OSTEOGENESIS IMPERFECTA (Brittle bone disease; Fragilitas ossium)

Dept.of Oral Pathology & Microbiology

Learning Objectives

At the end of the lecture student should be able to describe

- Clinical features, oral manifestations,

  radiographic features, histopathological features,&

  surgical management of Osteogenesis Imperfecta &

  Osteopetrosis.

- Heritable C.T. disorder characterized by impairement of collagen (type I) maturation & bone fragility.
- Collagen forms a major portion of bone ,dentin, sclerae, ligaments & skin; O.I. demonstrates a variety of changes that involve these sites.
- Both autosomal dominant & recessive hereditary patterns are seen

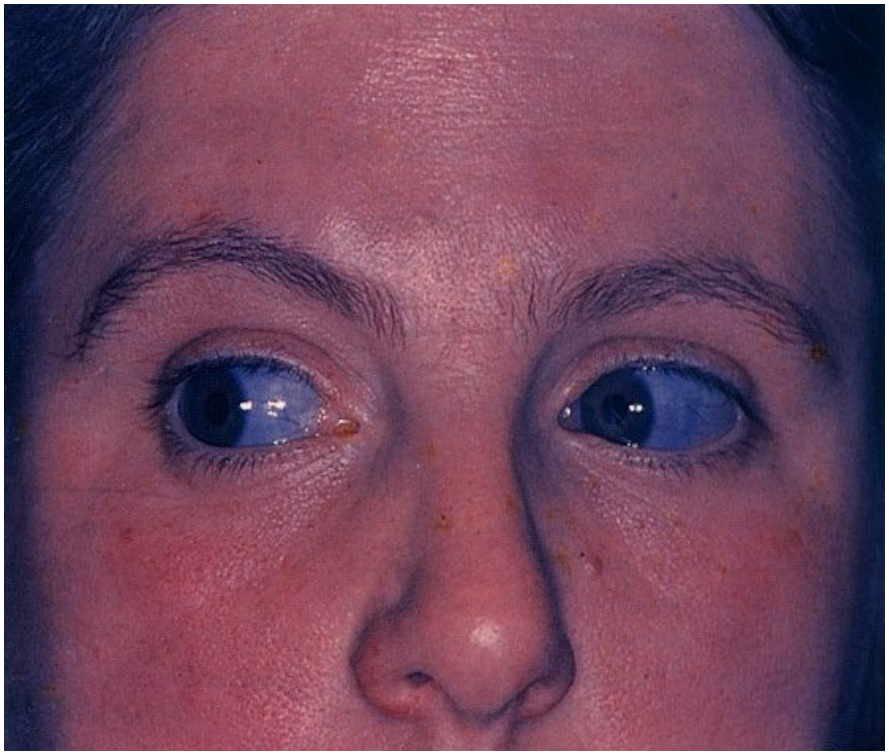
CLINICAL FEATURES:

- The chief clinical characteristic of OI is the extreme fragility & porosity of the bones, with increased proneness to fracture.
- The severity of the disease varies widely, even in the affected members of a single family.

- Disease in its usual form is present at birth (Congenita or Vorlik's type)
- Although some cases do not arise or are not recognized until later in the childhood (Tarda or Lobstein's type or Osteosathyrosis)
- Many infants suffering from OI are stillborn.

- Pale blue sclerae

(the sclerae are abnormally thin ,so pigmented choroid shows through & produces the bluish colour).



ORAL MANIFESTATIONS

- Class III malocclusion (Large head size, frontal & temporal bossing)
- Impacted & ectopic teeth
- Unerupted permanent 1st & 2nd molar
- Dentinogenesis imperfecta

SILLENCE CLASSIFICATION

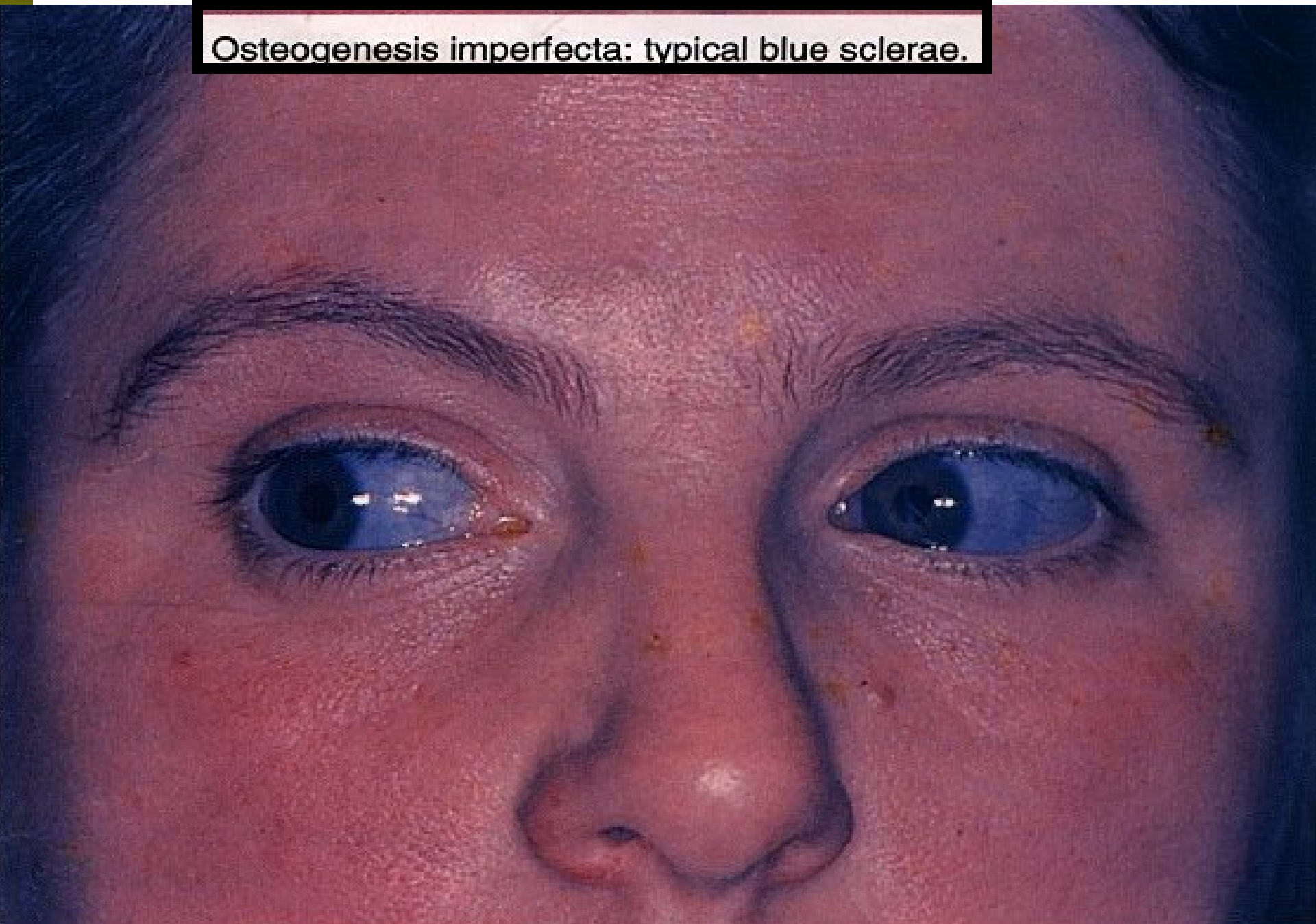
- Four major types of O.I.:
- **Type I**
- **Type II**
- **Type III &**
- **Type IV**

Type I:

- Most common & mildest form
- Autosomal dominant inheritance pattern
- Mild to moderate bone fragility
- Fractures at birth in 10% of the cases.
- Hyperlaxity of ligaments of knee, hand & feet.
- Hearing loss (due to otosclerosis) before age 30.
- Opalescent dentin (Dentinogenesis Imperfecta)
- Progressive kyphoscoliosis is seen in 20% patients

Type II:

- Both, an autosomal recessive & dominant pattern.
- Most of the patients are stillborn & 90% die before the age of 4 weeks.
- Extreme bone fragility & frequent fractures, which may occur during delivery.
- ***Blue sclera***.
- Tooth anomalies (Dentinogenesis Imperfecta) may be seen.

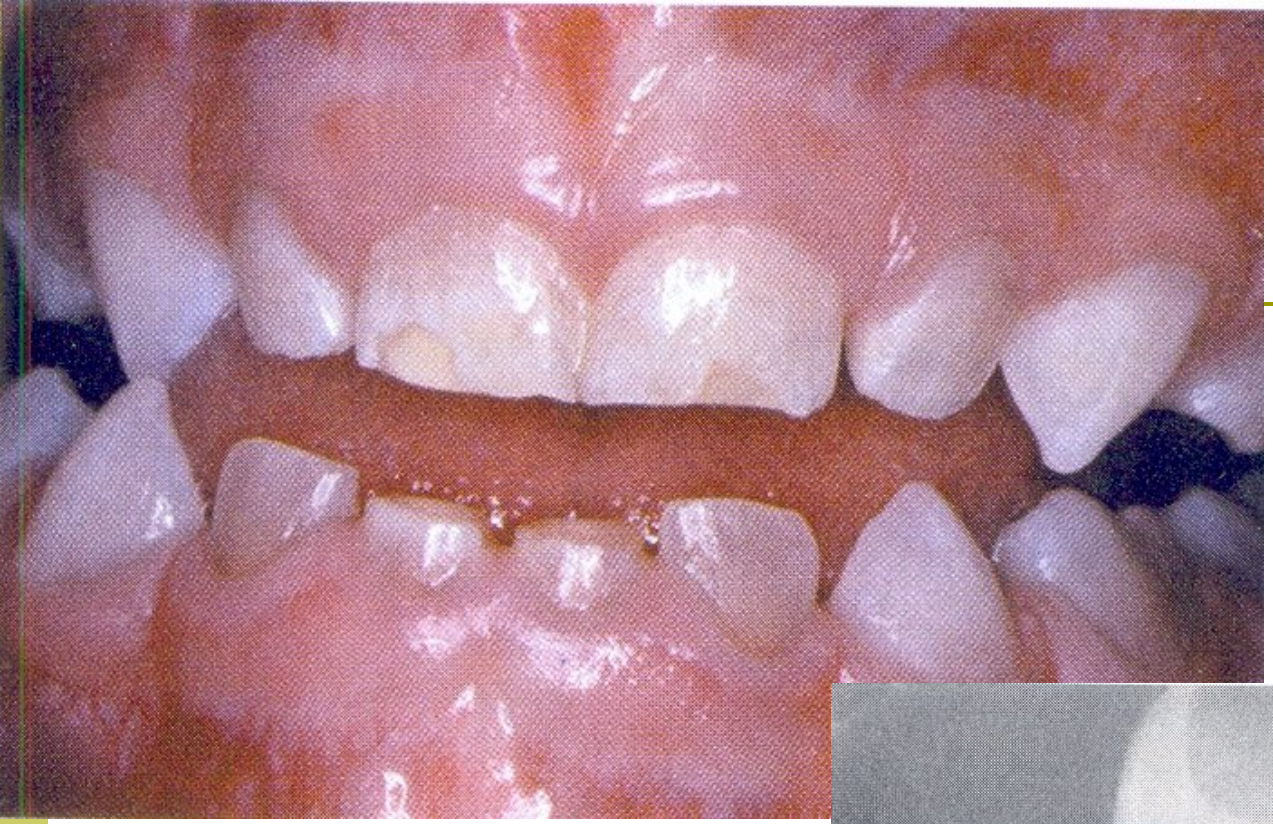

Osteogenesis imperfecta: typical blue sclerae.

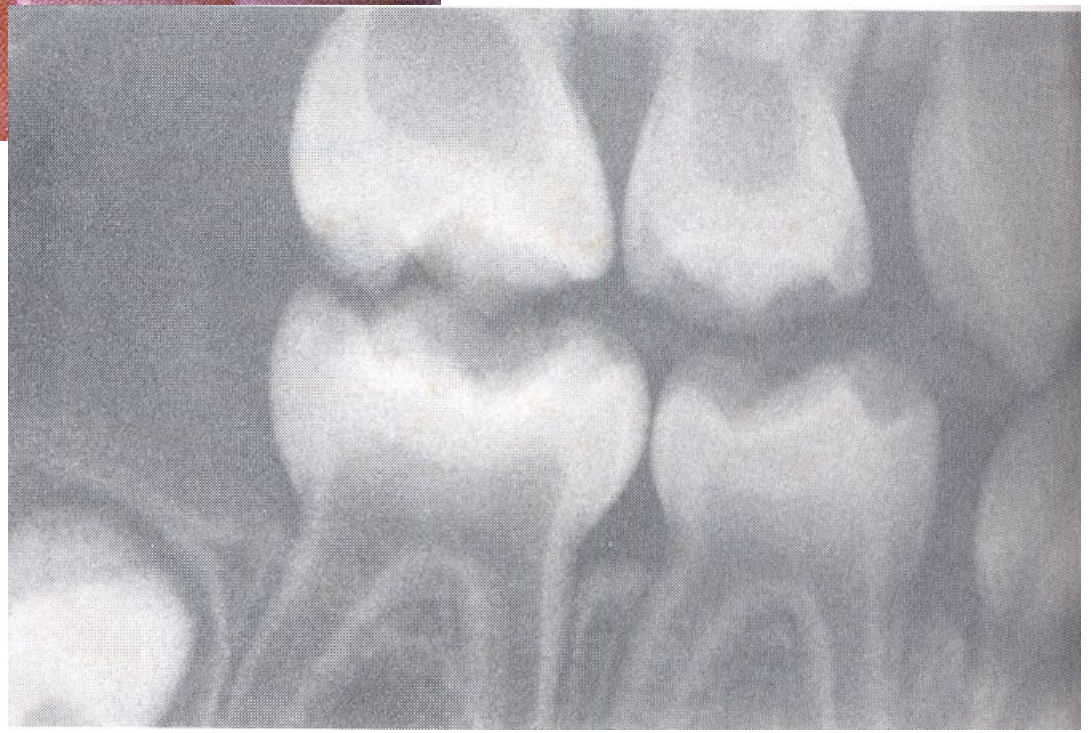
Type III:

- Associated with Dentinogenesis Imperfecta
- Both, an autosomal recessive & dominant pattern
- Sclera is ***Blue*** in infants, but the blue color fades with age.
- Patients have extreme bone fragility & frequent fractures, which may occur during delivery. In utero in 50% of cases
- Limb are short, curved & grossly deformed. The skin is thin ,frail & may be torn during delivery.

Type IV:

- **Type A** without dentinogenesis imperfecta
- **Type B** dentinogenesis imperfecta
- Normal sclera
- Normal hearing
- *In utero* # are rare





HISTOPATHOLOGY

•OI is a generalized disease of C.T. with abnormalities in collagen synthesis.

• Basic defect appears to lie in the organic matrix with failure of fetal collagen to be transformed into mature collagen.

•The mass of cortical & cancellous bone are greatly reduced.

- Osteoblasts are present, but bone matrix production is markedly reduced.
- The bone architecture remains immature throughout life, & there is a failure of woven bone to become transformed to lamellar bone.
- Qualitative & quantitative defects of collagen I formation
- The bone fractures heal with abundant callus formation.

OSTEOGENESIS IMPERFECTA

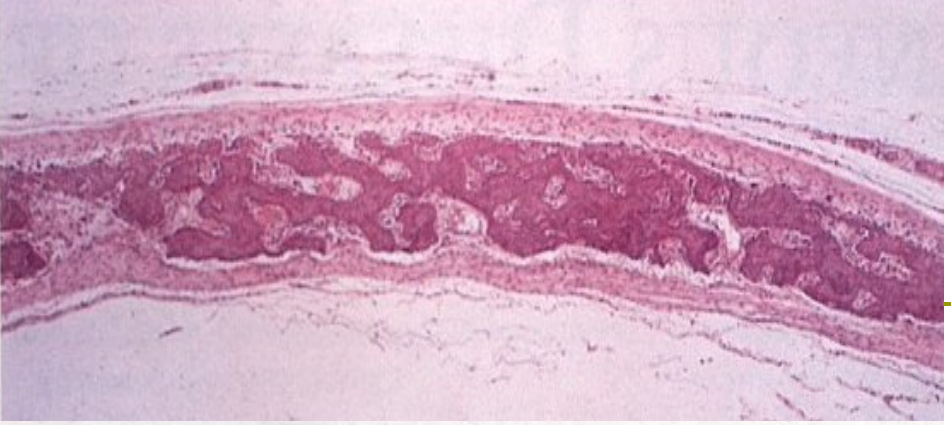
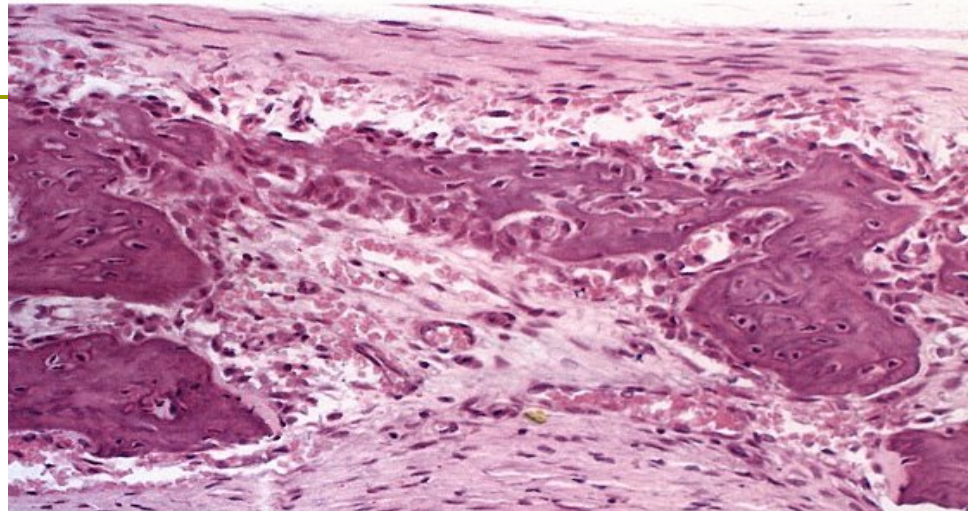


Fig. 8.2 Osteogenesis imperfecta: low power view of a section of the calvarium of an affected infant showing lack of differentiation between the inner and outer plates, and the extreme thinness of the bone overall.



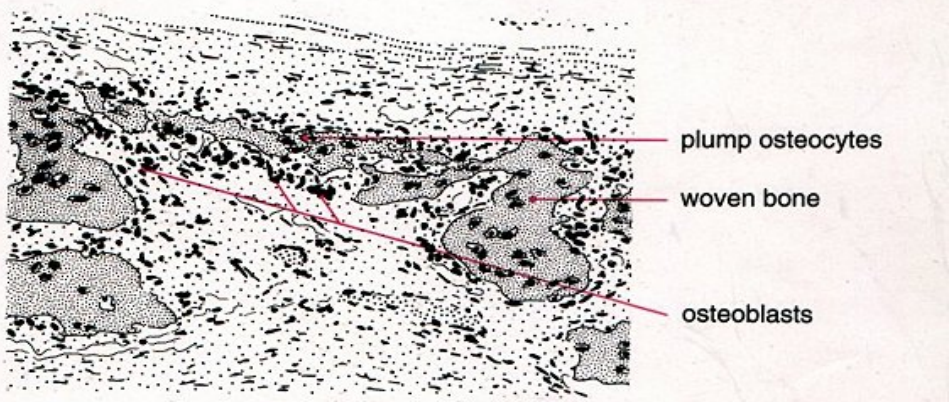
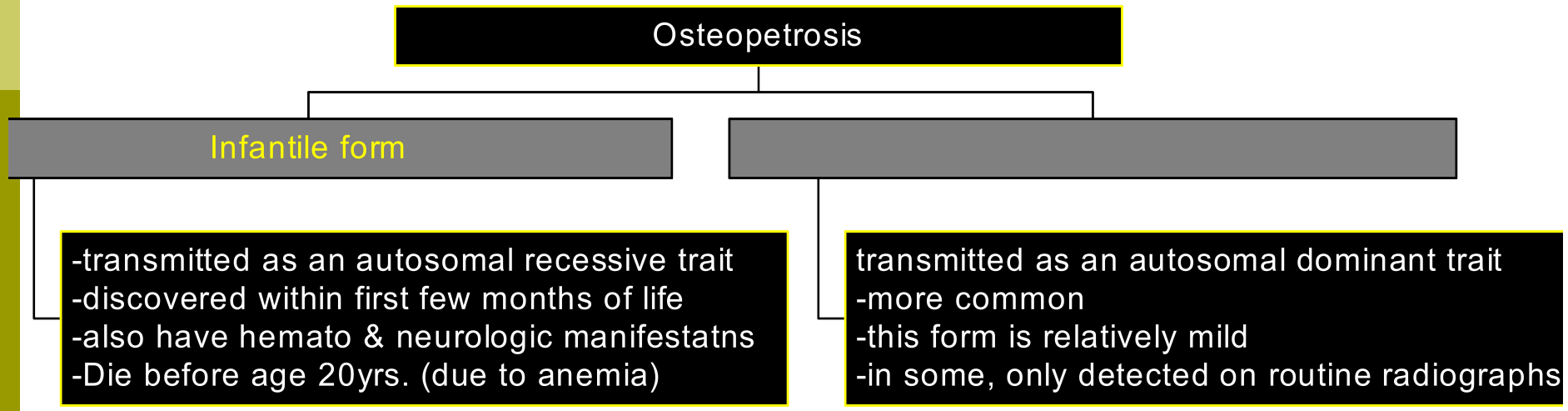


Fig. 8.3 Osteogenesis impertecta: higher power view of the section in Fig. 8.2 shows only woven bone and large numbers of osteoblasts but no compact bone.

OSTEOPETROSIS
(Marble Bone Disease; Albers-Schönberg Disease)

•Osteopetrosis is a rare hereditary skeletal disorder characterized by a marked increase in the bone density resulting from a defect in bone remodeling.



CLINICAL FEATURES:

INFANTILE:

- Initial signs are normocytic anemia & hepato-splenomegaly resulting from compensatory extramedullary hematopoiesis
- Increased susceptibility to infection is common
- Facial deformity develops in many of the children

-- broad face; hypertelorism; snub nose; frontal bossing

- Skull : failure of resorption & remodeling of skull bones produces narrowing of the various foramina (blindness, deafness & facial paralysis
- Dental : Tooth eruption is delayed, Osteomyelitis is common complication of extraction

ADULT:

- Detected at a later age than the infantile type
- Asymptomatic in early life & progressively involves more bones
- It is not life threatening
- **NO**- anemia, hepato-slenomegaly, blindness, deafness
- Dental : Fracture & Osteomyelitis are common complications of extraction

RADIOGRAPHY:

- Increase in skeletal density with defects in the metaphyseal remodeling
- Distinction between cancellous & cortical bone is lost

OSTEOPETROSIS

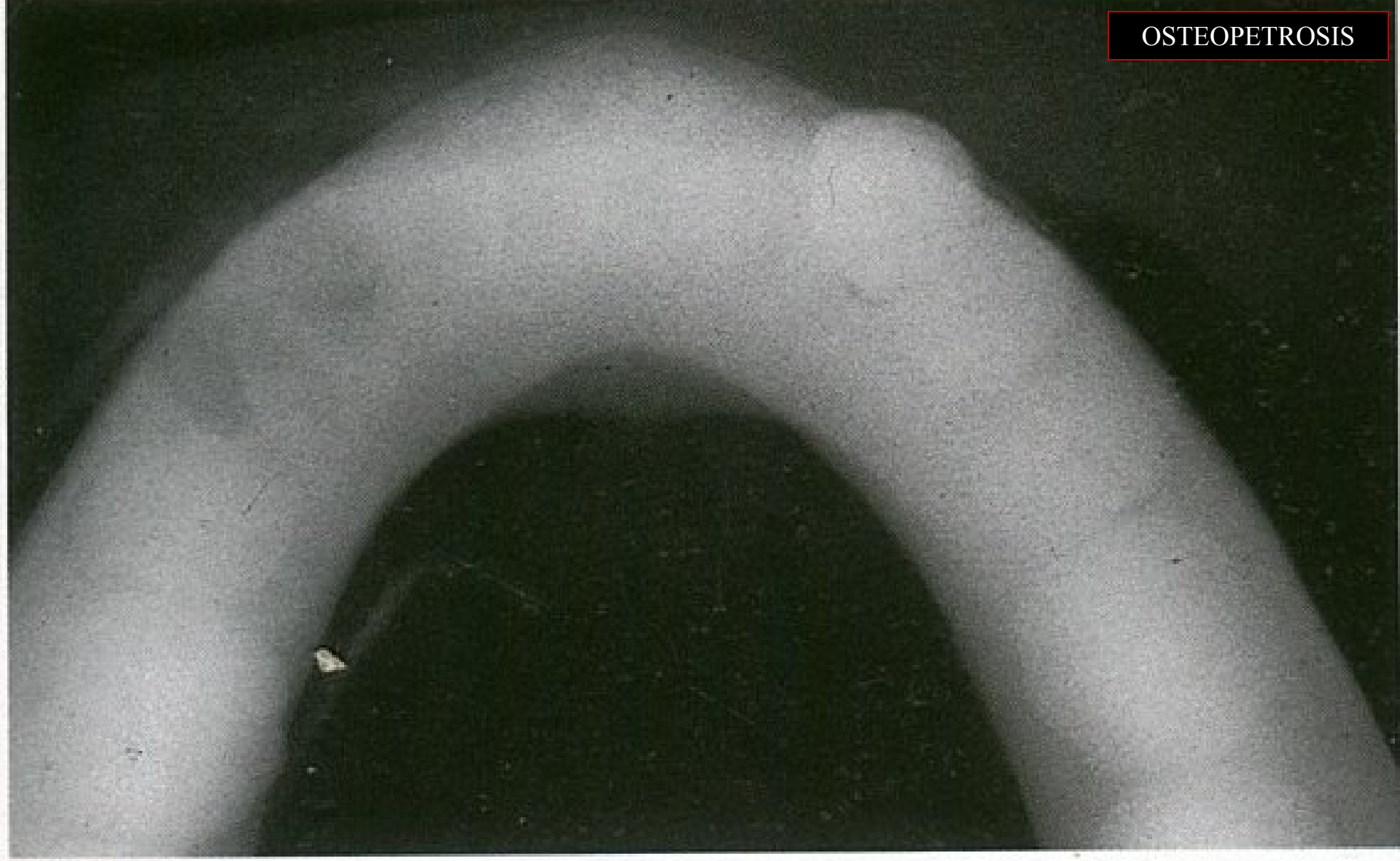


Fig. 8.5 Osteopetrosis: radiograph showing such extreme bone density that the teeth are barely visible.

OSTEOPETROSIS

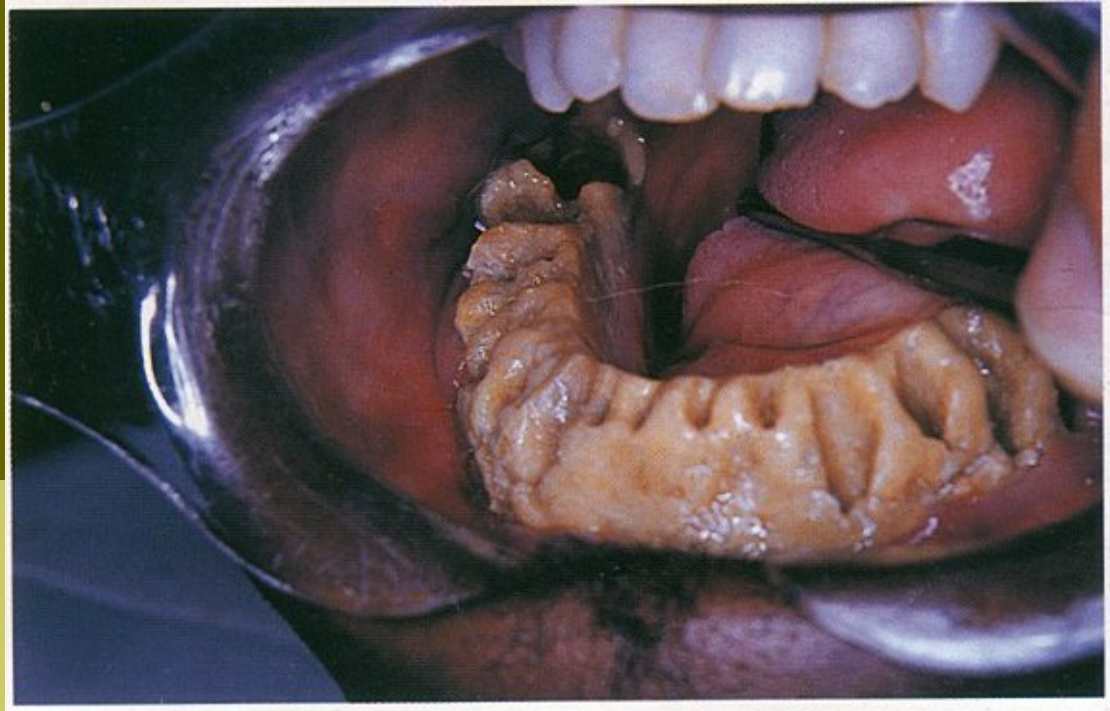


Fig. 8.6 Osteopetrosis: necrosis of the body of the mandible following extraction of the teeth from the same patient as in the previous picture.

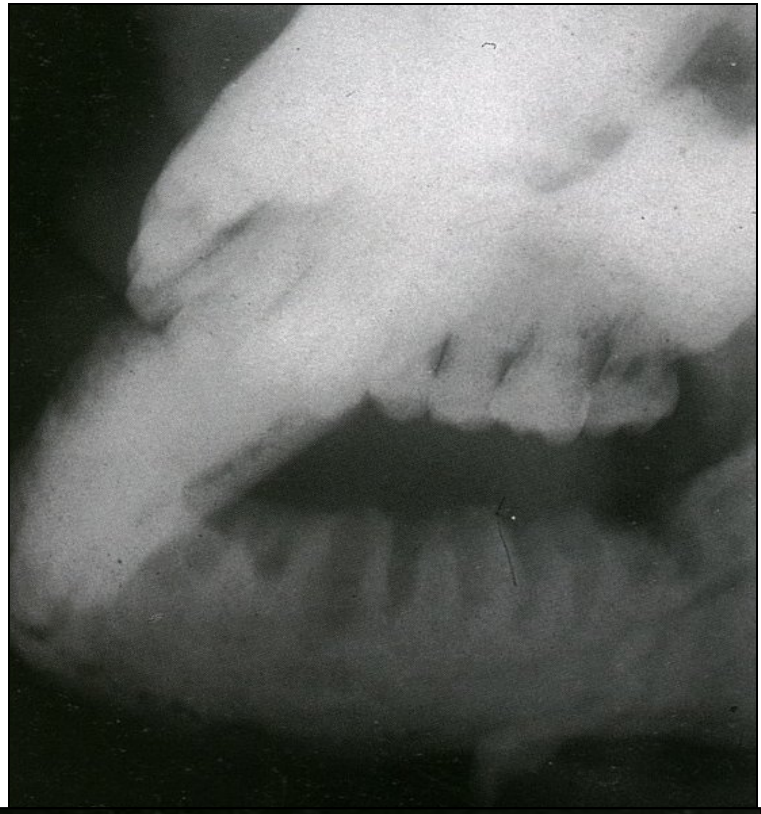


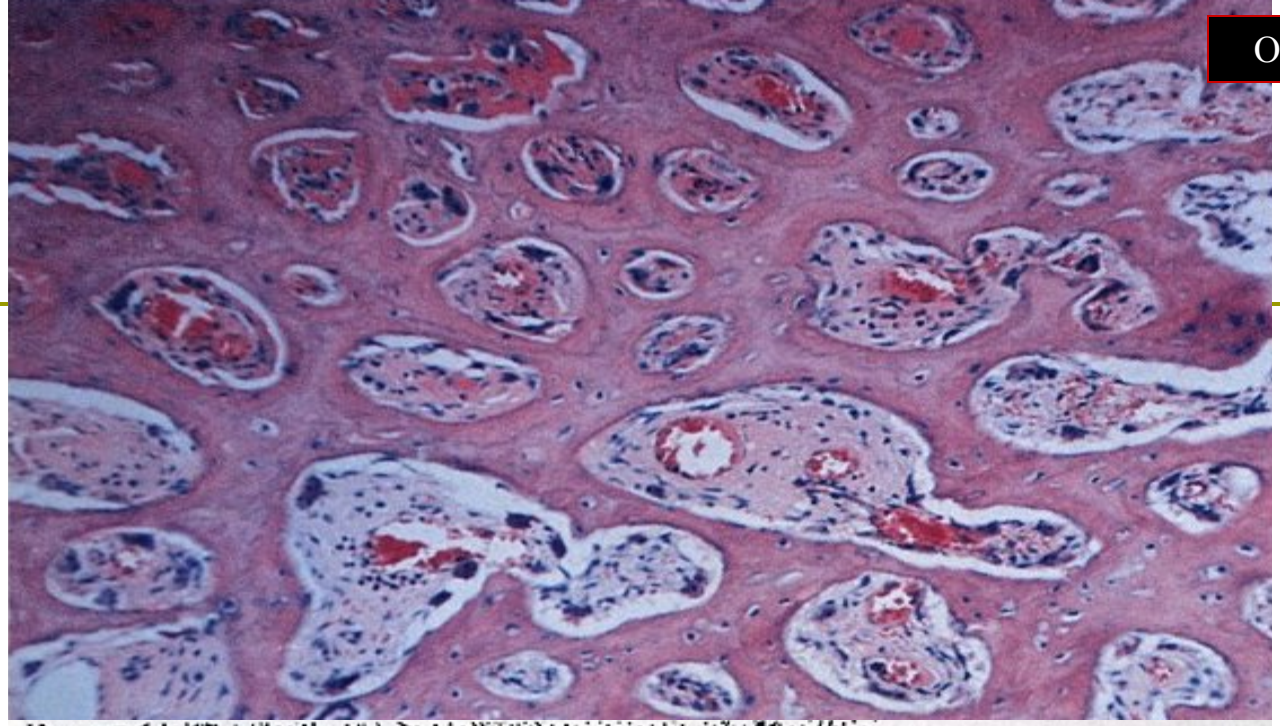
Fig. 8.7 Osteopetrosis: radiograph of the same patient as in the previous pictures showing the line of separation of the sequestrating alveolar bone.

HISTOPATHOLOGY

- Number of osteoclasts is increased, but their function to resorb bone is altered.
- Osteoclasts appear atypical.
- Defective resorption combined with continued bone formation & endochondral ossification is seen.

- Several patterns of abnormal endosteal bone formation have been described:
  - Tortuous lamellar trabeculae replacing the cancellous portion of the bone
  - Globular amorphous bone deposition in marrow spaces
  - Osteophytic bone formation
  - TREATMENT
- No specific treatment
- Prognosis for infantile is very poor (death results from anemia, infection usually before age 20.)

OSTEOPETROSIS



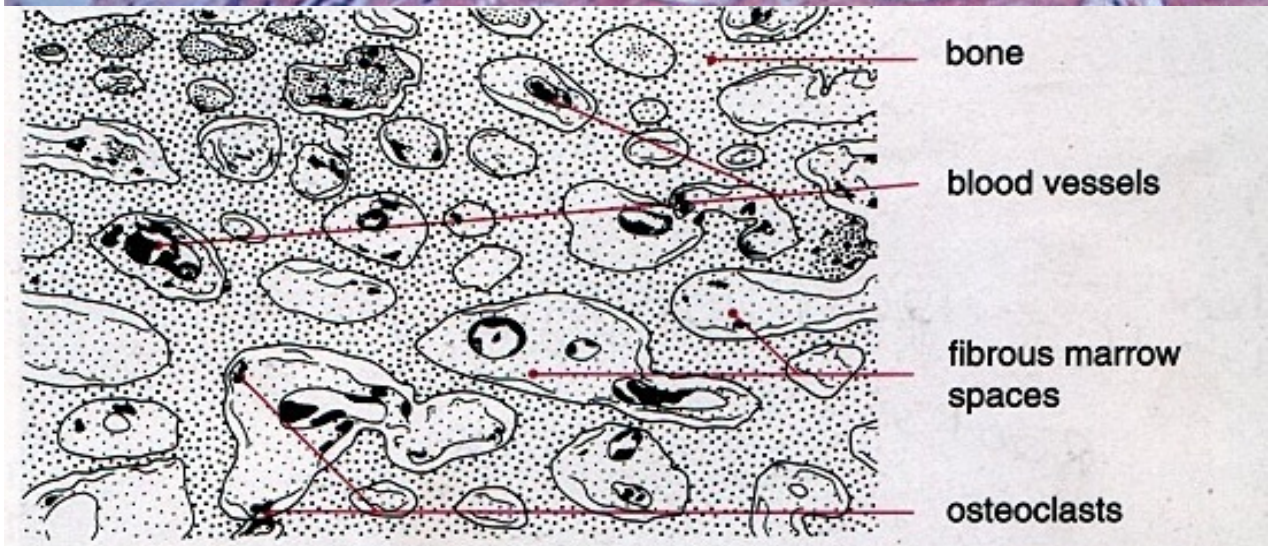


Fig. 8.8 Osteopetrosis: absence of differentiation into cortex and medulla. The bone is dense but penetrated by vascular channels and small amounts of fibrous marrow. Few osteoclasts are present.

Summary

- Clinical features, oral manifestations, radiographic features, histopathological features,& surgical management of Osteogenesis Imperfecta & Osteopetrosis

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Thank You