

Gastroenterology

A 7 year old girl presents with crampy lower abdominal pain, loose stools and anorexia of 3 days duration. The stools contain blood and slime and occur 20 times a day. There is no history of foreign travel. She was born at 38+4/40 weighing 3.2kg and there were no neonatal problems.

On examination she appears lethargic and pale. She has dry mucous membranes but no jaundice. Her temperature is 37.6°C (tympanic), HR 100/min, RR 15/min. She has a few purpuric spots on her legs.

What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Marrow failure [0]
- 2) Haemaglobinopathies eg Sickle, Thalassaemia [0]
- 3) Haemolytic uraemic syndrome [100]
- 4) Iron deficiency [100]
- 5) Malignancy [0]

Comments:

The history suggests a dysenteric illness followed by haemolytic uraemic syndrome. This is now the commonest cause of acquired acute renal failure in childhood. The likely organism is *E. coli* 0157:H7 in the UK, although *Shigella*, *Campylobacter* and a variety of viruses have also been described. These trigger the triad of microangiopathic haemolytic anaemia, thrombocytopenia and acute renal failure. A FBC showed Hb 6.8 g/dl, WC 16.8×10^9 , Platelets of 36. Blood film showed schistocytes, spherocytes, helmet cells and polychromasia. Careful attention to fluid balance is essential to avoid fluid and electrolyte-related complications.

A neonate does not pass meconium for 48 hrs after birth. A fortnight later his mother states that he is not passing stool regularly. He has been bottle fed since discharge. Investigations reveal massive dilatation of the colon proximal to the rectum and manometry shows increased internal anal pressure on rectal distension with a balloon. Where is the developmental abnormality responsible for this child's presentation?

Available marks are shown in brackets

- 1) Ectoderm [0]
- 2) Endoderm [0]
- 3) Neural crest [100]
- 4) Neural ectoderm [0]
- 5) Splanchnic mesoderm [0]

Comments:

This is Hirschsprung's disease which is due to absence of the ganglion cells supplying the colonic smooth muscle. Neural crest contribute to the formation of the postganglionic neurones of the autonomic nervous system and the sensory neurones of the peripheral nervous system.

At 2 months of age a boy is referred with abdominal mass. This has been found at the routine 6 week surveillance review. He was born at term weighing 3.7 kg, and the parents were first cousins.

On examination he has low-set ears, frontal bossing, a depressed nasal bridge and long philtrum. He has 4 cm hepatomegaly and 3 cm splenomegaly. He has poor head control. Although he smiled at 6 weeks he has not made further developmental progress.

What is the most likely diagnosis?

Available marks are shown in brackets

- | | |
|---|-------|
| 1) Inborn errors of metabolism | [100] |
| 2) Constipation/encopresis | [0] |
| 3) Pregnancy | [0] |
| 4) Malignancy eg Neuroblastoma, Wilm's, Lymphoma | [0] |
| 5) Renal anomalies | [0] |

Comments:

The consanguinity, dysmorphic features, hepatosplenomegaly and developmental arrest suggest a storage disease. These include lipidoses, mucopolysaccharidoses and glycogenoses.

In this case the features are most characteristic of infantile GM₁ Gangliosidosis, 50% of whom have a macular cherry red spot.

Which of the following statements applies to infants with gastroenteritis:

Available marks are shown in brackets

- 1) In most instances require treatment with an antibiotic which is not absorbed from the gut [0]
- 2) May have blood in their stools [0]
- 3) Should be admitted to hospital if they are unable to tolerate fluid orally [100]
- 4) Always develop lactose intolerance [0]
- 5) Should have a barium meal if the initial refeeding with milk is unsuccessful [0]

Comments:

The majority of children do not require antibiotics as the cause is viral. Formula feeds should not be changed. Intravenous fluid therapy will be required if they are unable to tolerate oral fluids. Lactose intolerance is common, but not inevitable. Barium meals are not useful in the investigation of gastroenteritis

Which of the following statements regarding kernicterus is incorrect?

Available marks are shown in brackets

- 1) Usually occurs in the first month of life [0]
- 2) The diagnosis of kernicterus requires the histological confirmation of yellow staining of brain tissue on autopsy caused by fat soluble unconjugated hyperbilirubinaemia [100]
- 3) May cause a chronic syndrome of athetosis, gaze disturbance and hearing loss [0]
- 4) Is associated with Gastrointestinal haemorrhage [0]
- 5) May cause convulsions [0]

Comments:

Intracellular crystal is noted in intestinal mucosa of affected infants. This can be related to GI haemorrhage. the chronic syndrome of neurologic sequelae observed following marked hyperbilirubinemia includes athetosis, gaze disturbance, and hearing loss. If the affected infants survives the neonatal period and subsequently dies, the yellow staining of neural tissue may no longer be present, but the basal ganglia will display microscopic evidence of cell injury, neuronal loss, and glial replacement.

A 16 year old boy with cystic fibrosis presents with abdominal pain.

Which of the following is most likely to be the cause?

Available marks are shown in brackets

- 1) Ulcerative colitis [0]
- 2) Irritable Bowel Syndrome [0]
- 3) Pyelonephritis [0]
- 4) Meconium Ileus Equivalent Syndrome [100]
- 5) Renal Calculi [0]

Comments:

Meconium ileus equivalent or distal intestinal obstruction syndrome occurs in older children and adults with CF and presents with colicky abdominal pain, distension, vomiting and failure to pass faeces. The plain AXR confirms small bowel obstruction. Initial management includes rehydration with IV fluids and oral N- acetyl cysteine. Other GI complications of / associations with CF include liver cirrhosis, gall bladder disease, pancreatitis, peptic ulceration, hiatus hernia, coeliac disease and Crohns disease.

Which one of the following statements is true concerning subjects with untreated celiac disease?

Available marks are shown in brackets

- 1) They are likely to have normal jejunal villi [0]
- 2) Usually absorb glucose normally [0]
- 3) Usually have anorexia [100]
- 4) May safely resume a normal diet in adult life [0]
- 5) Characteristically have abdominal distension [0]

Comments:

They are likely to have subtotal or total villous atrophy. Glucose absorption and hydrolysis of lactose and sucrose is impaired in florid coeliac disease. Anorexia and weight loss are common.

The gluten free diet is required lifelong in this condition. Abdominal distension may develop, but it is not characteristic.

Which of the following statements is characteristic of acute hepatitis B infection?

Available marks are shown in brackets

- 1) Most patients present with splenomegaly. [0]
- 2) It confers immunity to hepatitis A. [0]
- 3) It commonly presents with distal joint arthritis. [0]
- 4) There is increased infectivity in the presence of the e antigen. [100]
- 5) Pruritis is an important early symptom. [0]

Comments:

Clinical features of hepatitis B are as follows:

1. Most are asymptomatic.
2. Symptoms: Lethargy, anorexia, arthralgia, rash (any type), papular acrodermatitis (Gianotti Crosti), polyarthritis, glomerulonephritis, aplastic anaemia. 25 % have jaundice.
3. Complications: · Acute fulminant hepatitis. · Chronic hepatitis. · Membranous glomerulonephritis. Hepatitis E antigen is present in the acute phase and indicates a highly infectious state. Pruritis is characteristic of chronic hepatitis.

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A 17-year-old male is admitted to casualty after a night out with friends at a nightclub. After drinking eight pints of beer, he felt nauseated and vomited. Vomitus was seen to contain bright red blood after vomiting a second time. Investigations:
Oesophago-gastro-duodenoscopy: Normal
What is the next step in his management?

Available marks are shown in brackets

- 1) Coeliac axis angiography [0]
- 2) Discharge the patient from hospital [100]
- 3) Prescribe a proton pump inhibitor and admit the patient overnight for observation [0]
- 4) Refer to the surgeons for review [0]
- 5) Discharge the patient and arrange a repeat endoscopy in one month [0]

Comments:

The history is compatible with a Mallory-Weiss tear. No additional treatment or follow-up is required and the patient can be discharged from hospital.

A male child weighing 4.2kg is admitted at age 4 weeks with a one week history of forceful vomiting after feeds. He is alert and eager for food but exhibits a loss of skin elasticity and has apparent distension in the left hypochondrium. Which of the following statements is correct for this patient?

Available marks are shown in brackets

- 1) The most likely diagnosis is hiatus hernia [0]
- 2) He has hyperosmolar dehydration. [0]
- 3) Metabolic acidosis would be anticipated. [0]
- 4) Initial fluid replacement should comprise normal saline 10-20 ml/kg in one hour [100]
- 5) He should be started on an anticholinergic drug, e.g. Atropine Methylnitrate (Eumydrin), before feeds [0]

Comments:

The most likely diagnosis is pyloric stenosis, which is more common in males (4:1 M:F ratio) and characteristically presents 3-6 weeks after birth. This situation will cause a proportionate loss of fluid and electrolytes. Characteristically he will have hypokalaemic, hypochloraemic metabolic

alkalosis. Initial resuscitation should be with normal saline, at twice the maintenance volume. This will ensure he is volume replete and prevent secondary activation of the renin/aldosterone system, which would cause more alkalosis and hypokalaemia. Once volume replete, the fluid can be changed to half strength saline with added potassium.

Although there are a few emerging reports showing satisfactory short term and long term outcome with medical treatment, the preferred treatment is currently pylorotomy. This should be performed after successful resuscitation and electrolyte correction.

Which of the following is true of Gilbert's syndrome?

Available marks are shown in brackets

- | | |
|---|-------|
| 1) inheritance is autosomal recessive | [0] |
| 2) serum conjugated bilirubin levels are elevated | [0] |
| 3) serum bilirubin levels are decreased by fasting | [0] |
| 4) serum bilirubin levels are decreased by liver enzyme inducers | [100] |
| 5) there is bilirubinuria | [0] |

Comments:

Gilbert's syndrome is inherited in autosomal dominant fashion and affects 2-5% of the population. UDP glucuronyl transferase levels are reduced leading to an unconjugated hyperbilirubinaemia. Whilst serum bilirubin levels are elevated the other LFTs are normal. Jaundice deepens after a period of fasting or intercurrent illness but bilirubin levels are reduced by enzyme inducers such as phenobarbitone. As unconjugated bilirubin is tightly bound to albumin it cannot cross the glomerulus and so is not found in the urine. This contrasts with the bilirubin-glucuronide-albumin complex formed in patients with cholestatic jaundice (and raised conjugated bilirubin levels) where 1% of the complex is dialysable and although most of the bilirubin is reabsorbed in the proximal tubule some bilirubin is detectable in the urine.

Which of the following statements correctly applies to a baby of 2 months who is considered to be 10% clinically dehydrated with gastroenteritis:

Available marks are shown in brackets

- 1) He may have blood and mucus in his stools [100]
- 2) He will have a full fontanelle [0]
- 3) He will have a low packed cell volume [0]
- 4) Urea level will be normal [0]
- 5) Should not be given milk orally for at least 48 hours [0]

Comments:

Blood and mucus may be seen with dysentery and *E.coli* infection. The fontanelle will be sunken and haematocrit will be raised. An increased urea will be seen in this degree of dehydration. Oral feeding should be continued whenever possible.

Which of the following conditions may give a false/positive sweat test?

Available marks are shown in brackets

- 1) Non-classic Congenital adrenal hyperplasia [0]
- 2) Hyperthyroidism [0]
- 3) Hyperparathyroidism [0]
- 4) Obesity [0]
- 5) Glucose-6-phosphatase deficiency [100]

Comments:

Non-cystic fibrosis conditions associated with elevated concentrations of sweat electrolytes include:

- Endocrine: Untreated adrenal insufficiency, hereditary nephrogenic diabetes insipidus, hypothyroidism, hypoparathyroidism.
- Metabolic: Glucose-6-phosphatase deficiency, mucopolysaccharidoses, fucosidosis.
- Other: Ectodermal dysplasia, familial cholestasis, pancreatitis, malnutrition.

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A 17 year old male student attends clinic complaining of right knee pain and left ankle pain. These pains have deteriorated when associated with episodic diarrhoea. What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Amoebic dysentery [0]
- 2) Coeliac disease [0]
- 3) Chronic appendicitis [0]
- 4) Ulcerative colitis [100]
- 5) Whipple's disease [0]

Comments:

This patient has oligoarthropathy associated with episodic diarrhoea which suggests a diagnosis of inflammatory bowel disease.

A 5 year old girl presents with pallor and lethargy. This has been worsening over the last few weeks. Apart from occasional abdominal pain with dark stools from time to time, she has otherwise been well. She was born at 40+5/40 weighing 3.5kg and there were no neonatal problems.

On examination she has pale conjunctivae, but looks generally well. Her height and weight are on the 25%. Temperature is 36.5°C, HR is 95/min and RR 15/min. She has a 2/6 ejection murmur at the left sternal edge. Abdominal examination is unremarkable.

What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Marrow failure [0]
- 2) Iron deficiency [0]
- 3) Occult blood loss [100]
- 4) Haemaglobinopathies eg Sickle, Thalassaemia [0]
- 5) Malignancy [0]

Comments:

The history suggests isolated anaemia, progressing over several months. The dark stools and abdominal pain suggest an occult source of bleeding from the upper GI tract. Potential sources include oesophagitis, peptic ulcer and Meckel's diverticulum, and non-steroidal anti-inflammatories. Investigation may include FBC and film, Meckel's scan and upper GI endoscopy.

A 7 month old boy presents in Spring-time with severe vomiting and diarrhoea. This began overnight, with vomiting of stomach contents on 6 occasions. He has started passing offensive loose stools every 2 hours containing no blood or mucus. He was a term 3.6kg infant, and there were no neonatal problems. He is fully immunised and there is no FH/SH of note.

On examination he has a dry mouth and slightly reduced skin turgor. His temperature is 37.9°C, RR 35/min, HR 130/min. His peripheries are warm and capillary refill time 1.5 seconds. There are no other findings.

What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Coeliac disease [0]
- 2) Gastroenteritis [100]
- 3) Inflammatory bowel disease [0]
- 4) Malabsorbtion [0]

5) Toddler diarrhoea [0]

Comments:

The history suggests an acute gastroenteritis, presumed viral. The most likely organism at this time of year is Rotavirus, in winter Norwalk agent. Adenovirus, astrovirus and calicivirus are other common agents. The key to management is preventing dehydration by using oral rehydration therapy to promote optimal absorption of fluid.

A new diagnostic test for malabsorption has been analysed and the results have yielded the following 2x2 contingency table.

	Disease present	
test result	yes	no
+ve	0.9	0.1
-ve	0.2	0.8

Applying this test to a case of chronic diarrhoea from a patient group where the prevalence of malabsorption is known to be 20% (probability = 0.2) what is the probability of a patient having malabsorption if they have a positive test?

Available marks are shown in brackets

- 1) 0.16 [0]
 - 2) 0.24 [0]
 - 3) 0.48 [0]
 - 4) 0.64 [100]
 - 5) 0.8 [0]
-

Comments:

This is tough but the College are putting more and more Evidence Based Medicine Questions into the exam. This question tests understanding of pre-test and post-test odds, likelihood ratios, sensitivity and specificity. The calculation is as follows.

$$\text{Sensitivity} = 0.9 / (0.9 + 0.2) = 0.818$$

$$\text{Specificity} = 0.8 / (0.1 + 0.8) = 0.889$$

$$\text{Likelihood ratio for a positive test (LR+)} = 0.818 / (1 - 0.889) = 7.2$$

$$\text{Pre-test odds} = 0.2 / (1 - 0.2) = 0.25$$

$$\text{Post-test odds} = \text{pre-test odds} \times \text{LR+} = 0.25 \times 7.2 = 1.8$$

$$\text{Post-test probability} = 1.8 / (1.8 + 1) = 0.64$$

	Disease present	
test result	yes	no
+ve	true positive (A)	false positive (B)

-ve	false negative (C)	true negative (D)
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Sensitivity (how much a test is positive in disease) = $A / (A + C)$

Specificity (how much a test is negative in health) = $D / (B + D)$

Positive Predictive Value = $A / (A + B)$

Negative Predictive value = $D / (C + D)$

Pre-test odds = the odds of having the disease before you do the test (e.g. your rule-of-thumb guesstimate or the prevalence of the disease in the population or based on clinical findings etc.)

Post-test odds = the odds of having the disease after you did the test

Systematic Error = $(A + B) / (A + C)$ = good statistic for - 1) breaking the ice at a party of epidemiologists, 2) confusing your fellow SpRs at meetings

Likelihood Ratio (LR) + (the ratio of the chance of having a +ve test if the disease is present to the chance of having a positive test if the disease is absent) = $\text{sensitivity} / (1 - \text{specificity})$

LR- = $(1 - \text{sensitivity}) / \text{specificity}$

WHAT? ... Aghh! I knew I hated stats. Of what use is an LR?

Likelihood Ratios are good for

- directly calculating post-test odds
- tests with multiple levels (i.e. not just +ve or -ve). Calculate the LR at each level by taking the ratio of true +ves to false +ves both expressed as percentages of the total number tested.
- diseases requiring multiple tests. The post-test odds after one test is the pre-test odds for the next.
- bluffing your way in statistics, especially when talking to Evidence Based Medicine boffins

A 10 month old girl presents with abdominal pain and diarrhoea, which has recently had pink staining. She was well until 12 hours ago, when she had some loose stools and episodes of crying and holding her abdomen. She was a full term normal delivery and there were no neonatal problems. On examination she looks intermittently uncomfortable. Her heart rate is 110/ min and respiratory rate 20/min. She has a temperature of 37.4°C and is well perfused. Her abdomen is slightly distended and she has a curved firm mass 10 cm long extending from the right iliac fossa towards the hepatic flexure.

What is the most likely diagnosis?

Available marks are shown in brackets

1) Inborn errors of metabolism [0]

2) Constipation/encopresis [0]

3) Intussusception [100]

4) Renal anomalies [0]

5) Malignancy eg Neuroblastoma, Wilm's, Lymphoma [0]

Comments:

The history suggests an intussusception in the typical ileo-colic location, associated with intermittent colic. At this stage air enemas are usually successful in reducing them completely

A 6 week old infant presents with vomiting and failure to thrive. A diagnosis of pyloric stenosis is considered. Which one of the following statements is correct regarding congenital hypertrophic pyloric stenosis?

Available marks are shown in brackets

- 1) A "double bubble" is likely to be seen on abdominal X-ray [0]
- 2) A hyperchloraemic alkalosis would be expected [0]
- 3) Refusal of feeds is a feature [0]
- 4) Vomitus is typically bile stained [0]
- 5) Vomiting would have typically started between 2 to 4 weeks [100]

Comments:

The characteristic radiological feature is the 'string sign', which comprises a thin contracted pyloric canal, containing a central streak of barium.

The clinical symptoms commonly appear in this time frame. The vomiting is not bile stained but consists of large volumes of curdled milk. Appetite and feeding are normal or increased. The characteristic metabolic abnormality is hypochloraemic hypokalaemic alkalosis.

A 3 year old boy is referred because of an abdominal mass. He otherwise is well apart from sluggish bowel movements and sleep disturbance, which is causing stress in the family. He was a full term normal delivery weighing 3.4kg. He had gastroenteritis at 13 months of age.

On examination he is on the 75th centile for height and the 90th for weight. He has tympanic abdominal distension with no organomegaly. He has large non-tender irregular indentable masses in the left iliac fossa and suprapubic region.

What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Inborn errors of metabolism [0]
- 2) Constipation/encopresis [100]
- 3) Pregnancy [0]
- 4) Malignancy eg Neuroblastoma, Wilm's, Lymphoma [0]
- 5) Renal anomalies [0]

Comments:

The history suggests chronic idiopathic constipation. Often this begins following gastroenteritis, when poor fluid intake results in hard stools. When these are passed they can tear the delicate anal mucosa, causing a fissure. There is then voluntary stool retention, completing the vicious circle. As in this case, psychological stresses are common

An infant is admitted with diarrhoea and a diagnosis of Rotavirus is suspected. Which one of the following is correct regarding Rotavirus infection?

Available marks are shown in brackets

- 1) It typically affects infants older than 18 months [0]
- 2) The Rotazyme test to detect virus particles is a direct enzyme-linked immunosorbent assay [100]
- 3) Blood and mucus is found in the stools of about 50% of affected babies [0]
- 4) It occurs most often in the summer months [0]
- 5) Protection is not conferred by breast feeding [0]

Comments:

The peak incidence of Rota virus infection is 3-15 months. ELISA is performed on the stool samples to detect virus particles. Watery diarrhoea is common and blood and mucus is rare. Epidemics occur in the cooler months. Breast feeding protects against Rota virus diarrhoea and other infections eg pneumonia.

A young infant presents with a suspected diagnosis of pyloric stenosis. Which of the following is a feature of this diagnosis?

Available marks are shown in brackets

- 1) Projectile vomiting [100]
- 2) Anorexia [0]
- 3) Loose stools [0]
- 4) More frequent in girls [0]
- 5) Hyperchloraemic acidosis [0]

Comments:

Projectile vomiting of large quantities of curdled milk is characteristic. Anorexia and loose stools are not clinical features. It is commonest in first-born male children. The classical biochemical picture is of hypokalaemic, hypochloraemic metabolic alkalosis.

Which one of the following is a typical feature of toddler's diarrhoea.

Available marks are shown in brackets

- | | |
|-----------------------------------|-------|
| 1) Abdominal cramps | [0] |
| 2) Failure to thrive | [0] |
| 3) Pale stools | [0] |
| 4) Poor growth | [0] |
| 5) Undigested food in the stools | [100] |
-

Comments:

Toddler's diarrhoea affects children between the age of 1-5 years. It is not serious, the child is well and grows normally. It is characterised by 3 or more watery motions per day. Bits of undigested vegetable matter are usually present in the stool, which is offensive. Abdominal pain is unusual. Pale stools suggest malabsorption and more serious underlying pathology.

Whilst palpating the abdomen of a newborn an abdominal mass is felt. There are no other symptoms of note. She is apyrexial and feeding well.

On examination she is well, and on the 25% for height and weight. There are no abnormalities to find except an 8cm lobulated firm mass in the right flank.

What is the most likely diagnosis?

Available marks are shown in brackets

- | | |
|---|-------|
| 1) Inborn errors of metabolism | [0] |
| 2) Constipation/encopresis | [0] |
| 3) Pregnancy | [0] |
| 4) Malignancy eg Neuroblastoma, Wilm's, Lymphoma | [0] |
| 5) Renal anomalies | [100] |
-

Comments:

The likely diagnosis is a dysplastic kidney, which can be confirmed by ultrasound. Full renal investigations, including MCUG and DMSA scan are needed to excluded associated reflux, which occurs in 15% of cases.

These defects are not inherited, in contrast to polycystic kidney diseases.

Which one of the following is a reliable method of assessing dehydration in infants: Useful indices of dehydration in infants include:

Available marks are shown in brackets

- 1) Assessment of intra-ocular tension by palpation [0]
 - 2) Skin turgor over the dorsum of the hand [0]
 - 3) Palpation of the anterior fontanelle [100]
 - 4) Blood pressure [0]
 - 5) Serum sodium concentration [0]
-

Comments:

Signs of dehydration are very unreliable in the infant and always lag behind symptoms. Other than palpation of the anterior fontanelle there are few useful clinical signs. Thirst and oliguria may occur if the child is > 5% dehydrated . Tachycardia and low blood pressure only occur in severe cases of > 10% dehydration. Skin turgor is unreliable in overweight infants. Serum sodium may be entirely normal, or it may be low if there is electrolyte loss in the stool. However raised serum sodium can be seen when water loss is in excess of electrolytes.

A 6 week old infant presents with vomiting and failure to thrive. A diagnosis of pyloric stenosis is considered. Which one of the following statements is correct regarding congenital hypertrophic pyloric stenosis?

Available marks are shown in brackets

- 1) A "double bubble" is likely to be seen on abdominal X-ray [0]
 - 2) A hyperchloraemic alkalosis would be expected [0]
 - 3) Refusal of feeds is a feature [0]
 - 4) Vomitus is typically bile stained [0]
 - 5) Vomiting would have typically started between 2 to 4 weeks [100]
-

Comments:

The characteristic radiological feature is the 'string sign', which comprises a thin contracted pyloric canal, containing a central streak of barium. The clinical symptoms commonly appear in this time frame. The vomiting is not bile stained but consists of large volumes of curdled milk. Appetite and feeding are normal or increased. The characteristic metabolic abnormality is hypochloraemic hypokalaemic alkalosis.

A 10 year old child with Down's syndrome presents with chronic cough. He was diagnosed at birth and has always been a difficult feeder, with such frequent vomiting that mother keeps his bibs on even between meals. On examination he is well below the 3rd centile for weight and head circumference. He looks underweight, His chest is hyperinflated and he has coarse crackles symmetrically.

What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Gastroesophageal reflux [100]
 - 2) Allergic rhinitis [0]
 - 3) Sinusitis [0]
 - 4) Asthma [0]
 - 5) Croup [0]
-

Comments:

The picture suggests significant gastro-oesophageal reflux, associated with chronic aspiration and failure to thrive. Assessment is often difficult in children with developmental delay because of lack of cooperation with investigations.

Medical treatment is with antacids (eg Gaviscon), prokinetics (eg Domperidone) and acid suppression (eg Ranitidine, Omeprazole). Surgical fundoplication may be necessary in resistant cases.

A 16 year old mentally handicapped boy is brought to clinic by his parents who are concerned that he has lost 3kg over the last 6 weeks and has had some problems with lower abdominal pain. He has attended day centres for the last 1 year and has a long history of severe epilepsy for which he takes lamotrigine and carbamazepine. He had one fit two weeks ago. On examination he appears well, afebrile and no abnormalities are noted on abdominal examination. Examination of his faeces reveals fine, thin white worms approximately 1-2cm in length. What is the most appropriate treatment for this patient?

Available marks are shown in brackets

- 1) Levamisole [0]
 - 2) Mebendazole [100]
 - 3) Niclosamide [0]
 - 4) Piperazine [0]
 - 5) Praziquantel [0]
-

Comments:

This patient has threadworms, which is not uncommon amongst institutionalised individuals. The most appropriate treatment is mebendazole. Other drugs that are used include piperazine but this drug is not recommended for use in epilepsy. Therefore mebendazole is most appropriate. Praziquantel may be used for tapeworms.

A neonate does not pass meconium for 48 hrs after birth. A fortnight later his mother states that he is not passing stool regularly. He has been bottle fed since discharge. Investigations reveal massive dilatation of the colon proximal to the rectum and manometry shows increased internal anal pressure on rectal distension with a balloon. What is the most likely diagnosis?

Available marks are shown in brackets

- 1) Cystic Fibrosis [0]
- 2) Cow's milk allergy [0]
- 3) Hirschsprung's disease [100]
- 4) Hypothyroidism [0]
- 5) Idiopathic Constipation [0]

Comments:

The child has Hirschsprung's disease characterised by the early constipation and radiology. The disorder is due to an absence of the ganglion cells in the wall of the colon. Treatment is through resection of the atonic segment.

Which one of the following suggest a diagnosis of Hirschsprung's disease?

Available marks are shown in brackets

- 1) A contrast-study showing dilatation of the aganglionic bowel segment. [0]
- 2) Early presentation with vomiting. [0]
- 3) neonatal large bowel obstruction. [100]
- 4) Presentation after 1 year of age. [0]
- 5) Red current jelly stools. [0]

Comments:

Hirschsprung's disease is a common cause of neonatal large bowel obstruction. It results from failure of migration of ganglion cells to the affected segment of bowel. This always involves the distal colon but the proximal extent of the involvement is variable and in rare cases may involve the whole of the large bowel. Histologically, the affected segment has absent ganglion cells in the Meissner's and Auerbach's plexus but immunohistochemical evidence of increased ACE activity. 80% of cases present in the neonatal period. Contrast studies show the affected segment to be tonically contracted.

Rectal irrigation or an emergency colostomy may be required before a definitive 'pull-through' procedure. Patient's present typically with constipation and present late with vomiting and obstruction.

A 17 year old student returns from a back-packing trip to Nepal with a two-week history of offensive diarrhoea and weight loss. What is the most likely infective organism?

Available marks are shown in brackets

- | | |
|--|-------|
| 1) <i>Escherichia coli</i> 0157 | [0] |
| 2) <i>Giardia intestinalis</i> (<i>G.lamblia</i>) | [100] |
| 3) <i>Shigella flexneri</i> | [0] |
| 4) <i>Samonella typhi</i> | [0] |
| 5) <i>Yersinia enterocolitica</i> | [0] |

Comments:

The best bet here is Giardiasis as it presents as chronic diarrhoeal illness due to duodenal infestation by the faeco-oral route. Malabsorption can occur along with epigastric discomfort and flatulence. *E. coli* does not have a chronic illness (neither does *Shigella*) and like *Yersinia* causes bloody diarrhoea. *Salmonella typhi* is likely to cause a particularly serious systemic illness in this patient.

An infant boy is brought to clinic with a short history of vomiting and diarrhoea. Which of the following is most applicable concerning this infant?

Available marks are shown in brackets

- 1) Do not show signs of dehydration until more than 5% of the body weight is depleted [0]
- 2) Bloody diarrhoea would be expected with Staph Aureus food poisoning [0]
- 3) If dehydrated, he is likely to be hyponatraemic [0]
- 4) Should be given a broad-spectrum antibiotic [0]
- 5) Stool culture will typically reveal pathogenic E coli [100]

Comments:

Antibiotics are of little value as the condition is commonly caused by viruses. Haemorrhagic colitis may develop following E.Coli O157:H7 infections but Staph Aureus enterotoxin would not be expected to cause bloody diarrhoea. The commonest cause is Rota virus. If severely dehydrated hypernatraemia may also develop if there has been disproportionate water loss. Clinical signs of dehydration are generally absent unless body weight has reduced by >5%, but they are not always present either, hence history is important.

Which of the following statements applies to infants with gastroenteritis:

Available marks are shown in brackets

- 1) In most instances require treatment with an antibiotic which is not absorbed from the gut [0]
- 2) May have blood in their stools [0]
- 3) Should be admitted to hospital if they are unable to tolerate fluid orally [100]
- 4) Always develop lactose intolerance [0]
- 5) Should have a barium meal if the initial refeeding with milk is unsuccessful [0]

Comments:

The majority of children do not require antibiotics as the cause is viral. Formula feeds should not be changed. Intravenous fluid therapy will be required if they are unable to tolerate oral fluids. Lactose intolerance is common, but not inevitable. Barium meals are not useful in the investigation of gastroenteritis.

Which one of the following statements correctly applies to this case scenario:

An 8 month old child has been passing frequent watery stools for 5 days. His serum biochemistry revealed the following:

Na 158 mmol/l
K 3.5 mmol/l
Urea 13 mmol/l
Creatinine 120 umol/l
pH 7.08
PCO₂ 4.5 KPa
PO₂ 13 KPa
Base excess 18 mmol/l

Available marks are shown in brackets

- 1) The high serum sodium might have resulted from his being given only plain congee for the past 3 days at home [0]
- 2) A rapid infusion of sodium bicarbonate should be given to correct the acidosis [100]
- 3) Skin turgor is a good guide in assessing hydration state [0]
- 4) The condition might have resulted from taking concentrated feed before the onset of diarrhoea [0]
- 5) Replacement fluid to correct dehydration should be given over 6 hours to replenish the intravascular volume [0]

Comments:

The hypernatraemic dehydration will be a result of disproportionate loss of water and low volume fluid intake. Congee (rice water) is an effective rehydration solution and prevents further diarrhoea. The acidosis should be corrected by intravascular volume expansion. Skin turgor is not an effective means of assessing hydration as it will be preserved in overweight infants. Concentrated high sodium feeds and high sodium breast milk have been reported to cause hypernatraemic dehydration. If there is evidence of hypotension and shock, resuscitation should be fairly rapid, otherwise correction of electrolyte deficits should be over 24-48hours.

Which one of the following is a typical feature of toddler's diarrhoea.

Available marks are shown in brackets

- | | | |
|-----|-------------------------------|-------|
| 1) | Abdominal cramps | [0] |
| 2) | Failure to thrive | [0] |
| 3) | Pale stools | [0] |
| 4) | Poor growth | [0] |
| 5) | Undigested food in the stools | [100] |
-

Comments:

Toddler's diarrhoea affects children between the age of 1-5 years. It is not serious, the child is well and grows normally. It is characterised by 3 or more watery motions per day. Bits of undigested vegetable matter are usually present in the stool, which is offensive. Abdominal pain is unusual. Pale stools suggest malabsorption and more serious underlying pathology.

Which one of the following conditions has abnormal chromosome karyotype?

Available marks are shown in brackets

- | | | |
|-----|-------------------|-------|
| 1) | Cystic fibrosis | [0] |
| 2) | Phenylketonuria | [0] |
| 3) | Turner's syndrome | [100] |
| 4) | Spina bifida | [0] |
| 5) | Omphalocele | [0] |
-

Comments:

Of the conditions list only Turner's syndrome (45 XO) has an abnormal karyotype. 45XO/XX mosaicism may also be present.

A young infant presents with a suspected diagnosis of pyloric stenosis. Which of the following is a feature of this diagnosis?

Available marks are shown in brackets

- | | | |
|-----|--------------------------|-------|
| 1) | Projectile vomiting | [100] |
| 2) | Anorexia | [0] |
| 3) | Loose stools | [0] |
| 4) | More frequent in girls | [0] |
| 5) | Hyperchloraemic acidosis | [0] |
-

Comments:

Projectile vomiting of large quantities of curdled milk is characteristic.

Anorexia and loose stools are not clinical features. It is commonest in first-born male children. The classical biochemical picture is of hypokalaemic, hypochloraemic metabolic alkalosis.

Which of the following statements is true concerning: Intussusception in children:

Available marks are shown in brackets

- | | | |
|-----|---|-----|
| 1) | Is more common between the age of 3 months to 8 years | [0] |
| 2) | Bile stain vomiting is an early feature | [0] |

- 3) Bleeding per rectum is the most common presenting symptom [0]
4) Is usually caused by a polyp or pedunculated tumour [0]
5) Hydrostatic reduction plays an important role in the management [100]
-

Comments:

Intussusception in children can occur between 3 months and six years of age, but it is more common in the first 3 years. Colicky abdominal pain, straining and lethargy are clinical features. Bloody mucus and vomiting occur late when the bowel becomes strangulated and ischaemic. The condition involves the telescoping of one segment of the bowel into an adjacent segment. Most cases are idiopathic, however in a minority of cases there is a 'mechanical leading segment' which predisposes to the abnormality eg polyposis, Peutz-Jegher's syndrome. A contrast enema is therapeutic in most cases which present within 24 hours.

A 6 week old infant presents with vomiting and failure to thrive. A diagnosis of pyloric stenosis is considered.

Which one of the following statements is correct regarding congenital hypertrophic pyloric stenosis?

Available marks are shown in brackets

- 1) A "double bubble" is likely to be seen on abdominal X-ray [0]
2) Vomiting would have typically started between 2 to 4 weeks [100]
3) Vomitus is typically bile stained [0]
4) Refusal of feeds is a feature [0]
5) A hyperchloraemic alkalosis would be expected [0]
-

Comments:

The characteristic radiological feature is the 'string sign', which comprises a thin contracted pyloric canal, containing a central streak of barium. The clinical symptoms commonly appear in this time frame. The vomiting is not bile stained but consists of large volumes of curdled milk. Appetite and feeding are normal or increased. The characteristic metabolic abnormality is hypochloraemic hypokalaemic alkalosis.

Which one of the following statements is true regarding giardia lamblia.

Available marks are shown in brackets

- 1) May cause intestinal malabsorption [100]
2) Often causes a bloody diarrhoea [0]
3) May cause of haemolytic uraemic syndrome [0]
4) May be excluded by stool microscopy [0]
5) May be treated with Co-Trimoxazole (Septrin) [0]
-

Comments:

Giardia is a common cause of intestinal malabsorption. Blood loss is not a feature of this condition. It is a common cause of traveller's diarrhoea. Stool microscopy to detect cysts and oocysts is labour intensive and lacks the sensitivity of the current test of choice which involves detection of antigens on the surface of organisms in the stool specimen. It is treated with metronidazole. HUS may be caused by E Coli 0157 infection, but not Giardiasis.

An infant is suspected of having gastroesophageal reflux.

Which one of the following statements relating to this diagnosis is correct?

Available marks are shown in brackets

- | | |
|--|-------|
| 1) He is unlikely to present with vomiting. | [0] |
| 2) Vomitus will be bile stained. | [0] |
| 3) He is likely to require drug treatment | [0] |
| 4) He may have apnoeic episodes | [100] |
| 5) A barium swallow would be the investigation of choice to diagnose it | [0] |

Comments:

Gastro-oesophageal reflux is a common, usually benign and self-limiting cause of vomiting in children. It is not associated with bile stained vomiting. It can be treated in many cases with positioning after feeds and smaller frequent feeds. Drug treatment is reserved for severe cases. Oesophageal stricture, cough, apnoeic episodes, oesophagitis, pneumonia and failure to thrive are sequelae. Barium swallow is an unreliable tool as there may be intermittent reflux of barium, which is not identified when the x-ray is taken. Other more reliable methods include endoscopy and oesophageal pH probe.

Which of the following statements correctly applies to a baby of 2 months who is considered to be 10% clinically dehydrated with gastroenteritis:

Available marks are shown in brackets

- | | |
|---|-------|
| 1) He may have blood and mucus in his stools | [100] |
| 2) He will have a full fontanelle | [0] |
| 3) He will have a low packed cell volume | [0] |
| 4) Urea level will be normal | [0] |
| 5) Should not be given milk orally for at least 48 hours | [0] |

Comments:

Blood and mucus may be seen with dysentery and E.Coli infection. The fontanelle will be sunken and haematocrit will be raised. An increased urea will be seen in this degree of dehydration. Oral feeding should be continued whenever possible.

A young child is admitted with a two day history of feeling unwell, fever and has developed bloody diarrhoea. Which of the following is the most likely cause?

Available marks are shown in brackets

- | | |
|----------------------------|-------|
| 1) Crohn's disease | [0] |
| 2) E.Coli 0157 | [100] |
| 3) Giardiasis | [0] |
| 4) Polio | [0] |
| 5) Threadworm infestation | [0] |

Comments:

Enteropathogenic E coli is the most likely cause and is a cause of Haemolytic uraemic syndrome. It would be unusual for Crohn's to present as acutely as this. Polio usually first manifests as a non-bloody

diarrhoea as does giardiasis.

A male child weighing 4.2kg is admitted at age 4 weeks with a one week history of forceful vomiting after feeds. He is alert and eager for food but exhibits a loss of skin elasticity and has apparent distension in the left hypochondrium.

Which of the following statements is correct for this patient?

Available marks are shown in brackets

- 1) The most likely diagnosis is hiatus hernia [0]
- 2) He has hyperosmolar dehydration. [0]
- 3) Metabolic acidosis would be anticipated. [0]
- 4) Initial fluid replacement should comprise normal saline 10-20 ml/kg in one [100]
- 5) He should be started on an anticholinergic drug, e.g. Atropine Methylnitrate (Eumydrin), before feeds [0]

Comments:

The most likely diagnosis is pyloric stenosis, which is more common in males (4:1 M:F ratio) and characteristically presents 3-6 weeks after birth. This situation will cause a proportionate loss of fluid and electrolytes. Characteristically he will have hypokalaemic, hypochloraemic metabolic alkalosis. Initial resuscitation should be with normal saline, at twice the maintenance volume. This will ensure he is volume replete and prevent secondary activation of the renin/aldosterone system, which would cause more alkalosis and hypokalaemia. Once volume replete, the fluid can be changed to half strength saline with added potassium. Although there are a few emerging reports showing satisfactory short term and long term outcome with medical treatment, the preferred treatment is currently pylorotomy. This should be performed after successful resuscitation and electrolyte correction.

The following laboratory results were returned in a 6 week old boy admitted with 6 days of severe projectile vomiting:

Ph 7.51
PO₂ 12 KPa (95 mmHg)
PCO₂ 4.7 KPa (35 mmHg) Blood Urea 11 mmol/l
Na⁺ 131 mmol/l
K⁺ 3 mmol/l
Chloride 83 mmol/l

Which of the following is true concerning this patient?

Available marks are shown in brackets

- 1) He has respiratory alkalosis [0]
- 2) He is likely to have a bulging anterior fontanelle [0]
- 3) He should be resuscitated immediately with normal saline [100]
- 4) He should be commenced immediately on half strength soy protein, low lactose formula [0]
- 5) X-ray of abdomen is likely to show dilated loops of small bowel [0]

Comments:

He has a metabolic rather than respiratory alkalosis as CO₂ is not reduced. He is likely to have a sunken fontanelle as he is very dehydrated. Pyloric stenosis would highly likely as because of the age and hypochloraemic, hypokalaemic metabolic alkalosis. He should be resuscitated with normal saline first. Dilated bowel would not be expected, as pyloric rather than small bowel obstruction is present.

An infant is admitted with diarrhoea and a diagnosis of rota virus is suspected. Which one of the following is correct regarding Rota virus infection?

Available marks are shown in brackets

- | | |
|---|-------|
| 1) It typically affects infants older than 18 months | [0] |
| 2) The Rotazyme test to detect virus particles is a direct enzyme-linked immunosorbent assay | [100] |
| 3) Blood and mucus is found in the stools of about 50% of affected babies | [0] |
| 4) It occurs most often in the summer months | [0] |
| 5) Protection is not conferred by breast feeding | [0] |

Comments:

The peak incidence of Rota virus infection is 3-15 months. ELISA is performed on the stool samples to detect virus particles. Watery diarrhoea is common and blood and mucus is rare. Epidemics occur in the cooler months. Breast feeding protects against Rota virus diarrhoea and other infections eg pneumonia.

A 2 year old child is admitted with a 2 day history of diarrhoea.

Which of the following statements correctly applies to his condition?

Available marks are shown in brackets

- | | |
|---|-------|
| 1) Hyponatraemia is expected. | [0] |
| 2) Adenovirus is the commonest pathogen isolated. | [0] |
| 3) He should be treated with anti-diarrhoeal agents. | [0] |
| 4) Dehydration is best assessed clinically rather than biochemical | [100] |
| 5) He should be treated with boiled water alone for 24 hours | [0] |

Comments:

Hypernatraemic and hyponatraemic dehydration are both infrequent complications of childhood gastroenteritis. Rota virus is the commonest pathogen. Anti diarrhoeal agents are not recommended in the treatment, as they may cause complications such as respiratory depression, bowel obstruction and bacterial overgrowth. Biochemical tests may not be abnormal till the dehydration is advanced. Clinical signs such as decreased urine output, dry mucous membranes and sunken fontanelle may appear when the child is >5% dehydrated. Oral rehydration with a glucose electrolyte solution (sodium, glucose, chloride,potassium) is recommended.
