If you disagree with any of the answers, please contact Matthew Saxonhouse at msaxonhouse@gmail.com. As with any examination, there may be an alternative answer that you may feel should be correct. If this is the case, please contact the email address provided and Dr. Saxonhouse would be happy to discuss the question with you. The purpose of this examination is not to see how high of a score you can achieve, but rather to develop an understanding of the material that will be covered during the course and areas of weakness that you have going into the course.

1). D
One of the more common malformations, single umbilical arteries are observed in 0.5 – 1.0% of all infants. They are 3-4 times more common in twins compared with singletons and they may be associated with urogenital tract or cardiac anomalies. The presence of other anomalies on exam does warrant a more detailed evaluation consisting of chromosomal analysis and/or renal US.

2). D
The woman presented in the vignette has demonstrated multiple diastolic BPs ≥ 90mmHg. Two of these are ≥ 110 mm Hg thus supporting a diagnosis of severe preeclampsia. The definition of preeclampsia is: DBP ≥ 90 mmHg or SBP ≥ 140 mm Hg documented more than twice; severe if DBP ≥ 110 mmHg or SBP ≥ 160 mm Hg. She does not have any prior medical history thus not supporing a diagnosis of superimposed preeclampsia.

3). C
The ultrasound (US) findings demonstrate an endocardial cushion defect. Of note, US images displayed on a sheet of paper may not be of the best quality but it is important to be familiar with the appearance of the more common cardiac lesions. Being that this question is a 3rd level question, one must also be familiar with the triple screen findings that support Trisomy 21. Of the choices, choice C is correct. If you could not identify the lesion, than just knowing that only Trisomy 21 or 18 are supported by triple screen findings, this could narrow your choices to either C or D.

4). C
The fetal heart rate tracing and mother’s contractions represent late decelerations. Late decelerations are usually due to uteroplacental sufficiency and demonstrate fetal hypoxemia. Thus, choice C is the best answer.
5). D
The demonstration of higher post-ductal saturations supports a diagnosis of Transposition of the great arteries with elevated pulmonary pressures. This finding in combination with the others listed strongly supports fetal exposure to Isotretinoin (Retinoic Acid).
Reference: Jones KL. Smith’s Recognizable Patterns of Human Malformation, 6th edition pgs. 660-661.

6). B
Cardiac abnormalities (structural or arrhythmias) represent ~ 25% of all cases of non-immune hydrops fetalis. The fetus presented in the vignette may have had an arrhythmia that has recently resolved during the ultrasound but resulted in the findings.

7). B
When performing resuscitation in any newborn infant, proper guidelines published from the neonatal resuscitation program should be followed. For this particular infant, you do not know what was performed prior to your arrival. Rather, you need to start from the beginning and from the choices listed, choice B is the best option.

8). C
Intrauterine hypoxia initially results in increased blow flow to the heart, brain, and adrenal glands.

9). C
Based on the one-minute examination, the infant’s APGAR score is 6. See Table below:

<table>
<thead>
<tr>
<th>Clinical Sign</th>
<th>Score = 0</th>
<th>Score = 1</th>
<th>Score = 2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Respirations</td>
<td>None</td>
<td>Gasping, poor, irregular</td>
<td>Strong cry</td>
</tr>
<tr>
<td>Heart Rate</td>
<td>None</td>
<td>&lt; 100 bpm</td>
<td>&gt; 100 bpm</td>
</tr>
<tr>
<td>Color</td>
<td>Cyanotic</td>
<td>Acrocyanosis</td>
<td>Pink</td>
</tr>
<tr>
<td>Muscle Tone</td>
<td>Floppy</td>
<td>Some flexion of extremities</td>
<td>Active</td>
</tr>
<tr>
<td>Reflex Irritability</td>
<td>None</td>
<td>Grimace</td>
<td>Cough, sneeze, or cry</td>
</tr>
</tbody>
</table>

10). C
Although following standard NRP guidelines would recommend initiating positive pressure ventilation via bag and mask ventilation, the infant presented in the vignette is unique in that you are suspecting a diagnosis of congenital diaphragmatic hernia. NRP recommendations for these infants are to place an endotracheal tube immediately with appropriate gastric decompression.

11). B
A. Blood returning from the placenta to the fetus by way of the umbilical vein, normally has a pO₂ of 20-30 mmHg.
B. The most oxygenated blood in the fetus travels from the placenta via the umbilical vein into the inferior vena cava and ~ 1/3 of this blood travels through the foramen ovale to the left atrium then to the left ventricle and out the aorta to the major blood vessels of the brain.
C. Blood returning from the brain, via the superior vena cava, is directed primarily from the right atrium to the right ventricle, through the pulmonary artery and the patent ductus arteriosus to the descending aorta.
D. Approximately 45% of the descending aorta blood flow goes to the placenta by way of the umbilical arteries.
E. The pulmonary circulation receives approximately 7% of the fetal blood volume.

12). B
The infant presented in the vignette clearly has findings of cyanotic congenital heart disease. Although all of the choices may present with cyanosis, the single second heart sound and left superior axis on ECG strongly support the diagnosis of tricuspid atresia.

13). D
The findings listed support a congenital infection with either Rubella or CMV. Infants with congenital rubella syndrome, if infected < 8-weeks of infection, have a 50% chance of cardiac disease with patent ductus arteriosus being most common.
Reference: see below

14). C
The clinical findings presented support a diagnosis of Williams Syndrome. The most common cardiac lesion in infant with Williams Syndrome is supravalvular subaortic stenosis.
Reference for 13 and 14: Jones KL. Smith’s Recognizable Patterns of Human Malformation, 6th edition pgs. 120-123.
15). D

Although the infant presented in the vignette most likely has a ductal dependent congenital heart lesion, such as the hypoplastic left heart syndrome or severe coarctation of the aorta, one should not assume that this is the diagnosis until proper evaluation by a Cardiologist is performed (unless a prenatal diagnosis was obtained). It is important for the outside physician to also entertain the diagnoses of severe neonatal sepsis or an inborn error of metabolism. To hopefully begin stabilization of this critical infant, the following procedures should be performed (if the clinician is able to): stabilization of airway (if PGE is to be started prior to transfer, then intubation is recommended), proper fluid resuscitation, IV infusion of PGE, termination of all protein intake with IV infusion of carbohydrates only, and immediate start of Ampicillin and Gentamicin. By following this algorithm, you have entertained all three diagnoses and initiated proper initial stabilization of this potentially very critical infant.


16). D

The formula for oxygen consumption is the Fick principle

\[ \text{Oxygen consumption} = \text{CO} \times (\text{CaO}_2 - \text{CvO}_2) \]

\[ \text{CO} = \text{cardiac output} \]
\[ \text{CaO}_2 = \text{oxygen content of arterial blood} \]
\[ \text{CvO}_2 = \text{oxygen content of mixed venous blood} \]

For this question, all of the answers provided are in dL/minute reminding you to change the units from what you are given. You are provided with the cardiac output in L/min and need to convert this to dL/min before proceeding. In addition, the saturation values provided should be converted to decimal points to answer the question properly. Using the values provided, the question should be answered as:

1.2 L/min = 12 dL/min

12 x (1.34 x 15) (.98-.68) = 12 x 20.1 x .3 = 72.4 dL/min


17). B

D-Transposition of the Great Arteries is the most common cyanotic heart defect identified in the first week of life. This fact alone allows you to answer the question correctly. The additional findings of higher saturations in the right foot compared with the right hand also strongly supports this diagnosis. Elevated pulmonary pressures with shunting of oxygenated blood from the pulmonary artery to the descending aorta create these clinical findings.


18). C

Central cyanosis occurs when the concentration of deoxygenated hemoglobin exceeds 3g/dL. Therefore, the infant in this question has a hemoglobin of 16g/dL. If this infant would first appear cyanotic if > 3g/dL was deoxygenated, than the saturation would be 16 – 3 / 16 = 13/16 = 81%.

19). C
The EKG presented demonstrates a prolonged QT interval. Of the choices provided, hypocalcemia is the best answer, as hypercalcemia may result in a shortened QT interval.

20). A
The infant presented in the vignette likely has either esophageal atresia with a distal tracheo-esophageal fistula or isolated esophageal atresia. The esophagus and trachea first appear at the 21st day of gestation as a median ventral diverticulum of the primitive pharynx. By 34-36 days’ gestation, lateral ridges divide the diverticulum to form the trachea and esophagus. Tracheoesophageal anomalies occur at this age, during the embryonic stage of lung development.

21). B
The infant in the vignette is displaying symptoms of respiratory distress that immediately respond to supplemental oxygen. Choices C and D represent fixed shunts that would not respond to oxygen therapy. If the infant was sedated and not breathing, raising the oxygen concentration would not necessarily raise the oxygen saturations. Therefore, choices B and E remain. Respiratory distress syndrome (RDS) is rare in a term infant plus no maneuvers were provided to improve the compliance of the stiff lungs that are caused by RDS. Thus, choice B is the best answer.

22). B
The effect of altitude on the pO\textsubscript{2} (arterial oxygen) can best be solved by the following equation:
\[
(\text{pB#1} - \text{pH2O}) \times \text{FiO}_2\#1 = (\text{pB#2} - \text{pH2O}) \times \text{FiO}_2\#2
\]
Using the values provided:
\[
(687 - 47) \times .35 = (760-47) \times (? \text{ Value}) = 224 = 713 (?)
\]
\[
(?) = 0.314 = 31\% \text{ oxygen}
\]

23). B
24). D
To properly answer this question, the following formulas should be used:
Oxygen delivery to alveoli (cc/kg/minute) = alveolar minute ventilation x FiO$_2$
alveolar minute ventilation = tidal volume x respiratory rate

Using the values provided from the question:
Alveolar minute ventilation = 4 x 40 = 160
Oxygen delivery to alveoli = 160 x 0.21 = 33.6 = 34 ml oxygen per minute

25). E

26). A
SP-A is the most abundant of the surfactant proteins constituting nearly 5% by weight of the surfactant.

27). A
Oxygen carrying capacity is equal to the oxygen content of blood. The formula for oxygen content is equal to oxygen bound to hemoglobin (Hgb) plus dissolved oxygen or O$_2$ bound to Hgb plus dissolved O$_2$

O$_2$ bound to Hgb = [(1.34 cc O$_2$/g Hgb) x Hgb (mg/dL) x oxygen saturation
Dissolved O$_2$ = [0.003 cc O$_2$/dL torr) x pa O$_2$(torr)]

Increasing cardiac output will not change the oxygen content, only oxygen delivery. This eliminates choices C and D. Using the values provided in the question, the oxygen content is:
1.34 x 10 x 0.92 + .003 x 60 = 12.3 + 0.18 = 12.5.

If we change the Hgb to 15 mg/dL, the content increases to:
1.34 x 15 x 0.92 + .003 x 60 = 18.5 + 0.18 = 18.7 (choice A).

If we increase to PaO$_2$ to 400, the content increases to:
1.34 x 10 x 1 (assume saturation 100% if PaO$_2$ 400) + 0.003 x 400 = 13.4 + 1.2 = 14.6 (choice B).

Therefore, choice A is the best answer.
28). D
Factors affecting Oxyhemoglobin Dissociation Curve

<table>
<thead>
<tr>
<th>Shift Curve to Left</th>
<th>Shift Curve to Right</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Impair oxygen delivery</td>
</tr>
<tr>
<td>Alkalosis</td>
<td>Acidosis</td>
</tr>
<tr>
<td>Decreased temperature</td>
<td>Increased temperature</td>
</tr>
<tr>
<td>Decreased 2,3-DPG concentrations</td>
<td>Increased 2,3-DPG concentrations</td>
</tr>
<tr>
<td>Increased fetal Hemoglobin</td>
<td>Decreased fetal Hemoglobin</td>
</tr>
</tbody>
</table>


29). D
Oxygenation Index =
\[
\left\{ \frac{\text{Mean airway pressure} \times \text{inspired oxygen concentration}}{\text{PaO}_2} \right\} \times 100
\]
\[
\left\{ \frac{18 \times 1}{42} \right\} \times 100 = 43
\]

30). D
The prenatal risk factors and emergent delivery plus the infant’s clinical symptoms support a diagnosis of respiratory distress syndrome. In addition, the chest x-ray demonstrates ground glass appearance, air bronchograms, and decreased lung volumes. However, it is important to pay close attention to all of the details of the chest x-ray as the ETT is also in too far and probably contributing to the infant’s distress.

31). C
Close attention to the clinical history and CXR findings demonstrate a pleural effusion on the patient’s right side as the cardiac silhouette is displaced towards to the left. The PICC line on the right is not in good position thus supporting the diagnosis of a pleural effusion from TPN given the infant’s NPO status.
32). C
The CXR demonstrates cystic changes on the infant’s left side. Although this can be mistaken for intestines thus supporting a diagnosis of a left sided congenital diaphragmatic hernia, review of the CXR demonstrates a normally located stomach bubble with NG tube in place. In addition, a normal bowel gas pattern is demonstrated. These findings support a diagnosis of a CPAM.

33). D
Please see the table below describing the laboratory values for the different urea cycle defects:

<table>
<thead>
<tr>
<th>Enzyme Deficiency</th>
<th>Orotic Acid</th>
<th>Glutamine</th>
<th>Alanine</th>
<th>Citrulline</th>
<th>Arginine</th>
</tr>
</thead>
<tbody>
<tr>
<td>N-acetylglutamate Deficiency</td>
<td>Normal or low</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Carbamyl phosphate synthetase</td>
<td>Normal or low</td>
<td>Increased</td>
<td>Increased</td>
<td>Decreased</td>
<td>Decreased</td>
</tr>
<tr>
<td>Ornithine carbamyl transferase</td>
<td>Increased</td>
<td>Increased</td>
<td>Increased</td>
<td>Decreased</td>
<td>Decreased</td>
</tr>
<tr>
<td>Arginosuccinic acid synthetase (citrullinemia)</td>
<td>Increased</td>
<td></td>
<td></td>
<td>Increased</td>
<td>Decreased</td>
</tr>
<tr>
<td>Arginase</td>
<td>Increased</td>
<td></td>
<td></td>
<td>Normal</td>
<td>Decreased</td>
</tr>
<tr>
<td>Arginosuccinic lyase</td>
<td>Increased</td>
<td></td>
<td></td>
<td>Normal</td>
<td>Decreased</td>
</tr>
</tbody>
</table>


34). B
The infant described in the vignette has features consistent with one of the Mucopolysaccharidoses. The majority of these are inherited in an autosomal recessive pattern. However, the family history provided supports an inheritance consistent with X-linked inheritance. Hunter syndrome is the only Mucopolysaccharidosis inherited in a X-linked pattern thus supporting choice B.

35). B
See below

36). D
37). B
The mother of the infant has a sister with Carpenter syndrome. We can assume that she does not have the syndrome; therefore, we need to determine her risk of being a carrier for the syndrome. Because it is an autosomal recessive disorder, her chance of being a carrier is 2/3 or 0.67.

The chance that the father of the baby is a carrier (we can also assume that he does not have the syndrome as that information was not provided to us) can be calculated by using the Hardy-Weinberg equation. The disease incidence of an autosomal recessive disorder is $q^2$.

Therefore:
$q^2 = 1/5000$, so $q = 1/70.7$ and $p = 69.7/70.7$. The carrier frequency is $2pq = 2 (69.7/70.7)$ $(1/70.7) = 0.03$ is the probability he is a carrier.

Applying the fact that the mother’s chance of being a carrier is 0.67 and the father’s chance is 0.03, they then have a $\frac{1}{2}$ probability of passing the defective gene.

So: $0.67 \times 0.03 \times 0.5 = 0.01 = 1\%$.

References: 1. Genetics in Medicine Thompson and Thompson, Chapter 9
2. Clinical Genetics by Andrew Read and Dian Donnai Chapter 10

38). C
See below

39). A

40). C
Homocysteine is not considered an essential amino acid.

41). B

42). B
A 10-day old, 34-week infant with symptoms of respiratory distress should be receiving anywhere from 100-140 ml/kg/day. Based on this, choices D and E can be eliminated. Choice A is on the restricted side but not totally incorrect. If an infant receiving only TPN, than there caloric intake should be about 70-90 kcal/kg/day and should follow the rule that about 50% of the total calories should be from carbohydrates, 40% should be from fat, and ~ 10% from protein. Applying these principles, choice B is the best answer. Choice C is close but provides 150 ml/kg/day and 103 total kcal with 62% of total calories from carbohydrates.
43). B

44). E

45). C

46). D

47). C

48). A
Na+ deficit (mEq) = [Na+ desired (mEq/L) – Na+ current (mEq/L)] x 0.6 x weight (kg) = 128 – 116 x 0.6 x 0.98kg = 7.0 mEq.

49). C
Point a = respiratory alkalosis
Point b = respiratory acidosis with metabolic compensation
Point c = normal
Point d = metabolic acidosis with respiratory compensation
Point e = metabolic acidosis
Point f = respiratory acidosis
Point g = metabolic alkalosis

50). C
The infant described in the vignette has a normal anion gap acidosis.  
Na + K – Cl + HCO₃⁻ = 125 + 5 – 110 + 15 = 5.
The infant’s urine pH is 4.5 supporting a diagnosis of type II or proximal renal tubular acidosis. The pathology is due to decreased or absent proximal tubular reabsorption of HCO₃⁻ with normal distal acidification. If the urine pH was > 6.2, than this would support a diagnosis of type I or distal renal tubular acidosis in which H⁺ cannot be secreted in the distal tubule.
51). B
The calculation for the fractional excretion of Na or \( FE_{\text{Na}} = \frac{(\text{Urine Na} \times \text{Plasma Cr})}{(\text{Urine Cr} \times \text{Plasma Na})} \times 100 \)
Using the values provided = \( \frac{2 \times 1}{147 \times 1} = 0.014 \times 100 = 1.4\% \)

Interpretation for neonates: < 1% is normal; 1-2.5% is pre-renal; > 3% is intrinsic renal failure.
Thus, choice B.

52). C
At first, it appears that the infant has congenital adrenal hyperplasia based on the low Na and elevated K. However, the infant’s aldosterone level is elevated supporting a diagnosis of pseudohypoaldosteronism.
In CAH, aldosterone levels are low. K is also usually elevated to a greater extent than in this example.

53). C
The infant presented in the vignette has signs and symptoms consistent with Galactosemia. Galactosemia is a deficiency in the enzyme galactose-1-phosphate-uridyltransferase. Galactokinase deficiency may present with cataracts, but elevated blood glucose values exist.

54). D
The profound metabolic acidosis and slightly elevated ammonia level are most consistent with an organic acidemia. A urea cycle, if caught early, would present with a respiratory alkalosis and severely elevated ammonia level. The lack of renal failure and elevated ammonia level go against a ductal dependent congenital heart lesion.

55). C
Placing an infant in a double-walled incubator, compared with a single-walled incubator reduces the amount of radiant heat loss.

56). B
The infant has a relatively normal serum calcium value but low serum phosphorous value. In addition, the alkaline phosphatase level is significantly elevated. The infant is also only on fortified breast milk feedings and not a multivitamin. These findings are most consistent with vitamin D deficiency. The infant is likely receiving a normal amount of phosphorous but likely using it for bone growth reflecting the low serum value.
<table>
<thead>
<tr>
<th>Nephrogenic DI</th>
<th>Neurogenic DI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Increased UOP due to failure of kidneys to respond to ADH</td>
<td>Increased UOP due to inadequate production of ADH</td>
</tr>
<tr>
<td>Increased plasma osmolality</td>
<td>Increased plasma osmolality</td>
</tr>
<tr>
<td>Decreased urine osmolality</td>
<td>Decreased urine osmolality</td>
</tr>
<tr>
<td>Normal to high ADH levels</td>
<td>Low ADH levels</td>
</tr>
<tr>
<td>No change in urine osmolality after fluid restriction</td>
<td>No change in urine osmolality after fluid restriction</td>
</tr>
<tr>
<td>Increase in ADH levels after fluid restriction</td>
<td>No change in ADH levels after fluid restriction</td>
</tr>
<tr>
<td>No change in urine osmolality after ADH administration</td>
<td>Increase in urine osmolality after ADH administration</td>
</tr>
</tbody>
</table>


58). D
The findings of hypocalcemia and hyperphosphatemia suggest primary hypoparathyroidism. However, the infant has an elevated PTH level supporting pseudohypoparathyroidism.

59). C
The clinical findings support a diagnosis of Zellweger Syndrome or cerebro-hepato-renal syndrome. This is a rare autosomal recessive syndrome marked by the absence of hepatic and renal peroxisomes. Increased levels of very long chain fatty acids confirms the diagnosis in suspected infants.

60). A
LAD-1 deficiency may present in the neonatal period. A history of recurrent infections, delayed separation of the umbilical cord, leukocytosis, and absence of pus supports this diagnosis. Confusion may be made with Chronic Granulomatous Deficiency as this may also present in the neonatal period. However, recurrent infections with catalase positive bacteria or fungi strongly supports this diagnosis.

61). C
Although all of the immunoglobulin levels evaluated are low, they fall within the expected ranges for neonates of this post-conceptional age. Whether are not they predispose the infant to infection remains to be determined. Thus, these values represent the expected nadir of immunoglobulin levels for neonates of this post-conceptional age.
62). D
The persistent symptoms of diarrhea, failure to thrive, and scaly eruption combined with the eosinophilia strongly support a diagnosis of severe combined immunodeficiency disease (SCID).

63). C
Due to the fact that gonococcal ophthalmia or disseminated infection occasionally can occur in infants born to mothers with gonococcal infections, infants born to mothers known to have gonorrhea should receive a single dose of IV Cefotaxime. Ceftriaxone used to be the drug of choice, but to the risk for kernicterus, Cefotaxime should be used.

64). D
Due to difficulties in eradication Candidal sepsis when a central line is in place, it is recommended that all central lines be removed if an infant has candidal sepsis. Another central catheter may be placed after 3-negative blood cultures.

65). D
Newborn infants are at minimal risk for syphilis if the are born to mothers who completed appropriate penicillin treatment for syphilis more than 4-weeks before delivery, the mother had an appropriate response to treatment (documented fourfold or greater decrease in VDRL, and mother has no evidence of reinfection or relaps. The mother presented in the vignette received adequate treatment with an appropriate VDRL response.

66). C
The infant presented in the vignette is very concerning for neonatal sepsis. Based on the infant’s age, Ampicillin and Gentamicin are recommended. However, the mother’s history of fever and chills is concerning for a viral infection. In addition, the infant is lethargic, febrile, and jaundiced raising suspicions for HSV infection. Therefore, adequate fluid resuscitation, antibiotics, and antiviral therapy is strongly recommended.

67). D
Infants with congenital toxoplasmosis typically present with hydrocephalus and cortical calcifications. Infants with congenial CMV demonstrate periventricular calcifications. Both may demonstrate mild hepatitis and thrombocytopenia.
68). E
The first four choices, although theoretical risks for breast feeding are not contraindications. Choice E though is an absolute contraindication for breast feeding. Mother’s with active TB and an abnormal CXR should be isolated from their infants until the mother has been appropriately evaluated and the both the mother an infant are receiving appropriate antituberculosis therapy, the mother wears a mask, and the mother understands and is willing to adhere to infection control measures.

69). B
Most abdominal masses presenting in the neonatal period are of renal origin. They include hydronephrosis, renal dysplasia, and polycystic kidney disease.

70). C
The abdominal wall defect described is a gastroschisis. Unlike neonates with omphalocele, infants with gastroschisis usually do not have other nongastrointestinal abnormalities. However, infants with gastroschisis due have about a 16% chance of having other gastrointestinal abnormalities such as intestinal atresia, midgult volvulus, and intestinal stenosis.

71). C
Lactase levels reach adult levels by 36-weeks gestation.

72). D
The infant presented in the vignette is a late preterm infant with significant hyperbilirubinemia due to hemolytic anemia. In addition, the infant demonstrates findings consistent with the intermediate phase of acute bilirubin encephalopathy. Infants who demonstrate physical findings at this point should receive a double volume exchange transfusion to prevent the irreversible, advance phase of acute bilirubin encephalopathy and kernicterus.

73). C
The optimal volume for an exchange transfusion is twice the infant’s blood volume. For a term infant, blood volume is about 80-85 ml/kg. For a preterm infant, it is 100 ml/kg. The infant in the vignette is a full term infant. Using 80 ml/kg, the appropriate volume for an exchange transfusion would be 480 ml/kg. An exchange transfusion results in removal of about 85% of the neonate’s red blood cells.
74). C
Line A is alpha globin chain; Line B is the gamma globin chain, Line C is the beta globin chain, and line D is the delta globin chain.

75). D
Complete absence of Protein S or C activity may result in purpura fulminans in the neonate. In addition, factor V Leiden mutation may also present in this fashion.

76). C
The infant presented in the vignette most likely has late hemorrhagic disease of the newborn. The combination of the mother being a vegetarian, solely breast feeding, and not allowing the infant to receive vitamin K after birth is the likely reason for this illness. The lack of retinal hemorrhages also goes against shaken baby syndrome. In addition, late hemorrhagic disease of the newborn mostly presents with intracranial bleeding. Hemorrhagic disease of the newborn is also supported by an isolated prolongation of the Prothrombin time, with normal partial Thromboplastin times and normal platelet counts. Thus, choice C. Choice A represents hemophilia A or B; Choice B represents DIC and/or liver disease; Choice represents isolated thrombocytopenia; and choice E demonstrates possible liver disease with thrombocytopenia or sepsis but is not specific for one illness.

77). C
Although there really is no standard of care for management of neonates with thromboses, the infant presented in the vignette has a definite organ/life threatening thrombosis. Current recommendations are to initiate low dose systemic rTPA and low dose heparin for life/orgam/limb threatening thromboses.

78). B
The infant presented in the vignette likely has methemoglobinemia likely due to cytochrome-b5 reductase deficiency. If the level is > 40% (as in this infant), treatment should be with methylene blue at a dose of 1-2 mg/kg. Although methylene blue should not be given to infants with G6PD, the infant is stressed but demonstrates no evidence of hemolysis with a normal hemoglobin value.

79). B
The infant presented in the vignette likely has Staphylococcal scalded skin syndrome. The majority of cases are caused by Staph. Aureus exotoxin. The history of facial erythema and a positive Nikolsky sign with a sterile culture from the fluid from the bullae strongly support the diagnosis.
80). D
The infant in the vignette has a complete gastric outlet obstruction as there is no air below the stomach bubble. A possible cause of this finding is pyloric atresia. The multiple blisters on the arms and legs are concerning for infection but could also be associated with a form of epidermolysis bullosa. In fact, a genetic cause has been identified for some cases of pyloric atresia that occur in association with epidermolysis bullosa lethalis (Herlitz and Carmi syndromes).

81). A
Premature closure of the sagittal suture results in Scaphocephaly or dolichocephaly. This is the most common type of craniosynostosis. Surgery is for cosmetic improvement and if no other lesions are present, usually not associated with increased intracranial pressure or other neurologic complications.

82). D
If the birth injury resulted in involvement of C4/5, then the resulting phrenic nerve paralysis will result in respiratory distress and decreased diaphragm movement with elevation of the hemidiaphragm on CXR.

83). C

84). C
A = Caput succedaneum; B = Cephalohematoma; C = Sugaleal hemorrhage; D = Extradural hematoma

85). D
Selective neuronal necrosis is the most common pattern of cerebral injury after hypoxic-ischemic encephalopathy. The clinical outcome is mental deficiency, seizures, ataxia, feeding difficulties, and pyramidal cerebral palsy.

86). D
Extrapyramidal or athetoid cerebral palsy is classified as having mixed tone in the same muscle with gross and fine motor skills affected. Hearing deficits and speech abnormalities also exist.
87). B
McCune-Albright Syndrome is characterized by multiple areas of fibrous dysplasia of the long bones but also may include the ribs and spine. Irregular brown pigmentation usually involves the sacrum, buttocks, and upper spine. In addition, infants suffer from hyperthyroidism, hyperparathyroidism, and pituitary adenomas.

88). C
The infant described in the vignette has findings consistent with spastic diplegia.

89). C
Biphase stridor is usually the result of laryngeal obstruction. It tends to worsen with agitation and it is the most common type of stridor in the neonatal period. Of the various causes of laryngeal obstruction, laryngomalacia is the most common laryngeal anomaly.

90). B
A decrease in cerebral blood flow may be caused by decreased paCO_2_, increased paO_2_, increased hemoglobin concentrations, and decreased fetal hemoglobin concentrations.

91). B
First-order kinetics is characterized by the excretion of a certain percentage of drug per unit time and the rate of drug elimination is directly proportional to the serum drug concentration. Zero-order kinetics is characterized by the excretion of a constant amount of time regardless of the serum drug concentration.

92). B
Ranitidine inhibits the cytochrome P_{450} system. Theophylline is metabolized by the cytochrome P_{450} system. Due to the inhibition of the system by starting ranitidine, the infant’s theophylline level would likely increase.

93). D
As in most ethics questions, there may be several answers that seem very reasonable. Providing comfort care and the administration of narcotics is done in many units and hospice centers. However, these procedures are usually carried out after some effort has been made between families, nursing staff, and ethics committees. Therefore, choice D is the most logical and safest answer and addresses the interests of the family, infant, and staff.

94). C
95). A
A type I error is when the null hypothesis is rejected when the null hypothesis is really true. This usually occurs when the sample size is very small.

96). E
Sensitivity is the probability of a test being positive when true disease is present.

<table>
<thead>
<tr>
<th>Test</th>
<th>Disease</th>
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<tbody>
<tr>
<td>+</td>
<td>5</td>
</tr>
<tr>
<td>-</td>
<td>0</td>
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\[
\frac{5}{5 + 0} = 100\%
\]

97). D
The earliest finding of congenital glaucoma is an enlarged cornea.

98). C
Odds ratio can be calculated by the following formula:
True positives x true negatives / false positives x false negatives
\[
\frac{5 \times 100}{2 \times 50} = 5
\]

99). D
Cumulative incidence is the number of new cases in a given time period divided by the total population at risk. For this question, there were 6 cases in 15,000 infants born.

100). E

101). C

102). D

103). B
104). A

105). C
The ultrasound suggests bilateral lower urinary tract obstruction. The most common cause of lower urinary tract obstruction is posterior urethral valves. Immediate goals for treatment are to relieve the obstruction through the placement of a drainage catheter.

106). C
Remember it is the hCG from the placenta that initiates genital differentiation. Around 12 weeks the fetus begins to produce GnRH and the LH and FSH stimulates the fetal gonads to continue with genital development. The hypothalamus needs to provide GnRH so that the pituitary can secrete LH and FSH.

107). E
An MRI of the brain and cervical spine may show a defect intracranial or at the craniocervical junction causing the tongue fasciculation and hypotonia. Congenital myotonic dystrophy can cause severe neonatal hypotonia however the tongue fasciculations would not be expected. A high arched palate, “myopathic” facies with ptosis and facial weakness would be expected. Grip or percussion myotonia are often absent at this age but will appear later in childhood. EMG/NCS can help determine if it is a lower motor neuron problem at the anterior horn level, axon, myelin, neuromuscular junction, or muscle level. Spinal muscular atrophy is a common cause of severe hypotonia and tongue fasciculations especially in light of a normal mental status. Although Prader Willi syndrome can cause profound hypotonia, tongue fasciculations would not be expected.

108). B
The patient in this case has a classic history and symptomatology of a Vein of Galen malformation. Other causes of high output congestive heart failure need to be excluded. Carnitine deficiency may present with a cardiomyopathy but is typically associated with other findings such as muscle weakness and GI issues. Auscultation of the head and neck may detect a loud bruit.

109). D
The crossed adductor sign is present until 7-8 months. The Moro, palmar, and Galant responses should disappear by 6 months of age. The pupillary light response is variably present by 30 weeks gestation and definitely present by 34 weeks gestation.
While autosomal dominant polycystic kidney disease has a higher overall incidence than multicystic dysplastic kidneys (MCDK), it is uncommon in neonatal period. MCDK is the most common cystic renal disease in the newborn period.


We thank you for completing the Specialty Review in Neonatology Course Pretest for 2014. If you have questions about the course, suggestions, or comments, please feel free to email Dr. David L. Weisoly, Course Director, at dweisoly@yahoo.com or David_Weisoly@pediatrix.com. For questions or input specifically regarding this pretest, please contact Dr. Matthew Saxonhouse, Associate Course Director, at msaxonhouse@gmail.com.