RACP Written Examination February 2018

Paediatrics & Child Health

Clinical Applications questions 1–100
1. A 7-year-old boy whose parents separated 3 months ago was brought in by his mother, who he lives with. He has been sleeping in his mother's bed and refusing to go to school. He loses his temper often and has been verbally aggressive towards his mother's new partner. He refuses to comply with requests made by his mother. When he was last at school, he was verbally aggressive and punched a wall before running home. He is settled in his grandmother's presence. He spends most of his time playing video games and does not want to play with his friends, despite having enjoyed this in the past.

What is the most likely diagnosis?

A. Attention deficit hyperactivity disorder.
B. Conduct disorder.
C. Major depressive disorder.
D. Oppositional defiant disorder.
E. Separation anxiety disorder.
2. A baby is delivered urgently because of fetal tachycardia during labour. She is well at birth, but has a rapid and irregular heart rate. The following ECG is recorded. The rhythm proves unresponsive to repeated doses of intravenous adenosine.

What treatment is most appropriate to achieve reversion to sinus rhythm?

A. Amiodarone infusion.
B. Electrical cardioversion.
C. Electrophysiologic ablation.
D. Oral sotalol.
E. Overdrive pacing.
3. A 13-year-old boy diagnosed with ulcerative colitis is admitted with increased bloody diarrhoea and fevers. An abdominal X-ray is performed.

What is the most likely explanation for the boy’s clinical deterioration?
A. Ileus.
B. Large bowel ischaemia.
C. Spontaneous colonic perforation.
D. Toxic megacolon.
E. Volvulus.
4. A 2-year-old girl presents with a 4-week history of progressively enlarging submandibular lymphadenopathy as shown below. She is otherwise well. The parents recall that she grazed her chin prior to the lymphadenopathy developing.

What is the most likely infectious agent to cause this condition?

A. *Actinomyces* species.
B. *Bartonella henselae*.
C. *Mycobacterium avium intracellulare*.
D. *Nocardia* species.
E. *Staphylococcus aureus*. 
5. A 5-month-old boy presents with fevers, poor feeds and worsening breathlessness. He has failure to thrive with head sparing. His saturation is 92% in air and normalises in oxygen. He is tachycardic and tachypnoeic. He has widespread crepitations and a harsh systolic murmur at the left sternal edge. Polymerase chain reaction (PCR) from a nasal swab is positive for respiratory syncytial virus (RSV). His chest x-ray is shown below.

What is the most likely diagnosis?

A. Acute myocarditis.
B. Atrial septal defect.
C. Dilated cardiomyopathy.
D. Tetralogy of Fallot.
E. Ventricular septal defect.
6. A 2-year-old boy presents with a 3-day history of cough and fever. He was admitted to hospital 6 weeks ago with a left lower lobe pneumonia which was treated appropriately with antibiotics. Chest x-ray demonstrates persistent opacity in the left lower lobe. A CT chest is requested and the results demonstrate a lung lesion with an arterial connection to the descending aorta as shown below.

What is the most likely diagnosis of the lesion?

A. Bronchogenic cyst.
B. Congenital cystic adenomatoid malformation.
C. Congenital lobar emphysema.
D. Pulmonary sequestration.
E. Teratoma.
7. A 12-year-old southeast Asian boy with a past history of a small ventricular septal defect presents in heart failure. He is afebrile. He is thin, has a bulging precordium and a prominent apical impulse. He is not cyanosed or clubbed. He has a pansystolic murmur and a high pitched early diastolic murmur at the left sternal edge. His chest x-ray shows cardiomegaly without plethora. His blood count and inflammatory markers are normal.

What complication of his original lesion best explains his heart failure?

A. Acute rheumatic fever.
B. Aortic valve regurgitation.
C. Eisenmenger syndrome.
D. Increasing left to right shunt.
E. Subacute bacterial endocarditis.
8. An infant born at 27 weeks is being ventilated for respiratory distress syndrome on the following settings.

Mode: Synchronised Intermittent Positive Pressure Ventilation + Volume Guarantee

- Peak inspiratory pressure (PIP) limit: 25 cm H$_2$O
- Positive end-expiratory pressure (PEEP): 5 cm H$_2$O
- Inspiratory time (Ti): 0.3 seconds
- Back up rate: 40 breaths/minute
- Set tidal volume (V$_t$): 5 mL/kg
- FiO$_2$: 28%

A blood gas done at 12 hours of age shows:

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>pH</td>
<td>7.35</td>
<td>[7.25–7.35]</td>
</tr>
<tr>
<td>CO$_2$</td>
<td>37 mmHg</td>
<td>[40–60]</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>22</td>
<td>[18–22]</td>
</tr>
<tr>
<td>Base excess</td>
<td>-2</td>
<td>[-2 to 4]</td>
</tr>
<tr>
<td>Lactate</td>
<td>1.2</td>
<td>[&lt; 2]</td>
</tr>
</tbody>
</table>

What is the most appropriate change to the ventilator settings?

A. Decrease peak inspiratory pressure limit.
B. Decrease respiratory rate.
C. Decrease tidal volume.
D. Increase inspiratory time.
E. Increase peak end-expiratory pressure.
9. An 8-year-old boy presents with a 3-day history of fever, lethargy and nausea. On examination he is alert, febrile (37.7 °C), and has vague abdominal tenderness with a palpable liver. He has a widespread macular rash with several petechial spots on his legs. The remainder of his examination is normal.

Investigation results are shown:

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alanine aminotransferase (ALT)</td>
<td>85 U/L</td>
<td>[&lt; 45]</td>
</tr>
<tr>
<td>Alkaline phosphatase (ALP)</td>
<td>390 U/L</td>
<td>[80–350]</td>
</tr>
<tr>
<td>Bilirubin</td>
<td>8 μmol/L</td>
<td>[&lt; 25]</td>
</tr>
<tr>
<td>Albumin</td>
<td>34 g/L</td>
<td>[34–48]</td>
</tr>
<tr>
<td>Total protein</td>
<td>68 g/L</td>
<td>[66–84]</td>
</tr>
<tr>
<td>Activated partial thromboplastin time (aPTT)</td>
<td>48 seconds</td>
<td>[25–37]</td>
</tr>
<tr>
<td>aPTT 1+1/mixing study</td>
<td>46 seconds</td>
<td></td>
</tr>
<tr>
<td>Prothrombin ratio</td>
<td>1.1</td>
<td>[0.8–1.2]</td>
</tr>
<tr>
<td>Fibrinogen</td>
<td>3 g/L</td>
<td>[1.5–4]</td>
</tr>
</tbody>
</table>

Full blood count is normal.

What is the most likely cause of his prolonged aPTT?

A. Disseminated intravascular coagulation.
B. Mild haemophilia A.
C. Transient antiphospholipid antibodies.
D. Viral hepatitis.
E. von Willebrand disease.
10. Which of the following neurodevelopmental tests is most predictive of long-term motor outcomes in a preterm infant?

A. Amiel-Tison neurological assessment.
B. Dubowitz neurological examination.
C. General movement assessment.
D. NICU Network Neurobehavioural Scale.
E. Test of infant motor performance.

11. An 8-year-old girl is short and has two café-au-lait spots on her back. She is below the 3rd centile for height and weight. On a full blood count she has a mild normocytic anaemia, a platelet count of $109 \times 10^9/L$ [150–600], normal white cell count and a mild neutropaenia of $0.9 \times 10^9/L$ [1.0–8.0].

On further questioning her parents say she had a urinary tract infection as a toddler and on ultrasound had the incidental finding of a single kidney.

What is the most likely diagnosis?

A. Dyskeratosis congenita.
B. Fanconi anaemia.
C. Neurofibromatosis.
D. Noonan syndrome.
E. Turner syndrome.
12. An 8-year-old boy with known tuberous sclerosis is referred to hospital for review of recurrent abdominal pain. Renal ultrasound shows a 2 cm mass in the left kidney, with multiple smaller (< 4 mm) lesions bilaterally.

What is the most likely diagnosis of the larger lesion?

A. Angiomyolipoma.
B. Arteriovenous malformation.
C. Haemangioblastoma.
D. Renal cell carcinoma.
E. Wilms tumour.

13. A 9-year-old girl is referred for investigation of 10 days of abdominal pain and diarrhoea. Her general practitioner has performed an ultrasound which reveals ileitis. On examination she has multiple red tender nodules on her shins.

What is the most likely causative organism?

A. *Bacillus cereus*.
B. *Campylobacter jejuni*.
C. *Salmonella enteriditis*.
D. *Staphylococcus aureus*.
E. *Yersinia enterocolitica*. 
14. A 14-year-old girl, with known type 1 diabetes and poor glycaemic control presents to the emergency department acutely unwell after spending 3 days at her friend’s house. She has been vomiting for 24 hours.

On examination, she is tachypnoeic, tachycardic and her blood pressure is 85/60 mmHg. She is cool peripherally and has dry lips. She responds to pain and voice, but is very tired.

A venous gas taken after intravenous cannulation shows the following results:

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>pH</td>
<td>6.82</td>
<td>[7.32–7.43]</td>
</tr>
<tr>
<td>pCO₂</td>
<td>27 mmHg</td>
<td>[37–50]</td>
</tr>
<tr>
<td>pO₂</td>
<td>38 mmHg</td>
<td>[34–44]</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>4 mmol/L</td>
<td>[22–28]</td>
</tr>
<tr>
<td>Base excess</td>
<td>-30 mmol/L</td>
<td>[-3–3]</td>
</tr>
<tr>
<td>Sodium</td>
<td>131 mmol/L</td>
<td>[133–144]</td>
</tr>
<tr>
<td>Potassium</td>
<td>5.8 mmol/L</td>
<td>[3.5–5.0]</td>
</tr>
<tr>
<td>Glucose</td>
<td>41.6 mmol/L</td>
<td>[3.0–5.4]</td>
</tr>
</tbody>
</table>

What is the most appropriate initial management?

A. Intravenous bicarbonate infusion.
B. Intravenous dextrose and insulin infusion.
C. Intravenous mannitol.
D. Intravenous normal saline bolus.
E. Subcutaneous insulin.
15. You are asked to see an 8-year-old boy with retained deciduous and supernumerary teeth. On further examination you note he has short stature, his fontanelle is still open, and he has hypermobile shoulders as shown.

What is the diagnosis?

A. Cleidocranial dysostosis.
B. Ehlers–Danlos syndrome.
C. Facioscapulohumeral dystrophy.
D. Klippel–Feil syndrome.
E. Osteogenesis imperfecta type 1.
16. What disorder of sexual development is associated with McCune–Albright syndrome?

A. Androgen resistance.
B. Hyperandrogenism and virilisation.
C. Hypergonadotrophic hypogonadism.
D. Hypogonadotrophic hypogonadism.
E. Precocious puberty.
17. A 9-year-old boy is referred for assessment of cough and shortness of breath on exertion. Lung function testing is performed as shown below.

What abnormality is shown?

A. Mixed restrictive and obstructive lung disease.
B. Obstructive lung disease.
C. Poor technique.
D. Restrictive lung disease.
E. Upper airway obstruction.
18. A previously well 15-year-old boy presents with a 12-hour history of central chest pain. There is some, but little, variation with respiration and posture. He is not febrile.

On examination, he is anxious and tachycardic. He has normal heart sounds, no murmur and no evidence of heart failure. Blood tests reveal a mild leucocytosis and troponin elevation to 386 ng/L [< 30 ng/L]. The following ECG is recorded.

What is the most likely diagnosis?

A. Costochondritis.
B. Endocarditis.
C. Myocardial infarction.
D. Myocarditis.
E. Pericarditis.
19. A 6-month-old boy presents to hospital critically unwell with 10 days of progressive tachypnoea and cough. He has been previously well and is thriving. *Pneumocystis jiroveci* is identified on bronchoalveolar lavage. His blood count is normal.

Immunoglobulin results are:

<p>| | | |</p>
<table>
<thead>
<tr>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>IgG</td>
<td>0.2 g/L</td>
<td>[1.6–5.8]</td>
</tr>
<tr>
<td>IgA</td>
<td>&lt; 0.07 g/L</td>
<td>[0–0.8]</td>
</tr>
<tr>
<td>IgM</td>
<td>5.2 g/L</td>
<td>[0.3–1.3]</td>
</tr>
</tbody>
</table>

Inherited deficiency or dysfunction of which protein best explains this pattern of immune deficiency?

A. Bruton's tyrosine kinase.
B. CD40 ligand.
C. Common gamma chain.
D. NF-kappa B.
E. STAT5b.

20. A previously well 10-year-old girl presents to hospital acutely unwell with lobar pneumonia and persistent vomiting. She has a history of reaction to cefaclor at age 5 with 5 days of maculopapular rash including target lesions, swollen feet and arthralgia. She has never been prescribed penicillins due to a family history of penicillin anaphylaxis affecting her father.

What is the most appropriate antibiotic for treatment of her pneumonia?

A. Ceftriaxone.
B. Ciprofloxacin.
C. Clindamycin.
D. Penicillin.
E. Vancomycin.

What is the most appropriate test to investigate the possibility of fructose intolerance contributing to her symptoms?

A. Faecal calprotectin.
B. Hydrogen breath test.
C. Serum aldolase B enzyme measurement.
D. Small bowel biopsy with disaccharidase level measurement.
E. Stool reducing substances.

22. Which of the following fracture types has the highest specificity for inflicted injury?

A. Anterior rib fractures.
B. Metaphyseal corner fracture of tibia.
C. Parietal skull fracture.
D. Spiral fracture of long bone.
E. Supracondylar fracture of humerus.

23. A 14-year-old girl presents complaining of daytime tiredness. She falls asleep at school at her desk. When her friends share a joke, she feels her "legs give way" and can slump to the floor. She falls asleep when she gets home from school, sleeping for 2 hours before waking to have dinner and complete her homework. She goes to bed at 9 pm, sleeps through the night and wakes at 7 am. Her family report that she does not snore.

What is the most likely diagnosis?

A. Chronic fatigue syndrome.
B. Gelastic seizures.
C. Narcolepsy.
D. Obstructive sleep apnoea.
E. Sleep phase delay syndrome.
24. What is the most effective treatment for oppositional defiant disorder?

A. Cognitive behavioural therapy directed at the child.
B. Multisystemic therapy aimed at family and school.
C. Parent management training.
D. Social-emotional skills training directed at the child.
E. Stimulant medication.

25. In which genetic condition does Hirschsprung disease occur more commonly than in the general population?

A. 22q11.2 microdeletion.
B. Angelman syndrome.
C. Charcot–Marie–Tooth disease.
D. Congenital central hypoventilation syndrome.
E. Rett syndrome.
26. An 8-year-old boy is seen in the emergency department with a 24 hour history of pruritic rash associated with swollen lips, hands and feet. He was not on any medications and there is no history of known allergy. He was unwell with a viral URTI 1 week prior to this illness.

On examination, the child appeared uncomfortable and was afebrile with no respiratory distress. The rash is shown in the picture below.
What is the diagnosis?

A. Anaphylaxis.
B. Erythema multiforme.
C. Pityriasis rosea.
D. Stevens–Johnson syndrome.
E. Urticaria multiforme.

27. A 7-year-old girl is referred to outpatients for parental concern about the development of pubic hair. On examination she has Tanner P2 pubic hair and Tanner B1 breast development. Her examination is otherwise normal. She is on the 75th percentile for height and weight, with normal growth velocity.

The following investigations are performed:

Bone age: skeletal age is 7 years.

Androgen profile:

<table>
<thead>
<tr>
<th>Androgen</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>17-OHP progesterone</td>
<td>1.2 nmol/L</td>
<td>[0.2–1.9]</td>
</tr>
<tr>
<td>Dehydroepiandrosterone sulfate (DHEAS)</td>
<td>1.7 µmol/L</td>
<td>&lt; 1.5</td>
</tr>
<tr>
<td>Androstenedione</td>
<td>0.6 nmol/L</td>
<td>[0.1–1.5]</td>
</tr>
<tr>
<td>Testosterone</td>
<td>0.2 nmol/L</td>
<td>&lt; 0.5</td>
</tr>
<tr>
<td>Estradiol (E2)</td>
<td>63 pmol/L</td>
<td>&lt; 100</td>
</tr>
</tbody>
</table>

What is her likely outcome with respect to growth and puberty?

A. Early puberty and reduced final height.
B. Early puberty and normal final height.
C. Late puberty and normal final height.
D. Late puberty and reduced final height.
E. Normal puberty and normal final height.
28. A 7-year-old girl presents with a 6-week history of lower limb discomfort and increasing limp, particularly on the right. In the 2 weeks prior to presentation she has missed a significant amount of school as a result of her musculoskeletal symptoms. Her physical examination is normal except for a small effusion of her right knee.

Investigations reveal:

Full blood examination:

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Haemoglobin</td>
<td>111 g/L</td>
<td>[115–155]</td>
</tr>
<tr>
<td>White cell count</td>
<td>$4.5 \times 10^9$/L</td>
<td>[5.0–14.5]</td>
</tr>
<tr>
<td>Neutrophils</td>
<td>$0.68 \times 10^9$/L</td>
<td>[1.5–8.0]</td>
</tr>
<tr>
<td>Lymphocytes</td>
<td>$3.6 \times 10^9$/L</td>
<td>[1.5–7.0]</td>
</tr>
<tr>
<td>Monocytes</td>
<td>$0.14 \times 10^9$/L</td>
<td>[0.1–1.0]</td>
</tr>
<tr>
<td>Eosinophils</td>
<td>$0.09 \times 10^9$/L</td>
<td>[0–0.5]</td>
</tr>
<tr>
<td>Platelets</td>
<td>$102 \times 10^9$/L</td>
<td>[150–400]</td>
</tr>
<tr>
<td>Erythrocyte sedimentation rate (ESR)</td>
<td>90 mm/hr</td>
<td>[&lt; 10]</td>
</tr>
<tr>
<td>C-reactive protein</td>
<td>40 mg/L</td>
<td>[&lt; 8]</td>
</tr>
<tr>
<td>Urea and electrolytes</td>
<td>Normal</td>
<td></td>
</tr>
<tr>
<td>Liver function tests</td>
<td>Normal</td>
<td></td>
</tr>
<tr>
<td>Antinuclear antibodies</td>
<td>Positive 1:160 speckled.</td>
<td></td>
</tr>
</tbody>
</table>

An x-ray of the right knee (shown below) showed multiple ill-defined areas of lucency within both the distal femoral and proximal fibula and tibial metaphyses extending into the proximal diaphyses.
What is the most likely diagnosis?

A. Acute leukaemia.
B. Oligoarticular juvenile idiopathic arthritis.
C. Osgood–Schlatter disease.
D. Reactive arthritis.
E. Subacute osteomyelitis.
29. A 9-year-old child is referred with arachnodactyly and an increased arm span to height ratio (> 1.05). Further examination reveals hypertelorism and a bifid uvula. Cognitive development is normal. Echocardiography shows marked dilatation of the aortic root (z-score + 4.0).

What is the most likely diagnosis?

A. Familial thoracic aortic aneurysm syndrome.
B. Homocystinuria.
C. Loeys–Dietz syndrome.
D. Marfan syndrome.
E. Shprintzen–Goldberg syndrome.

30. A 13-year-old girl had been diagnosed with liver disease secondary to Wilson disease. Two weeks after commencing penicillamine, she has developed a widespread itchy and erythematous rash. You believe this to be drug-related and discontinue the penicillamine.

What treatment should you commence for ongoing copper chelation?

A. Citrate.
B. Desferrioxamine.
C. Dimercaprol.
D. EDTA.
E. Trientine.
31. A 9-year-old boy has developed nausea, vomiting and abdominal pain 4 hours after eating a ham sandwich.

Which organism is most likely to have caused his symptoms?

A. Campylobacter jejuni.
B. Escherichia coli.
C. Salmonella enterica.
D. Staphylococcus aureus.
E. Yersinia enterocolitica.
32. An 18-month-old female is brought to the emergency department with haematuria. She has been unwell the past 2 days with rhinorrhoea and four episodes of diarrhoea. Her examination is unremarkable.

The urine sample is shown below and is sent for culture which is negative.

What is the most likely organism?

A. Adenovirus.
B. Enterovirus.
C. Norovirus.
D. Parvovirus.
E. Rhinovirus.
33. A term newborn presents with respiratory distress and a chest x-ray confirms the presence of bilateral pleural effusions. The infant is dysmorphic with hypertelorism, posteriorly rotated ears and redundant nuchal skin. At 2 weeks of age the infant develops marked pallor and petechiae and is diagnosed with juvenile myelomonocytic leukaemia.

What is the most likely underlying condition?

A. Cockayne syndrome.
B. Fanconi anaemia.
C. Noonan syndrome.
D. Omenn syndrome.
E. Velo-cardio-facial syndrome.
34. The changes seen in the nailfold of this 11-year-old girl are most typically found in which condition?

A. Infective endocarditis.
B. Juvenile dermatomyositis.
C. Linear scleroderma.
D. Microscopic polyangiitis.
E. Systemic lupus erythematosus.
35. Following acute bronchiolitis, which virus is associated with the long-term complication of bronchiolitis obliterans?

A. Adenovirus.
B. Human metapneumovirus.
C. Influenza virus.
D. Parainfluenza virus.
E. Respiratory syncytial virus.

36. A 12-year-old girl with cystic fibrosis is reviewed in outpatients following a hospital admission with her first episode of renal colic. She has pancreatic insufficiency being treated with pancreatic enzyme replacement. Urinary results are shown below.

<table>
<thead>
<tr>
<th>Random urine</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Citrate</td>
<td>0.70 mmol/L</td>
</tr>
<tr>
<td></td>
<td>[0.40–3.40]</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>24 hr urine</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Volume</td>
<td>630 mL</td>
</tr>
<tr>
<td>Creatinine</td>
<td>5.2 mmol/24 hr</td>
</tr>
<tr>
<td></td>
<td>[1.5–10.0]</td>
</tr>
<tr>
<td>Calcium</td>
<td>5.5 mmol/24 hr</td>
</tr>
<tr>
<td></td>
<td>[2.5–7.5]</td>
</tr>
<tr>
<td>Urate</td>
<td>1.3 mmol/24 hr</td>
</tr>
<tr>
<td></td>
<td>[1.5–4.4]</td>
</tr>
<tr>
<td>Oxalate</td>
<td>0.55 mmol/1.73 m²/24 hr</td>
</tr>
<tr>
<td></td>
<td>[&lt; 0.5]</td>
</tr>
</tbody>
</table>

Which of the following strategies is most effective in decreasing the risk of further calculi?

A. Decreasing dietary calcium intake.
B. Decreasing dietary potassium intake.
C. Increasing daily fluid intake.
D. Increasing dietary citrate intake.
E. Increasing dietary protein intake.
37. A 9-year-old girl presents with her third episode of bilateral parotitis. On examination, she has a low grade fever and firm, tender, enlarged parotid glands with no other abnormalities.

Investigations reveal:

<table>
<thead>
<tr>
<th>Test</th>
<th>Result</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Haemoglobin</td>
<td>128 g/L</td>
<td>[115–155]</td>
</tr>
<tr>
<td>White cell count</td>
<td>4.5 × 10⁹/L</td>
<td>[5.0–14.5]</td>
</tr>
<tr>
<td>Neutrophils</td>
<td>3.68 × 10⁹/L</td>
<td>[1.5–8.0]</td>
</tr>
<tr>
<td>Lymphocytes</td>
<td>0.6 × 10⁹/L</td>
<td>[1.5–7.0]</td>
</tr>
<tr>
<td>Monocytes</td>
<td>0.14 × 10⁹/L</td>
<td>[0.1–1.0]</td>
</tr>
<tr>
<td>Eosinophils</td>
<td>0.09 × 10⁹/L</td>
<td>[0–0.5]</td>
</tr>
<tr>
<td>Platelets</td>
<td>235 × 10⁹/L</td>
<td>[150–400]</td>
</tr>
<tr>
<td>Erythrocyte sedimentation rate (ESR)</td>
<td>30 mm/hr</td>
<td>[&lt; 20]</td>
</tr>
<tr>
<td>C-reactive protein</td>
<td>22 mg/L</td>
<td>[&lt; 10]</td>
</tr>
<tr>
<td>Antinuclear antibodies</td>
<td>1:640 speckled</td>
<td></td>
</tr>
<tr>
<td>ENA</td>
<td>Ro detected</td>
<td></td>
</tr>
<tr>
<td>Rheumatoid factor</td>
<td>90 IU/mL</td>
<td>[&lt; 40]</td>
</tr>
<tr>
<td>IgG</td>
<td>30 g/L</td>
<td>[4.95–16.56]</td>
</tr>
<tr>
<td>ACE negative</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

What is the most likely diagnosis?

A. HIV infection.
B. Recurrent mumps infection.
C. Recurrent parotitis of childhood.
D. Sarcoidosis.
E. Sjögren syndrome.
38. A 4-year-old boy is assessed for failure to thrive, having not gained weight over the preceding year. He was becoming increasingly irritable and lethargic and is now refusing foods. His mother reported abdominal bloating and excess wind over recent months. A symmetrical rash is noted on his elbows and knees. As part of a nutritional screen, his transglutaminase IgA antibodies were positive.

Which of the following is the most likely diagnosis?

A. Dermatitis herpetiformis.
B. Discoid eczema.
C. Henoch–Schönlein purpura.
D. Impetigo.
E. Papular acrodermatitis.

39. You are called after hours by the mother of a 14-year-old girl with well-controlled type 1 diabetes, who has been recently started on insulin pump therapy. The mother states that she has recorded a capillary blood glucose level of 20.2 mmol/L, and ketones of 1.2 mmol/L [< 0.5]. Earlier in the day her blood glucose level was normal at 6.5 mmol/L, and she had been correctly given insulin with her dinner meal through the insulin pump. Her mother reports that apart from feeling thirsty, her daughter is well, with no nausea or vomiting.

With respect to her hyperglycaemia, what is the best advice you should give?

A. Deliver a correction bolus of rapid acting insulin through the insulin pump.
B. Deliver a correction bolus of rapid acting insulin via an insulin syringe or pen.
C. Deliver a dose of long acting insulin.
D. Perform 30 minutes of exercise and drink 500 mL of water.
E. Start a temporary increased basal insulin rate.
40. Which renal condition is most commonly associated with congenital hepatic fibrosis?

A. Autosomal recessive polycystic kidney disease.
B. Horseshoe kidney.
C. Multicystic dysplastic kidney.
D. Nephronophthisis.
E. Renal hypodysplasia.

41. An 8-year-old boy develops a severe rash and mucositis 10 days after commencing carbamazepine for the treatment of epilepsy. What is the main feature that differentiates between Stevens–Johnson syndrome and toxic epidermal necrolysis?

A. Extent of skin detachment.
B. Number of sites of mucosal involvement.
C. Presence of purpuric macules.
D. Response to intravenous steroids.
E. Time to onset of rash.
42. A 3-year-old girl is treated for a sore throat and otitis media with oral amoxycillin. Nine days after commencing treatment she develops a widespread rash as shown below, fever and swollen knees.

What is the most likely diagnosis?

A. Acute rheumatic fever.
B. Epstein–Barr virus infection.
C. Henoch–Schönlein purpura.
D. Serum sickness-like reaction.
E. Systemic-onset juvenile idiopathic arthritis.
43. What is the most important serious side effect that parents should be warned about before commencing atomoxetine?

A. Cardiac toxicity.
B. Depression.
C. Potential for abuse.
D. Seizures.
E. Suicidal ideation.

44. Night terrors typically occur at what time during sleep?

A. 60–90 minutes after going to sleep.
B. 60–90 minutes before waking.
C. During REM sleep.
D. On waking.
E. Within 30 minutes of going to sleep.

45. What is the most common early visual finding in a child with idiopathic intracranial hypertension?

A. Afferent pupillary defect.
B. Diplopia.
C. Loss of colour vision.
D. Reduced visual acuity.
E. Visual field deficit.
46. A clinician managing a child with abdominal pain places greater weight on clinical findings that support his working diagnosis of mesenteric adenitis, and ignores laboratory evidence which is consistent with the correct diagnosis of appendicitis. This is consistent with which type of bias?

A. Confirmation.
B. Confounding.
C. Information.
D. Recall.
E. Selection.

47. A proactive fasting test is organised for a 3-year-old with a history of episodic hypoglycaemia. Results at 6 hours are shown below:

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glucose</td>
<td>2.9 mmol/L</td>
<td>[3.4–6.53]</td>
</tr>
<tr>
<td>3 hydroxybutyrate</td>
<td>&lt; 50 µmol/L</td>
<td>[50–100]</td>
</tr>
<tr>
<td>Free fatty acids</td>
<td>573 µmol/L</td>
<td>[&lt; 100]</td>
</tr>
<tr>
<td>Insulin</td>
<td>&lt; 1 mIU/L</td>
<td>[2–23]</td>
</tr>
<tr>
<td>Growth hormone</td>
<td>12 mU/L</td>
<td>[post-stimulation &gt; 10 mU/L, post-suppression &lt; 0.5 mU/L]</td>
</tr>
<tr>
<td>Cortisol</td>
<td>350 nmol/L</td>
<td>[60–570]</td>
</tr>
<tr>
<td>Ammonia</td>
<td>55 µmol/L</td>
<td>[&lt; 50]</td>
</tr>
<tr>
<td>Lactate</td>
<td>&lt; 1 mmol/L</td>
<td>[0.5–2.2]</td>
</tr>
</tbody>
</table>

What is the likely diagnosis?

A. Fatty acid oxidation disorder.
B. Glycogen storage disorder.
C. Hyperinsulinism.
D. Idiopathic ketotic hypoglycaemia.
E. Mitochondrial respiratory chain defects.
48. Which condition is caused by maternal uniparental disomy of chromosome 7?

A. Angelman syndrome.
B. Beckwith–Wiedemann syndrome.
C. Neonatal diabetes.
D. Prader–Willi syndrome.
E. Russell–Silver syndrome.

49. A 12-year-old boy is seen in outpatients for review following an admission to hospital with glomerulonephritis. He had initially presented with macroscopic haematuria, oedema, mild hypertension and investigations showed hypocomplementaemia. He was treated with frusemide with improvement in oedema and on review one month after discharge had been well with no abnormal clinical findings.

On review 12 weeks after his admission, his mother reports ongoing intermittent facial oedema, occurring more commonly in the morning. On examination, blood pressure is 120/80 mmHg and he has mild peripheral oedema to the knees. Dipstick testing of urine shows heavy proteinuria and haematuria. Results of repeat biochemistry and serology are shown below.

**Serum**

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Level</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sodium</td>
<td>137 mmol/L</td>
<td>[137–147]</td>
</tr>
<tr>
<td>Potassium</td>
<td>4.3 mmol/L</td>
<td>[3.5–5.0]</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>22 mmol/L</td>
<td>[25–33]</td>
</tr>
<tr>
<td>Creatinine</td>
<td>70 µmol/L</td>
<td>[10–70]</td>
</tr>
<tr>
<td>Albumin</td>
<td>15 g/L</td>
<td>[35–50]</td>
</tr>
<tr>
<td>Cholesterol</td>
<td>10.4 mmol/L</td>
<td>[3.1–6.5]</td>
</tr>
<tr>
<td>Complement C3</td>
<td>0.20 g/L</td>
<td>[0.70–1.60]</td>
</tr>
<tr>
<td>Antinuclear antibodies (ANA)</td>
<td>negative</td>
<td></td>
</tr>
</tbody>
</table>

What is the most likely diagnosis?

A. Alport syndrome.
B. IgA nephropathy.
C. Membranoproliferative glomerulonephritis.
D. Post-streptococcal glomerulonephritis.
E. Systemic lupus erythematosus.
50. A 28-day-old male presents to the emergency department with increasing vomiting and listlessness on the background of poor weight gain. He weighs 3.3 kg (z-score -2.2) and has not regained his birth weight of 3.7 kg (z-score 0.7). On examination, he is cachectic with dry mucous membranes and had a capillary refill time of 4 seconds and blood pressure of 76/38 mmHg. Fluid resuscitation was instituted with normal saline.

Investigations are shown below:

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>pH</td>
<td>7.29</td>
<td>[7.35–7.45]</td>
</tr>
<tr>
<td>pCO₂</td>
<td>35 mmHg</td>
<td>[34–43]</td>
</tr>
<tr>
<td>Sodium</td>
<td>111 mmol/L</td>
<td>[133–144]</td>
</tr>
<tr>
<td>Potassium</td>
<td>6.7 mmol/L</td>
<td>[3.3–4.9]</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>18 mmol/L</td>
<td>[19–28]</td>
</tr>
<tr>
<td>Lactate</td>
<td>2.8 mmol/L</td>
<td>[&lt; 1.3]</td>
</tr>
<tr>
<td>Cortisol</td>
<td>550 nmol/L</td>
<td>[80–600]</td>
</tr>
<tr>
<td>Renin</td>
<td>1048 mU/L</td>
<td>[5–100]</td>
</tr>
<tr>
<td>Aldosterone</td>
<td>7.05 nmol/L</td>
<td>[0.3–1.5]</td>
</tr>
</tbody>
</table>

Urine dipstick showed leucocytes, trace ketones, and no glucose.

What is the most likely diagnosis?

A. Aldosterone synthase deficiency.
B. Congenital adrenal hyperplasia.
C. Pyloric stenosis.
D. Syndrome of inappropriate antidiuretic hormone secretion.
E. Urinary tract infection.
51. A term infant becomes increasingly unwell from day 2 with feeding difficulty, hypotonia, vomiting, lethargy and seizures.

Initial investigations are shown:

<table>
<thead>
<tr>
<th></th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glucose</td>
<td>4.5 mmol/L</td>
</tr>
<tr>
<td>pH</td>
<td>7.35</td>
</tr>
<tr>
<td>Anion gap</td>
<td>normal</td>
</tr>
<tr>
<td>Ammonia</td>
<td>580 μmol/L</td>
</tr>
</tbody>
</table>

Feeds are stopped and IV dextrose commenced.

Which other specific therapy is indicated to treat the hyperammonaemia?

A. Cobalamin (B₁₂).
B. Folic acid.
C. Glycine.
D. L-carnitine.
E. Sodium benzoate.
52. A 2-year-old with acute abdominal pain has been diagnosed with pancreatitis. An ultrasound is performed which reveals a significantly dilated common bile duct, tapering distally with dilatation of intrahepatic ducts. Results of liver function tests are shown below.

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alanine aminotransferase</td>
<td>65 U/L</td>
<td>[5–35]</td>
</tr>
<tr>
<td>Alkaline phosphatase</td>
<td>580 U/L</td>
<td>[80–250]</td>
</tr>
<tr>
<td>Gamma glutamyltransferase</td>
<td>749 U/L</td>
<td>[5–40]</td>
</tr>
<tr>
<td>Total bilirubin</td>
<td>39 µmol/L</td>
<td>[0–20]</td>
</tr>
<tr>
<td>Lipase</td>
<td>1200 U/L</td>
<td>[0–60]</td>
</tr>
</tbody>
</table>

What is the most likely cause for her pancreatitis?

A. Bile duct adenoma.
B. Choledochal cyst.
C. Neonatal sclerosing cholangitis.
D. Sphincter of Oddi dysfunction.
E. Viral hepatitis.

53. Which of the following is the most common clinical feature at presentation in acute rheumatic fever (ARF)?

A. Arthritis.
B. Carditis.
C. Erythema marginatum.
D. Subcutaneous nodules.
E. Sydenham's chorea.
54. A 3-year-old girl is admitted with right middle lobe pneumonia and commenced on intravenous antibiotics. After 3 days of treatment she remains febrile with worsening shortness of breath. A repeat chest x-ray is performed and is shown below.

What complications of pneumonia are demonstrated in this x-ray?

A. Bronchiectasis and empyema.
B. Lung abscess and empyema.
C. Lung abscess and pneumothorax.
D. Pneumatocoele and empyema.
E. Pneumatocoele and pneumothorax.
55. A 10-month-old boy is referred to clinic for investigation of hepatomegaly. He had hypoglycaemia in the neonatal period thought to be secondary to neonatal sepsis, but has otherwise been well and is developing and growing normally. An ultrasound shows marked hepatomegaly with increased echogenicity and no splenomegaly.

Biochemistry is shown below.

<table>
<thead>
<tr>
<th></th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sodium</td>
<td>138 mmol/L</td>
<td>[135–145]</td>
</tr>
<tr>
<td>Potassium</td>
<td>4.6 mmol/L</td>
<td>[3.5–5.0]</td>
</tr>
<tr>
<td>Chloride</td>
<td>101 mmol/L</td>
<td>[101–111]</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>22 mmol/L</td>
<td>[22–32]</td>
</tr>
<tr>
<td>Urea</td>
<td>2.5 mmol/L</td>
<td>[1.8–6.0]</td>
</tr>
<tr>
<td>Creatinine</td>
<td>14 µmol/L</td>
<td>[15–30]</td>
</tr>
<tr>
<td>Lactate</td>
<td>4.2 mmol/L</td>
<td>[0.5–2.2]</td>
</tr>
<tr>
<td>Uric acid</td>
<td>501 µmol/L</td>
<td>[80–300]</td>
</tr>
<tr>
<td>Alkaline phosphatase (ALP)</td>
<td>212 U/L</td>
<td>[80–310]</td>
</tr>
<tr>
<td>Gamma glutamyltransferase (GGT)</td>
<td>45 U/L</td>
<td>[5–50]</td>
</tr>
<tr>
<td>Alanine transaminase (ALT)</td>
<td>120 U/L</td>
<td>[5–40]</td>
</tr>
<tr>
<td>Total bilirubin</td>
<td>5 µmol/L</td>
<td>[0–20]</td>
</tr>
<tr>
<td>Albumin</td>
<td>43 g/L</td>
<td>[35–45]</td>
</tr>
</tbody>
</table>

What is the most likely diagnosis?

A. Congenital hepatic fibrosis.
B. Glycogen storage disease.
C. Hepatoblastoma.
D. Lysosomal storage disease.
E. Non-alcoholic steatohepatitis.
56. Which tumour commonly presents with a mediastinal mass?

A. Ewing sarcoma.
B. Germ cell tumour.
C. Neuroblastoma.
D. Pre-B acute lymphoblastic leukaemia.
E. T-cell acute lymphoblastic lymphoma.

57. A 7-year-old boy presents with a history of bedwetting at night time, at least 3–4 times per week. He has no associated daytime urinary symptoms. Friends have started inviting him over for sleepovers, but he is too embarrassed to go as he is worried that he will wet the bed. You discuss treatment options with his parents.

Which treatment approach has the lowest relapse rates?

A. Bedwetting alarm.
B. Desmopressin (DDAVP).
C. Imipramine.
D. Oxybutynin.
E. Psychotherapy.

58. What are the recognised facial features of fetal alcohol spectrum disorder in early childhood?

A. Broad forehead, flat nasal bridge, malar flattening and wide mouth.
B. Long and narrow face with prominent forehead and chin (prognathism) and large ears.
C. Low-set and posteriorly rotated ears, ocular hypertelorism, and a bulbous nasal tip.
D. Short palpebral fissure, thin vermilion border, smooth philtrum.
E. Upslanting palpebral fissure, flat facial profile/flat nasal bridge and low set small ears.
59. Which antiepileptic is associated with the highest risk of congenital malformation?

A. Carbamazepine.
B. Lamotrigine.
C. Levetiracetam.
D. Sodium valproate.
E. Topiramate.

60. A previously well 3-year-old boy arrives in the emergency department following a collapse at home. CPR is in progress but he has been given no medications prior to arrival. His rhythm strip taken just prior to arrival at hospital is shown.

What is the correct initial management?

A. 10 mcg/kg of adrenaline 1:1000.
B. 10 mcg/kg of adrenaline 1:10000.
C. 5 mg/kg intravenous amiodarone.
D. Immediate DC shock at 2 J/kg.
E. Immediate DC shock at 4 J/kg.
61. A 3-day-old baby girl presents to the emergency department with vaginal bleeding. The baby looks well on examination.

What is the most likely cause?

A. Haematocolpos.
B. Hymen tear.
C. Neonatal alloimmune thrombocytopenia.
D. Normal physiological response.
E. Vitamin K deficiency.

62. A 12-year-old girl in the orthopaedic ward undergoes investigation for prolonged bleeding from the wound post-operatively. The following coagulation studies are obtained:

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prothrombin time</td>
<td>28 s</td>
<td>8–14</td>
</tr>
<tr>
<td>International normalised ratio</td>
<td>2.7</td>
<td>0.8–1.2</td>
</tr>
<tr>
<td>Activated partial thromboplastin time</td>
<td>33 s</td>
<td>24–38</td>
</tr>
<tr>
<td>Thrombin clotting time</td>
<td>11 s</td>
<td>11–14</td>
</tr>
<tr>
<td>Fibrinogen</td>
<td>2.8 g/L</td>
<td>2.0–6.0</td>
</tr>
</tbody>
</table>

Deficiency of which coagulation factor best explains this pattern of results?

A. Factor II.
B. Factor VII.
C. Factor VIII.
D. Factor X.
E. Factor XII.
63. An 18-month-old girl has a 4-week history of progressively unsteady gait. She also cries when her mother lifts her legs to change her nappy. An MRI is reported as showing lumbosacral discitis and osteomyelitis.

What is the most likely organism?

A. *Bartonella henselae.*
B. *Escherichia coli.*
C. *Kingella kingae.*
D. *Staphylococcus aureus.*
E. *Streptococcus pyogenes.*
64. A 7-month-old boy presents with abdominal distension and increased crying. He has a large right-sided mass and undergoes a CT examination. He also has mildly abnormal liver function tests and normal beta-HCG and alpha-fetoprotein is extremely high.

What is the most likely tumour given the images below?

A. Adrenocortical carcinoma.
B. Burkitt's lymphoma.
C. Hepatoblastoma.
D. Neuroblastoma.
E. Wilms tumour.
65. A 3-year-old boy is referred for assessment of mild microcytic anaemia. He is clinically well, and his general examination is unremarkable. The following results were obtained 4 weeks after the local General Practitioner advised commencement of an oral iron supplement:

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Haemoglobin</td>
<td>90 g/L</td>
<td>[95–125]</td>
</tr>
<tr>
<td>Mean cell volume</td>
<td>72 fL</td>
<td>[76–98]</td>
</tr>
<tr>
<td>Serum iron</td>
<td>27 µmol/L</td>
<td>[5–25]</td>
</tr>
<tr>
<td>Transferrin</td>
<td>3.3 g/L</td>
<td>[1.9–3.1]</td>
</tr>
<tr>
<td>TIBC</td>
<td>86 µmol/L</td>
<td>[47–77]</td>
</tr>
<tr>
<td>Saturation</td>
<td>31%</td>
<td>[20–45]</td>
</tr>
<tr>
<td>Ferritin</td>
<td>5 µg/L</td>
<td>[30–200]</td>
</tr>
</tbody>
</table>

What is the likely explanation for these results?

A. Chronic inflammation.
B. Congenital hypoferritinaemia.
D. Non-adherence to therapy.
E. Thalassaemia trait.
66. A 7-year-old previously well female is airlifted to hospital following a motor vehicle accident in a rural area. A secondary survey has identified her major injury to be bilateral compound femoral fractures. Upon her arrival at a tertiary facility she is administered intravenous antibiotics and given a transfusion of packed red blood cells. Within 10 minutes of commencement of transfusion she is noted to have worsening tachycardia, and shortly afterwards develops profound hypotension. There are no cutaneous findings and her chest is clear on auscultation. She is blood group A Rh D negative and the transfused bag is group O Rh D positive.

What is the most likely cause of the patient's shocked state?

A. Anaphylaxis.
B. Bacterial sepsis.
C. Hypovolemia.
D. Incompatible transfusion.
E. Raised intracranial pressure.

67. A 16-month-old infant presents with a history of irritability and delayed development. He has knee hyperextension causing genu recurvatum and is unable to walk. Deep tendon reflexes are diminished. An MRI scan shows symmetric confluent areas of high signal intensity in the periventricular white matter.

What is the most likely diagnosis?

A. Farber disease.
B. Krabbe disease.
C. Metachromatic leukodystrophy.
D. Multiple sulfatase deficiency.
E. Type C Niemann–Pick disease.
68. Which intervention is most effective in preventing nosocomial infections in the neonatal intensive care unit?

A. Antibiotic impregnated catheters.
B. Chlorhexidine for neonatal skin cleansing.
C. Nursing in an incubator.
D. Probiotics for the neonate.
E. Strict hand hygiene.

69. A 2-day-old term neonate is found to have bilious vomiting.

What would be the most appropriate diagnostic investigation?

A. Endoscopy.
B. Lower GI contrast study.
C. Plain x-ray of the abdomen.
D. Ultrasound of the abdomen.
E. Upper GI contrast study.

70. An 8-day-old preterm infant on CPAP is found to have bilateral yellow discharge from the eyes with hyperemic conjunctiva.

What is the most appropriate management?

A. Benzylpenicillin.
B. Erythromycin.
C. Massage of inner canthus.
D. Normal saline wash.
E. Silver nitrate eye drops.
71. A 9-month-old previously well child who has received three doses of conjugate pneumococcal vaccine is hospitalised with pneumococcal meningitis. Her mother asks why she has this infection despite being vaccinated.

What is the most likely explanation?

A. Agammaglobulinaemia.
B. A non-vaccine serotype.
C. Complement deficiency.
D. Hyposplenism.
E. Occult CSF leak.

72. You wish to determine the extent to which smoking during pregnancy reduces birth weight. Which study design best answers this question?

A. Case-control.
B. Ecological.
C. Prospective cohort.
D. Randomised controlled trial.
E. Retrospective cohort.
73. A 4-month-old girl is seen in outpatients for follow up of antenatally detected left hydronephrosis. Initial renal ultrasound at 4 days of age showed a left duplex kidney with dilatation of the left upper pole moiety, dilatation of the left lower pole ureter and ureterocoele. Since birth, she had been well and thriving. Repeat renal ultrasound at 3 months of age reports increasing dilatation of the left upper pole moiety (antero-posterior pelvic diameter 15 mm), with no change in the ureterocoele and a normal right kidney.

What is the most appropriate investigation?

A. CT pyelogram.
B. Dimercaptosuccinic acid (DMSA) scan.
C. Mercaptoacetyltriglycine (MAG3) scan.
D. Micturating cystourethrogram.
E. MR urogram.
A 3-year-old girl is investigated for episodic ataxia and epilepsy. She has myoclonic and generalised tonic clonic seizures. She is taking sodium valproate.

On examination, she is not dysmorphic, her height and weight are on the 75th centile and her head circumference is on the 3rd centile.

The following metabolic investigations were performed:

**Cerebrospinal fluid**

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>White blood cells</td>
<td>0 × 10⁶/L</td>
<td></td>
</tr>
<tr>
<td>Red blood cells</td>
<td>0 × 10⁶/L</td>
<td></td>
</tr>
<tr>
<td>Protein</td>
<td>0.2 g/L</td>
<td>[0.15–0.45]</td>
</tr>
<tr>
<td>Glucose</td>
<td>1.9 mmol/L</td>
<td></td>
</tr>
<tr>
<td>Lactate</td>
<td>1.0 mmol/L</td>
<td></td>
</tr>
<tr>
<td>Amino acids</td>
<td>normal</td>
<td>[0.9–2.1]</td>
</tr>
</tbody>
</table>

**Urine**

Organic acids normal

**Blood**

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Calcium</td>
<td>2.5 mmol/L</td>
<td>[2.1–2.8]</td>
</tr>
<tr>
<td>Glucose</td>
<td>5.2 mmol/L</td>
<td>[3.5–5.4]</td>
</tr>
<tr>
<td>Ammonia</td>
<td>32 μmol/L</td>
<td>[&lt; 40]</td>
</tr>
<tr>
<td>Lactate</td>
<td>0.8 mmol/L</td>
<td>[0.5–2.2]</td>
</tr>
<tr>
<td>Pyruvate</td>
<td>0.07 mmol/L</td>
<td>[0.06–0.19]</td>
</tr>
<tr>
<td>pH</td>
<td>7.37</td>
<td>[7.35–7.45]</td>
</tr>
<tr>
<td>Bicarbonate</td>
<td>25 mmol/L</td>
<td>[22–31]</td>
</tr>
<tr>
<td>Acylcarnitine profile</td>
<td>normal</td>
<td></td>
</tr>
<tr>
<td>Amino acids</td>
<td>normal</td>
<td>mildly elevated glycine</td>
</tr>
</tbody>
</table>

What is the diagnosis?

A. Glucose transporter 1 (GLUT1) deficiency.
B. Methylmalonic acidaemia.
C. Myoclonic epilepsy with red-ragged fibres (MERRF).
D. Neuronal ceroid lipofuscinosis.
E. Non-ketotic hyperglycinaemia.
75. You are called to the postnatal ward to assess a 4-day old male neonate who has red discolouration noted in the nappy. The infant was born at 42 weeks gestation with a birth weight of 4.3 kg, after a pregnancy complicated by gestational diabetes. Apgar score at birth was 5 at 1 minute and 8 at 5 minutes. The infant had mild respiratory distress at birth that had settled over the subsequent 24 hours and had otherwise been well, aside from some difficulty in establishing feeds. On examination, there were no dysmorphic features. Cardiorespiratory examination was normal. Examination of the abdomen revealed a firm mass in the left flank.

What is the most likely diagnosis?

A. Multicystic dysplastic kidney.
B. Pelviureteric junction obstruction.
C. Renal vein thrombosis.
D. Teratoma.
E. Wilms tumour.

76. Conduct disorder is a known precursor to which personality disorder?

A. Antisocial.
B. Borderline.
C. Histrionic.
D. Narcissistic.
E. Schizotypal.
77. A 7-year-old boy is referred to clinic with a 2-month history of urticaria occurring most days without apparent reason, sometimes associated with marked angioedema of his lips. Individual lesions persist for several days. He is on regular antihistamine with cetirizine 5 mg daily. He has dermatographism on examination.

Which feature best differentiates urticarial vasculitis from chronic idiopathic urticaria?

A. Angioedema.
B. Dermatographism.
C. Lack of trigger.
D. Persistence of individual lesions.
E. Poor response to antihistamine.

78. You are requested to urgently review a 15-year-old boy with autism spectrum disorder presenting with acute onset torticollis. He has recently commenced risperidone 2 mg for behavioural disturbance.

What is the most appropriate treatment for his torticollis?

A. Benztropine.
B. Ibuprofen.
C. Lorazepam.
D. Olanzapine.
E. Promethazine.
79. A 17-year-old boy with first-episode psychosis who has been prescribed risperidone is observed to have orofacial tardive dyskinesia involving repetitive minor tongue protrusion. This has not bothered him.

What is the most appropriate management?

A. Change to an alternative antipsychotic.
B. Continue current treatment.
C. Prescribe benztropine.
D. Prescribe vitamin E.
E. Reduce the dose of risperidone.

80. What is the most appropriate diagnostic molecular genetic test in a child where there is a strong clinical suspicion of Angelman syndrome?

A. Exome sequencing.
B. Methylation sensitive multiplex ligand probe amplification.
C. Microarray.
D. Sequencing of the target gene.
E. Uniparental disomy studies.
81. A 3-year-old girl with acute lymphoblastic leukaemia is recovering from a prolonged episode of febrile neutropenia and bacteraemia with *Klebsiella pneumoniae* for which she has been treated with 2 weeks of broad spectrum antibiotics. She develops abdominal pain and distension with hepatosplenomegaly.

The abdominal CT scan (shown below) is reported as showing multiple lesions in the liver, spleen and kidney.

![Abdominal CT scan](image)

What is the most likely organism responsible for her current illness?
A. *Aspergillus fumigatus.*
B. *Candida albicans.*
C. Cytomegalovirus.
D. Methicillin-resistant *Staphylococcus aureus.*
E. *Pseudomonas aeruginosa.*

82. A 3-year-old child has a large boil on her thigh with worsening cellulitis despite 4 days of treatment with oral cephalexin. Her siblings have also had boils in the past 3 months.

Which antibiotic is indicated for empiric treatment while awaiting culture results of a swab sent from the purulent discharge?

A. Amoxicillin-clavulanate.
B. Ciprofloxacin.
C. Erythromycin.
D. Flucloxacillin.
E. Trimethoprim-sulphamethoxazole.

83. A 16-year-old boy experiences acute visual loss and is found to have severe bilateral optic atrophy, pseudoedema of the optic disc and evidence of circumpapillary telangiectatic microangiopathy. He is diagnosed with Leber optic neuropathy.

What is the inheritance of this condition?

A. Autosomal dominant.
B. Autosomal recessive.
C. Mitochondrial.
D. Multifactorial.
E. X-linked recessive.
84. A 4-year-old girl has been diagnosed with hereditary angioedema after recurrent bouts of severe abdominal pain and vomiting. She presents with a 4 hour history of progressive swelling of her face and lips with concern about possible respiratory obstruction.

What is the most appropriate treatment?

A. Intramuscular adrenaline.
B. Intravenous purified C1 inhibitor.
C. Oral prednisolone.
D. Oral promethazine.
E. Oral tranexamic acid.
85. A 7-year-old girl presents with a 4-week history of cough, shortness of breath on exertion and recent haemoptysis. Over the last 12 months she has been treated for persistent iron deficiency anaemia. She has normal renal function. A chest x-ray demonstrates diffuse alveolar opacity.

What is the most likely diagnosis?

A. Churg–Strauss syndrome.
B. Goodpasture syndrome.
C. *Pneumocystis jiroveci* pneumonia.
D. Pulmonary fibrosis.
E. Pulmonary haemosiderosis.
86. An 8-year-old girl returned 4 days ago from 3 weeks in a rural area in the Philippines. She presents with 3 days of fever to 39 °C and complains of severe pain all over her body. On examination she is very miserable but has no specific abnormal findings.

A full blood count shows:

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Haemoglobin</td>
<td>9.9 g/L</td>
<td>[110–140]</td>
</tr>
<tr>
<td>Platelet count</td>
<td>45 × 10^9/L</td>
<td>[150–400]</td>
</tr>
</tbody>
</table>

Malaria screen is negative.

What is the most likely diagnosis?

A. Chikungunya.
B. Dengue fever.
C. Hepatitis A.
D. Leptospirosis.
E. Typhoid fever.

87. Which central nervous system tumour is commonly associated with multiple cranial nerve palsies?

A. Diffuse intrinsic pontine glioma.
B. Ependymoma.
C. Glioblastoma multiforme.
D. Medulloblastoma.
E. Pilocytic astrocytoma.
88. A 10-year-old boy presents acutely with painful itchy eyes with photophobia, ropey discharge, a feeling of grit in his eyes and decreased vision and has been diagnosed with vernal keratoconjunctivitis.

What acute complication of this condition mandates urgent ophthalmology review?

A. Acute glaucoma.
B. Corneal shield ulcer.
C. Giant papillae of the tarsal conjunctivae.
D. Horner–Trantas dots.
E. Posterior uveitis.

89. A 6-month-old boy is investigated for lethargy and pallor. His parents have fed him exclusively with goat’s milk from 4 weeks of age due to issues with colic. He is gaining weight, and the history is otherwise unremarkable. Examination is normal apart from pallor.

The full blood count is shown:

<table>
<thead>
<tr>
<th></th>
<th>Normal values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Haemoglobin</td>
<td>51 g/L</td>
</tr>
<tr>
<td>Mean cell volume</td>
<td>96 fL</td>
</tr>
<tr>
<td>White cell count</td>
<td>5.5 x 10^9/L</td>
</tr>
<tr>
<td>Neutrophils</td>
<td>1.82 x 10^9/L</td>
</tr>
<tr>
<td>Platelets</td>
<td>110 x 10^9/L</td>
</tr>
<tr>
<td>Reticulocytes</td>
<td>15 x 10^9/L</td>
</tr>
</tbody>
</table>

Deficiency of which micronutrient best explains this clinical picture?

A. Folate.
B. Iron.
C. Vitamin B12.
D. Vitamin E.
E. Zinc.
90. A journal reports that therapeutic hypothermia for moderate-severe neonatal hypoxic ischaemic encephalopathy leads to a reduction in combined outcome of mortality and major neurodevelopmental disability (typical relative risk (RR) 0.75, 95% confidence interval (CI) 0.68 to 0.83; typical risk difference (RD) −0.15, 95% CI −0.2 to −0.1).

What is the number needed to treat?

A. \( \frac{1}{0.75} \)

B. \( \frac{1}{1 - 0.75} \)

C. \( \frac{1}{1 - 0.15} \)

D. \( \frac{1}{0.15} \)

E. \( 1 - 0.15 \)

91. A 14-year-old girl who broke up with her boyfriend 6 weeks ago is brought in by her concerned parents. She has barely left her bedroom since the break-up. She reports feeling tired, needing to sleep more than usual, and not wanting to spend time with friends. She has not been attending school regularly because she has difficulty concentrating in class. Her parents report she has been uncharacteristically irritable when they try to encourage her to go to school. She protests when her parents leave her at home alone. She denies suicidal ideation.

What is the most likely diagnosis?

A. Adjustment disorder with depressed mood.

B. Bipolar affective disorder.

C. Borderline personality disorder.

D. Major depressive disorder.

E. Separation anxiety disorder.
92. What is the specific antidote for a tricyclic antidepressant overdose?

A. Flumazenil.
B. Fomepizole.
C. Insulin euglycaemic therapy.
D. Sodium bicarbonate infusion.
E. Sodium Ethylenediaminetetraacetic acid (EDTA).
QUESTIONS 93 AND 94 REFER TO THE FOLLOWING INFORMATION
What is the most likely diagnosis for this infant?

93. A 5-day-old baby is found to be in severe heart failure with marked tachycardia, tachypnoea and hepatomegaly. He is poorly perfused and mottled. All of his pulses are poor. He has an active precordium. On auscultation he has a gallop rhythm, a click and an ejection systolic murmur at the base.

A. Arteriovenous malformation.
B. Atrioventricular septal defect.
C. Coarctation of the aorta.
D. Critical aortic stenosis.
E. Hypoplastic left heart.
F. Patent duct.
G. Truncus arteriosus.
H. Ventricular septal defect.

94. A 1-month-old baby presents with slow feeds and poor weight gain. She is small and has dysmorphic features suggestive of 22q11.2 deletion syndrome. She has an oxygen saturation of 91% in air. She has prominent systolic and diastolic murmurs and a large liver. Her chest x-ray demonstrates marked cardiomegaly and plethora.

A. Arteriovenous malformation.
B. Atrioventricular septal defect.
C. Coarctation of the aorta.
D. Critical aortic stenosis.
E. Hypoplastic left heart.
F. Patent duct.
G. Truncus arteriosus.
H. Ventricular septal defect.
QUESTIONS 95 AND 96 REFER TO THE FOLLOWING INFORMATION
The following phenotype is most suggestive of which neuromuscular disease?

95. A 12-year-old girl has weakness which worsens over the course of a day. At times, she has also noted difficulty climbing stairs. On examination she has mild ptosis and grade 4/5 power proximally. She has normal reflexes.

A. Collagen VI related myopathy.
B. Duchenne muscular dystrophy.
C. Fascioscapulohumeral muscular dystrophy.
D. Myasthenia gravis.
E. Myotonia congenita.
F. Myotonic dystrophy.
G. Nemaline myopathy.
H. Spinal muscular atrophy.

96. A 2-year-old girl has episodic difficulty in opening her eyes. Her mother has noticed that this is worse during cold weather. On examination, she has generalised muscular hypertrophy. Her creatine kinase is mildly elevated.

A. Collagen VI related myopathy.
B. Duchenne muscular dystrophy.
C. Fascioscapulohumeral muscular dystrophy.
D. Myasthenia gravis.
E. Myotonia congenita.
F. Myotonic dystrophy.
G. Nemaline myopathy.
H. Spinal muscular atrophy.
QUESTIONS 97 AND 98 REFER TO THE FOLLOWING INFORMATION

For this clinical presentation select the most likely infectious agent.

97. A 7-year-old boy has had fever and lethargy for 3 days. He is pale with petechiae over his chest, and has a temperature of 39.4 ºC, widespread lymphadenopathy and hepatosplenomegaly. He is pancytopenic with raised ferritin and triglycerides. A bone marrow biopsy confirms haemophagocytic lymphohistiocytosis. There is a family history of a maternal uncle on immunoglobulin replacement for hypogammaglobulinaemia.

A. Adenovirus.
B. *Burkholderia cepacia*.
C. *Cytomegalovirus*.
D. Epstein–Barr virus.
E. *Haemophilus* group B.
F. *Pneumocystis jiroveci*.
G. *Staphylococcus aureus*.
H. *Streptococcus pneumoniae*.

98. A 6-month-old boy presents with 4-days of spiking fever. Investigation shows a dense left-sided pneumonia and abdominal ultrasound shows multifocal liver abscesses. His dihydrorhodamine (DHR) result is markedly abnormal.

A. Adenovirus.
B. *Burkholderia cepacia*.
C. *Cytomegalovirus*.
D. Epstein–Barr virus.
E. *Haemophilus* group B.
F. *Pneumocystis jiroveci*.
G. *Staphylococcus aureus*.
H. *Streptococcus pneumoniae*. 
QUESTIONS 99 AND 100 REFER TO THE FOLLOWING INFORMATION
What is the correct interpretation of the thyroid function tests for the following clinical presentation?

99. A 10-year-old girl with trisomy 21 is seen for routine follow up. She is well, growing appropriately and examination of her thyroid is normal. Health surveillance includes thyroid function tests which are shown below.

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal Range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Thyroid-stimulating hormone (TSH)</td>
<td>6.8 mU/L</td>
<td>[0.5–4.5]</td>
</tr>
<tr>
<td>Free thyroxine</td>
<td>13 pmol/L</td>
<td>[11–22]</td>
</tr>
<tr>
<td>Free T3</td>
<td>4.3 pmol/L</td>
<td>[3.5–5.8]</td>
</tr>
</tbody>
</table>

A. Central hyperthyroidism.
B. Central hypothyroidism.
C. Euthyroid.
D. Primary hyperthyroidism.
E. Primary hypothyroidism.
F. Sick euthyroid syndrome.
G. Subclinical hypothyroidism.
H. Thyroid resistance syndrome.
100. A 2-week-old infant presents with poor feeding, prolonged jaundice and lethargy. On arrival the baby has a temperature of 38.2 °C, heart rate 140, respiratory rate of 40/minute, and mottled skin. He is mildly jaundiced. Blood cultures are positive at 18 hours for Group B *Streptococcus*.

Thyroid function tests were performed to investigate jaundice and results are shown below.

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>T₃</td>
<td>1.2 pmol/L</td>
<td>[3–10]</td>
</tr>
<tr>
<td>Free thyroxine</td>
<td>10 pmol/L</td>
<td>[10–40]</td>
</tr>
<tr>
<td>Thyroid-stimulating hormone (TSH)</td>
<td>10 mU/L</td>
<td>[0.4–16]</td>
</tr>
</tbody>
</table>

A. Central hyperthyroidism.
B. Central hypothyroidism.
C. Euthyroid.
D. Primary hyperthyroidism.
E. Primary hypothyroidism.
F. Sick euthyroid syndrome.
G. Subclinical hypothyroidism.
H. Thyroid resistance syndrome.