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

21 February 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 15-year-old girl presents to a primary health care clinic with thrush. On questioning, she says that she has had polyuria and polydipsia for a week. She is 165cm tall (Z-score 0.47), with a BMI of 30.1 (Z-score 2.30)
The medical officer has sent a formal laboratory glucose, which is 16mmol/l
The HbA1c is 9%
The medical officer has started metformin 500mg bd and referred her for further management

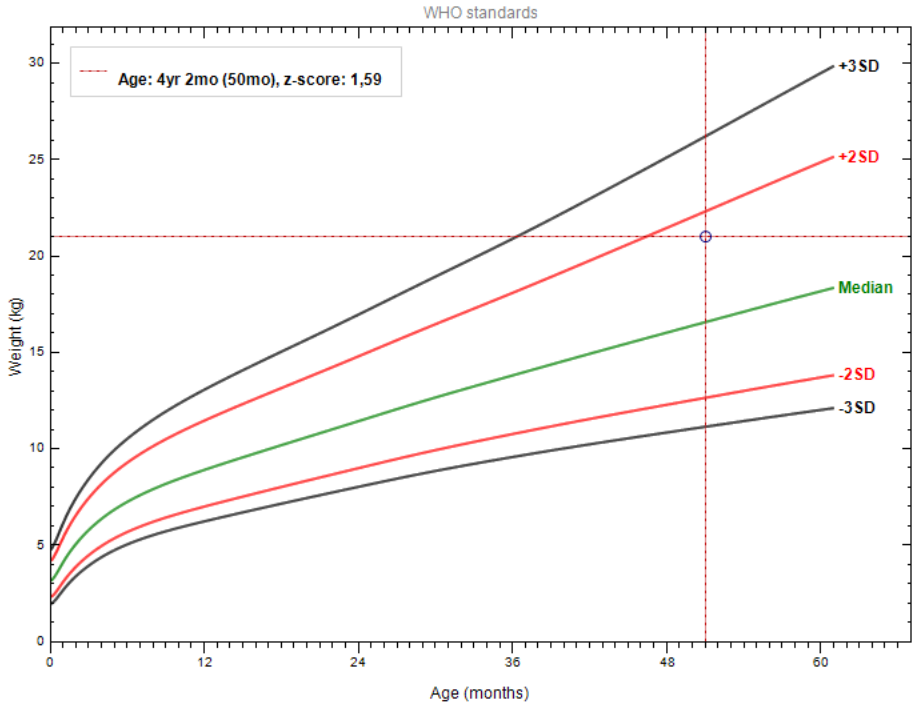
a) Discuss how insulin resistance impacts the development and presentation of Type 1 (ONE) diabetes, and how you would decide whether this girl has Type 1 or Type 2 diabetes. (10)
b) Discuss her comprehensive management plan, comparing the possible diagnoses. (15)
[25]

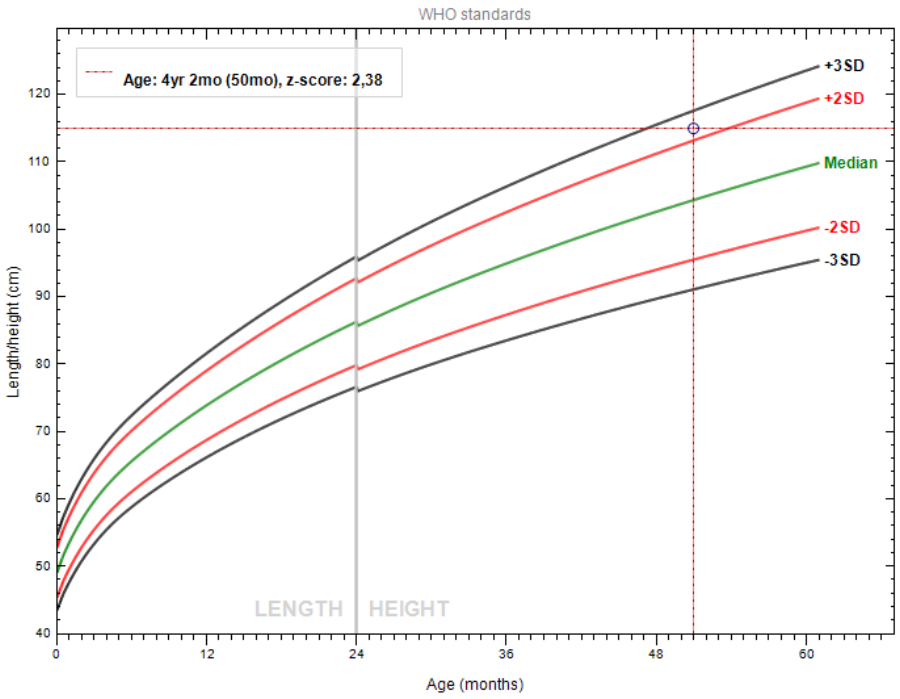
2 a) Hypopituitarism can be due to mutations in genes involved in pituitary development. List at least 5 of these genes. (5)
b) Discuss the embryology of the pituitary gland. (10)
c) A 4-year-old child presents with signs and symptoms of multiple anterior and posterior pituitary hormone deficiency. Discuss the signs and symptoms and discuss emergency management. (10)
[25]

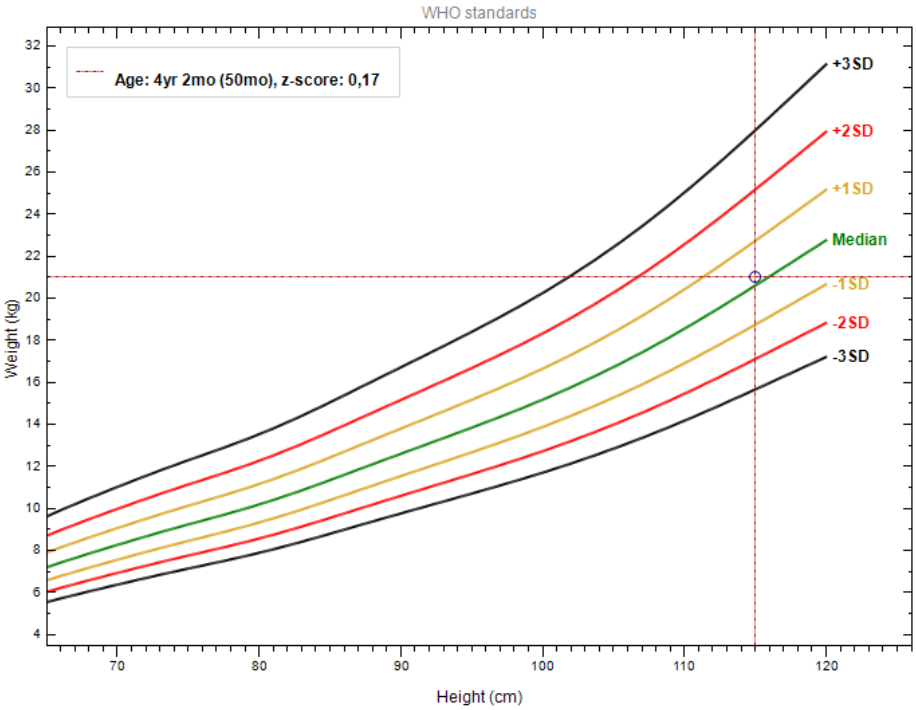
3 A 27-year-old patient known with Graves' Disease gives birth to a term neonate via caesarean section. Mom is currently on Carbimazole (40mg daily). On day 2 of life, baby is noted to have mild proptosis, persistent tachycardia as well as pedal and periorbital oedema. The baby is assessed as having clinical features in keeping with high-output cardiac failure.

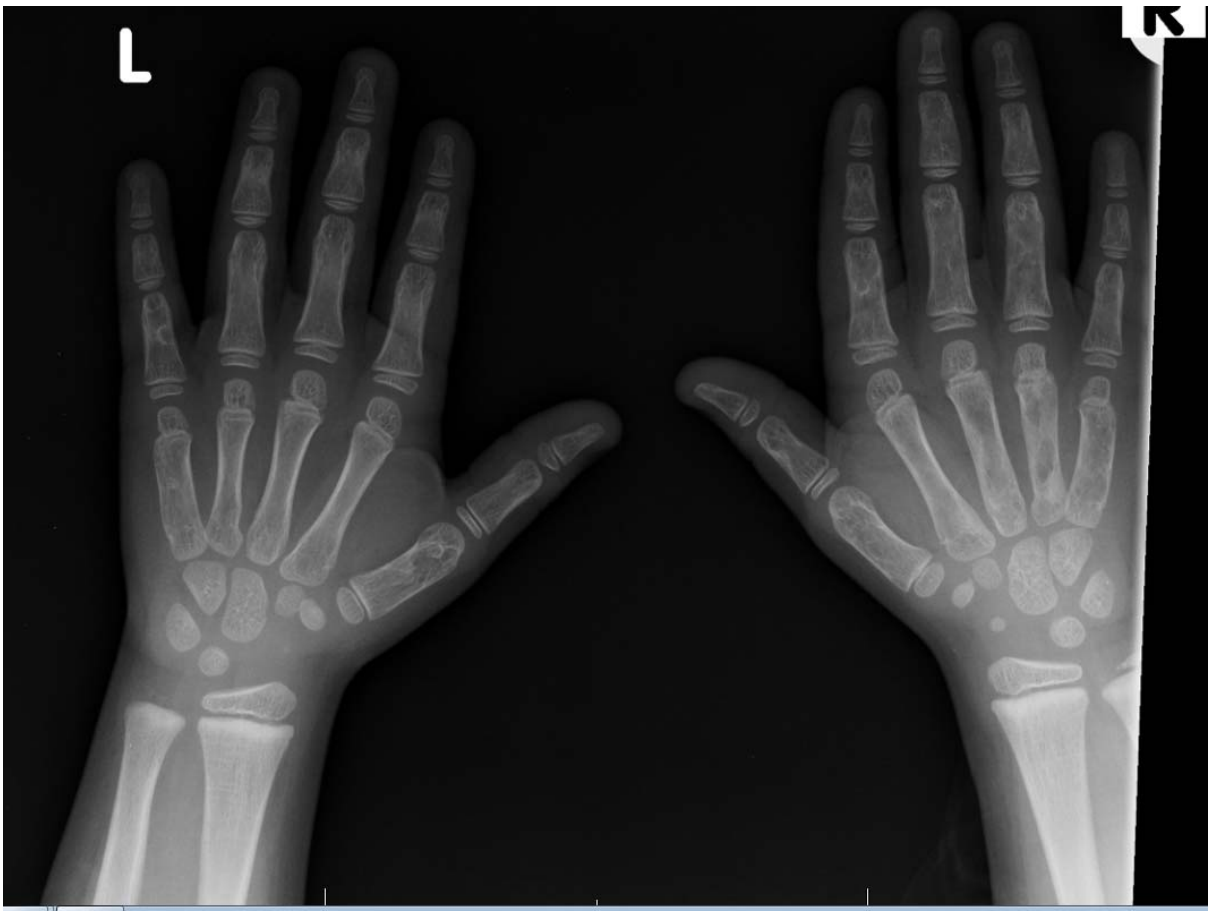
a) What is the most likely diagnosis in the neonate? (2)
b) Explain the pathophysiology behind this diagnosis. (4)
c) What confirmatory tests would you request in the neonate? (4)
d) What would the emergency treatment be in the neonate? (13)
e) Would you advise mom to breastfeed? Explain your answer (2)
[25]

4 A paediatrician has referred a 4-year-old girl with vaginal bleeding. This is the 3rd such episode in 8 months. There are breast buds but no secondary sexual hair. She has an extensive birthmark on her left leg.
Anthropometry:






WHO standards
Age: 4yr 2mo (50mo), z-score: 0,17
+3SD
+2SD
+1SD
Median
-1SD
-2SD
-3SD
Weight (kg)
Height (cm)
32
30
28
26
24
22
20
18
16
14
12
10
8
6
4
70
80
90
100
110
120


L
R

Skeletal maturity: reported as 5y9m (Greulich and Pyle Atlas)
Biochemistry:

CHEMICAL PATHOLOGY

Tel: 041 395 6183, Fax: 041 374 1996

Specimen received: Clotted blood
Tests requested: b-HCG @, AFP @, TSH @, FT4 @, FSH @, LH @, E2 @, Prog @, Prol @, PTH
@ Test referred to another laboratory

Blood chemistry:			
Beta-HCG	**<1 L**	IU/L	1 - 3
Alpha-feto protein (AFP)	1.0	ug/L	0.6 - 4.2
Thyroid function tests:			
Thyroid stimulating hormone	0.17	mIU/L	
Thyroxine (free T4)	8.6	pmol/L	
Endocrinology:			
Follicle stimulating hormone	**<0.2 L**	IU/L	0.2 - 3.3
Luteinising hormone	**<0.2 L**	IU/L	0.3 - 1.9
Oestradiol (E2)	286	pmol/L	
Progesterone	1.0	nmol/L	
Prolactin	31.7	ug/L	

Endocrinology:			
Parathyroid hormone (PTH)	2.7	pmol/L	1.3 - 9.3

FBC and Differential Count:<Continued>

White Cell Count	10.79	x 10^9/L			6.00 - 18.00
Red Cell Count	4.42	x 10^{12}/L			3.70 - 5.30
Haemoglobin	12.3	g/dL			10.7 - 13.1
Haematocrit	0.349	L/L			0.330 - 0.390
MCV	79.0	fL			70.0 - 86.0
MCH	27.8	pg			23.0 - 31.0
MCHC	35.2	g/dL			30.0 - 36.0
RDW	13.3	%			
Platelet Count	428	x 10^9/L			180 - 440
MPV	9.1	fL			7.0 - 11.4
Neutrophils	36.90	%	3.98	x 10^9/L	2.00 - 5.50
Lymphocytes	47.60	%	5.14	x 10^9/L	3.60 - 12.00
Monocytes	**10.30 H**	%	**1.11 H**	x 10^9/L	0.00 - 0.90
Eosinophils	**4.30 H**	%	0.46	x 10^9/L	0.00 - 0.50
Basophils	0.90	%	0.10	x 10^9/L	0.00 - 0.20
Immature Cells	0.40	%	0.04	x 10^9/L	

White Cell morphology comment:
Lymphocytosis
Monocytosis

CHEMICAL PATHOLOGY

Tel: 041 395 6183, Fax: 041 374 1996

Specimen received: Clotted blood
Tests requested: Na, K, Cl, Urea, Creat, Ca, Mg, PO4, TP, Alb, T bili, C bili, ALT, AST ALP, GGT, LD

Blood chemistry:			
Sodium	**135 L**	mmol/L	136 - 145
Potassium	**4.9 H**	mmol/L	3.4 - 4.7
Chloride	101	mmol/L	98 - 107
Urea	2.1	mmol/L	1.4 - 5.0
Creatinine and estimated GFR:			
Creatinine	**21 L**	umol/L	23 - 37

Estimation of GFR using the MDRD (4-variable) equation is not performed if patient's age is less than 18 years or unknown, or if patient's sex is unknown.

Blood chemistry:			
Calcium	2.31	mmol/L	2.12 - 2.64
Magnesium	0.79	mmol/L	0.70 - 0.91
Inorganic phosphate	**1.82 H**	mmol/L	1.05 - 1.80
Liver function tests:			
Total protein	66	g/L	57 - 80
Albumin	**43 H**	g/L	29 - 42
Total bilirubin	**4 L**	umol/L	5 - 21
Conjugated bilirubin (DBil)	2	umol/L	0 - 5
Alanine transaminase (ALT)	**28 H**	U/L	5 - 25
Aspartate transaminase (AST)	26	U/L	0 - 59
Alkaline phosphatase (ALP)	**405 H**	U/L	96 - 297
Gamma-glutamyl transferase (GGT)	**23 H**	U/L	4 - 22
Lactate dehydrogenase (LD)	228	U/L	110 - 295

a) In general, which conditions can present with bleeding from the perineal area in girl children? (5)
b) Discuss the diagnosis in this child with respect to the information available. (10)
c) Which further investigations would you request? (5)
d) What are the treatment options for this girl? (5)

[25]

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22 February 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)

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1 A 16-year-old girl presents to your clinic with a 2-month history of excessive weight gain, headaches and easy bruising. On further enquiry she admits that she has been to a dermatologist who has prescribed "steroid" creams and medication for her acne. She has been on this treatment for 4 months. On examination, she is noted to have moon facies, with truncal obesity, violaceous striae and is hypertensive (BP = 155/99).

a) What is the most likely diagnosis in the above patient? (2)
b) What is the most likely cause for the above condition in this patient? (2)
c) What screening tests could you do to confirm your diagnosis? (4)

The patient further discloses that she is pregnant and is concerned about her baby.

d) Based on the above diagnosis, what are the likely risks to the neonate? (2)

[10]

2 Discuss the investigation and monitoring of hypophosphataemic rickets. [10]

3 A 12-year-old boy presenting with generalised tonic-clonic convulsions and altered level of consciousness is diagnosed with hypertensive encephalopathy and managed accordingly. Ultrasound abdomen is normal. Doppler ultrasound to rule out renal artery stenosis identifies normal blood flow in the renal arteries but a left adrenal mass. MRI imaging of the abdomen confirms a left adrenal mass (3.8x3.2x2.7cm) and a right adrenal mass (1.9x1.5x2.7) both of which are T2 lesions suggestive of bilateral pheochromocytoma.

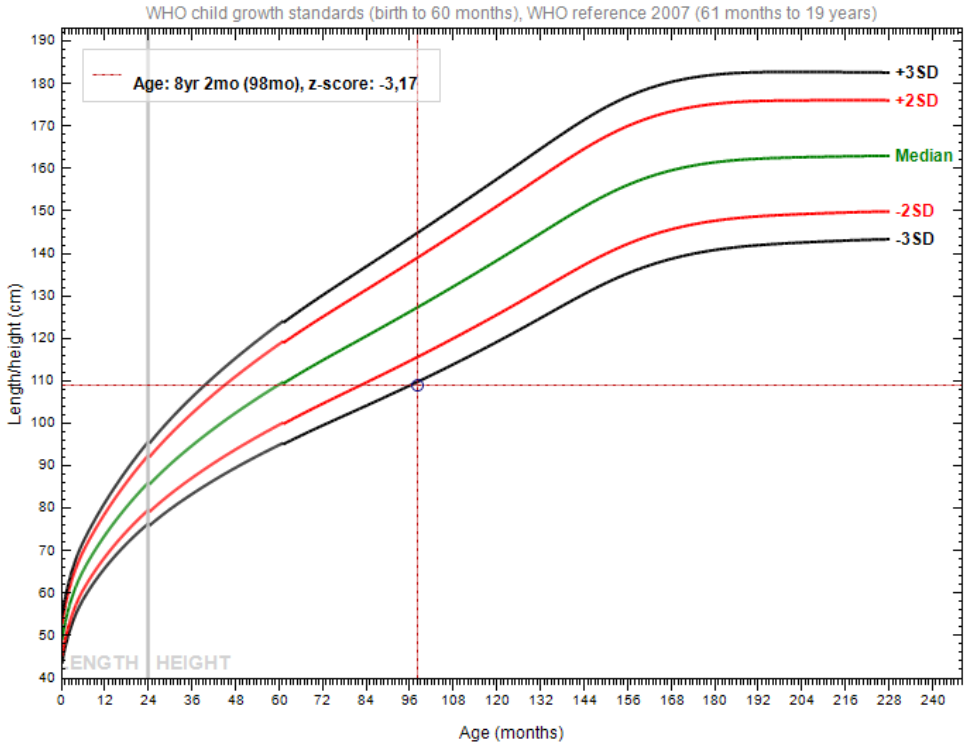
a) List the familial syndromes associated with pheochromocytoma and briefly describe their clinical features. (5)
b) Discuss the post-operative complications and long-term management of this patient. (5)

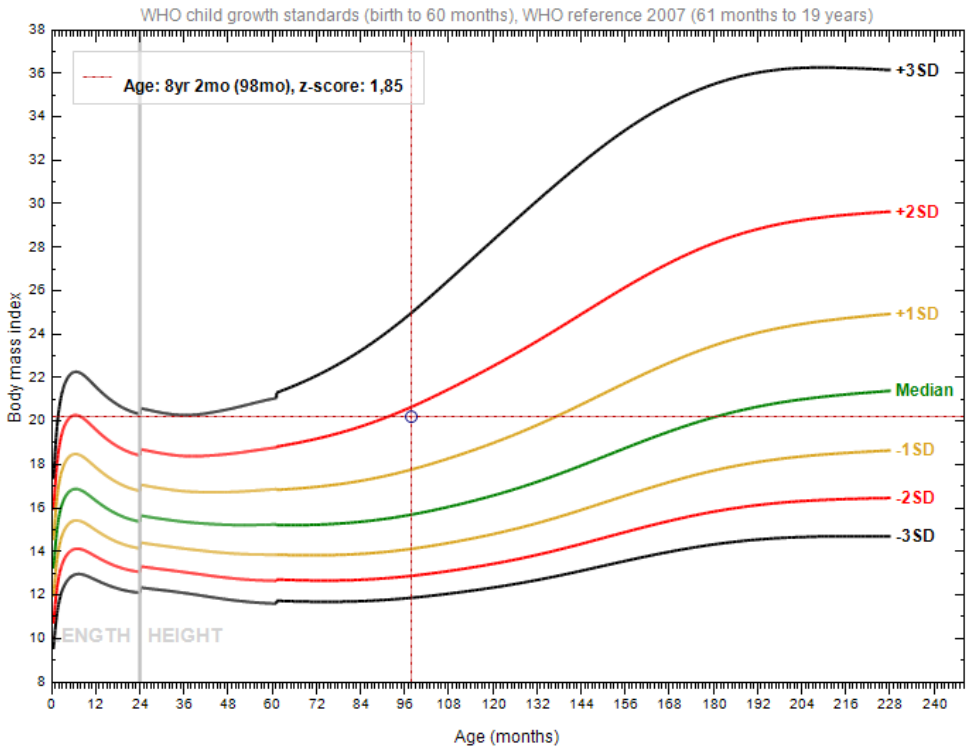
[10]

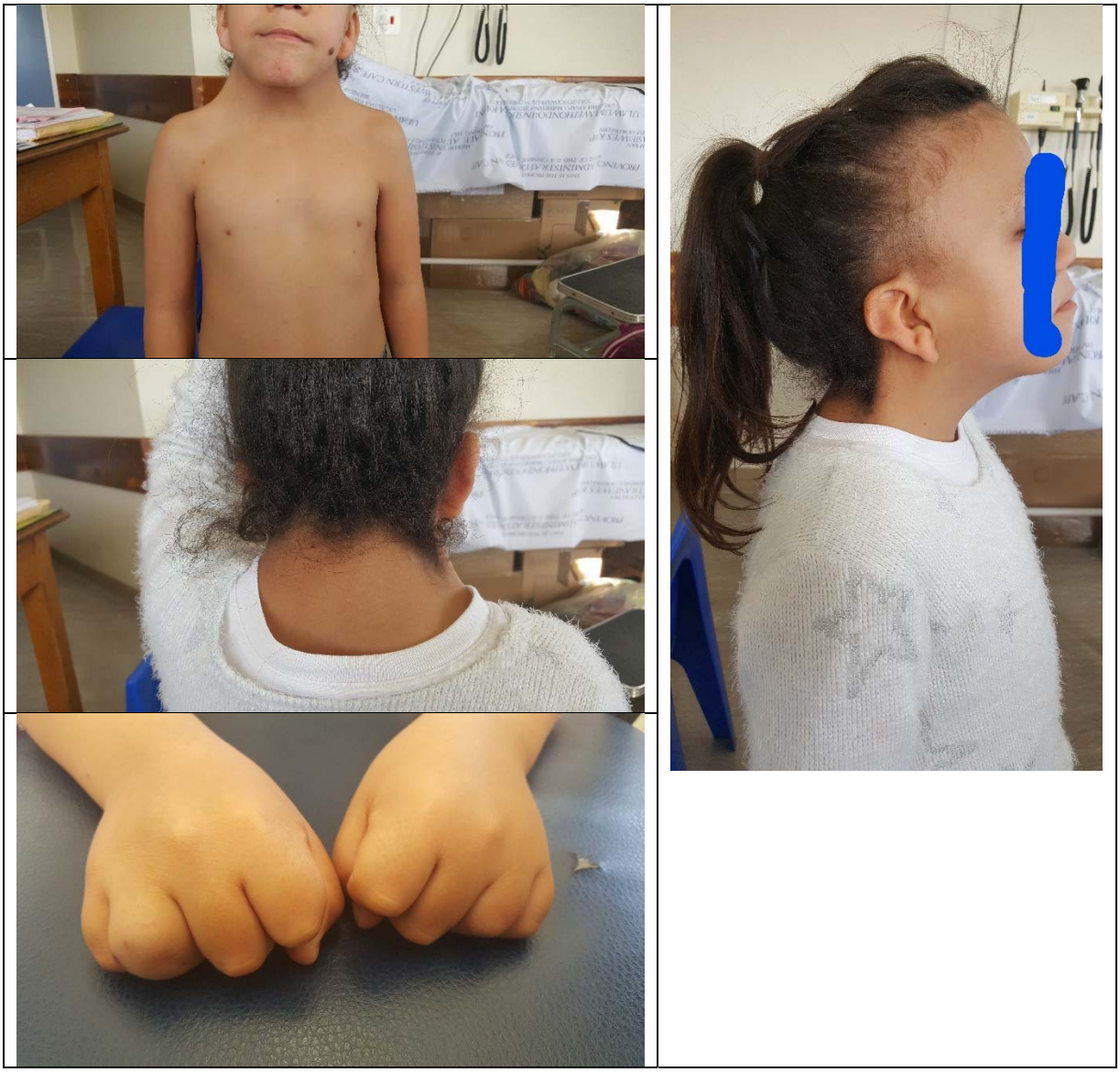
4 This 8-year-old girl has been referred to you for evaluation of short stature.

a) What is her diagnosis? Describe the clinical features evident that confirm this. (4)
b) What chromosome analysis results cause this syndrome? (3)
c) Does she need further genetic analysis? Explain your answer. (3)

[10]


WHO child growth standards (birth to 60 months), WHO reference 2007 (61 months to 19 years)
Age: 8yr 2mo (98mo), z-score: -3,17
+3 SD
+2 SD
Median
-2 SD
-3 SD
Length/height (cm)
LENGTH
HEIGHT
Age (months)


WHO child growth standards (birth to 60 months), WHO reference 2007 (61 months to 19 years)
Age: 8yr 2mo (98mo), z-score: 1,85
+3 SD
+2 SD
+1 SD
Median
-1 SD
-2 SD
-3 SD
Body mass index
LENGTH
HEIGHT
Age (months)



5 A 5-day-old term neonate presents with features in keeping with significant dehydration (10% dehydration), acidosis and poor weight gain since birth. No history of diarrhoea or vomiting is reported. On examination, it is noted that the baby has a prominent clitoris with hyperpigmented labioscrotal folds, no rugae and no gonads palpable. PCR aneuploidy result is "46XX".

Laboratory investigations reveal:

Na	122 mmol/l
K	6.8 mmol/l
CO_2	9 mmol/l
Urea	7.6 mmol/l
Creatinine	78 umol/l

a) What is the most likely cause of this clinical scenario above? (2)

b) How would you verify this? (4)

c) In point form, highlight the embryology of the urogenital system from bipotential gonad to internal and external female genitalia. (4)

[10]

6 A 10-year-old boy presents with generalised lethargy.
The paediatrician has done the following investigations:

Sodium	124 mmol/L	136 – 145
Potassium	4.8 mmol/L	3.5 – 5.1
Urea	7.0 mmol/L	2.1 – 7.1
TSH	2.65 mIU/L	0.48 – 4.67
Hb	13.9 g/dL	11.8 – 14.6
WCC	5.95 x 10^9/L	3.92 – 10.40
PLT	277 x 10^9/L	180 – 440

He has treated the boy with a normal saline infusion for 24 hours and now calls to ask you if this could be Addison's disease.

a) Which history and physical findings would you enquire about? Explain their significance. (5)

b) How would you monitor treatment? (5)

[10]

7 a) How would you screen for CFRD? (2)

b) Discuss dietary therapy in patients with CFRD. (4)

c) Discuss the basics of insulin therapy in CFRD. (4)

[10]

8 A 13-year-4-month-old girl presents with a 3-week history of blurred vision, chronic headaches, and deterioration in schoolwork. Clinically, she is not syndromic, but is hypertensive (BP = 145/98) and is noted to have a squint (esotropic) and is unable to move her right eye laterally to the right side. She has globally increased tone and brisk reflexes (3/4). Her Tanner Staging for breast development and pubic hair is Stage I and her stature plots on the – 1 z score, with normal weight and HC for age.
As part of her investigations, CT imaging of the brain is done which reveals:
"A suprasella mass which is cystic in nature, with impingement on the optic chiasm and features of mild subacute hydrocephalus".
Patient is further cared for by the neurosurgeons who take the patient to theatre. Findings include:
"A cystic mass (measuring 15cm x 12 cm) with brownish oil-like fluid that was aspirated. An Omaya reservoir was inserted within the cyst. Mild hydrocephalus noted, not amenable to V-P shunt as yet. To monitor closely".

a) Based on the above, what is the likely diagnosis? (1)

Further Investigations:

Na	156 mmol/l	**Serum Osmolality**	406 mosm/kg	**8am cortisol**	82 nmol/l
K	4.5 mmol/l	**Urine Osmolality**	126 mosm/kg	**LH**	0.1 IU/l
CO2	17 mmol/l	**FT4**	6.2 pmol/l	**FSH**	0.2 IU/l
Urea	1.8 mmol/l	**TSH**	<0.01 Mu/l	**Estradiol**	16 pmol/l
Creat	29 umol/l	**Urine Output**	6 ml/kg/hr	**IGF1**	135µg/L

b) Interpret the above results. (5)

c) From an endocrine perspective, what management would you institute and in what order? (4)

[10]

9 A term neonate presents with hypoglycaemia at day 2 of life. There is no history of Diabetes Mellitus in the mom and the baby's birth weight was 3.8kg. There is a 2cm hepatomegaly and the rest of the examination is normal. The hypoglycaemia persists in spite of being placed on a 15 percent glucose solution and intravenous hydrocortisone. The critical sample done at the time of the hypoglycaemic episode is as follows:

Formal blood glucose	1.7 mmol/l	Lactate	1.0 mmol/l
Insulin	12 mU/l	Pyruvate	Rejected
C-peptide	5 ng/ml		
Growth Hormone	25 ng/ml		
Cortisol	550 nmol/l		

a) Based on the clinical scenario and above results, what is the most likely cause for the hypoglycaemia? (1)
b) Explain your answer. (3)
c) What are the possible genetic causes? (3)
d) On day 7 of life, the baby has a seizure and is noted to be hypoglycaemic at the time. Elaborate on the emergency therapy at this point. (3)

[10]

10 A 15-year-old adolescent with Thalassaemia Major is referred to the endocrine clinic. She has had multiple blood transfusions since childhood. Her height is -3SD and she is pre-pubertal. Which associated endocrine complications would you look for in this patient? [10]