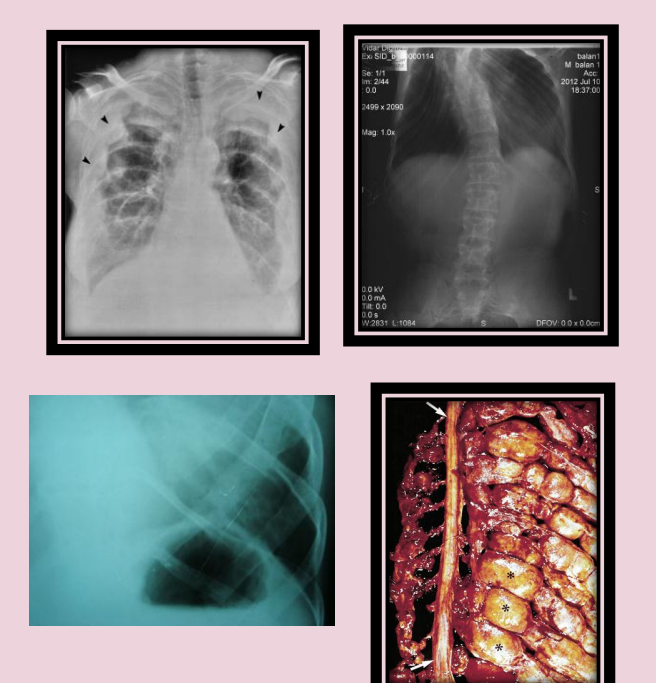
Most bizarre and fascinating aspect of NF1. The hypertrophy may involve a single bone or the entire extremity. Results from chronic hyperemia owing to hemangiomas and lymphangiomatous lesions associated with NF1.



Rib defect

Ribbon ribs

Thin, irregular, scalloped, attenuated ribs. Produced by erosions on the inferior surface of the ribs by the intercostal neurofibromas



Reference:

1. Yochum essentials of skeletal radiology
2. www.Radiographics
3. Durbach, Nadja (2009). "Monstrosity, Masculinity, and Medicine: Reexamining the Elephant Man". *The Spectacle of Deformity: Freak Shows and Modern British Culture*. Berkeley