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

25 July 2019

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)

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1 A 14-year-old boy with Type 1 Diabetes Mellitus diagnosed at 3-years of age is followed up at the clinic. His treatment includes regular human insulin and isophane insulin.
He administers regular human insulin before breakfast and before supper, and the isophane insulin at breakfast and in the evening. His HbA1C values over the last 2-years, have been 7.6% (60 mmol/mol), 9.1% (76 mmol/mol), 9.9% (85 mmol/mol), 10.8% (95 mmol/mol) and most recently 11.6% (103 mmol/mol). Discuss the factors contributing to the deterioration in his glycaemic control, and how you would address these in your management strategy. [25]

2 A 9-year-10-month-old female had visual problems since Grade 1 and was treated by an optometrist. For the past 2-years, the mum was still unhappy as her child continued to have visual problems. She was recently reassessed and diagnosed with esotropia for which she received surgery. However, as part of her investigations, a CT brain was done to screen for underlying causes of the esotropia. A cystic suprasellar mass was noted with calcifications.

a) Explain the embryology and genetics behind the development of the mass. (5)
b) Discuss the early and late onset endocrinopathies of this condition and investigations that are required to manage these complications. (10)
c) Discuss the immediate post-operative complications and emergency treatment options that may be anticipated for this patient. (6)
d) Discuss the late effects of treatment that this patient should be monitored for. (4)

[25]

3 A panicked mother and her 14-day-old baby boy are seen at the General Paediatric Outpatient clinic following a telephone call she received from a doctor stating that her baby had an abnormal new-born screen. The mother reports that apart from heartburn and palpitations, she was well during her pregnancy. The baby was born at term, with a birthweight of 3200g and normal Apgar scores. The baby's TSH result is 65 mIU/L.

a) Discuss the screening tests, including the advantages, disadvantages, sensitivity and specificity, used to detect congenital hypothyroidism in the neonatal period. (5)
b) Justify the need for implementing a newborn screening programme for diagnosing congenital hypothyroidism in South Africa. (2)

On examination, this baby is lethargic with facial puffiness, cool peripheries and low tone. The results of the investigations performed are as follows:

Test	Result	Reference Range
TSH	70 mIU/L	0.72-11.00
T4	3 pmol/L	11.5-28.3
Thyroid Receptor Antibodies	10 U/L	<1

Tc- 99m Pertechnetate Thyroid Scan	There is no uptake of pertechnetate noted.

c) What is the most likely diagnosis and discuss the pathophysiology of this condition? (4)
d) Discuss the management of this baby. (4)
e) Discuss the potential neurodevelopmental and cognitive outcomes and discuss how these could be managed. (4)

The baby, now two-months-old, is routinely reviewed at the paediatric endocrine clinic. His mother reports that this baby does not sleep very well and is often startled. On examination, his weight is 4000g, his pulse is 170 beats per minute, and he does appear quite jittery.

f) How would you manage this baby further? (3)

The mother informs that you that she would like to have another baby soon.

g) i) Discuss the risk of recurrence of this condition in future pregnancies. (1)
ii) Briefly discuss the screening strategies available for foetal hypothyroidism. (2)

[25]

4 Discuss the pathophysiology, clinical manifestations and management of the endocrine complications associated with anorexia nervosa. [25]

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26 July 2019

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)

Please answer questions 1 – 3 in one book.

1 An 11-year-old male presents to casualty with a history of falling off a chair after trying to help his mum change a light bulb. He is tender over his right lower limb and x-rays confirm a right tibia fracture. The paediatric orthopaedic surgeon noted that the child had blue sclera and he was of normal stature. On further history, the mom reports two previous fractures of both forearms; sustained after playing outside at the age of 2-years and the other at 5-years after falling off the jungle gym.

a) What is the most likely diagnosis of this patient? (2)

b) What are the possible genetic causes of this condition and discuss the underlying pathophysiology? (8)

[10]

2 A 3-year-old male presented to the paediatric outpatient clinic with abdominal distention. The clinical history revealed that the child had become progressively unwell over the last 6-months with abdominal pain and tiredness. He was an only child with no significant family history. On examination, he had severe hepatomegaly and growth retardation. There was no muscle weakness, skeletal abnormalities or cardiomegaly. A few blood tests were sent as an initial diagnostic screen, as the consultant paediatrician had a diagnosis in mind. Some of the non-fasting biochemistry tests were as follows:

Investigations	**Results**	
Sodium	142 mmol/l	136 - 145
Potassium	3.8 mmol/l	3.4 - 4.7
Chloride	101 mmol/l	98 - 107
Urea	4.5 mmol/l	1.8 - 6.4
Creatinine	60 µmol/l	27 - 54
CO_2	15 mmol/l	21 -29
Urate	0.63 mmol/l	0.21 – 0.42
Random glucose	2.0 mmol/l	4.4 -8.9
Lactate	6.5 mmol/l	0.1 – 2.2
Cholesterol	5.9 mmol/l	< 4.4
Triglyceride	7.9 mmol/l	< 0.85

PTO/Page 2 Question 2a)…

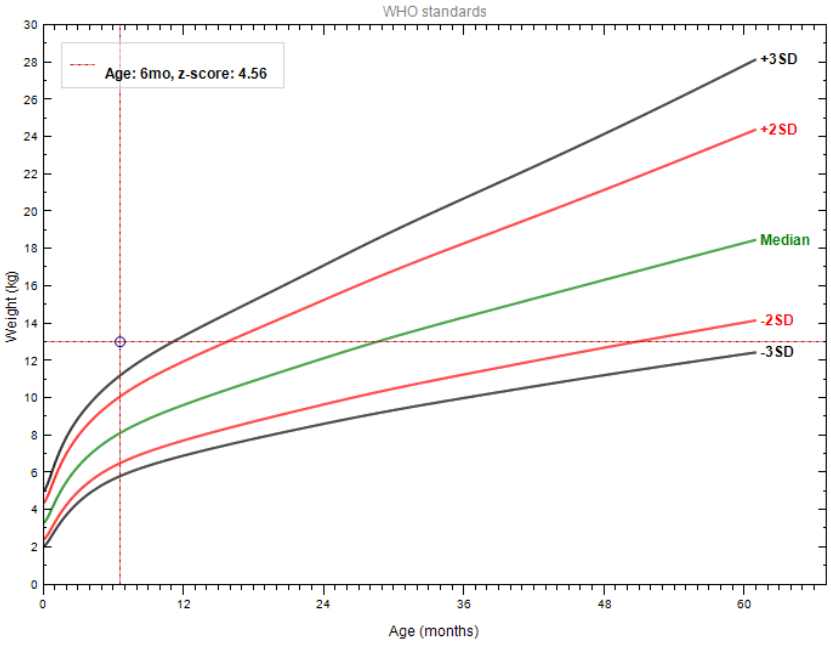
a) What is the most likely diagnosis? (2)
b) Explain the pathophysiology of the abnormal blood investigations. (4)
c) Which stimulation test is recommended and discuss the principles and findings of this test. (4)
[10]

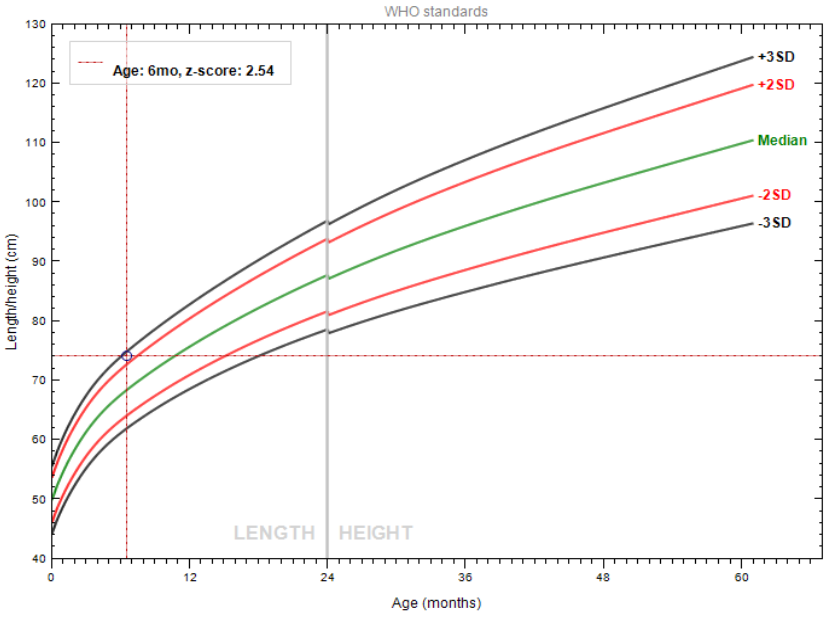
3 A 3-month-old baby with clinical features of Down Syndrome is screened for thyroid disease. Result: TSH = 15 mIU/l (0.35-4.94).

a) Discuss the pathogenesis of thyroid abnormalities in children with Down Syndrome. (5)
b) Discuss your approach to the management of this patient. (5)
[10]

Please answer questions 4 – 5 in one book.

4 A 6-month-old boy is reviewed for rapid weight gain. He is breastfeeding, and no complimentary feeds have been introduced. He was born at term and had a rather uneventful delivery. His mother reports that he does cry often and that she must breast feed to calm him. He has been treated at least three times for upper respiratory tract infections. Apart from a phallus that measures 2 cm, the clinical examination is unremarkable.





Bone Age	16 months Tanner White House

a) Briefly explain how the deficiency of this hormone will result in
   i) Hyperphagia. (1)
   ii) Recurrent upper respiratory tract infections. (1)
   iii) Hypogonadism. (1)
   iv) Skeletal Maturation. (1)

b) What other clinical symptoms and signs may become apparent as this child grows? (4)

c) How would you manage this child further? (2)

[10]

5 You are asked to review a 3-day-old neonate with atypical looking genitalia. On clinical examination, this child is adequately hydrated and there is no hyperpigmentation. The cliterophallus measures 2 cm, there is single opening noted at the base of cliterophallus, and the gonads are palpable in the inguinal canal.

FSH	10 IU/L	0.4 – 1.6
LH	12 IU/L	< 0.5
Testosterone	23 nmol/L	0.5 – 1.5
Chromosome Analysis	46XY, SRY detected	
17 OH Progesterone	4 nmol/L	< 19

a) In diagrammatic form, highlight the genetic and hormonal factors involved in the embryology of the urogenital tract from the bipotential gonad to the male genitalia. Indicate on this diagram where common disorders of sex development can occur. (3)
b) What is the most likely diagnosis of this neonate? (1)
c) Discuss the genetics of the above condition. (1)
d) What additional investigations can be done to substantiate the diagnosis and how would you interpret them? (2)
e) Briefly discuss the principles of gender assignment in this child. (3)
[10]

Please answer questions 6 – 10 in separate books.

6 Hypertension
a) List the endocrine causes of hypertension in childhood and adolescence. (5)
b) A 14-year-old girl presents with a blood pressure of 165/100. Discuss your clinical approach to investigating this girl for an endocrine cause of hypertension. Use a primary and secondary screening approach to the investigations. (5)
[10]

7 Diabetes Technologies
a) Discuss the advantages and disadvantages of insulin pump therapy in young children. (6)
b) Discuss the use of diabetes technology to improve time in range. (2)
c) Discuss the role of continuous glucose monitoring in resource limited settings. (2)
[10]

8 A 2-month-old girl presents to the emergency department with increased working of breathing and poor feeding. Her mother reports that she had flu-like symptoms preceding her admission. On examination, her anthropometry is appropriate for her age. Her pulse is 180 bpm and are of small volume. Her respiratory rate is 60 breathes per minute with moderate respiratory distress. Her capillary refill time is 5 seconds. Her apex beat is localised to the 6th intercostal space, anterior axillary line. No murmurs are audible. Her breath sounds are normal. A soft liver is palpable 3 cm below the costal margin. There is reduced peripheral tone.

pH	7.23	7.38-7.42
PCO_2	3.2 kPa	5.1 - 5.6
PO_2	9.0 kPa	10.5 – 13.5
Bicarbonate	12.8 mmol/L	22 -28
BE	-10	
Lactate	4.5 mmol/L	0.1 – 2.2
Blood glucose	1.9 mmol/L	
Betahydroxybutyrate	0.1 mmol/L	< 0.3mmol/L
Insulin	0.1 Miu/L	2.2 - 10.1
Cortisol	565 nmol/L	101 - 535
ALT	5000 U/L	0 - 30
CK	15000 U/L	39 - 308

a) Use a diagram to briefly outline an approach to hypoglycaemia in neonates. (4)
b) What is the most likely diagnosis in this child? (1)
c) Provide reasons for the biochemical abnormalities listed above. (2)
d) Outline the emergency management plan for this child. (3)
[10]

9 A 15-year-old girl is referred to you for assessment of delayed puberty. She has had no secondary sexual development. She has a background history of acute lymphoblastic leukaemia treated at the age of 7-years. Discuss your approach to the investigation and management of this girl. [10]

10 Baby AA is delivered at term by emergency caesarean section for foetal distress. Birth weight is 2300g. He presents at 3-months of age with failure to thrive, dehydration and difficulty breathing. Initial laboratory evaluation reveals hyperglycaemia (serum glucose 33.6 mmol/l) and a severe metabolic acidosis (pH 6.87, bicarbonate 3 mEq/L) with ketonuria.

a) Discuss the pathophysiology of neonatal diabetes. An annotated diagram may be used. (5)
b) Discuss the role of molecular genetic testing in the management and prognosis of this condition. (5)
[10]