



Welcome to the MRCPCH Theory and Science Specimen Paper

This document provides a specimen Theory and Science (TAS) paper, designed to illustrate the types of questions used in that examination. It includes 20 sample questions with accompanying reasoning and references.

Important Note for Candidates

This specimen paper illustrates the format and style used in the TAS examination. Although the examination is based on current UK practice, the questions are written to ensure international candidates will understand the context. Please note that this paper is for illustrative purposes only and cannot reflect future examination papers in terms of distribution of questions (the Blueprint) or level of difficulty. While it can be a helpful resource for exam preparation, it should not be used as a definitive guide to the actual exam content.

All questions that provide laboratory results will also include the age-adjusted normal ranges. These may differ slightly from local laboratories but are appropriate for interpretation of a given question

Expected exam conduct

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Question 1 – Best answer and reasoning

Stem question:

What is the most appropriate investigation to confirm the diagnosis?

Correct answer E: Ultrasound of the upper abdomen

Reasoning:

The clinical information describes the typical presentation of biliary atresia, which is the commonest cause of neonatal cholestatic jaundice. An early diagnosis is critical in preventing future chronic liver disease.

In this context, the most appropriate investigation to confirm the diagnosis is an Ultrasound scan of the upper abdomen.

This imaging modality can help visualize the liver and biliary tree, allowing the assessment of any structural abnormalities, such as biliary atresia or other causes of obstructive jaundice

Answer A is not the best answer

While intrauterine infections (e.g. CMV) can cause neonatal hepatitis and hepatosplenomegaly, ruling out biliary atresia takes priority and is the most likely cause of this scenario.

Answer B is not the best answer

Measuring the fractionated (conjugated vs. unconjugated) bilirubin level is the critical initial investigation to establish whether the jaundice is obstructive but does not confirm the underlying diagnosis.

Answer C is not the best answer

Glucose-6-phosphate dehydrogenase deficiency causes a haemolytic anaemia leading to unconjugated hyperbilirubinemia, which presents with normal-coloured stools and no hepatomegaly.

Answer D is not the best answer

Eosin-5-maleimide (EMA) binding test is used to diagnose hereditary spherocytosis, which causes a hemolytic (unconjugated) jaundice rather than obstructive jaundice.

Further Reading

Jaundice in newborn babies under 28 days NICE Clinical guideline CG98

Published: 19 May 2010, Last updated: 31st October 2023

The Science of Paediatrics MRCPC Mastercourse: Chapter 21

Nelson's Textbook of Pediatrics, Edition 22: Chapter 137

Question 2 – Best answer and reasoning

Stem question:

Which is the principal mechanism by which vitamin D maintains calcium homeostasis?

Correct answer A: Increases calcium absorption from the gut

Reasoning:

Vitamin D binds to an intracellular receptor and regulates gene transcription. It stimulates active calcium absorption in the small intestine (duodenum and jejunum) by upregulating the expression of epithelial calcium channels (TRPV6) and calcium-binding proteins.

Answer B is not the best answer:

Vitamin D does aid phosphate absorption but the effect is less significant because most dietary phosphate absorption is vitamin D independent.

Answer C is not the best answer:

Vitamin D promotes bone resorption via osteoclast regulation but this effect on calcium is less important than absorption from the gut.

Answer D is not the best answer:

Vitamin D reduces renal loss of calcium (and phosphate) but this effect is less important than absorption from the gut.

Answer E is not the best answer:

Vitamin D actively suppresses parathyroid hormone (PTH) release. PTH release is also inhibited by the vitamin D induced rise in serum calcium.

Further Reading: Nelson Textbook of Pediatrics, 22nd Edition, Chapter 69

Question 3 – Best answer and reasoning

Stem question:

What is the most likely pathophysiological mechanism to account for these findings?

Correct answer A: Dysfunction of the levator palpebrae superioris

Reasoning

An isolated ptosis is most commonly due to dysfunction of the levator palpebrae superioris.

Histologically, the levator muscles of patients with congenital ptosis are dystrophic. The levator muscle and aponeurosis tissues appear to be infiltrated or replaced by fat and fibrous tissue.

In severe cases, little or no striated muscle can be identified at the time of surgery.

This suggests that congenital ptosis is secondary to local developmental defects in muscle structure. Because it is a local muscular defect, pupil responses and extraocular eye movements remain entirely normal.

Answer B is not the best answer:

The zygomatic branch of the Facial nerve innervates upper and lower eyelids (orbicularis oculi muscle), and is thus responsible for eye closure. An injury to this nerve causes an inability to close your eyelids rather than a ptosis.

Answer C is not the best answer:

Transplacental acetylcholine receptor antibodies cause transient neonatal myasthenia gravis which is likely to cause bilateral ptosis alongside generalised hypotonia and muscular weakness.

Answer D is not the best answer:

Disruption of the sympathetic nerve supply to the eye (Horner's syndrome) is caused by disruption of the sympathetic nerve supply to the eye and would also result in miosis (constricted pupil).

Answer E is not the best answer: Congenital third nerve palsy causes marked ptosis and restriction of eye movements.

The eye is typically turned down and outwards.

Further Reading:

Nelson Textbook of Pediatrics Ed 22: Chapters 662-664

The Science of Paediatrics MRCPC Mastercourse 2017 Chapter **30**

Question 4 – Best answer and reasoning

Stem question:

Which is the most likely causative organism?

Correct answer B: Escherichia coli

Reasoning:

Escherichia coli is one of the leading causes of bacterial meningitis and sepsis in neonates (alongside Group B Streptococcus). It is the most likely causative organism in this scenario given the clinical presentation and the presence of Gram-negative bacilli identified in the CSF Gram stain results.

Answer A is not the best answer:

Pseudomonas aeruginosa is a gram-negative bacillus but a rare cause of neonatal meningitis and usually only seen in immunocompromised patients.

Answer C is not the best answer:

Haemophilus influenza is a gram-negative coccobacillus. It is more commonly associated with meningitis in older infants and unvaccinated children rather than neonates.

Answer D is not the best answer: Listeria monocytogenes is an important cause of neonatal meningitis and is a gram-positive bacillus.

Answer E is not the best answer: Neisseria meningitidis is a gram-negative diplococcus. It is more commonly associated with meningitis in older infants and unvaccinated children rather than neonates.

Further Reading: The Science of Paediatrics MRCPCH Mastercourse: pg 279
Nelson Textbook of Pediatrics, Edition 22, Chapter 148

Question 5 – Best answer and reasoning

Stem question:

Which of the following parameters will have the greatest influence on oxygenation?

Correct answer C: Mean airway pressure

Reasoning

Oxygenation is determined by mean airway pressure (MAP) and fraction of inspired oxygen. MAP represents the average pressure applied to the lungs during the entire respiratory cycle. By maintaining a steady average pressure, it keeps alveoli open (recruitment), increases the surface area for gas exchange, and directly improves oxygenation.

Answer A is not the best answer: Prolonging inspiratory: expiratory ratio will help oxygenation, but only indirectly by its effects on MAP.

Answer B is not the best answer: Alveolar minute ventilation aids carbon dioxide clearance but will not directly affect oxygenation.

Answer D is not the best answer:

Inspiratory time constant is a function of the mechanics of the respiratory system, and is not changed by ventilatory adjustments, except possibly by alveolar recruitment.

Answer E is not the best answer:

Tidal volume controls alveolar ventilation and primary clearance of carbon dioxide not oxygenation.

Further Reading:

The Science of Paediatrics, MRCPCH Mastercourse, Chapter 6.
Nelson Textbook of Pediatrics 22nd edition Chapter 126

Question 6 – Best answer and reasoning

Stem question:

Which of the following statements regarding a randomised clinical trial (RCT) is the most accurate?

Correct answer E: The power of the study is the likelihood of rejecting the null hypothesis given the alternative hypothesis.

Reasoning: Statistical power is defined as the probability of correctly rejecting a false null hypothesis. In other words, it is the likelihood that the study will detect a true treatment effect if one actually exists.

Answer A is not the best answer:

A double-blind study will not eliminate the placebo effect, it will just ensure that the placebo effect is applied equally across both groups.

Answer B is not the best answer:

Increasing the sample size will increase the power of a study (reduce random errors) but does not affect bias that may have been introduced for example by poor study design.

Answer C is not the best answer:

Subjects should not be excluded from the final analysis even if they do not complete the study. Intention to treat is the gold standard i.e. all participants who were randomised must be included in the final analysis, regardless of whether they completed the study.

Answer D is not the best answer:

The number of participants should be guided by a sample size calculation. Enrolling more participants than necessary to prove an effect is considered unethical because it may unnecessarily expose more patients to potential experimental risks.

Further Reading:

The Science of Paediatrics MRCPC Mastercourse, Chapter 37 and 38

Question 7 – Best answer and reasoning

Stem question:

What is the mode of action of atenolol as an anti-hypertensive agent?

Correct answer B: Beta-adrenoreceptor antagonist

Reasoning

Atenolol is a pure selective Beta-adrenoreceptor blocker. It lowers blood pressure by binding to and blocking Beta 1 receptors located primarily on cardiac smooth muscle and kidneys. This reduces heart rate, myocardial contractility, and the release of renin from the kidneys.

Answer A is not the best answer:

Angiotensin converting enzyme (ACE) inhibitor Angiotensin converting enzyme (ACE) inhibitors act on the Renin-Angiotensin system to reduce blood pressure e.g. Captopril.

Answer C is not the best answer:

Calcium channel blockers differ in their predilection for the various possible sites of action and

therefore their therapeutic effects are disparate e.g. Verapamil, Nifedipine.

Answer D is not the best answer: Alpha and beta-

adrenoreceptor antagonist Alpha and beta-

adrenoreceptor antagonist- Alpha and beta dual receptor blockers are a subclass of beta blockers

which are commonly used to treat high blood pressure (BP). Drugs in this class include carvedilol and labetalol.

Answer E is not the best answer:

Alpha adrenoreceptor antagonist- (Alpha blockers) relax certain muscles and help small blood

vessels remain open. They work by keeping the hormone norepinephrine (noradrenaline) from

tightening the muscles in the walls of smaller arteries and veins, which causes the vessels to remain

open and relaxed. This improves blood flow and lowers blood pressure. E.g. Doxazosin and Prazosin.

Further Reading:

Practical Paediatric Prescribing 2021

The Science of Paediatrics MRCPC Mastercourse, Chapter 19

Question 8 – Best answer and reasoning

Stem question:

What is the most likely diagnosis?

Correct answer D: Cow's milk-sensitive enteropathy

Reasoning:

Although duodenal histologic changes in patients with cow's milk protein allergy vary and in individuals with a T-cell mediated hypersensitivity reaction, the duodenal biopsies show changes similar to those seen in coeliac disease (low- grade villous blunting and intraepithelial lymphocytosis). The presence of increased numbers of eosinophils means that this is the best answer alongside changes being patchy rather than diffuse. The histology should still be interpreted in the context of medical history and oral challenges.

Answer A is not the best answer: Abetalipoproteinaemia is not the best answer as there is no triglyceride accumulation in villous enterocytes.

Answer B is not the best answer:

Duodenal involvement in Crohn's is uncommon. Findings on colonoscopy can include patchy, nonspecific inflammatory changes, aphthous ulcers, linear ulcers, nodularity, and strictures. Findings on biopsy may be only nonspecific chronic inflammatory changes. Noncaseating granulomas, similar to those of sarcoidosis, are the most characteristic histologic findings

Answer C is not the best

Although the histologic appearances of coeliac disease and cow's milk protein allergy can be similar the presence of increased numbers of eosinophils and the fact the enteropathy is patchy rather than diffuse suggests cows milk protein enteropathy rather than coeliac disease.

Answer E is not the best answer:

Giardiasis may cause villous blunting, but the pathognomonic feature is the presence of trophozoites attached to the brush border, often accompanied by normal or minimally disrupted architecture.

Further Reading:

BMJ Gut , March 2026 Patchy enteropathy in childhood: P D Manuel et al
The Science of Paediatrics, MRCPCH Mastercourse, Chapter 14

Question 9 – Best answer and reasoning

Stem question:

What is the most likely pathogen to be isolated from her blood cultures?

Correct answer E: Staphylococcus aureus

Reasoning:

The stem describes a case of osteomyelitis. The most common causative organism in children is Staphylococcus aureus. Whilst the other pathogens can also cause osteomyelitis, there is nothing further in the history/stem to suggest that this case will be atypical and make a less common pathogen more likely. In a three-year old the commonest organisms are Staphylococcus aureus, Haemophilus influenzae, Group A Streptococcus, Streptococcus pneumoniae. MRSA is an increasing problem.

Answer A is not the best answer:

The introduction of vaccination against Haemophilus influenzae type b (Hib) has virtually eliminated osteomyelitis caused by this organism in countries where the vaccination is available.

Answer B is not the best answer:

Whilst Mycobacterium tuberculosis can cause osteomyelitis, there is nothing further in the history to suggest that this is likely here.

Answer C is not the best answer:

Group B streptococcus is seen predominately in neonatal osteomyelitis; not at 3 years of age.

Answer D is not the best answer:

Salmonella osteomyelitis in children is an uncommon condition, typically associated with hemoglobinopathies or other underlying disorders. Only a few cases have been reported in children without predisposing factors.

Further Reading: Nelson Textbook of Pediatrics, Ed 22, Chapter 725

Question 10 – Best answer and reasoning

Stem question:

What is the most likely underlying pathophysiology of this condition?

Correct answer B: Autoimmune pancreatic beta cell destruction

Reasoning:

The clinical scenario is a child presenting in diabetic ketoacidosis (DKA). DKA is predominately secondary to Type 1 Diabetes Mellitus caused by autoimmune beta cell destruction leading to complete insulin deficiency.

Answer A is not the best answer:

Genetic defect in pancreatic beta cell function is seen in MODY (Maturity onset diabetes of the young). While MODY occurs in young people, it typically presents with mild, non-ketotic hyperglycemia rather than presenting acutely with severe DKA. There is often a strong autosomal dominant family history of diabetes.

Answer C is not the best answer:

Obesity-induced insulin resistance describes Type 2 Diabetes mellitus. This patient's weight (38 kg at 12 years old) does not indicate obesity, and DKA is far less common as an initial presentation in Type 2 diabetes.

Answer D is not the best answer:

Glucocorticoids can be associated with diabetes in patients with impaired insulin secretion eg Cystic fibrosis. This patient, however, is only on inhaled beclomethasone with no other co morbidities. Regular use of standard inhaled corticosteroids does not cause absolute insulin deficiency or acute DKA.

Answer E is not the best answer:

Genetic mutation in HNF (hepatocyte nuclear factor) gene is a type of MODY (see Answer A).

Further Reading:

Nelson Textbook of Paediatrics, Ed 22, Chapter 629

Question 11 – Best answer and reasoning

Stem question:

What is the most likely anatomical site of the lesion?

Correct answer C: Pituitary fossa

Reasoning:

Tumours arising in the pituitary gland or the suprasellar region can extend to compress the optic chiasm leading to bitemporal hemianopia and problems with visual acuity.

Answer A is not the best answer:

Children with brain stem tumours characteristically present with motor weakness, cranial nerve dysfunction, cerebellar dysfunction and/or raised ICP.

Answer B is not the best answer:

Children with cerebellopontine angle tumours characteristically present with hearing loss, problems with speech, ataxia, nystagmus and involvement of local cranial nerve nuclei.

Answer D is not the best answer:

Children with posterior fossa tumours characteristically present with symptoms and signs of raised ICP and cerebellar dysfunction.

Answer E is not the best answer:

Children with temporal lobe tumours characteristically present with seizures, usually focal.

Further Reading:

Nelson Textbook of Pediatrics 22nd ed: Chapter 546

The Science of Paediatrics MRCPC Mastercourse: pg 501-503

Question 12 – Best answer and reasoning

Stem question:

What is the best interpretation of the findings?

Correct answer D: Restrictive lung disease

Reasoning:

The results of this spirometry show a reduction in both FEV1 and FVC and the FEV1/FVC ratio is not reduced. An increase of 8 to 12% following bronchodilatation would be expected in asthma. In restrictive lung disease there is a reduction in both FEV1 and FVC, FEV1/FVC ratio is not reduced and there is no response to bronchodilator, so this is a restrictive pattern, in keeping with the child's presentation of muscle weakness.

Answer A is not the best answer:

Bronchial hyperreactivity should be apparent following bronchodilator. There was no significant change post bronchodilator in peak flow measurement.

Answer B is not the best answer:

Hyperinflation may be due to air trapping, secondary to obstructive airway disease such as COPD, asthma, Cystic fibrosis. Hyperinflation is not easily diagnosed using spirometry but rather whole body plethysmography.

Answer C is not the best answer:

Impairment of gas exchange is not diagnosed using spirometry.

Answer E is the best answer:

Ventilation perfusion mismatch Impairment of ventilation perfusion mismatch is not diagnosed using spirometry

Further Reading:

The Science of Paediatrics MRCPC Mastercourse: p325-326,
Nelson textbook of Pediatrics Ed 22: Pulmonary Function testing, Chapter 421

13 – Best answer and reasoning

Stem question:

What is the most likely diagnosis?

Correct answer E: Von Willebrand disease

Reasoning:

Low factor VIII and an abnormal APPT (demonstrating an abnormality in the intrinsic coagulation pathway) are consistent with Von Willebrand disease. A deficiency in von Willebrand factor leads to reduced plasma Factor VIII levels, which consequently prolongs the APTT. VWD is the commonest inherited coagulation disorder affecting up to 1% of the population.

Answer A is not the best answer:

Glanzmann thrombasthenia is an autosomal recessive disorder due to deficiency of the platelet glycoprotein GPIIb leading to failure of platelet aggregation. This results in a severe bleeding tendency despite a normal platelet count. Other clotting factors would be normal.

Answer B is not the best answer:

Factor IX is not low as in Haemophilia B.

Answer C is not the best answer:

Protein C deficiency is a congenital or acquired condition that leads to increased risk for thrombosis.

Answer D is not the best answer:

Normal Prothrombin time excludes extrinsic pathway defects (like Vitamin K deficiency)

Further Reading:

The Science of Paediatrics MRCPC Mastercourse: p 455-458

Nelson Textbook of Pediatrics ed 22: Chapter 526

Question 14 – Best answer and reasoning

Stem Question:

In the sequence of events after birth, which of the following processes has the greatest impact on closure of the ductus arteriosus?

Correct Answer C: Reduction in prostaglandin E2

Reasoning:

During fetal life the ductus arteriosus remains open due to low oxygen tension and due to high levels of Prostaglandin E2 (PGE2), which is a potent vasodilator on the smooth muscle of the ductal wall. PGE2 levels fall sharply after birth due to separation of the placenta and increased clearance via neonatal lungs. The ductal smooth muscle constricts as a result of these falling levels (and increased oxygen levels).

Answer A is not the best answer:

The increase in left atrial pressure with decrease in right atrial pressure occurs as a result of the decrease in pulmonary vascular resistance but this leads to closure of the foramen ovale rather than the ductus arteriosus.

Answer B is not the best answer:

The reduction in blood flow through the ductus venosus occurs following the clamping of the umbilical cord, but it has no direct mechanical or chemical impact on the musculature of the ductus arteriosus.

Answer D is not the best answer:

Decrease in pulmonary vascular resistance leads higher left atrial to right atrial pressure gradient leading to closure of the foramen ovale rather than the ductus arteriosus.

Answer E is not the best answer:

While separation from the umbilical-placental circulation triggers the systemic drop in prostaglandins, it is the specific loss of prostaglandin E2 itself (the chemical mediator) that has the direct, greatest impact on the smooth muscle spasm required to close the shunt.

Further Reading:

Nelson Textbook of Paediatrics, Ed 22; Chapter 124

The Science of Paediatrics MRCPC Mastercourse p176

Question 15 – Best answer and reasoning

Stem question:

Which toxin is most frequently involved in the pathogenesis of bullous impetigo caused by staphylococcus?

Correct answer D: Exfoliative toxin A

Reasoning:

Bullous impetigo is due to strains of *Staphylococcus aureus* that produce exfoliative toxin A.

This causes loss of cell adhesion in the superficial epidermis by targeting the protein desmoglein 1.

Answer A is not the best answer:

Bullous impetigo and staphylococcal scalded skin syndrome (SSSS) are both caused by exfoliative toxin released by *Staphylococcus aureus*. Of the two main serotypes, A and B, toxin B circulates through the blood stream causing blistering distant from the infection and thus SSS.

Answer B is not the best answer:

Panton-

Valentine leukocidin is a cytotoxic protein produced by some strains of *Staphylococcus aureus* that increases the virulence. It causes neutrophil destruction and tissue necrosis leading to conditions such as cutaneous abscesses.

Answer C is not the best answer:

Toxic Shock Syndrome Toxin 1 (TSST-

1) is a superantigen that induces production of interleukin-

1 and tumour necrosis factor causing fever, multisystem failure and shock. It is linked to tampon use.

Answer E is not the best answer:

Staphylococcus aureus produces several enterotoxins ingestion of which cause vomiting and diarrhoea with a short incubation time. The commonest is Type A, a frequent cause of food poisoning.

Further Reading:

Nelson Textbook of Pediatrics 21st edition Chapter 685 (Page 3549)

Clinical Cases for MRCPCH Theory and Science 2013 p37-40

Question 16 – Best answer and reasoning

Stem question:

What is the most likely cause of the laboratory abnormalities in this patient?

Correct answer A: Disseminated intravascular coagulation (DIC)

Reasoning:

The prolonged APTT and severely reduced fibrinogen reflect consumption of coagulation factors and fibrinogen, typical of DIC. In addition, the low platelets and reduced haemoglobin suggest a microangiopathic process. DIC occurs in sepsis which would be in keeping with the described clinical picture.

Answer B is not the best answer:

Haemolytic uraemic syndrome (HUS) will not cause coagulopathy, and the renal function would be severely deranged.

Answer C is not the best answer: Haemophilia will cause only prolonged APTT and will not cause a consumption coagulopathy or thrombocytopenia.

Answer D is not the best answer:

Immune thrombocytopenic purpura ITP will not cause coagulopathy or renal impairment.

Answer E is not the best answer: Salicylate ingestion does not cause acute consumption of fibrinogen and prolonged APTT.

Further Reading:

The Science of Paediatrics MRCPCH Mastercourse: pg 457

Nelson Textbook of Pediatrics, Ed 22; Chapter 532

Question 17 – Best answer and reasoning

Stem question:

Which study design is most appropriate?

Correct answer D: Crossover trial

Reasoning:

Cross over trial is the best answer A cross over study is appropriate in view of the absence of blinding and small numbers. In such a study, individuals provide both control and experimental arm by sequentially receiving multiple/all interventions at different time-points, interspersed with periods for intervention wash-out.

Answer A is not the best answer:

In a case-controlled study, patients with a certain outcome or disease and an appropriate group of controls without the outcome or disease are selected and then information is obtained on whether the subjects have been exposed to the factor under investigation.

Answer B is not the best answer:

A case series is a nonanalytic or descriptive study, which measures the frequency of several factors, and hence the size of a problem.

Answer C is not the best answer:

In a cohort study, data are obtained from groups who have been exposed, or not exposed, to the new technology or factor. No allocation of exposure is made by the researcher.

Answer E is not the best answer:

Blinding is not possible, and numbers are small. The gold standard approach for assessing the benefit of a new investigation or treatment approach is a randomized controlled trial (RCT). In this setting, each patient is randomly allocated to receive either standard therapy/placebo or the new treatment.

This removes the potential for biasing the outcome by allocating one particular group of patients, e.g. a more severe pattern of disease, to one arm or other.

Further Reading:

The Science of Paediatrics MRCPC Mastercourse: Chapter 37

Question 18 – Best answer and reasoning

Stem question:

Which of the following is a recognised pathophysiological effect of histamine?

Correct Answer A: Stimulates airway mucus production.

Reasoning

Histamine is released following immunoglobulin E (IgE) mediated mast cell and basophil degranulation. It acts via H1 receptors to stimulate mucus secretion in the airway. It also causes bronchoconstriction and increases vascular permeability.

Answer B is not the best answer:

Histamine causes the contraction of bronchial and gastrointestinal smooth muscle, though it relaxes vascular smooth muscle (causing vasodilation).

Answer C is not the best answer:

IgE is produced by plasma cells (B lymphocytes) in response to allergens, a process driven by cytokines like Interleukin-4 and Interleukin-13, not histamine.

Answer D is not the best answer:

While histamine can modulate immune responses, its primary immediate hypersensitivity effects are mediated directly through pre-formed mediator release rather than enhancing interleukin synthesis as the main acute mechanism.

Answer E is not the best answer:

Histamine stimulates (rather than reduces) gastric acid secretion by binding to H2 receptors on the parietal cells of the stomach.

Further Reading:

Nelson Textbook of Pediatrics 22nd ed: Chapter 104

The Science of Paediatrics MRCPC Mastercourse, Chapter 16

Question 19 – Best answer and reasoning

Stem question:

Given the clinical presentation and lab results, which of the following is the most likely abnormal investigation?

Correct answer D: Urine methylmalonic acid

Reasoning

The blood gas demonstrates a severe metabolic acidosis and a mild hyperammonaemia in keeping with an organic aciduria, such as methylmalonic aciduria. The fact that parents are consanguineous increases the chances of an autosomal recessive condition.

Answer A is not the best answer:

At 7 days of age, it is very unlikely that accumulation of plasma phenylalanine will have any overt clinical manifestations. However, detection by newborn screening is essential to prevent future consequences of the metabolic derangement.

Answer B is not the best answer:

A child with Galactosemia will have reduced blood cell galactose-1-phosphate uridyl transferase activity.

Within a few days after birth, they present with persistent jaundice, vomiting, diarrhoea and failure to thrive, typically without a profound metabolic acidosis.

There may be a history of *E. coli* septicaemia and neonatal death of siblings.

Answer C is not the best answer:

Defects in acylcarnitine present with attacks of fasting induced hypoglycaemia, hyperammonaemia and cardiorespiratory collapse.

Answer E is not the best answer:

Serum lactate It is likely to be raised in mitochondrial disorders, disorders of gluconeogenesis and glycogen storage disorders but will not be diagnostic for any of them.

Further Reading: Nelson Textbook of Pediatrics 22nd ed: Chapter 104
The Science of Paediatrics MRCPC Mastercourse: Chapter 29

Question 20 – Best answer and reasoning

Stem question:

Which of the following conditions would be the most likely cause of this child's clinical presentation?

Correct answer D: Galactosaemia

Reasoning:

Galactosaemia symptoms typically manifest within the first few days of life after the infant ingests lactose which is broken down into glucose and galactose. Deficiency of the galactose-1-phosphate uridylyl transferase (GALT) enzyme leads to a toxic accumulation of galactose-1-phosphate. This causes acute liver dysfunction, vomiting, poor feeding, and increased intracranial pressure. Infants with classic galactosaemia have a profoundly increased risk of life-threatening *Escherichia coli* (*E. coli*) septicaemia, which explains the previous sibling's death at 2 weeks of age.

Answer A is not the best answer

Gestational alloimmune liver disease presents with acute liver failure at or shortly after birth and intrauterine growth restriction. There is no association with raised intracranial pressure or tendency to *E. coli* septicaemia.

Answer B is not the best answer

Symptoms of Fructosaemia only occur after introduction of sucrose or fructose (e.g. fruit juice) in the diet.

Answer C is not the best answer

Biliary Atresia usually presents later with prolonged jaundice and there is no predisposition to *E. coli* septicaemia

Answer E is not the best answer

Congenital cytomegalovirus infection presents at birth with intrauterine growth restriction, microcephaly, prolonged jaundice with hepatosplenomegaly and petechiae/purpura.

Further reading:

The Science of Pediatrics, MRCPCH Mastercourse: Chapter 21
Nelson's Textbook of Paediatrics 22nd ed: Chapter 107 and 137