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SICOT e-Newsletter

Exam Corner

Paediatrics

Prepared by Mohamed Sukeik

1) **Which of the following statements is incorrect regarding achondroplasia?**
    a) Inheritance is autosomal dominant
    b) 80% of cases are caused by a spontaneous mutation in the fibroblast growth factor receptor 3 (FGFR3)
    c) Lumbar stenosis is the most likely cause of disability
    d) It is caused by failure in the cartilaginous proliferative zone of the physis
    e) It is a proportionate form of dwarfism caused by abnormal endochondral bone formation that is more affected than appositional growth

2) **Which of the following statements is incorrect regarding pseudoachondroplasia?**
    a) Pseudoachondroplasia is a disorder clinically similar to achondroplasia
    b) The inheritance pattern is autosomal recessive with a defect on chromosome 19 within the cartilage oligometric matrix protein (COMP)
    c) Radiographic findings include metaphyseal flaring and delayed epiphyseal ossification
    d) Orthopaedic manifestations include cervical instability; scoliosis with increased lumbar lordosis; significant lower extremity bowing; and hip, knee, and elbow flexion contractures with precocious osteoarthritis
    e) Normal facies is characteristic of the condition

3) **Which of the following statements is incorrect regarding Kniest syndrome?**
    a) The inheritance pattern is autosomal dominant
    b) Otitis media and hearing loss are frequent
    c) Aetiology includes a defect within type III collagen
    d) Orthopaedic manifestations include osteopenia and dumbbell-shaped bones
    e) Early therapy for joint contractures is required

4) **Which of the following statements is incorrect regarding metaphyseal chondroplasia?**
    a) Heterogeneous group of disorders characterized by metaphyseal changes of tubular bones with normal epiphyses
    b) The defect appears to be in the proliferative and hypertrophic zones of the physis
    c) Jansen type is the most severe form with a genetic defect in parathyroid hormone–related peptide
    d) Schmid type is a less severe form with a genetic defect in type I collagen and is transmitted by autosomal dominant inheritance
    e) McKusick type is transmitted by autosomal recessive inheritance

5) **Which of the following statements is incorrect regarding metaphyseal epiphyseal dysplasia?**
    a) Short-limbed, disproportionate dwarfism that often is not manifested until between the ages of 5 and 14
    b) Varus knees, waddling gait, and early hip arthritis are common
    c) Most common gene mutation is in COMP
    d) The proximal femoral involvement can be confused with Perthes disease
    e) It is bilateral and symmetric, is characterized by early acetabular changes, and is not accompanied by metaphyseal cysts

6) **Which of the following statements is incorrect regarding Morquio syndrome?**
    a) It is the most common form of mucopolysaccharidosis
    b) Inheritance pattern is autosomal recessive
    c) It is characterised by urinary excretion of dermatan/heparan sulfate
    d) Bony changes include a thickening skull; wide ribs; anterior beaking of vertebrae; a wide, flat pelvis; coxa vara with unossified femoral heads; and bullet-shaped metacarpals
    e) C1-C2 instability can be seen with Morquio syndrome

7) **Which of the following statements is incorrect regarding Hurler syndrome?**
    a) It is the most severe form of mucopolysaccharidosis
    b) Inheritance pattern is autosomal recessive
    c) It is characterised by urinary excretion of dermatan/heparan sulfate
    d) Bone marrow transplantation has increased the life span for patients with this disorder
    e) Mental retardation is not a feature of Hurler syndrome

8) **Which of the following statements is incorrect regarding Hunter syndrome?**
    a) It is the least severe form of mucopolysaccharidosis
    b) Inheritance pattern is autosomal recessive
    c) It is characterised by urinary excretion of dermatan/heparan sulfate
    d) Bone marrow transplantation has increased the life span for patients with this disorder
    e) Mental retardation is a feature of Hunter syndrome

9) Which of the following statements is incorrect regarding Sanfilippo syndrome?

a) Inheritance pattern is sex-linked recessive
b) It is characterised by urinary excretion of heparan sulfate
c) Bone marrow transplantation has increased the life span for patients with this disorder
d) Mental retardation is a feature of Sanfilippo syndrome
e) Spinal cord involvement is a feature of Sanfilippo syndrome

10) Which of the following statements is incorrect regarding Diastrophic Dysplasia?

a) Inheritance pattern is autosomal recessive
b) It is characterised by severe, short-limbed dwarfism and hitchhiker's thumb
c) Cauliflower ears are seen in 20% of cases
d) It is related to a deficiency in sulfate transport protein
e) Severe joint contractures especially hip and knee are common

11) Which of the following statements is incorrect regarding Diastrophic Dysplasia?

a) Cleft palate is seen in around 60% of cases
b) Flexible clubfeet are common
c) Cervical kyphosis, thoracolumbar kyphoscoliosis, spina bifida occulta, and atlantoaxial instability are radiologic findings
d) Quadriplegia is a major concern with deformities of the cervical spine
e) Early fusion of the C-spine may be indicated

12) Which of the following statements is incorrect regarding Cleidocranial Dysplasia (Dysostosis)?

a) Inheritance pattern is autosomal dominant
b) It is characterised by proportionate dwarfism that affects bones formed by intramembranous ossification
c) It is related to a defect in transcription factor for osteocalcin (CBFA1)
d) Hypoplasia or aplasia of the clavicle is common
e) Coxa valga is common

13) Which of the following statements is incorrect regarding Down syndrome?

a) Trisomy 21 is the most common chromosomal abnormality; its incidence increases with maternal age
b) Mental retardation, heart disease with atrial septal defect, endocrine disorders (hypothyroidism and diabetes) and premature aging are all features
c) Abnormal type II collagen is thought to be cause for generalised joint laxity in Down syndrome
d) Orthopaedic abnormalities include ligamentous laxity, hypotonia, hip instability, patellar dislocation and symptomatic planovalgus feet
e) Spinal abnormalities include atlantoaxial instability, scoliosis and spondylolisthesis

14) Which of the following statements is incorrect regarding Turner syndrome?

a) Affected patients are female and have short stature, lack of sexual development, webbed neck, and cubitus valgus
b) Idiopathic scoliosis is uncommon
c) It is associated with 45, XO Genotype
d) Malignant hyperthermia is common with anaesthetic use
e) Noonan syndrome has a same appearance except for normal gonadal development and mental retardation

15) Which of the following is not associated with Prader-Willi syndrome?

a) Floppy, hypotonic infant who grows up to be an intellectually impaired, obese adult with an insatiable appetite
b) Growth retardation
c) Normal genitalia
d) Hip dysplasia and juvenile-onset scoliosis
e) Partial chromosome 15 deletion

16) Which of the following statements is incorrect regarding Menkes syndrome?

a) Autosomal recessive disorder of copper transport
b) Characteristic “kinky” hair
c) May be differentiated from occipital horn syndrome in that the latter is characterised by bony projections from the occiput of the skull
d) Radiological features include: skull (wormian bones), long bones (metaphysial spurring)
e) Anterior flaring and multiple fractures of the ribs are common

17) Which of the following statements is incorrect regarding Rett syndrome?

a) Progressive impairment and stereotaxic, abnormal hand movements
b) Manifests in girls at 6 to 18 years of age
c) Loss of developmental milestones that is rapid and then stabilises
d) It is associated with a family of deletion mutations of the X-linked gene encoding a protein called methyl-CpG-binding protein 2 (MECP2)
e) It is associated with scoliosis with a C-shaped curve that is unresponsive to bracing

18) Which of the following statements is incorrect regarding Beckwith-Wiedemann syndrome?

a) Clinical features include organomegaly, omphalocele, and a large tongue
b) Orthopaedic manifestations include hemihypertrophy with spastic cerebral palsy
c) There is a predisposition to Wilms tumor
d) Spasticity is thought to be the result of infantile hyperglycemic episodes secondary to pancreatic islet cell hypertrophy
e) Growth arrest may be necessary in large limb

19) Which of the following statements is incorrect regarding Gaucher syndrome?

a) Aberrant autosomal recessive, lysosomal storage disease characterised by accumulation of sphingomyelin in reticuloendothelial system cells
b) The cause is a deficiency of the enzyme β-glucocerebrosidase
c) Clinical features include osteopenia, bone pain and hepatosplenomegaly
d) Radiological findings include metaphyseal enlargement, femoral head necrosis, "moth-eaten" trabeculae, patchy sclerosis, and Erlenmeyer flask deformity of the distal femora
e) Treatment is supportive; new enzyme therapy is available but is extremely expensive

20) Which of the following statements is incorrect regarding Niemann-Pick Disease?

a) Inheritance pattern is autosomal recessive
b) It is caused by an accumulation of cerebroside in reticuloendothelial system cells
c) It occurs commonly in Jews of eastern European descent
d) Marrow expansion and cortical thinning are common in long bones
e) Coxa valga can be a manifestation of the disease

Answers:

1. Answer: e)
   It is a disproportionate, short limbed form of dwarfism caused by abnormal endochondral bone formation that is more affected than appositional growth
   Reference: Miller review of Orthopaedics

2. Answer: b)
   The inheritance pattern is autosomal dominant with a defect on chromosome 19 within the cartilage oligometric matrix protein (COMP)
   Reference: Miller review of Orthopaedics

3. Answer: c)
   Aetiology includes a defect within type II collagen not III
   References: Miller review of Orthopaedics

4. Answer: d)
   Schmid type is a less severe form with a genetic defect in type X collagen and is transmitted by autosomal dominant inheritance
   Reference: Miller review of Orthopaedics

5. Answer: b)
   Valgus knees, waddling gait, and early hip arthritis are common
   Reference: Miller review of Orthopaedics

6. Answer: c)
   It is characterised by urinary excretion of keratan sulfate
   Reference: Miller review of Orthopaedics

7. Answer: e)
   Mental retardation is a feature of Hurler syndrome
   Reference: Miller review of Orthopaedics

8. Answer: b)
   Inheritance pattern is sex-linked recessive inheritance
   Reference: Miller review of Orthopaedics

9. Answer: a)
   Inheritance pattern is autosomal recessive
   Reference: Miller review of Orthopaedics

10. Answer: c)
    Cauliflower ears are seen in 80% of cases
    Reference: Miller review of Orthopaedics

11. Answer: b)
    Rigid clubfeet are common
    Reference: Miller review of Orthopaedics

12. Answer: e)
    Coxa vara is common
    Reference: Miller review of Orthopaedics

13. Answer: c)
    Abnormal type VI collagen is thought to be cause for generalised joint laxity in Down syndrome
    Reference: Miller review of Orthopaedics

14. Answer: b)
    Idiopathic scoliosis is common
    Reference: Miller review of Orthopaedics

15. Answer: c)
    Hypoplastic genitalia is associated with Prader-Willi syndrome
    Reference: Miller review of Orthopaedics

16. Answer: a)
    Sex-linked recessive disorder of copper transport
    Reference: Miller review of Orthopaedics

17. Answer: b)
    Manifests in girls at 6 to 18 months of age
    Reference: Miller review of Orthopaedics

18. Answer: d)
    Spasticity is thought to be the result of infantile hypoglycemic episodes secondary to pancreatic islet cell hypertrophy
    Reference: Miller review of Orthopaedics

19. Answer: a)
    Aberrant autosomal recessive, lysosomal storage disease characterised by accumulation of cerebroside in reticuloendothelial system cells
    Reference: Miller review of Orthopaedics

20. Answer: b)
    It is caused by an accumulation of sphingomyelin in reticuloendothelial system cells
    Reference: Miller review of Orthopaedics