

DCH: Paper 3

Question 1

Jeannette, an 5 week old baby, is brought to a primary health clinic by her mother because of a high fever, for about six hours, and excessive crying. The infant's temperature is measured at 38°C. Mom says that Jeannette is still breastfeeding but not sucking as well as she usually does. She has not vomited.

- a) What additional IMCI danger signs should the clinic nurse enquire about? (2)

Convulsions

Lethargy/ decreased level of consciousness

Following a full assessment, the clinic nurse refers Jeannette to the hospital. She provides her with a dose of paracetamol before she leaves the clinic.

Jeannette arrives at your hospital an hour later. You are the doctor on call and examine her. You find her to be irritable but she seems to settle when comforted by her mother. Her temperature now is 37.1°C. You are unable to find any obvious source for the fever on examination. However, her fontanelle feels full, although there is no obvious neck stiffness.

- b) How would you manage this infant? (6)

<3 months of age and no obvious source of fever. Fever responding to antipyretics not indicative of cause of fever.

Children < 3months of age may not have localized source of infection.

Needs full septic workup.

Sterile Urine dipstick and MC&S (1)

Blood culture (1)

LP and Blood glucose (1)

CXR if tachypnoeic (1/2)

FBC and diff (1)

CRP if available (1/2)

Admit (1)

Jeannette is admitted to the ward. She is started on intravenous ceftriaxone 50mg/kg.

- c) Is this an appropriate antibiotic choice for the infant. Motivate your answer. (3)

No, dose too low for meningitis (1)

Should add ampicillin to cover for listeria 50mg/kg 6hrly (2)

An hour later, the laboratory phones the ward with a lumbar puncture result. It shows: polymorphs 600 units, lymphocytes 50 units, protein 0.8 g/L, glucose 0.2 mmol/l

d) Interpret the CSF result. (2)

Raised WCC with poly predominance (1/2)
Raised Protein (1/2)
Low Glucose (1/2)
Indicative of Bacterial meningitis (1/2)

e) List three organisms that are likely to be responsible for this infection. (3)

Group B Strp
Listeria
E coli
HiB and Pneumococc: shold have been immunized.

Gram positive cocci are seen on direct stain of the CSF (although after 72 hours the culture is negative).

You attempt to explain the diagnosis to the mother. She asks if there is anything she could have done to prevent the infection.

f) What do you tell her? (3)

Maternal screening and treatment of Group B strep prevents early onset disease, but does not prevent late onset disease. (1)
However it would be advisable to be screened during the next pregnancy and receive antibiotics if positive at the time of delivery (2)

g) With the additional information, would you modify your antibiotic treatment? Explain. (1)

Can use monotherapy, either ampicillin or ceftriaxone at meningitis doses (1)

h) How long would you continue the antibiotic(s) for? (1)

21 days (1)

A few hours later the nursing sister in the ward calls you because Jeannette is fitting.

i) Outline the steps you would take to manage this new problem. (6)

ABC (1)
Give oxygen (1)
Put in recovery position (1)
Stop the fit: valium I.V.I, lorazepam I.V.I. Buccal midazolam (1)
Check the glucose (1)
Load with phenobarbitone 20mg/kg (1)

You are able to stop the seizure. However, the infant remains sleepy. You manage to get a CT scan which shows extensive bilateral infarcts.

- j) What would you tell (counsel) the mother, based on this new information? (4)

Extensive damage to the brain as a result of the meningitis.

Able to treat the infection but unable to reverse the brain damage(1)

Prognosis is guarded (1)

Unlikely develop like other children/ will have delayed milestones (1)

If survives will need ongoing medical input and support in order to reach full potential(1)

Other areas that may be mentioned are concerns re vision and hearing, risk of repeated seizures and long term treatment

Jeannette's mother remains at the bedside overnight and is actively involved in the infant's care. She is 38 years old; this is her first baby. She has struggled to conceive.

The next day, Jeannette has a prolonged apnoea which responds to bag and mask ventilation. However, she has frequent, repeat apnoeic episodes during the next few hours.

- k) Would you motivate for Jeannette to be ventilated in ICU? Explain your answer. (3)

There are two views on this argument and answers should be well motivated.

Firstly: NO: There are limited resources in South Africa, especially for ICU/ventilation space. There is a guarded prognosis as the child has extensive brain damage and likely will have severe developmental delay. Whilst there may be other children with a much better prognosis turned down.

However: YES: This is an elderly mother who may not have further children since she has struggled to conceive ie a precious baby. She has shown herself to be a caring and involved mother. This is early in disease process and there may be some improvement. It may allow her time to come to terms with the process

You are able to successfully treat the infection, however her neurological condition remains concerning. She takes feeds slowly and you notice increased tone in all four limbs with clonus in the lower limbs.

- l) Outline a management plan for Jeannette on discharge. (6)

This child has evidence of permanent neurological scarring, she will need a multidisciplinary team approach on follow up. And preferably an identified primary doctor who can co ordinate care and follow up. Since this child will need ongoing care a good relationship between a primary doctor and care giver is ideal (1) She will need regular follow up and developmental assessment especially in her first year of life and school placement in the future(1/2)

At one year she should be assessed for a care dependency grant if her mother qualifies.(1/2)

Vision and hearing should be assessed. (1)

Physiotherapy to prevent contractures and to promote development. Mother could be taught a home programme.(1)

Ongoing assessment of feeding and growth. May need thickened feeds in poor swallowing/
pseudobulbar palsy (1)

She is at high risk of ongoing seizure and should be started on maintenance therapy (phenobarb vs
epilem) (1)

Other disciplines/ complications that may be mentioned

Occupational therapy if available. Especially as gets older for splints etc

Orthopaedics for strictures// dislocated hips

Prevention of constipation

Question 2

Bapendile is a 12 year old girl referred to you from a local clinic with a heart murmur and tachycardia. You are the attending medical officer at the hospital. Two years earlier (in 2009) she was diagnosed as having Sydenham's chorea. Her heart was normal at the time.

a) Define chorea. (2)

- *The term "chorea" is derived from the Greek word "choreia" for dancing (as is choreography). Chorea is the occurrence of continuous rapid, jerky, involuntary, irregular, and purposeless movements that may involve the face and limb and result in an inability to maintain a posture. They are brief, asymmetric, present at rest, and may persist during sleep. Children may attempt to hide the movements with quasi-purposeful actions (such as flinging hair back), or they may sit on their hands as an attempt to prevent these movements.*

b) Which part of the brain is affected in chorea?

- *Basal ganglia*

c) List one of the three cardinal signs of Sydenham's chorea (other than chorea). (1)

- *Milk maids grip; quasi-purposeful actions (such as flinging hair back); the pronator sign consists of hyperpronation of the hands, causing the palms to face outward when the arms are held over the head.*

d) What was the most likely cause of the Sydenham's chorea two years ago? (1)

- *Acute rheumatic fever*

e) What treatment should have been prescribed then (in 2009) and why? (4)

- *Haloperidol: control the chorea*
- *Continuous penicillin (monthly intramuscularly or oral daily): prevent acute rheumatic fever recurrence*

f) For how long should this treatment have been given and why? (4)

- *Haloperidol: until chorea resolved. Often needed for months (or years)*

- *Penicillin: until the age of 21 because there was no cardiac involvement*
- g) Suggest a good and sensitive marker for monitoring the response of Sydenham's chorea to treatment. (1)
- *Handwriting: simply writing her own name in her clinical record over time will be a good indicator of chorea control.*

On examining Bapendile, you identify a tachycardia, cardiomegaly, hepatomegaly, a 3/6 pansystolic murmur at the apex radiating to the left axilla, and a low pitched 1/4 diastolic murmur.

- h) What cardiac lesion(s) does she have? (1)
- *Mitral incompetence*
- i) How severe is the cardiac lesion? Explain. (4)
- *The low pitched diastolic murmur is heard when there is substantial flow of excessive blood back into the left ventricle in diastole, which has resulted from moderate to severe regurgitation. This has also resulted in heart failure as evidenced by the tachycardia, cardiomegaly and hepatomegaly.*
- j) What treatment should you start during this acute illness, and for how long? (4)
- *Intravenous penicillin for 10 days.*
 - *Antifailure treatment should be initiated and titrated to severity of lesion and response.*

Bapendile's mother died following this admission. She therefore moves in with an aunt who has no knowledge of Bapendile's medical history. Bapendile appears quite well to her aunt, so her treatment is discontinued.

Seven months later, she arrives at the hospital complaining of fever and joint pain.

- k) What is the likely diagnosis now? (1)
- *Acute rheumatic fever recurrence*
- l) Why has this illness occurred? (2)
- *Interruption of penicillin prophylaxis for acute rheumatic fever*
- m) What will you do to ensure that this illness does not recur? (5)
- *Once an episode of acute rheumatic fever has occurred, the most important intervention is to ensure proper monthly follow up for penicillin injections, to prevent recurrence. A patient held letter (or equivalent) documenting receipt of, and due dates for penicillin is essential. It is critical to make sure that*

the patient and caregiver understand its use and the importance of never getting acute rheumatic fever again. All patients with rheumatic heart disease, and Sydenham's Chorea, must be followed up in a cardiology service.

Level 1: for monthly penicillin, and other treatment

Level 2: for cardiac assessment, 6 monthly to annually

Level 3 if: for long term planning at disease onset, and if heart valve replacement is a possibility

Bapendile is about to turn 13.

- n) Outline important issues that you will have to deal with in managing her now (as an adolescent patient). (4)
- *During the transition from childhood to adulthood, adolescents and young adults experience pivotal biological, cognitive, emotional, and social changes. In addition, they establish patterns of behaviours and make lifestyle choices that affect both their current and future health.*
 - *Of importance in a child with rheumatic heart disease:*
 - *Encourage increasing autonomy and responsibility for taking medication and keeping appointments, understanding that this is a time of risk taking and rebellion*
 - *Discuss sexuality and reproductive health (including HIV), emphasizing the need for planned parenthood (and the danger of pregnancy)*
 - *Discuss nutrition and diet, and healthy lifestyle issues (such as alcohol and tobacco abuse)*
 - *Discuss schooling and physical exercise and body image*

Following discharge, and a period of relative stability with good penicillin adherence, Bapendile's condition deteriorates acutely once again.

- o) What are the possible causes? (5)
- *Infective endocarditis*
 - *Precipitous myocardial failure*
 - *irreversible – apoptosis,*
 - *reversible – treatment problem*
 - *Valve stenosis, especially mitral*
 - *A non-cardiac condition (e.g. severe ARI, APSGN, pregnancy)*

Question 3

You are the medical officer assigned to paediatrics in a district hospital. Your obstetric colleague has performed an antenatal ultrasound examination at 37 weeks gestation on an unbooked, primiparous, 28 year-old woman. The sonar noted polyhydramnios, breech presentation and an open thoraco-lumbar spine. He consults you to assist in the diagnosis and management.

- a) The mother-to-be asks for information on the aetiology of the condition. What advice can you offer in this respect?

The etiology of meningo-myelocele is thought to represent a classic example of multifactorial inheritance with environmental factors acting in concert with a polygenic genetic factors. The most important *environmental* hit is folate deficiency but other environmental associations include lower SES, younger and older mothers, hyperthermia in pregnancy and others. The genetic component is suggested by marked variation in incidence in different race groups and from many experimental animal models. The details of the specific candidate genes in humans remain complex.

(5)

- b) What advice would you offer your colleague regarding the mode of delivery? (1)

Most experts recommend the delivery of fetuses with meningomyelocele should be by caesarean section prior to- or early in labour. This is especially true in breech (common with spinal defects) and the presence of co-existing hydrocephalus

You are present at the delivery one week later. The baby weighs 2800 grams. The Apgar scores are 5, 7 & 8 at 1, 5 and 10 minutes respectively You note the following on examination of the baby: Term female Caucasian infant. Large ruptured cystic meningo-myelocele over lower thoracic and lumbar spine. Cerebro-spinal fluid is leaking. Skull circumference is 39 cm (>95th centile). There is moderate kypho-scoliosis present. A large midline abdominal mass is palpated, arising in the pelvis. This is dull to percussion. There is bilateral talipes equino-varus (club feet).

- c) What is the likely cause for the macrocephaly and discuss the pathogenesis of the macrocephaly in this infant?

The most likely cause for the large head is associated hydrocephalus. Almost all patients with MMC also have the Arnold–Chiari II malformation, characterized by descent of the cerebellar vermis through the foramen magnum, elongation and kinking of the medulla, caudal displacement of the cervical spinal cord and medulla, and obliteration of the cisterna magna. The Arnold – Chiari malformation is sequential due to linear growth of the spine in the face of tethering of the lumbar cord.

(5)

- d) What is the likely cause for the pelvic mass and how can you explain its pathogenesis?

It is most likely that the mass arising from the pelvis is a distended urinary bladder. Disordered innervation of the detrusor musculature and external sphincter in patients with MM adversely affects bladder function resulting in urinary retention.

(5)

- e) Discuss the neurological findings expected in the lower limbs. How do you explain the presence of the club feet? There is usually complete loss neural transmission across the lesion in open meningomyelocele. This results in poor neurological function of the lower limbs with areflexia, complete paralysis and sensory loss. Occasionally there is preservation of a distal spinal arch with reflexes present but there is loss of afferent pathways to the brain.

The talipes is explained by deformation in utero of the denervated lower limbs (5)

You refer the infant to a tertiary centre where neurosurgery is performed. You next see her at the six-week visit.

- f) What advice would you give to the parents regarding the likely long-term problems facing this child during infancy, childhood and adolescence?

The long-term problems in severe meningomyelocele are a multitude. In *infancy*, the main problems include lack of mobility with delayed motor milestones, urinary incontinence and UTIs, hydrocephalus and often delayed intellectual milestones. During *childhood* there continue to be serious mobility problems. On-going urinary problems result from the balance between striving for a capacitance bladder and urinary infections. Impaired renal function may become apparent. Intellectual challenge may be present as may behavioural disorders. Blocked VP Shunts are common. In *adolescence* there may be deterioration of renal function. Psychological problems due to body image perceptions, incontinence etc are common. Trophic ulcers may occur.

(10)

- g) What advice can you give the parents for primary, secondary and tertiary prevention regarding recurrence in any future pregnancy?

In future pregnancies;

Primary Prevention involves family planning and the administration of folate prior to, and during future pregnancies. Other risk factors (eg diabetes) may be amenable to intervention

Secondary Prevention involves screening for the defect (serum alpha feto protein, amniocentesis, ultrasound) and if requested after counselling, termination of an affected infant.

Tertiary Prevention is the term applied to early diagnosis and management of an affected child (or fetus) so as to optimize health and minimize complications (9)

Question 4

Mpho, a 7 month old boy, is brought to the primary health care clinic with a 2 day history of diarrhoea. The nurse at the clinic assesses him. He weighs 6 kg. She finds that he is lethargic, has sunken eyes and a very slow skin pinch. .

a) What is the IMCI classification for his dehydration? Explain. (2)

SEVERE DEHYDRATION

She puts up a drip and administers intravenous fluid according to plan C in IMCI.

b) What fluid and how much will she administer as a bolus? (2)

Normal Saline, 20ml/kg iv in the first half hour i.e 6 x 20ml = 120ml

After initiation of treatment for dehydration she asks the mother to take him to hospital. There is a 12 hour delay.

He arrives at the district hospital at 10 pm in the evening. You, the attending doctor, find that Mpho has a weak pulse, a heart rate of 170 beats/minute, sunken eyes and fontanelle, and a very slow skin pinch.

c) What is your immediate management? (4)

Child is in shock and has severe dehydration (> 10%)

Insert an iv line(if not possible an intraosseous line)

Administer a bolus of ivi fluids at 20ml/kg, (Normal Saline or Ringers lactate) over 10 – 20 minutes, and reassess pulse, circulation and capillary filling time afterwards, if child is still in shock repeat the bolus.

At the same time rapidly check

Airway

Breathing – administer oxygen as circulation is poor

Blood glucose with a disptix

d) What treatment will you prescribe when Mpho is sent to the paediatric ward? (4)

If still in shock, assess continuously and refer to ICU if shock does not resolve after 2nd bolus

If shock has resolved and child is passing urine then give 10 - 20ml/kg ivi of normal saline or ringers lactate for the next few hours (1 – 4 hours), and then reassess hydration in 4 hours. (guidelines vary, as long as participant displays evidence of consistent and safe practice) Ringers lactate can be replaced with ½ DD if the child is no longer in shock.

Monitor the pulse, RR, CFT, hydration regularly while shocked, hourly while severely dehydrated and thereafter 4 hourly.

Check electrolytes if this service is available

Record intake and output

If child is able to drink give sips of oral ORS 5ml/kg

The next morning, you review Mpho's condition. He is irritable, has a good pulse, and is thirsty. Mom notes that his eyes are less sunken. He still has a slow skin pinch.

e) Discuss the management of Mpho's dehydration and diarrhoea for that day. (4)

Mpho now has diarrhoea with some dehydration (around 5%). He is much better as he is thirsty and presumably able to drink.

Continue ivi fluids if the drip is still up, you can switch to ivi ½ dd, determine the amount of fluid to be given every 4 hours. Give 10 – 20ml/kg extra every 4 hours i.e 6 x 20 = 120ml/kg for next 4 hours. After 4 hours reassess, if improving Slow down drip, or increase drip if not improving. If child is drinking well this fluid can be given orally as ORS i.e. 480ml or ORS for the next 4 hours, and then reassess every 4 hours.

Monitor intake and output. If Mpho has diarrhoea give ~10ml/kg of ORS to drink with each loose stool

Restart feeds. If he is breastfed then breastfeed. If on formula feed give formula for his age. Give soft food as well

Mpho's weighs 6.4 kg (weight for age z score: -2.1), his height is 68 cm (length for age: -1 z score and weight for height: below -2 z score).

He looks a little pale, has generalised lymphadenopathy, but no hepatosplenomegaly. Other systems are normal. Mpho is getting formula milk as he is HIV exposed. Mom received AZT from 38 weeks.

f) Interpret (classify) Mpho's growth parameters. (2)

His weight for age is -2.1 z scores which means that he is underweight for age. His length for age is at -1z score is normal and weight for height below -2 z scores shows that he is wasted.

g) Explain the value of using z scores to classify anthropometric status. (2)

Z scores is based on the normal curve and describe the no of Standard deviations from the median. They are more sensitive than percentil changes and are useful in cases that lie outside the percentiles.

Mpho has had the following immunisations BCG and OPV0 at birth; OPV1, DTaP-IPV-Hib1 and HepB 1 at 6 weeks; and DTaP-IPV-Hib2 and Hep 2 at 10 weeks.

- e) What immunisations has he missed, what immunisations should he get now and before being discharged home. (4)

He missed Rotavirus at 6 and 14 weeks, and Pneumococcal conjugate vaccine at 6 and 14 weeks, as well as DtaP-IPV/Hib at 14 weeks.

He should be given measles immunisation immediately as this should be given to all children 6 months and older admitted to hospital.

On or before discharge he should be given DtaP-IPV/Hib (3) and Pneumococcal conjugate vaccine (1).

He should not be given Rotavirus vaccine as this should not be administered to children older than 6 months of age or 24 weeks.

A HIV DNA PCR test was done at 6 weeks with the first immunisation, but the result was not there at 10 weeks. You repeat the HIV DNA PCR test.

- e) The test comes back positive. Briefly outline the management of Mpho's HIV infection. (6)

Mpho is HIV infected

Counsel the mother about HIV and the need to start ARV therapy.

Provide ongoing HIV adherence counselling while she is in the ward, this includes

- *What HIV is and what HIV disease does in children*
- *Why he needs ARV treatment and that this will be lifelong*
- *How he will have to take the HIV treatment*
- *Demonstrate the doses and how to take and get her to practise*
- *Discuss storage and possible side effects that are rare*
- *Discuss disclosure in the family and who will help administer treatment to Mpho*

Do a confirmatory HIV test – in this case a viral load test and a baseline CD4 count and %
Start on cotrimoxazole prophylaxis

Start on ARV treatment – according to the national guideline (Abacavir, 3TC and Kaletra)

Ensure follow up at a Paediatric, HIV clinic convenient for the mother / caretaker

Document treatment given, and ensure that there is follow up confirming the child and caretakers attendance

The ward sister reminds you that Mpho must get zinc.

- f) Explain why Mpho needs zinc and the dose/duration to be prescribed? (4)

Zinc has been shown to decrease the duration of diarrhoea by 25%, the proportion of episodes lasting longer than 7 days, and reduce the stool volume by 30%. It may also prevent future episodes of diarrhoea by up to 3 months.

He should receive 10mg of elemental Zinc daily(or 2mg/kg/day) for 2 weeks

You review the admissions and deaths in your paediatric ward for the past year. You have had 720 children admitted to the ward; 200 were admitted with diarrhoeal disease and of these 30 died. You are concerned about the case fatality rate for diarrhoea.

j) Discuss the steps you put in place to decrease the case fatality rate for diarrhoeal disease in your hospital.

(6)

The case fatality rate is 15% and you would like to reduce it to 5% and even <1 %.

You decide on the following interventions

- *Update all doctors and nurses on the IMCI and EDL guidelines for the management of diarrhoea in children*
- *Based on the above guideline draw up a protocol for the management of diarrhoea in children in your hospital, including the initial management in casualty and in the paediatric ward. Be sure to include*
 - *Reassess children every 4 hours – when the nurses can do this and when the doctor should be called. What the nurses can prescribe and what the doctors are able to prescribe*
 - *What to do if the child cannot be dripped*
 - *Ensure that the correct fluids and feeds are available*
- *Review the deaths of all children the next morning in the doctors meeting*
- *Take steps to correct any problems in the management*
- *Ensure that all new staff are orientated on the management.*
- *Ensure that there are forms to record the hydration checks*
- *Conduct clinical audit, to review the management of children with diarrhoea in the ward.*
- *Meet with PHC nurses and traditional leaders to discuss the management of diarrhoea and early referral*
- *Review the HIV and infant feeding counselling in your area, if you suspect that patients not meeting AFASS criteria are being counselled to formula feed.*

Question 5:

5 day-old Onako is brought to your hospital. She was born at term weighing 2.2 kg. She appeared quite well on discharge from the Midwife Obstetric Unit (MOU).

- a) List THREE clinical problems/complications that babies with a similar birth weight are at risk for. (3)

Accept any 3:

Hypoglycaemia

Hypothermia

Infection

Nutritional-intolerance, NEC, poor weight gain

Anaemia

hyperbilirubinaemia

Respiratory (RDS) if premature

Onako's mother reports that all was well initially. However, since this morning Onako is refusing to breastfeed and has difficulty breathing. She does not have a fever.

Onako is pale and perfusing poorly. Her respiratory rate is 70 breaths/minute with subcostal recession. Her heart rate is 180 beats/min and her peripheral pulses are difficult to feel. Her extremities are cold and have a bluish discolouration. Capillary refill time is >5 seconds.

- b) How does IMCI define the upper limit of normal respiratory rate at this age? (1)

<60 breaths per minute (infant less than 2 months)

- c) What is your interpretation of the clinical problem? (3)

Infant has CCF and is shocked –poor perfusion, tachycardia, raised cap refill time with respiratory distress –tachypnoea and recession and needs urgent management

Onako is irritable on handling, she has a gallop rhythm and a 3/6 systolic murmur over the left praecordium. Her chest sounds clear on auscultation. Her liver is felt 5 cm below the costal margin. The 4 limb blood pressures in mm Hg are as follows: right arm: 95/40, left arm: 96/ 38; right lower limb: 45/30; left lower limb: 45/36.
The chest X-ray shows a cardiothoracic ratio of 65%.

- d) Considering this additional clinical information, what is the most likely diagnosis? Explain. (2)

(Duct dependent) Coarctation of the aorta- the upper limb BPs are higher than lower limbs-being perfused; murmur may be from flow through the constricted area; both ventricles are not coping(CCF)

e) How would you explain the blood pressure findings? (2)

-The blood flow to the upper limbs is being supplied by the preductal aorta, the blood flow to the lower limbs is severely compromised following closure of the duct
-Relative upper limb hypertension may also be related to renin release due to renal and lower body hypoperfusion

f) Considering cardiovascular haemodynamics, explain why Onako's condition presented on day 5 of life rather than at birth or soon thereafter. (2)

Duct still open in the first few days of life there was enough blood flow to the lower half of the body including the kidneys via the ductus; once duct closes severe aortic constriction and overload and failure of the Left ventricle with backward load on the R ventricle and severely reduced blood flow to the kidneys, liver and lower half of the body.

g) IGNORING the specific cardiac findings, the clinical presentation of this baby may have been the result of other serious newborn problems. Suggest TWO non-cardiac differential diagnoses that could have been considered. (2)

ANY 2:

Acute respiratory distress-pneumonia/ diaphragmatic hernia

Sepsis/septicaemia with shock

CNS:meningitis/seizures(subclinical/subtle)

Hypoglycaemia

Inborn error of metabolism

Hypothermia

h) Outline your initial immediate management. (3)

-Triage as needing immediate care- take to the emergency room
-A & B-give high flow O2 via face mask or provide ventilator support if distress severe
-C-Circulation- Establish IV access, ideally no large fluid boluses (CCF). Prostin to reopen duct- with airway support as indicated; inotropes, IV frusemide to reduce preload

i) What immediate blood test is required and why? (2)

Bedside glucose measurement as hypoglycaemia may present in this way, needs immediate management to prevent complications

Other blood tests are sent off and the results are telephoned through from the laboratory.

Blood gas: pH 7.16, pCO₂ 5.5 kPa (41 mm Hg), base excess -16 mmol/l, standard bicarbonate 11 mmol/l

Liver enzymes: ALT 160 IU/l, AST 80 IU/l

j) Interpret these results and explain the likely aetiology of the liver enzyme changes in the context of your most likely diagnosis. (3)

Severe metabolic acidosis
Normal PCO₂- uncompensated
Evidence of poor perfusion of the liver with hepatocellular damage

Serum electrolytes: Sodium 124 mmol/l; Potassium 7.9 mmol/l; Urea 13 mmol/l; Creatinine 140 µmol/l

- k) Interpret these results and explain the likely aetiology. (4)

Hyponatraemia
Hyperkalaemia
Azotaemia /Renal dysfunction.
All evidence of poor renal perfusion and failure

- l) What steps would you take to manage the serum potassium? (3)

Ensure specimen was taken without haemolysis, stat repeat if not
Stop any potassium containing fluids/medication
Establish IV line- Give IV calcium gluconate
Salbutamol by nebulisation fast and easy to set up
Sodabic bolus
10% glucose bolus (potassium free)
Kexelate rectally
Check levels hourly if renal output is established- restoring this is key; if not may need dialysis
Encourage urine output- difficult to give fluids as CCF therefore give IV lasix

- m) List your next management steps. (3)

H-get help/transfer to appropriate level ICU urgently- will need cardiology review with ECHO and surgery/balloon angioplasty for relief of the obstruction
Monitor urine output and renal function; and blood gases and BP
Exclude associated abnormalities
Manage hypertension
Attend to enteral nutrition as soon as possible
Arrange follow up

On more detailed examination Onako is found to have a webbed neck, non pitting oedema of both feet and widely spaced nipples.

- n) What syndrome do you suspect? What would the chromosomal results show if your suspicion is correct? What is the inheritance pattern of this condition? (3)

Turner Syndrome
45 XO Karyotype
Chromosomal disorder therefore no genetic inheritance pattern

- o) Outline some key facts that you would offer when counselling the parents in terms of this syndrome.
(4)

Lifelong condition

Girls only disorder- will need blood tests for chromosomal analysis

Infant will grow up and generally be well but will need long term medical follow up

Mental retardation not expected

Short stature prominent

Other problems to be monitored for, prevention and treatment of associated problems

Treatment options for complications

Infertility therefore not passed on