THE UNIVERSITY OF ZAMBIA

SCHOOL OF MEDICINE

DEPARTMENT OF PATHOLOGY & MICROBIOLOGY

SPECIAL SUPPLEMENTARY EXAMINATION FOR ACADEMIC YEAR 2021/2022

GENERAL AND SYSTEMIC PATHOLOGY (MB CHB) PTM 4210

DATE: 13/02/2023

COMPUTER NO:.....

TIME ALLOWED: ***THREE (3) HOURS***

INSTRUCTIONS

1. Answer all questions on this paper.
2. Each question should be answered on a separate answer sheet
3. Clearly indicate your computer number on every answer sheet you submit, failure to do so may lead to loss of marks.

CamScanner

QUESTION 1 (20marks)

Describe the following disorders, giving examples:

a. Autosomal dominant and Recessive
b. Sex linked dominant and Recessive

QUESTION 2 (20marks)

A couple with husband who has blood group AB+ and wife who has blood group 0+ have three children; Alice with A+, John with B+ and Clint with 0+ blood groups.

a. Which of the three children is adopted and explain why? (5marks)
b. What two theories of genetics explain which blood group can be seen in the phenotype? (5marks)
c. In case of an emergency, which parent can donate blood to all the children and why? (5marks)
d. Which member of the family cannot donate blood to anyone in the family and why?(5marks)

QUESTION 3 (20marks)

A 28 year old female patient of kanyama compound presented with fever and decreased urine output. On examination she has offensive vaginal discharge, and exhibits abdominal tenderness. One week earlier she had an abortion attended by non-medical personnel with metallic materials. The doctor found out that her blood pressure was 60/20 mmHg, that she has altered consciousness, & a temperature of 38.90 c.

a. What is the likely diagnosis? (2 marks)
b. What is the pathogenesis of the above diagnosis? (10 marks)
c. What investigations are you going to institute? (4 marks)
d. What are you going to do with the patient? (4 marks)

QUESTION 4 (20marks)

Briefly describe the mechanisms of apoptosis.

QUESTION 5(20marks)

You are a newly qualified medical doctor that receives a 30-year-old woman at UTH casualty with a significantly raised blood pressure (Hypertension). She tells you that she sometimes experiences these sudden onsets of racing heart (palpitations), headache, sweating and tremors (paroxysmal episodes). She also informs you that her local clinic which she attends has been following her up for her hypertension for the past 4 months, hence the referral. You order tests that include an ultrasound and the report informs you that she has a tumor in her left adrenal gland, in the medulla.

a. What is the most likely diagnosis for the tumor? [5 marks]
b. What cells is this tumor composed of? [5 marks]
c. Why does she experience these paroxysmal episodes? [5 marks]
d. List a (one) syndrome this tumor is associated with. [2.5 marks]
e. Apart from a biopsy, which other laboratory test would you order to confirm your diagnosis? [2.5 marks]

QUESTION 6 (20marks)

Answer the following questions

a. Define an acute phase response **(3 marks)**
b. What is the difference between positive acute phase proteins (PAPP) and negative acute phase proteins (NAPP)? **(3 marks)**
c. Give any 2 examples of PAPPs **(2 marks)**
d. Give any 2 examples of NAPPs **(2 marks)**
e. List the clinical uses of plasma albumin measurement **(10 marks)**

QUESTION 7 (20marks)

A one-month old girl was investigated for ambiguous genitalia. The pregnancy and birth had been normal, and the baby had gained weight normally and had shown no signs of illness. Ambiguous genitalia were noted at birth. The clitoris was enlarged and there was partial fusion of the labia. An ultrasound scan showed the presence of a uterus. An XX karyotype was found in peripheral blood.

Biochemical tests on plasma revealed the following (reference ranges in brackets):

- Electrolytes: Na+ : 135mmol/L, K+ : 4.3mmol//L
- Cortisol (11 am): 313nmol/L (9 am : 140-700), (11 pm: 0- 138)

- Testosterone: 6.6nmol/L (<1 prepubertal) (8.4-30 adult male) (<2.7 adult female)
- 17-OH-progesterone: 356nmol/L(<6)
- Androstenedione: 19.4nmol/L (<12)

a. What is the diagnosis? (work it out from the steroid synthetic pathway).
b. Explain the reason for the overproduction of androgens.
c. Would you expect the level of plasma ACTH to be elevated, decreased or normal?
d. Comment on the cortisol level.
e. Is it necessary to treat this condition? Why?
f. Explain the principles of therapy in this disorder.
g. What are the genetics of the condition? What advice would you give the parents regarding future children?

QUESTION 8 (20marks)

A 25 year old woman had been on an oral contraceptive pill for 4 years, then stopped taking the pill. 13 months later her periods had still not returned. A pregnancy test was negative.

The following investigations were performed:

	Patients results	Normal levels
Prolactin	346 ng/Ml	< 29
TSH	1.2	0.35 – 5.5
fT4	17.0	10 – 24
9 a.m. plasma cortisol	560 nmol/L	140 – 700

a. What diagnosis is likely?
b. Which drugs can cause a raised PRL level?
c. What is meant by the term "stalk effect", and how does it arise?

MRI scan of the pituitary showed a small tumour. The PRL level returned to normal and the menstrual cycle was restored following treatment with bromocriptine.

d. What symptom of this condition did this patient NOT have?
e. How can the normal thyroid and cortisol results in this patient be reconciled with the other abnormal findings?

QUESTION 9 (20marks)

A 19-year-old male presents with a two (2) month history of weakness, fatigue, intermittent fever, and epistaxis. Hewas previously well with no history of illness. On examination he is pale, febrile (Temp 38°C), mild hepatosplenomegaly and gum hypertrophy. Full blood count shows a total white cell count (TWCC) = $1.15x10^9/L$ ($4-10x10^9/L$), Haemoglobin(HB) = 4.2g/dL (13-17g/dL) and platelets = $24x10^9/L$ ($150-400x10^9/L$).

1. Give an interpretation of the full blood count results
2. What is the term used to describe the total white cell count in this patient?
3. Give two (2) causes of a low total white cell count.
4. What could the explanation be for the intermittent fever in this patient?
5. What could explain the epistaxis in this patient?

A bone marrow aspiration (BMA) and trephine biopsy are performed. The BMA is markedly hypercellular with predominance (67%) of intermediate to large cells with moderate cytoplasm with granules and Auer rods seen. Nuclei are of variable morphology with fine chromatin and prominent nucleoli. The trephine biopsy shows the same features. These cells express CD34, CD13, CD33, CD117 and are negative for TdT.

6. What could explain the gum hypertrophy in this patient?
7. What is the nature of the cells described in the BMA report?
8. What is the definitive diagnosis in this patient?
9. What further investigations would you request beyond what has already been provided?
10. What are the treatment options for your diagnosis in (8) above?

THE END

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