THE COLLEGES OF MEDICINE OF SOUTH AFRICA

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Examination for the Subspeciality Certificate in Paediatric Neurology of the College of Paediatricians of South Africa

27 February 2020

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) You are called to the neonatal ward to assess a female baby suspected to have tuberous sclerosis complex based on antenatal screens

i) What are the challenges in terms of confirming a diagnosis of Tuberous Sclerosis in this age group and how would you then approach confirming a diagnosis? (3)

ii) How would you manage the child if she develops epileptic spasms? (2)

iii) Which specialists and which interventions should be part of the care you would implement specifically for the brain complications during infancy and the school-going child? (5)

b) A 3-month-old infant presents with a respiratory illness and is noted to have tongue fibrillations. He is not coping on nasal oxygen and a decision needs to be taken re mechanical ventilation

i) What is the differential diagnosis? (2)

ii) How would you confirm the diagnosis? (3)

iii) How would you manage the discussion around ventilation and what are the future challenges with health care? (3)

iv) What are the "state of the art" interventions for 2 of the differential diagnoses? (2)

c) Discuss Neonatal Encephalopathy with specific reference to the ACOG 2014 criteria for intrapartum hypoxia under the following headings

i) Definition. (3)

ii) Clinical presentation. (5)

iii) Causes. (4)

iv) Diagnosis. (5)

v) Management. (5)

vi) MRI findings. (4)

vii) Morbidity. (4)

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2 a) Write short notes on the evidence for the use of Cannabis in paediatric epilepsy. (10)

b) Write short notes on the role of precision medicine in treating epilepsy under the following headings

i) What is precision medicine? (5)

ii) Discuss the role of precision medicine in management of

1. Glut 1 deficiency. (5)
2. Pyridoxine dependent epilepsy. (5)
3. Dravet Syndrome. (5)

c) Discuss Congenital Myopathies under the following headings

i) Clinical features that distinguish the sub-types. (10)

ii) Therapeutic aspects in congenital myopathies. (10)

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3 a) Comprehensively discuss the various co-morbidities in a patient with Duchenne's Muscular Dystrophy. (30)

b) As the treating paediatric neurologist at a tertiary hospital, formulate a long-term management plan for patients with Duchenne Muscular Dystrophy, excluding medical treatment. Be specific in terms of visit intervals, timing of investigations, involvement of other team members. You can tabulate your answer. (10)

c) You have just confirmed the diagnosis of Duchenne's Muscular Dystrophy in a 3-and-a-half-year-old boy. He has a heterozygous non-contiguous duplication of exons 5 to 9 and 22 to 34 of the DMD gene. In terms of medical therapy

i) What treatment if any would you consider starting at this point? (2)

ii) What do you regard as current standard of care in terms of chronic medical treatment for this patient in your setting? (3)

iii) Discuss novel treatments for Duchenne's Muscular Dystrophy. (5)

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4 A 14-month-old child presents with recurrent seizures

a) Explain how you would differentiate between whether the child has a developmental encephalopathy, an epileptic encephalopathy or a developmental and epileptic encephalopathy. How will this alter your management in each setting? (10)

b) What early clinical markers may indicate that the child could have Dravet syndrome? (10)

c) In the setting where the child has had 3 recurrent simple febrile seizures, what are your management options, based on existing guidelines and evidence. What would your recommendation to the parents be in terms of care? (10)

d) Summarise the 3 recently published multicenter randomised control trials assessing optimal second-line intervention for status epilepticus in children. How would this affect your approach to status in the South African setting? (10)

e) Discuss the role of epilepsy surgery as an option in an infant of this age. What is the evidence to support intervention in this age group? (10)

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