



THE COLLEGES OF MEDICINE OF SOUTH AFRICA

Incorporated Association not for gain  
Reg No 1955/000003/08

Examination for the Subspecialty Certificate in Endocrinology  
and Metabolism of the College of Paediatricians of South Africa



27 July 2017

Paper 1

(3 hours)

All questions are to be answered. Each question to be answered in a separate book (or books if more than one is required for the one answer)

- 1 Discuss polycystic ovary syndrome (PCOS) under the following headings
- a) Indications for evaluation. (6)
  - b) Basic evaluation. (10)
  - c) Further endocrine evaluation. (5)
  - d) Additional evaluation after the diagnosis of PCOS. (4)
- [25]
- 2 A 14-year-old male who was evaluated for a headache was found to be hypertensive (systolic blood pressure 176mmHg; diastolic 128mmHg). A diagnosis of a pheochromocytoma was entertained because of an abdominal mass reported (CT scan) as a soft tissue tumour with increased vascularity localised in the para-aortic area
- a) Describe the radiological and laboratory investigations necessary to confirm the presence of a pheochromocytoma. Detail the factors that influence the interpretation of the tests. (6)
  - b) Discuss the use, pharmacology and precautions of drugs deployed to manage blood pressure in a patient in whom you suspect to have pheochromocytoma. (6)
  - c) What peri-operative precautions are important? (3)
  - d) What acute complications should be anticipated during surgery and post-operatively? (3)
  - e) Discuss the hereditary syndromes associated with pheochromocytomas. (4)
  - f) Discuss the value of genetic testing in paediatric pheochromocytomas. (3)
- [25]
- 3 A 5-year-old girl presents with depressed level of consciousness. A glucometer reading was 2.1mmol/l. There is a history of increasing tiredness, diarrhoea and weight loss. On examination, she is thin and weight is less than the 3<sup>rd</sup> centile. Her height is appropriate for age and mid-parental height. She is not dehydrated or dysmorphic and has a normal systemic examination including normal female genitalia. Her urine has 3+ ketones
- a) Discuss your differential diagnosis based on the available clinical information. (3)
  - b) What biochemical investigations would you request? (2)
- Blood glucose was 2.2mmol/l and serum cortisol was 32nmol/l
- c) What is the most likely diagnosis? (1)
  - d) Discuss how would you confirm this diagnosis? (4)
  - e) What features help you identify the site of the pathology? (2)
  - f) Discuss investigations you would consider to confirm the underlying pathology. (4)
  - g) How would you treat and monitor therapy in this child. (5)
- 1 year after commencing treatment, she presents in a coma having complained of cough and fever
- h) How would you treat this situation? (2)
  - i) How would you prevent this from occurring again? (2)
- [25]

- 4 You work in an area which does not have a neonatal thyroid screening programme (5)
- a) List the points to be made in a motivation to establish such a programme. (5)
- Having established a thyroid screening programme you receive the following results
- TSH 85.9mIU/L
  - Free T4 13.1pmol/L
- b) What is the most likely diagnosis? (2)
- c) Give a differential diagnosis. (3)
- d) How would you investigate this patient? (3)
- e) List the goals of long term care of this patient. (6)

The patient is followed by his paediatrician. He returns to your clinic at age 4 years for review and you note that his speech is delayed and his mother expresses concern about his abnormal behaviour. Review of his thyroid functions reveals

Age	TSH (0.4 – 5.5 mIU/L)	Free T4 (11 – 22 pmol/L)
3 months	4.8	19.9
6 months	1.6	18.5
18 months	60.6	10.2
2.0 years	53.6	11.8
2.1 years	0.91	30.7
2.6 years	44.2	10.3
3.0 years	26.0	24.9
3.5 years	No Record	
4.0 years	24	30.1

- f) What is your interpretation of these results? (3)
- g) How would you advise the mother and attending paediatrician? (3)
- [25]



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28 July 2017

Paper 2

(3 hours)

*All questions are to be answered. Each question to be answered in a separate book (or books if more than one is required for the one answer)*

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- 1 A mother is concerned about her 1-year-old female child who has bowing of her legs which is getting worse since she started standing. The family history reveals that the mother and her sister have similar problems. The patient is short for age at  $-2.5$  SD and has mild frontal bossing, widened wrists, and moderate bowing with torsion at the tibia bilaterally. Radiology shows rachitic changes at the wrists and knees.
- Laboratory investigations indicates a serum phosphate of  $0.84$ mmol/L (normal reference for age  $1.45 - 1.8$ ), serum calcium of  $2.27$ mmol/L ( $2.19 - 2.64$ ), alkaline phosphatase  $645$  IU/L (normal reference for age  $102-400$ ), normal creatinine, 25-hydroxyvitamin D  $60$ nmol/L ( $50 - 75$ ), 1,25-dihydroxyvitamin D of  $100$ pmol/L (normal reference  $43- 168$ pmol/L), Parathyroid hormone (PTH)  $2.0$ pmol/L ( $1.6-6.0$ ). TmP/GFR is low for age.
- Give an interpretation of the results. (1)
  - What is the most likely diagnosis? (1)
  - Discuss the pathophysiology of the disease. (5)
  - Discuss the management of the patient. (3)
- [10]
- 2 The teenager (14-years-of age) with peri-natally acquired HIV infection currently on second line anti-retroviral therapy including tenofovir presents with foot pain precipitated by jumping off a 2m wall. He also complained of lower back pain for 3 months duration. Investigations revealed stress fractures of the fourth metatarsals of both feet but no vertebral fractures. His bone density scan was reported as a T score of  $-3.24$  (Z score  $-2.3$ ) at the lumbar spine and -

5.86 (Z score -3.24) at the femoral neck. The alkaline phosphatase was 353 IU/L (38-140), parathyroid hormone of 7.5pmol/L (1.6 -6.9) with a serum phosphate of 0.37mmol/L (age specific reference 0.8- 1.45). The 25-hydroxyvitamin D level was 21nmol/L (50 – 75). The creatinine was 60umol/L (15-31). Examination of the urine showed a protein excretion of 1.93 g/24 h and glycosuria (2+) on dipstick with venous gas showing metabolic acidosis.

- a) What is the most likely pathogenesis of the presentation? (2)
- b) How would you manage the patient? (2)
- c) Which unique factors in paediatrics impact on the assessment of bone density? (2)
- d) How would you counsel the family about the risk of future fractures? (2)
- e) What are the controversies regarding vitamin D deficiency? (2)
- [10]
- 3 Your views on surgical intervention in children with disorders in sex development. [10]
- 4 The emotional wellbeing of the adolescent with diabetes is important to maintain optimal control. Your views on possible causes, management and prevention of emotional disturbances. [10]
- 5 a) Define obesity in paediatric practice. (2)
- b) List the commonest endocrine causes of obesity. (3)
- c) Discuss management of 'simple' obesity (i.e. caused by life style factors) in South Africa. (5)
- [10]
- 6 a) Discuss indications for growth hormone therapy in the South African context. (5)
- b) Discuss the side effects and complications of growth hormone therapy. (5)
- [10]
- 7 As the duty endocrinologist you are consulted by a colleague who has admitted a 6-week-old infant following a generalised seizure. Also found to have an upper lobe pneumonia. Investigations on admission:
- Blood glucose 1.9mmol/L
  - Lactate 12.4mmol/L
  - Metabolic acidosis
  - White cell count of 1.2 with 20% neutrophils
- Subsequent investigations:
- Lactate / Pyruvate ratio normal
  - Amino acid profile normal
  - Urinary organic acids normal

- a) Provide a diagrammatic outline for the diagnostic evaluation of a metabolic acidosis. (3)
  - b) What is the most likely diagnosis in this infant? (2)
  - c) Name the enzyme defect. (1)
  - d) Which single test could confirm the diagnosis? (1)
  - e) Briefly outline the long term management goals. (3)
- [10]

8 A neonate with indeterminate gender is referred for assessment. There is a small clitorophallus with a gonad palpable in one labioscrotal fold and no gonad palpable on the other side. Pelvic ultrasound and genitography reveal a vagina and uterus. Karyotype is 45X/46XY.

- a) What is the most likely diagnosis? (2)
  - b) Which other investigations would be of value in managing this infant? (4)
  - c) Which gender would you assign? Justify your decision. (4)
- [10]