جامعة الملك سعود
King Saud University

Pathology

Team 435

Lecture (1,2): Congenital Disorders, Acquired diseases and Fractures of Bones

Summary & MCQs

Objectives:

- Be aware of some important congenital and developmental bone diseases and their principal pathological features.
- Understand the aetiology, pathogenesis and major clinical features of osteoporosis.
- Be familiar with the terminology used in some important developmental and congenital disorders.
- Appreciate the importance of road traffic accidents with resultant trauma as a major cause of death and disability in the Kingdom.
- Be aware of the mechanisms and stages of fracture healing and understand the difference between trauma induced and pathological fractures.
- Know the factors contributing to delayed fracture healing.

Red: Important Gray: Extra explanation

SUMMARY

Congenital Disorders of Bone and Cartilage

- Abnormalities in a single or group of bones are called *dysostoses* and can result in the absence of bones, supernumerary bones, or inappropriately fused bones; some of these result from mutations in homeobox genes affecting localized migration and condensation of primitive mesenchymal cells.
- Abnormalities in bone or cartilage organogenesis are called *dysplasias*; these can be caused by mutations that affect signal transduction pathways or components of the extracellular matrix:
  - Achondroplasia and thanatophoric dwarfism occur as a consequence of constitutive FGFR3 activation, resulting in defective cartilage synthesis at growth plates.
  - Osteogenesis imperfecta (brittle bone disease) is a group of disorders caused by mutations in the genes for type I collagen that interfere with its normal production, with resultant bone fragility and susceptibility to fractures.
  - Osteopetrosis is caused by mutations that interfere with osteoclast function and is associated with dense but architecturally unsound bone owing to defective bone resorption.

SUMMARY

Acquired Diseases of Bone Development and Mass

- Nutritional deficiencies can affect bone integrity by altering the quality of the organic matrix (e.g., vitamin C is involved in collagen cross-linking) or by influencing bone mineralization (e.g., vitamin D is involved in calcium uptake).
- Osteoporosis results from decreased bone mass and is clinically significant because it predisposes bone to fracture. Although osteoporosis is multifactorial, the two most common forms are *senile osteoporosis* due to aging-related losses of osteoblast function, and *postmenopausal osteoporosis* due to increased osteoclastic activity caused by the relative absence of estrogen.
- Paget disease may result from a paramyxovirus infection in genetically susceptible persons and is caused by aberrant and excessive osteoclast activity, followed by exuberant—but structurally unsound—osteoblast deposition of bone.
- Primary or secondary (due to renal failure) overproduction of PTH (*hyperparathyroidism*) results in increased osteoclast activity and bone resorption, leading to fractures and deformities.

Check your understanding:

1. **Which of the following diseases is caused by primary hyperparathyroidism?**
   A. osteoporosis
   B. milk alkali syndrome
   C. chronic renal failure
   D. adenoma of the gland

2. The main difference between type 1 and type 2 osteogenesis imperfecta is that type 2 is?
   A. Non lethal
   B. An acquired disorder.
   C. More common.
   D. More severe

3. Excessive stress on the feet can result in a fracture:
   A. True
   B. False

4. A scar which occurs in the bone following a fracture is called:
   A. Colles
   B. Callus

5. Osteoclasts are highly specialized cells capable of forming bone:
   A. True
   B. False

6. Lamellar bone is _____ woven bone:
   A. Much weaker & less efficient than
   B. Much stronger & more efficient than
   C. The same as
   D. None of above

7. Bone remodeling is a:
   A. Lifelong process
   B. Temporary process
   C. Special type of osteoclasts
   D. The organic matrix

8. A 45 years-old women diagnosed with breast cancer was walking on a wet floor and suddenly slipped acquiring a fracture in her right femur, what type of fracture is this?
   A. Traumatic
   B. Pathological
   C. Stress

1-A 2-D 3-A 4-B 5-B 6-B 7-A 8-B

9. Which one of the following is the main cause of secondary hyperparathyroidism?

A. Osteoporosis
B. Milk alkali syndrome
C. Chronic renal failure
D. Adenoma of the gland

10. Osteopetrosis has a problem in function and formation of which of the following?

A. Osteoblast
B. Osteoclast
C. Compact bone mass
D. Calcium level

11. An open fracture is a fracture that:

A. Result in multiple pieces of bone
B. Ruptures a blood vessel
C. Occurs when a broken bone pierces through the skin
D. Damages a nerve

12. Osteoporosis can result in a fracture:

A. True
B. False

13. A bone tumor can result in a fracture:

A. True
B. False

14. The longest part of the bone is the epiphysis:

A. True
B. False

15. The healing of a fractured bone in a wrong position is called:

A. Malunion
B. Nonunion

16. Secondary osteomyelitis mostly occurs in:

A. Compound fracture
B. Closed fracture
C. Both A & B
D. None of the above

9-C 10-B 11-C 12-A 13-A 14-B 15-A 16-A

17. Which one of the following is a feature of Thanatophoric Dwarfism?

A. Blue sclera
B. Deformed teeth
C. Small thorax
D. Loss of hearing

18. A patient with thanatophoric dwarfism could have a point mutation in:

A. FGFR3
B. COLIA2
C. COLIA1
D. NF-κB

19. The first step of fracture healing is:

A. callus formation
B. hematoma formation
C. Remodeling
D. Nonunion

20. A patient with Osteopetrosis could exhibit which one of the following :

A. Recurrent infections
B. Hypercalcemia
C. Loss of hearing
D. Loss of sight

21. Blue Sclera is a sign for type 1 and type 2 osteogenesis imperfecta :

A. True
B. False

22. What happens when OPG blocks RANKL?

A. Osteoclast activity will increase
B. Osteoclast activity will decrease

23. Open fractures are not prone to infection:

A. True
B. False

24. A 70-year-old male complains of right hip and thigh pain of several months duration. Radiographs of the pelvis and right leg reveal sclerotic, thickened cortical bone with a narrowed joint space near the acetabulum. the serum alkaline phosphatase level is elevated, but serum calcium and phosphorus concentrations are normal. the microscopic appearance of a bone biopsy is illustrated. the most likely condition accounting for these findings is:

A. Osteochondroma
B. Osteomalacia
C. Osteoarthritis
D. Osteitis deformans

17-C 18-A 19-B 20-A 21-A 22-B 23-B 24-D

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قال صلى الله عليه وسلم: من سلك طريقاً يلتمس به علماً سهل الله له به طريقاً إلى الجنة.

دعواتنا لكم بالتوفيق.