



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

23 August 2012

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 A three-year-old boy is referred to your clinic. His parents are concerned about his increasing pigmentation and the appearance of pubic hair. They have also noted that he is significantly taller than his peers at pre-school. Clinical examination confirms the increased pigmentation, height falls on the 97th centile (mid-parental height on 25th centile), body odour is noted and he has mild facial acne. Genital assessment reveals penile enlargement, the presence of pubic hair and small testes (2ml volume)
- a) What is the most likely diagnosis? Explain your answer. (3)
 - b) Explain the mechanism of
 - i) the increased pigmentation and
 - ii) the tall stature. (4)
 - c) Discuss the genetics of this condition. (4)
 - d) Which investigations should be performed to
 - i) confirm the diagnosis and
 - ii) assist with monitoring the response to treatment in the long term? (7)
 - e) Discuss the long term management of this patient. (7)
- [25]
- 2 A 9-year-old boy is admitted with diabetic ketoacidosis. The blood pressure 135/80 mmHg, Heart-rate is 140 beats per minute. Investigations reveal the following blood glucose 55mmol/L; Sodium 125 mmol/L, Potassium 6.7 mmol/L, Urea 12.0 mmol/L, creatinine 150 umol/L ; Venous blood gas : pH 6.97, pCO₂ 1.8 kPa, Base deficit 28, Standard bicarbonate 6 mmol/L; blood ketones 3+.
- Discuss
- a) The risk of cerebral oedema occurring in *this* boy. (5)
 - b) How you would try to prevent cerebral oedema from occurring, and the rationale for these treatment strategies. (10)
 - c) How you would manage cerebral oedema that does occur, and the rationale for these treatment strategies. (10)
- [25]
- 3 A 15-year-old girl is referred to the endocrine clinic with a history of amenorrhoea of 1 year duration. She achieved her menarche at 12-years of age. Clinically she is obese with excess body and facial hair
- a) Discuss the differential diagnosis and the investigations you would perform in this adolescent. (10)
 - b) Discuss the relationship between premature adrenarche and polycystic ovarian syndrome. (5)
 - c) Discuss the pathogenesis of the spectrum of abnormalities in polycystic ovarian syndrome. (5)

- d) Discuss the principles of management of this adolescent with polycystic ovarian syndrome. (5)
[25]
- 4 a) Describe the pathophysiology, prevention, clinical features and management of neonatal thyrotoxicosis. (12)
- b) Discuss the prevalence and management of thyroid abnormalities in children with Down's syndrome. (10)
- c) Describe the molecular basis for McCune Albright syndrome. (3)
[25]



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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)

- 1 A 2-year-old boy is referred for assessment of his increasing bowed legs. His mother also has a bone disease. Investigations: Calcium 2.7 mmol/L, Phosphate 1.2 mmol/L, Alkaline phosphatase 658 IU/L (ref range 100-300), PTH 1.3 pmol/L (ref range 0.9-5.5)
 - a) What is the diagnosis? (3)
 - b) What additional investigation would clarify the diagnosis? (3)
 - c) Briefly discuss the genetics of the condition. (4)[10]
- 2 A boy born at term with a birth weight of 3.3 kg has an apnoeic episode 12 hours after birth. Blood glucose at the time measured 1.1 mmol/L. On examination he is found to have a micropenis. Thyroid screen done at birth reveals a normal TSH (thyroid stimulating hormone) with a low free T4
 - a) What is your diagnosis? Justify your answer. (3)
 - b) What other system should be assessed carefully? (2)
 - c) How would you investigate this neonate? (5)[10]
- 3 With regard to HbA1c
 - a) Discuss its use in the diagnosis and management of diabetes. (5)
 - b) What are the limitations of the test? (5)[10]
- 4 Discuss the diagnostic utility of measuring Anti-Mullerian Hormone in the evaluation of the child with a suspected disorder of sex development. [10]
- 5 Discuss the dilemmas with gender assignment in the 46 XX completely virilised child (Prader stage 5) diagnosed with congenital adrenal hyperplasia – classic salt wasting type. [10]
- 6 Write short notes on glucagon-like peptide (GLP) 1. [10]
- 7 A 10-year-old girl with type 1 diabetes needs a dental procedure requiring a fast because she will need anaesthesia. The dentist and anaesthetist ask you how best to proceed. How do you advise them if
 - a) The child is on regular and NPH insulin (human insulin)? (4)
 - b) The child is on insulin analogues? (3)
 - c) The child is on continuous subcutaneous insulin infusion (CSH)? (3)[10]
- 8 A six-week-old baby presents with seizures. You are asked to see him because the serum total calcium (corrected) is 1.3 mmol/L.

- a) Which investigations would you request? (5)
b) What management would you suggest? Explain. (5)
[10]
- 9 a) How is hypoglycaemia defined when the cause is under investigation. Explain. (4)
b) How is hypoglycaemia defined in a child with diabetes? Explain. (3)
c) How is hypoglycaemia defined in a paediatric emergency unit. Explain. (3)
[10]
- 10 Discuss the biochemical features and molecular basis for apparent mineralocorticoid excess presenting with hypertension in a child. [10]
- 11 A 10-year-old child with severe hypertension (due to a pheochromocytoma) is to undergo surgery to remove the tumour.
a) Suggest how you would manage the patient pre-operatively. (5)
b) What precautions are necessary during the surgery? (5)
[10]
- 12 The history and physical examination are normal in a 11-year-old prepubertal girl presenting with short stature (height standard deviation score of -2.1). The height is significantly less than the mid-parental height
a) List the screening investigations that you would request in the child. (5)
b) Discuss the protocol for the growth hormone stimulation test. (5)
[10]