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SICOT e-Newsletter

EXAM CORNER

Paediatrics

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1) Which of the following statements is incorrect regarding sickle cell anaemia?

a) Sickle cell disease is more common than sickle cell trait
b) Bone infarction is more common than acute osteomyelitis in children with sickle cell disease who present with acute musculoskeletal pain
c) *Salmonella* infection is more commonly seen in children with sickle cell disease
d) Dactylitis (acute hand/foot swelling) is common
e) *Staphylococcus aureus* infection is the most common cause of osteomyelitis in affected patients

2) Which of the following statements is incorrect regarding sickle cell anaemia?

a) Mutation in the β-globin gene, results in sickle hemoglobin (HbS) production. When the cell becomes deoxygenated, HbS molecules assemble into fibers that produce a sickle-shaped red blood cell
b) Crises usually begin at ages 10 years, are caused by substance P, and may lead to characteristic bone infarctions
c) Growth retardation or skeletal immaturity, osteonecrosis of femoral and humeral heads, osteomyelitis, biconcave “fish” vertebrae, acetabular protrusio, and septic arthritis are common in this disorder
d) Aspiration and culture may be necessary to differentiate infarction from osteomyelitis
e) Hydroxyurea has produced dramatic relief of pain when used for bone crises

3) **Which of the following statements is incorrect regarding Thalassemia?**
    a) Thalassemias are a heterogenous group of autosomal dominant inherited disorders of hemoglobin synthesis
    b) Marrow hyperplasia can cause dramatic expansion of calvarial bones
    c) It is most commonly observed in people of Mediterranean descent
    d) Common symptoms include bone pain and leg ulceration
    e) Radiographic findings include long-bone thinning, metaphyseal expansion, osteopenia, and premature physeal closure
    f) Growth disturbance can result from the effects of transfusion-induced iron overload on the anterior pituitary gland and hypothalamus

4) **Which of the following statements is incorrect regarding Hemophilia?**
    a) Hemarthrosis manifests with painful swelling and decreased range of motion of affected joints
    b) The knee is the joint most commonly affected
    c) Deep intramuscular bleeding is also common and can lead to the formation of a pseudotumor, which can occur in soft tissue or bone
    d) X-linked recessive disorder with decreased amounts of factor VIII (hemophilia B, Leyden, or Christmas disease), abnormal factor VIII with platelet dysfunction (von Willebrand disease), or decreased amounts of factor VIII (hemophilia A)
    e) Intramuscular hematomas can lead to compression of adjacent nerves

5) **Which of the following statements is incorrect regarding Hemophilia?**
    a) Acute treatment of hemarthrosis is crucial and should begin immediately with administration of factor VIII or factor IX
    b) Aspiration of a hemarthrosis is controversial
    c) Radiographic findings include squaring of the patellae and condyles, epiphyseal overgrowth with leg-length discrepancy and increased osteoblastic activity
    d) Synovectomy is indicated for hemarthroses that recur despite optimal medical management
    e) Arthroscopy has better results with motion and duration of hospitalization than does open synovectomy

6) **Which of the following statements is incorrect regarding Osteogenesis Imperfecta?**
    a) Clinical features include bone fragility, scoliosis, tooth defects, hearing defects, blue sclerae in types I and II and ligamentous laxity
    b) Basilar invagination is common in more severe clinical phenotypes.
    c) Healing is normal, but bone typically does not remodel
    d) Fractures occur more frequently with advancing age
    e) Compression fractures (codfish vertebrae) are common

7) Which of the following statements is incorrect regarding Osteogenesis Imperfecta?

a) Defect in type I collagen (*COL1A2* gene) that causes abnormal cross-linking and leads to decreased collagen secretion
b) Histologic findings: increased diameters of haversian canals and osteocyte lacunae, increased numbers of cells, and replicated cement lines, which result in the thin cortices seen on radiographs
c) Sofield osteotomies are sometimes required for progressive bowing of long bones
d) Fractures in children younger than 2 years are treated similarly to those in children without osteogenesis imperfecta
e) Bisphosphonates have been shown to increase the number of fractures in these patients

8) Which of the following statements is incorrect regarding Idiopathic Juvenile Osteoporosis?

a) It appears between the ages of 8 and 14 years with osteopenia, growth arrest, and bone and joint pain
b) Typically, this disorder resolves spontaneously 2 to 4 years after the onset of puberty
c) Serum calcium and phosphorus levels are abnormal
d) It may be associated with multiple vertebral body microfractures
e) This disorder must be differentiated from other causes of osteopenia (e.g., osteogenesis imperfecta, malignancy, Cushing disease)

9) Which of the following statements is incorrect regarding Osteopetrosis?

a) Loss of the medullary canal can cause anemias and encroachment on the optic and oculomotor nerves, which in turn causes blindness
b) Failure of osteoclastic resorption, probably secondary to a defect in the thymus, leading to dense bone (so-called “marble” bone)
c) The mild form is autosomal recessive; the “malignant” form is autosomal dominant
d) In the spine, the centers of the vertebral bodies are relatively lucent, resulting in a “rugger jersey” (horizontally striped shirt) appearance on radiographs
e) Healing is normal, but amount of time for healing may be prolonged

10) Which of the following statements is incorrect regarding Infantile Cortical Hyperostosis (Caffey Disease)?

a) It is characterised by soft tissue swelling and bony cortical thickening (especially the jaw and ulna) that follow a febrile illness in infants 0 to 9 months old
b) This disorder may be differentiated from trauma and child abuse on the basis of single-bone involvement
c) Infection, scurvy, tumor, and progressive diaphyseal dysplasia may be included in the differential diagnosis
d) Periosteal reaction is not characteristic of this condition
e) The condition is benign and self-limiting

11) Which of the following statements is incorrect regarding Marfan syndrome?

a) Inheritance pattern is autosomal dominant
b) Defect is in fibrillin-1 (FBN1)
c) Arachnodactyly, pectus deformities, cardiac abnormalities, and ocular abnormalities are clinical manifestations
d) Scoliosis and acetabular protrusio are radiologic findings
e) Bracing for scoliosis is usually effective

12) Which of the following statements is incorrect regarding Ehlers Danlos Syndrome?

a) It is characterised by hyperextensibility of "cigarette paper" skin
b) It is associated with soft tissue and bone fragility, and soft tissue calcification
c) Of types I to XI, types II and III are the most common and least disabling
d) It is an autosomal recessive disorder of collagen V (co-expressed with collagen I)
e) Dislocation and kyphoscoliosis are radiologic findings

13) Which of the following statements is incorrect regarding Homocystinuria?

a) It is differentiated from Marfan syndrome on the basis of the direction of lens dislocation and the presence of osteoporosis in homocystinuria
b) Mental retardation is uncommon in this disorder
c) It is linked with an autosomal recessive inborn error of methionine metabolism (decreased enzyme cystathionine β-synthase)
d) The diagnosis is made by demonstrating increased homocysteine in urine (cyanide-nitroprusside test)
e) Early treatment with vitamin B_6 and a diet with decreased amounts of methionine are often successful

14) Which of the following statements is incorrect regarding Juvenile Idiopathic Arthritis?

a) It is characterised by a persistent noninfectious arthritis lasting 6 weeks to 3 months and diagnosed after other possible causes have been ruled out
b) It affects boys more than girls and typically manifests before age 4 years
c) Slit-lamp examination is required twice yearly, as progressive iridocyclitis can lead to rapid loss of vision if left untreated
d) Cervical spine involvement can lead to kyphosis, facet ankylosis, and atlantoaxial subluxation
e) Medical therapy involves less high-dose steroids and salicylates and more specific immunomodulating drugs

15) Which of the following is not associated with Ankylosing Spondylitis?

a) Typically affects adolescent boys with asymmetric, lower extremity, large-joint arthritis; heel pain; and sometimes eye symptoms
b) Hip and back pain may develop later
c) A positive HLA-B27 test result is a more specific finding than is a limitation of chest wall expansion
d) The HLA-B27 test yields positive results in 90% to 95% of patients with ankylosing spondylitis
e) Bilateral, symmetric sacroiliac erosion, followed by joint space narrowing, subsequent ankylosis, and late vertebral scalloping (bamboo spine) are radiologic findings

16) Which of the following statements is incorrect regarding Arthrogryposis?

a) A progressive disorder with multiple joints that are congenitally rigid
b) This disorder can be myopathic, neuropathic, or both
c) It is associated with a decrease in anterior horn cells and other neural elements of the spinal cord
d) Intelligence is normal
e) Evaluation should include neurologic studies, enzyme tests, and muscle biopsy at 3 to 4 months of age

17) Which of the following statements is incorrect regarding Arthrogryposis?

a) Affected patients typically have normal facies, normal intelligence, multiple joint contractures, and no visceral abnormalities
b) Upper extremity involvement usually includes adduction and internal rotation of the shoulder, flexion of the elbow, and flexion and ulnar deviation of the wrist. The elbow has prominent creases
c) Patients have an ability to use the feet as functional appendages
d) Lower extremity involvement includes teratologic hip dislocations, knee contractures, resistant clubfeet, and vertical talus
e) The spine may be involved, with characteristic C-shaped neuromuscular scoliosis

18) Which of the following statements is incorrect regarding Larsen syndrome?

a) It is characterised primarily by multiple joint dislocations including bilateral congenital knee dislocations, flattened facies, scoliosis, and clubfeet
b) Cervical kyphosis is important to recognise early and patient should be monitored for late myelopathy
c) Affected patients have normal intelligence
d) Similar to arthrogryposis in clinical appearance, but joints are more rigid
e) Knee reduction may necessitate femoral shortening and excision of collateral ligaments

19) Which of the following statements is incorrect regarding Myelodysplasia (Spina Bifida)?

a) Disorder of incomplete spinal cord closure or rupture of the developing cord secondary to hydrocephalus
b) Classification includes spina bifida occulta, meningocele, myelomeningocele and rachischisis
c) The myelodysplasia level is based on the lowest sensory level
d) Increased attention has been focused on latex sensitivity in myelodysplastic patients. A latex-free environment is necessary to prevent life-threatening allergic reactions
e) It can be diagnosed in utero by increased levels of α-fetoprotein

20) Which of the following statements is incorrect regarding Muscular Dystrophies?

a) They are noninflammatory, inherited disorders that are characterised by progressive muscle weakness
b) Duchenne muscular dystrophy is characterised by a markedly elevated creatine phosphokinase level and high levels of dystrophin protein on muscle biopsy and DNA testing
c) Inheritance pattern for Duchenne muscular dystrophy is sex-linked recessive
d) Physical findings for Duchenne muscular dystrophy manifest as muscle weakness (proximal groups weaker than distal), clumsy walking, decreased motor skills, lumbar lordosis, calf pseudohypertrophy and a positive Gowers sign
e) Patients usually die of cardiorespiratory complications before age 20

Answers:

1. Answer: a)
   Sickle cell disease (affects 1% of African Americans) is more severe but less common than sickle cell trait (8% prevalence).
   Reference: Miller review of Orthopaedics

2. Answer: b)
   Crises usually begin at ages 2-3 years, are caused by substance P, and may lead to characteristic bone infarctions
   Reference: Miller review of Orthopaedics

3. Answer: a)
   Thalassemias are a heterogenous group of autosomal recessive inherited disorders of hemoglobin synthesis
   References: Lovell & Winter's Pediatric Orthopaedics, Miller review of Orthopaedics

4. Answer: d)
   X-linked recessive disorder with decreased amounts of factor VIII (hemophilia A), abnormal factor VIII with platelet dysfunction (von Willebrand disease), or decreased amounts of factor IX (hemophilia B, Leyden, or Christmas disease)
   Reference: Miller review of Orthopaedics

5. Answer: c)
   Radiographic findings include squaring of the patellae and condyles, epiphyseal overgrowth with leg-length discrepancy and generalised osteopenia
   Reference: Miller review of Orthopaedics

6. Answer: d)
   Fractures occur less frequently with advancing age (usually cease at puberty)
   Reference: Miller review of Orthopaedics

7. Answer: e)
   Bisphosphonates have been shown to decrease the number of fractures in these patients
   Reference: Miller review of Orthopaedics

8. Answer: c)
   Serum calcium and phosphorus levels are normal
   Reference: Miller review of Orthopaedics

9. Answer: c)
   The mild form is autosomal dominant; the “malignant” form is autosomal recessive
   Reference: Miller review of Orthopaedics

10. Answer: d)
    Periosteal reaction is characteristic of this condition
    Reference: Miller review of Orthopaedics

11. Answer: e)
    Bracing for scoliosis is ineffective
    Reference: Miller review of Orthopaedics

12. Answer: d)
    It is an autosomal dominant disorder of collagen V (co-expressed with collagen I)
    Reference: Miller review of Orthopaedics

13. Answer: b)
    Central nervous system effects, including mental retardation, are common in this disorder
    Reference: Miller review of Orthopaedics

14. Answer: b)
    It affects girls more than boys and typically manifests before age 4 years
    Reference: Miller review of Orthopaedics

15. Answer: c)
    Limitation of chest wall expansion is a more specific finding than is a positive HLA-B27 test result. The HLA-B27 test yields positive results in 90% to 95% of patients with ankylosing spondylitis or Reiter syndrome, but the result is also positive in 4% to 8% of all white Americans; thus, its usefulness as a screening tool is limited.
    Reference: Miller review of Orthopaedics

16. Answer: a)
    A nonprogressive disorder with multiple joints that are congenitally rigid
    Reference: Miller review of Orthopaedics

17. Answer: b)
    Upper extremity involvement usually includes adduction and internal rotation of the shoulder, extension of the elbow, and flexion and ulnar deviation of the wrist. The elbow has no creases
    Reference: Miller review of Orthopaedics

18. Answer: d)
    Similar to arthrogryposis in clinical appearance, but joints are less rigid
    Reference: Miller review of Orthopaedics

19. Answer: c)
    The myelodysplasia level is based on the lowest functional level
    Reference: Miller review of Orthopaedics

20. Answer: b)
    Duchenne muscular dystrophy is characterised by a markedly elevated creatine phosphokinase level and absence of dystrophin protein on muscle biopsy and DNA testing
    Reference: Miller review of Orthopaedics