

The following are a selection of Clinical Biochemistry Part 1 exam questions retired from the College question bank in December 2020:

Question 1

Serum from a man who has dermatomyositis had LDH isoenzymes analysed using thin-layer agarose gel electrophoresis.

Which isoenzyme would be expected to have the highest peak?

- A. LD1
- B. LD2
- C. LD3
- D. LD4
- E. LD5

Question 2

A 62-year-old man presented to his GP with a painful, red big toe.

Blood testing showed:

Serum urate 0.74 mmol/L (0.23-0.46)

What would be the most useful additional biochemistry test to perform in this situation?

- A. Cholesterol
- B. Creatinine
- C. Glucose
- D. Lactate
- E. LDH

Question 3

A 30-year-old, obese male was found elevated ALT and ALP. Fatty liver was noted on ultrasound. After weight loss, ALT decreased but the elevated ALP persisted. Isoenzyme analysis revealed the presence of intestinal ALP.

Which ABO blood types is this patient most likely to have?

- A. A or B
- B. A or AB
- C. A or O
- D. B or AB
- E. B or O

Question 4

A junior doctor telephones for advice. He needs to take various samples from his patient and wants to know the most appropriate order in which to draw the samples.

The following tests have been requested

Full blood count, renal profile, plasma glucose, clotting studies, blood cultures.

Which sample should he take third?

- A. Blood cultures
- B. Clotting studies
- C. Full blood count
- D. Plasma glucose
- E. Renal profile

Question 5

A 42-year-old man was seen in the oncology clinic with a diagnosis of metastatic pancreatic cancer. His serum Ca19-9 concentration was not elevated.

This finding might be explained by variation in which blood type antigen?

- A. ABO
- B. Duffy
- C. Kell
- D. Lewis
- E. Rhesus

Question 6

A young boy was suspected of having mucopolysaccharidoses. A urine sample was sent for analysis and found to contain keratan sulphate.

Which type of mucopolysaccharidosis is he most likely to have?

- A. Type I
- B. Type II
- C. Type III
- D. Type IV
- E. Type V

Question 7

A pharmacist contacted you to discuss how laboratory reporting of antiepilepsy drugs might be improved

Requests for which anti-epilepsy drug are most likely benefit from reflex addition of albumin to allow correction for the effects of hypoalbuminaemia?

- A. Carbamazepine
- B. Levetiracetam
- C. Phenytoin
- D. Topiramate
- E. Valproate

Question 8

A baby had a positive result on the newborn screening for cystic fibrosis. After assessment by a paediatrician, a sweat test was arranged.

The sweat test was performed when the baby is three weeks old.

Following iontophoresis, the sweat was collected over 30 minutes using the Macroduct system.

Results: Sweat volume 14 mg Sweat chloride 37 mmol/L

What interpretative comment should be added to these results?

- A. Result not consistent with a diagnosis of cystic fibrosis
- B. Result consistent with a diagnosis of cystic fibrosis
- C. Result suggestive but not diagnostic of cystic fibrosis
- D. Insufficient sweat volume collected. Repeat sweat test is required.
- E. Intermediate sweat chloride result. Repeat sweat test is required.

Question 9

A 76-year-old man had urinalysis performed as part of a research trial – it was positive for ketones. Blood ketones are within the reference range.

He is prescribed amlodipine for hypertension, atorvastatin for dyslipidaemia, etodolac for arthritis, levodopa for Parkinson's and lamotrigine and brivaracetam for epilepsy, Which of his medications is most likely to be responsible for this finding?

- A. Amlodipine
- B. Brivaracetam
- C. Etodolac
- D. Lamotrigine
- E. Levodopa

Question 10

PCSK9 (protease convertase subtilisin kexin type 9) is a protein involved in cholesterol metabolism. PCSK9 inhibitors are used in the treatment of hypercholesterolaemia.

What is the mechanism of action of these drugs?

- A. Inhibition of absorption of cholesterol from the gut
- B. Inhibition of conversion of HMG CoA to mevalonate
- C. Modulation of degradation of the LDL receptor in the liver
- D. Reduction of hepatic VLDL synthesis
- E. Up regulation of lipoprotein lipase in the liver

Question 11

A child with severe developmental delay and dysmorphic features is being investigated. Very long chain fatty acids have been requested

What type of disorder does this test investigate?

- A. Lysosomal
- B. Mitochondrial
- C. Nuclear
- D. Peroxisomal
- E. Ribosomal

Question 12

In the UK, it is recommended that men and women who drink regularly should consume no more than 14 units a week

In the UK unit system, how much pure alcohol is there in 1 unit?

- A. 8mL (12g)
- B. 10mL (8g)
- C. 12 mL (15g)
- D. 15mL (20g)
- E. 20mL (10g)

Question 13

A 57-year-old woman gave a history of recent weight loss with weakness and limb pain, which had worsened progressively over the past two years.

Results showed:

Calcium 2.25 mmol/L

Phosphate 0.48 mmol/L

ALP 379 U/L

A diagnosis of tumour induced osteomalacia was suspected

Which laboratory test is most likely to confirm the diagnosis?

- A. ALP isoenzymes
- B. 1,25-dihydroxyvitamin D
- C. FGF-23
- D. PTH-RP
- E. TMP/GFR

Question 14

A 57-year-old woman presented with hypertension, abdominal obesity and striae

Her prescribed medications were carbamazepine, furosemide, omeprazole, rosuvastatin and thyroxine.

A serum cortisol concentration measured at 9am, after taking dexamethasone at midnight, was 62 nmol/L. Cushing syndrome was excluded after further investigation.

Which of her medications would be most likely to underlie the false positive dexamethasone test result?

- A. Carbamazepine
- B. Furosemide
- C. Omeprazole
- D. Rosuvastatin
- E. Thyroxine

Question 15

A 2-month-old baby was found to have ambiguous genitalia

Which analyte is likely to be most useful in assessing for the presence of testicular tissue?

- A. AMH
- B. FSH
- C. hCG
- D. Inhibin B
- E. Testosterone

Question 16

A 28-year-old man presented to the gastroenterology clinic with a 3 month history of weight loss and mild iron deficiency anaemia. On colonoscopy he was found to have melanosis coli. Investigations showed

Serum Sodium	144 mmol/L	Urine
	Potassium	2.6 mmol/L
	Creatinine	96 µmol/L
	Ferritin	9 µg/L
Urine	Potassium	12 mmol/L

What is the most likely explanation for the hypokalaemia?

- A. Conn Syndrome
- B. Gitelman Syndrome
- C. Laxative abuse
- D. Renal tubular acidosis
- E. Villous adenoma

Question 17

A 34-year-old woman with a diagnosis of irritable bowel syndrome had on-going diarrhoea. Bile acid malabsorption was being considered as a cause.

Which blood test would you suggest to further evaluate?

- A. Chenodeoxycholic acid
- B. Delta bilirubin fraction
- C. Fibroblast growth factor 9
- D. 7 α -hydroxy-4-cholesten-3-one
- E. 27-hydroxycholesterol

Question 18

A 3-year-old boy was investigated for short stature.

Results showed:

Sodium	137 mmol/L
Potassium	4.5 mmol/L
Creatinine	28 µmol/L (23-37)
Adjusted calcium	2.25 mmol/L
Albumin	38 g/L
ALP	19 U/L (60-425)
Phosphate	1.0 mmol/L
TSH	2.7 mU/L
25-OH vitamin D	35 nmol/L
Zinc	3.2 µmol/L (6-25)

What is the most likely cause of the low ALP?

- A. EDTA contamination
- B. Hypophosphatasia
- C. Magnesium deficiency
- D. Vitamin D deficiency
- E. Zinc deficiency

Question 19

A 28-year-old woman with no previous history of thyroid disease is seen in the endocrine clinic. Her thyroid function tests are shown:

TSH <0.1 mU/L

FT4 46.3 pmol/L

FT3 21.6 pmol/L

Thyroglobulin <0.1 ng/mL (3.3-77)

Thyroid peroxidase antibodies: weak positive

What is the most likely diagnosis?

- A. Graves' disease
- B. Hashimoto's thyroiditis
- C. Multinodular goitre
- D. Papillary thyroid cancer
- E. Thyrotoxicosis factitia

Question 20

D-lactic acidosis may occur in some patients and is difficult to diagnosis in the laboratory

What type of surgery predisposes patients to this complication?

- A. Anterior resection of the rectum and colon
- B. Laparoscopic gastric banding
- C. Percutaneous endoscopic gastrostomy
- D. Removal of small bowel with preservation of colon
- E. Ureterosigmoidostomy

Question 21

When the kidneys fail, urea accumulates

Which breakdown product of urea can carbamylate proteins?

- A. Carbonate
- B. Cyanate
- C. Hyaluronate
- D. Nitrate
- E. Uronate

Question 22

A 41-year-old woman had a routine health check.

Results on a fasting serum sample showed

Cholesterol 8.1 mmol/L

LDL cholesterol 2.2 mmol/L

HDL cholesterol 5.4 mmol/L

Triglycerides 1.2 mmol/L

What is the most likely diagnosis?

- A. Cholesterol ester transfer protein deficiency
- B. Familial dysbetalipoproteinaemia
- C. Familial hypercholesterolaemia
- D. Lecithin-cholesterol acyltransferase deficiency
- E. Tangier disease

Question 23

Serum concentrations of cancer antigens may be increased in malignancy. Which cancer antigen is particularly used as a marker in breast cancer?

- A. CA 15-3
- B. CA 19-5
- C. CA 19-9
- D. CA 50
- E. CA 72-4

Question 24

The free androgen index (FAI) has been suggested to be useful in the assessment of polycystic ovarian syndrome.

The following results were obtained from a 27 year old woman with amenorrhoea

Testosterone 2.0 nmol/L
SHBG 27 nmol/L
Androstenedione 6.5 nmol/L
Albumin 38 g /L

What is her FAI?

- A. 0.315
- B. 0.77
- C. 7.4%
- D. 14.6%
- E. 31.5

Question 25

A 40-year-old man is referred to the neurology clinic for investigation of burning pain in his fingers. He is known to have renal dysfunction.

Fabry disease is suspected

Which enzyme should be tested to further investigate him?

- A. Arylsulphatase A
- B. α -Galactosidase
- C. Galactosylceramidase
- D. Glucocerebrosidase
- E. Sphingomyelinase

Question 26

A 36-year-old woman complains of a lifelong problem with body odour resembling rotten fish.

Urine testing confirms the presence of trimethylaminuria

If the cause is genetic, which enzyme is likely to be affected?

- A. Dimethylglycine dehydrogenase
- B. Fatty alcohol NAD⁺ oxidoreductase
- C. Flavin-containing mono-oxygenase 3
- D. Methane mono-oxygenase
- E. Succinate semialdehyde dehydrogenase

Question 27

A 56-year-old woman presents feeling “tired all the time”
The results of investigations by her GP show

Haemoglobin	105 g/L
Ferritin	15 µg/L
Iron	25 µmol/L
TIBC	50 µmol/L
Transferrin	2500 mg/L

What is her transferrin saturation?

- A. 1%
- B. 6%
- C. 25%
- D. 30%
- E. 50%

Question 28

Smith-Lemli-Opitz Syndrome is suspected in a child with growth retardation, developmental delay and behavioural problems. Examination reveals microcephaly and dysmorphism. Which metabolite is the diagnostic test for this condition?

- A. Campesterol
- B. Cholestanol
- C. 7-Dehydrocholesterol
- D. Sitosterol
- E. Stigmasterol

Question 29

Serum indices are calculations of absorbance measurements that provide a semiquantitative representation of levels of icterus, haemolysis or lipaemia in patient samples
At what wavelength (nm) is detection of lipaemia best performed?

- A. 300-410
- B. 400-510
- C. 500-610
- D. 600-710
- E. 700-810

Question 30

A 4-year-old child was diagnosed with Tyrosinaemia Type 1 and commenced on treatment with Nitisinone.
Which enzyme does this drug inhibit?

- A. Fumarylacetoacetate
- B. Homogentisic acid oxidase
- C. 4-Hydroxyphenylpyruvate dioxygenase
- D. Maleylacetoacetate isomerase
- E. Tyrosine aminotransferase

Question 31

A 52-year-old man with diabetes mellitus was prescribed a thiazolidinedione to assist in optimising glycaemic control

With which entity does the drug act to bring about this effect?

- A. AMP-activated protein kinase
- B. Dipeptidyl peptidase-4
- C. PPAR γ
- D. Sodium glucose co-transporter 2
- E. Sulphonylurea receptor

Question 32

The Friedewald formula is used to calculate serum LDL-cholesterol concentration. The formula is not valid if the serum fasting triglycerides concentration is raised.

What is the concentration of serum triglycerides above which the formula is considered invalid?

- A. 1.7 mmol/L
- B. 2.0 mmol/L
- C. 4.5 mmol/L
- D. 7.0 mmol/L
- E. 10.5 mmol/L

Question 33

A 25-year-old woman attended the A&E department complaining of a sudden onset severe headache. Twelve hours after the onset, a lumbar puncture was performed and the CSF sent for xanthochromia testing. The scan showed a single peak

At what wavelength would you expect a bilirubin peak to appear?

- A. 403 – 410 nm
- B. 410 – 418 nm
- C. 450 – 460 nm
- D. 510 – 518 nm
- E. 550 – 560 nm

Question 34

In enzyme kinetics, what is the Michaelis-Menton constant?

- A. The enzyme concentration at which half the maximum reaction velocity has been achieved under given conditions
- B. The lowest substrate concentration at which the enzyme reaction can proceed under given conditions
- C. The maximum velocity of the reaction under given conditions
- D. The substrate concentration at which half the maximum reaction velocity has been achieved under given conditions.
- E. The substrate concentration at which the maximum reaction velocity has been achieved under given conditions.

Question 35

The paediatric team enquire whether there might be an analytical reason for a possible anomalous CRP result.

The kit insert for the CRP assay describes a detector measuring light at 840nm placed at 90° to the main light path

What analytical technique is likely to be in use?

- A. Bichromatic spectrophotometry
- B. Derivative spectroscopy
- C. Nephelometry
- D. Turbidimetry
- E. Reflectance spectroscopy

Question 36

Phosphate can be measured by the formation of a colourless complex with ammonium molybdate

At what wavelength (nm) would such a complex generally be measured?

- A. 330-390
- B. 410-470
- C. 480-540
- D. 550-610
- E. 620-680

Question 37

A 27 year old woman with long standing Crohn's disease, who has been receiving home parenteral nutrition for several years, presents with a skin rash. Her clinical team think that its appearance is typical of that of pellagra

Deficiency of which vitamin is most likely to be the cause?

- A. Biotin
- B. Niacin
- C. Pyridoxine
- D. Riboflavin
- E. Thiamine

Question 38

Measurement of the faecal osmotic gap may be helpful in differentiating secretory from osmotic diarrhoea.

The following results were obtained on a faecal sample:

Osmolality 280 mOsm/Kg

Faecal sodium 20 mmol/L

Faecal potassium 65 mmol/L

What is the faecal osmotic gap?

- A. 40 mOsm/kg
- B. 110 mOsm/kg
- C. 130 mOsm/kg
- D. 175 mOsm/kg
- E. 195 mOsm/kg

Question 39

The following are antipsychotics:

Aripiprazole
Chlorpromazine
Flupenthixol
Risperidone
Sulpiride

Which of these drugs is least likely to elevate prolactin?

- A. Aripiprazole
- B. Chlorpromazine
- C. Flupenthixol
- D. Risperidone
- E. Sulpiride

Question 40

Various parameters can be calculated to assess biochemical test performance in different clinical circumstances.

In screening, how is the diagnostic specificity of a test defined?

- A. All affected individuals identified, divided by total of positive results.
- B. All affected individuals identified, divided by total of those with condition.
- C. All correct results, divided by total number of subjects studied.
- D. All healthy individuals identified, divided by total number unaffected.
- E. All healthy individuals identified, divided by total number of normal results

Question 41

A 32-year-old man underwent drug screening as part of a pre-employment medical examination. His urine tested positive for morphine and 6-monocetylmorphine. Which drug is he most likely to have taken?

- A. Buprenorphine
- B. Codeine
- C. Heroin
- D. Methadone
- E. Tramadol

Question 42

Elevated urinary levels of sulphite, thiosulphate and sulphocysteine are associated with a group of inherited metabolic disorder which may present with infantile epilepsy, encephalopathy and progressive psychomotor retardation.

What is the most likely diagnosis in patients with positive urinary sulphocysteine and low plasma or urine uric acid?

- A. Classical Homocystinuria
- B. Cystathionuria
- C. Lesch-Nyhan Disease
- D. Molybdenum co-factor deficiency
- E. Sulphite Oxidase deficiency

Answers

Question 1
E LD5

Question 2
B Creatinine

Question 3
E B or O

Question 4
E Renal profile

Question 5
D Lewis

Question 6
D Type IV
Question 7
C Phenytoin

Question 8
D Insufficient sweat volume collected. Repeat sweat test is required

Question 9
E Levodopa

Question 10
C Modulation of degradation of the LDL receptor in the liver

Question 11
D Peroxisomal

Question 12
B 10mL (8g)

Question 13
E FGF-23

Question 14
A Carbamazepine

Question 15
A AMH

Question 16
C Laxative abuse

Question 17
D 7α -hydroxy-4-cholesten-3-one

Question 18
E Zinc deficiency

Question 19
E Thyrotoxicosis factitia

Question 20
D Removal of small bowel with reservation of the colon

Question 21
B Cyanate

Question 22
A Cholesterol ester transfer protein deficiency

Question 23
A Ca15-3

Question 24
C 7.4%

Question 25
B α -Galactosidase

Question 26
C Flavin-containing mono-oxygenase 3

Question 27
E 50%

Question 28
C 7-dehydrocholesterol

Question 29
D 600-710

Question 30
C 4-Hydroxyphenylpyruvate dioxygenase

Question 31
C PPAR γ

Question 32
C 4.5 mmol/L

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C 450-460 nm

Question 34
D The substrate concentration at which half the maximum reaction velocity has been achieved under given conditions

Question 35
C Nephelometry

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A 330-390

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D All healthy individuals identified, divided by total number unaffected

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D Molybdenum co-factor deficiency