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

19 March 2014

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 3-and a half-year old girl presents with a generalised seizure. On examination her weight is 20 kg and her height 85 cm. She has round faces and brachydactyly with noticeable shortening of the 3rd and 4th metacarpals and metatarsals. Subcutaneous ossification is found at multiple sites

a) What is your clinical diagnosis? Explain your answer. (3)
b) Discuss the genetics and pathophysiology of the phenotype described above. (7)
c) Discuss the investigations that you would do to confirm your clinical diagnosis. (5)
d) Your investigations included a CT scan. What would you expect to find? (2)
e) In this condition there may be resistance to a number of other hormones. List these hormones. (4)
f) Briefly discuss the principles of treating this condition. (4)

[25]

2 Growth

a) Explain the process of catch-up growth within the growth plate. (5)
b) Discuss the potential side effects of growth hormone therapy, how to monitor for them and their treatment. (6)
c) Explain the hormones required for normal growth throughout childhood and their temporal influences on growth and the growth plate. (10)
d) What are the treatment options, covering potential side-effects for boys and girls who wish to limit their final height? (4)

[25]

3 Manipulating puberty in girls with severe learning difficulty has ethical implications. Write short notes on the following

a) Clinical approach. (5)
b) Intellectual and physical abilities of the child. (5)
c) Impact of puberty in a girl with severe learning disabilities. (5)
d) Interventions to manipulate puberty: benefits and risks. (8)
e) Who decides. (2)

[25]

4 a) An 11-year-old patient presents with 6 kg weight loss over the last 4 months, deteriorating school performance, difficulty sleeping and palpitations- The TSH is <0.01 mmol/l and the Free T4 48mmol/l

i) What tests could you do to confirm Grave's disease from cheapest to most expensive? (5)

ii) What are the pro's and con's of RAI therapy vs. medical therapy? (4)

iii) The family decide on medical therapy, outline a plan for them. (3)

iv) The family decide on RAI therapy, outline a plan. (3)

b) An infant is referred to you at 9-months for hypotonia, delayed development. He has a longstanding history of constipation and prolonged neonatal jaundice

i) What is the most likely diagnosis? (2)

ii) What are the long term cognitive concerns in this patient? (3)

c) You are investigating a short 15-year-old girl with features of hypothyroidism, a TSH of 436 mmoll and muscle pseudohypertrophy

i) What is the name of this syndrome? (1)

ii) What are the risks of hormone replacement? (2)

iii) Her MPH sits on the 75th %, she is currently below the 3rd % and her BA is 10y 6 mo. What is her final height likely to be and why? (2)

[25]

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20 March 2014

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 Write short notes on two of the following
- a) The genetics of ovarian differentiation. (5)
- b) 46XX males. (5)
- c) Confirmation of the diagnosis of 21-hydroxylas deficiency (CYP21). (5)

[15]

2 A 15-year-old boy is referred to you with short stature. On examination you confirm that he is short. He is pre-pubertal with testicular volumes of 3 ml bilaterally. He is also noted to be anosmic
- a) What is the likely diagnosis? (1)
- b) How is the condition inherited? (1)
- c) What is the molecular basis of the condition? (2)
- d) Explain the patient`s anosmia and pre-pubertal status. (3)
- e) Briefly discuss the management of this patient. (3)

[10]

3 Write short notes on the following monogenic causes of obesity
- a) Prader-Wili syndrome. (4)
- b) Leptin mutation. (3)
- c) MC4R mutation. (3)

[10]

4 Surviving childhood cancer is common. Describe the endocrine consequences of
- a) Chemotherapy. (2)
- b) Cranial radiation. (4)
- c) Spinal radiation. (2)
- d) What are the potential pre-existing risks in childhood cancer patients that increase the risk of growth hormone related complications? (2)

[10]

5 An infant presents with hypoglycaemia. In a flow diagram discuss your approach and differential diagnosis. [10]

6 A 3-year-old boy is diagnosed with Diabetes Mellitus, What are the clinical options and difficulties in managing a child of this age? [10]

7 A 7-year-old with JRA who has been on prednisone for 4 years presents with short stature
a) List the possible mechanisms of growth failure. (5)
b) What are the 2 most common features of Cushing syndrome in children? (2)
c) The Rheumatologist wants to wean the steroids and try disease modifying agents. How would you advise them? (3)
[10]

8 A 5-year-old male presents with precocious puberty. His testes are 4cc, slightly increased but not to the degree for enlarged phallus of 8cm. His bone age is 8 years. Testosterone increased, LH and FSH low and remain low with GnRH stimulation testing. HCG not increased. Dad also had early puberty
a) What is the likely diagnosis? (2)
b) How would you manage him? (4)
c) Discuss Van Wyk-Grumbach syndrome. (4)
[10]

9 A young family brings their daughter to you diagnosed with 45XO Turner syndrome on amniotic screening. She was born with lymphoedema and a webbed neck
a) What other tests would you order on this girl? (2)
b) What are the growth prospects and what therapies are available to improve them? (2)
c) How and when should pubertal induction be carried out? (2)
d) What screening tests would you perform and at what intervals? (3)
e) Describe the school and psychosocial difficulties that this child might have. (1)
[10]

10 A 14-year-old boy presents with a 3 year history of intermittent nausea and vague abdominal discomfort. He has occasional palpitations and is reportedly an anxious child. He has a family history of a pheochromocytoma in an uncle
a) List the tests that can be performed to screen for a pheochromocytoma. (2)
b) What imaging studies can be performed to localise a tumour following a positive screen? (2)
c) What pre-op preparation is required? (2)
d) List 4 genetic syndromes that are associated wih familial pheochromocytomas. (4)
[10]