A 3-hour-old term infant has cyanosis, no cardiac murmur, tachypnea, and no retraction. Pulse oximetry reveals an oxygen saturation of 60% in the right hand and 75% in the right foot.

Of the following, the MOST likely cause of the cyanosis is

- group B streptococcal sepsis
- persistent fetal circulation
- pulmonary valve atresia
- tetralogy of Fallot
- transposition of the great vessels

You selected 2, the correct answer is 5.

The absence of a cardiac murmur does not exclude serious cyanotic congenital heart disease. Abnormal murmurs result from high blood flow velocities causing turbulent blood flow, which is produced when blood flows across an obstruction, such as with valvular stenosis. Flow from a higher-pressure chamber or vessel to a lower-pressure chamber or vessel through a restrictive communication also causes high-velocity, turbulent blood flow.

Transposition of the great arteries and pulmonary atresia are two types of congenital heart diseases in which there are no abnormal murmurs. With transposed great arteries, blood flow from the right ventricle to the aorta and flow from the left ventricle to the pulmonary artery are unobstructed. In pulmonary atresia, there is no flow across the right ventricular outflow tract, which would be the case in pulmonary stenosis. Both of these congenital lesions can result in severe cyanosis in the newborn. The findings on physical examination that suggest cardiac disease may be very subtle and may include a single second heart sound of increased intensity and an abnormally active precordial impulse to palpation. The ductus arteriosus usually is patent, but it does not result in a cardiac murmur because the pressure in the pulmonary artery is almost the same as that in the aorta when affected infants present with cyanosis.

Some infants who have transposition of the great vessels will have early pulmonary hypertension, with shunting of pulmonary arterial blood into the descending aorta across a patent ductus arteriosus. Because the pulmonary artery carries oxygenated blood from the left ventricle, the ductal shunting results in reverse differential cyanosis. In this type of cyanosis, oximetry values measured in the feet are consistently higher than the preductal oximetry readings from the right hand, as described for the infant in the vignette. Not all cyanotic infants who have transposition of the great vessels will have reverse differential cyanosis, but its presence makes the diagnosis extremely likely. In contrast, although pulmonary atresia can mimic transposition with cyanosis, a single second heart sound, and no cardiac murmur, ductal shunting in either direction will not cause reverse differential cyanosis.

Primary pulmonary hypertension, sometimes called persistent fetal circulation
(PFC), also causes cyanosis in newborns who do not have congenital heart disease. Severely increased pulmonary vascular resistance results in inadequate pulmonary blood flow and in right-to-left shunting of deoxygenated pulmonary arterial blood into the descending aorta through the open ductus arteriosus and from the right atrium to the left atrium through a patent foramen ovale.

PFC may follow perinatal asphyxia or significant meconium aspiration at the time of delivery. It also may occur in newborns who have sepsis, such as with early-onset group B streptococcal infection. PFC may occur in apparently well term infants discovered to have cyanosis in the newborn nursery after an uneventful delivery. As with the cyanotic infant who has transposed great vessels or pulmonary atresia, there may be an abnormal second heart sound and precordial impulse, in this case due to pulmonary and right ventricular hypertension. Ductal patency in these infants also results in no murmur because the pulmonary artery pressure is essentially the same as the aortic pressure. If there is significant shunting of pulmonary arterial blood into the descending aorta across a patent arterial duct, differential cyanosis will occur. Pulse oximetry values will be lower in the feet than preductal oximetry values measured in the right hand. This may suggest that the cause of cyanosis is isolated pulmonary hypertension rather than congenital heart disease. Nevertheless, any infant who has cyanosis and is suspected of having PFC must undergo echocardiography to exclude cyanotic congenital heart disease.

Group B streptococcal sepsis and PFC without obvious severe meconium aspiration can mimic transposition of the great vessels in clinical presentation, but reverse differential cyanosis will not be present. If there is an oximetry difference, postductal oxygen saturation measurements will be lower.

Infants who have **tetralogy of Fallot** almost always have a loud murmur and no obvious cyanosis in the early neonatal period. This is because the degree of infundibular pulmonic stenosis present in the right ventricular outflow tract usually is not severe enough to result in significant right-to-left shunting through the ventricular septal defect. Normal pulmonary blood flow through the stenotic outflow causes a loud murmur. In the infrequent case of tetralogy of Fallot that also involves pulmonary atresia, the clinical presentation may include cyanosis and no appreciable murmur, but reverse differential cyanosis will not be present.

References:

Abman SH. Abnormal vasoreactivity in the pathophysiology of persistent pulmonary hypertension of the newborn. Pediatr Rev. 1999;20:e103-e109. Available at: http://pedsinreview.aapjournals.org/cgi/content/full/20/11/e103


Content Specification(s):

Understand the pathophysiology, recognize the clinical, laboratory, and radiographic features, and formulate a differential diagnosis of a cyanotic neonate
An infant is delivered by emergent cesarean section because of fetal distress from acute placental abruption. The Apgar scores are 1 and 3 at 1 and 5 minutes, respectively. The cord pH is 6.98, and the base deficit is 20 mEq/L. The infant is resuscitated and admitted to the neonatal intensive care unit for observation of potential injury to the brain and other organs.

Of the following, the MOST frequent complication of perinatal asphyxia is

- hepatic cholestasis
- myocardial dysfunction
- necrotizing enterocolitis
- pulmonary hemorrhage
- renal failure

You selected 1, the correct answer is 5.

The clinical history, cord blood acidemia, and low Apgar scores reported for the infant in the vignette suggest the diagnosis of perinatal asphyxia. In addition to the brain, the organ most frequently affected by perinatal asphyxia is the kidney. Renal failure occurs in 40% to 70% of cases. The clinical spectrum varies from oliguria to azotemia, depending on the severity of the insult. Monitoring of urine output, serum electrolytes and creatinine, and urinalysis for hematuria and myoglobinuria are important in the evaluation and management of affected infants.

Hepatic cholestasis occurs in approximately 10% of cases of perinatal asphyxia. Liver injury from perinatal asphyxia results in an increase in liver enzymes, hypoprothrombinemia, and abnormal liver-dependent coagulation factors. A rise in liver enzyme levels indicates hepatocyte necrosis or dysfunction. Hypoprothrombinemia is caused by impaired hepatic albumin production. Consumption of coagulation factors, thrombocytopenia from decreased platelet production or increased consumption, platelet dysfunction, and vascular endothelial damage are consequences of perinatal asphyxia that result in disseminated intravascular coagulopathy.

Myocardial dysfunction occurs in approximately 30% of cases of perinatal asphyxia. Early right heart failure with tricuspid regurgitation or global heart failure with cardiogenic shock characterizes the myocardial dysfunction. The dysfunction can be evaluated with a chest radiograph for cardiomegaly, arterial blood sampling for metabolic acidosis, and electrocardiography for evidence of myocardial hypoxia. An echocardiogram is useful for evaluating contractility of the heart, valvular incompetence, cardiac shunts, filling pressures, and volume status.

Necrotizing enterocolitis from hypoxic-ischemic injury of the gut is a potential complication of perinatal asphyxia. The onset of necrotizing enterocolitis is typically delayed until after enteral feedings are initiated. Accordingly, enteral feedings usually are delayed for several days after the initial injury to ensure recovery of the gut.

Pulmonary hemorrhage is an infrequent complication of perinatal asphyxia. Pulmonary edema from left heart failure, disruption of the pulmonary capillary bed from hypoxia, and coagulopathy associated with perinatal asphyxia contribute to
pulmonary hemorrhage.

References:

Abstract


Content Specification(s):
Understand the pathophysiology, recognize the clinical, laboratory, and radiographic features, and formulate a differential diagnosis of a cyanotic neonate
Please remember that you must answer all 10 of the questions in order to claim CME credit for this month.

A term newborn has tachypnea, rales, tachycardia, audible gallop, and diminished arm and leg pulses. Echocardiography shows enlargement of both ventricular chambers, with good systolic function and no congenital heart disease.

Of the following, the MOST likely diagnosis is

- carnitine deficiency
- hyperthyroidism
- hypoglycemia
- intracranial arteriovenous malformation
- pheochromocytoma

You selected 3, the correct answer is 4.

Congenital arteriovenous malformations are unusual vascular malformations that allow excessive shunting of systemic arterial blood to the systemic venous system, bypassing the vascular capillary bed. If the malformation is significant, the work of the heart is increased markedly by the increased return to the right heart. Both the normal systemic venous return and the excess oxygenated blood that bypassed the capillary bed in the organ where the malformation is located return to the right heart. Pulmonary blood flow, therefore, is excessive, resulting in pulmonary vascular congestion and tachypnea. This excess flow returns to the left ventricle, which becomes enlarged from the excess preload. Affected infants have signs and symptoms of congestive heart failure but no evidence of congenital heart disease on echocardiography. In the early stages of congestive heart failure, left ventricular function will be normal; in latter stages, left ventricular function may decrease, but in many cases function is preserved.

The most common sites of significant systemic arteriovenous malformation in neonates are the liver and the cerebral vasculature. Abnormal bruits over the head or liver may occur in some, but not all infants who have systemic arteriovenous malformation. When the vascular shunt is in the head, the systemic output of the aorta is "stolen" up the carotid artery, and downstream pulses in the arms and legs may be diminished, as described for the newborn in the vignette. The left ventricular output is high, but the cardiac output to the body downstream of the head is diminished.

Preserved systolic function in the presence of clinical congestive heart failure is not consistent with a dilated cardiomyopathy due to a metabolic defect, such as carnitine deficiency.

Cardiomegaly and signs of congestive heart failure may occur in newborns who have significant hypoglycemia. In some cases, echocardiography reveals that the cardiomegaly is due to a hypertrophic cardiomyopathy that has developed in the fetus in response to maternal hyperglycemia. Fetal hyperinsulinemia appears to
contribute to the abnormal hypertrophy of the heart and is responsible for the neonatal hypoglycemia. Postnatally this form of hypertrophic cardiomyopathy resolves as insulin levels decrease in the infant. The lack of severe left ventricular hypertrophy in the infant described in the vignette makes hypoglycemia unlikely. Some infants who have neonatal hypoglycemia without transient hypertrophic cardiomyopathy have poor systolic function that improves with effective treatment of the hypoglycemia.

Hyperthyroidism can cause a high-output form of neonatal heart failure. Late findings may include diminished ventricular function, but increased systolic performance and chamber enlargement would be evident before this occurs. In the early stages of hyperthyroid-associated high-output heart failure, pulses are brisk rather than diminished.

Pheochromocytoma is extremely unlikely in a newborn. Severe, often paroxysmal hypertension is the hallmark finding in patients who have pheochromocytomas, which are adrenal tissue tumors that secrete excessive catecholamine and catecholamine precursors. Symptomatic congestive heart failure can occur in newborns who have severe hypertension, but this usually is due to etiologies other than pheochromocytoma. One such cause is renal arterial stenosis or occlusion after umbilical artery catheter use. When neonatal hypertension causes heart failure, the cardiac systolic performance is poor. Pheochromocytoma is not a likely possibility for the infant in the vignette.

References:


Content Specification(s):

Understand the pathophysiology, recognize the clinical, laboratory, and radiographic features, and formulate a differential diagnosis of an acyanotic neonate with a left-to-right shunt lesion

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A 2-day-old 4.1-kg male infant is referred to you for evaluation of tachypnea that began 15 hours after birth. Pregnancy, labor, and vaginal delivery were uncomplicated. The respiratory rate is 75 breaths/min, but there are no retractions or nasal flaring. The infant appears pink, and the oxygen saturation is 97% in both upper and lower extremities. Physical examination reveals a grade II/VI systolic murmur, mild hepatomegaly, equal pulses, bobbing of the head, petechiae, and irritability. When he is awake, he can fix and follow movement, and muscle tone appears normal. The abdomen is slightly distended but soft and has active bowel sounds. You place an umbilical venous line and confirm that it is located just into the right atrium. Results of laboratory studies are: platelet count, 44 × 10^3/mcL (44 × 10^9/L); hematocrit, 31% (0.31); white blood cell count, 8 ×10^3/mcL (8 × 10^9/L) with 38% polymorphonuclear lymphocytes and 62% lymphocytes. Arterial blood gases with an Fio2 of 1.0 by mask are: pH, 7.33; Pco2, 36 mm Hg; Po2, 240 mm Hg; base excess, -1 mEq/L. Umbilical venous blood gases with an Fio2 of 1.0 by mask are: pH, 7.28; Pco2, 41 mm Hg; Po2, 190 mm Hg; base excess, -3 mEq/L.

Of the following, the MOST likely diagnosis for this infant is:

1. meconium aspiration syndrome
2. subgaleal hemorrhage
3. transient tachypnea of the newborn
4. transposition of great arteries
5. vein of Galen aneurysm

You selected 2, the correct answer is 5.

Approximately 40% to 60% of vein of Galen aneurysms present during the neonatal period. Anatomically, the aneurysm involves the persistent embryologic median prosencephalic vein of Markowski, which lies immediately anterior to the vein of Galen. Despite this finding, the term "vein of Galen" malformation is entrenched in the literature and applied to this entity. The most common feeding arteries are the posterior choroidal, anterior cerebral, middle cerebral, anterior choroidal, and posterior cerebral. Persistence of the transient venous structure called the falcine sinus often accompanies this malformation and, with the straight sinus, serves to drain the aneurysm. Neurologic effects frequently are associated with ischemic infarction, hemorrhage, or mass effect on brain structures. Congestive heart failure develops to compensate for the large proportion of cardiac output that may flow through the aneurysm. In fact, approximately 95% of affected neonates present with congestive heart failure; the remainder present with hydrocephalus or intracranial hemorrhage. Physical findings include bounding carotid pulses that may cause bobbing of the head, cranial bruit, and signs of congestive heart failure, as reported for the infant in the vignette. Older infants usually present with seizures and other neurologic findings. Among neonatal patients who survive, developmental delays, abnormal neurologic signs, and seizures occur due to mass effect and intracranial hemorrhage. The diagnosis is established in neonates who have unexplained high-output heart failure with head ultrasonography, computed tomography, or preferentially, magnetic resonance imaging. Thrombocytopenia and disseminated intravascular coagulation may result from consumption of platelets and clotting factors within the aneurysm. Right atrial oxygenation may be significantly elevated.
due to the return of arterial blood that has flowed through the aneurysm without passing through capillary beds where oxygen is consumed. Improved embolization techniques have resulted in increasing survival rates, but the prognosis remains guarded, with only 40% to 65% of survivors having favorable outcomes.

Meconium aspiration often presents with hypoxic respiratory failure and pulmonary hypertension. Although congestive cardiomyopathy may accompany meconium aspiration, hypotension usually is due to poor myocardial function, not a hyperdynamic high-output state. The absence of meconium, complicated delivery, hypoxic respiratory failure, and preductal/postductal oxygenation difference for the infant described in the vignette argues against meconium aspiration complicated by pulmonary hypertension. Right atrial oxygenation is not significantly increased above venous levels in meconium aspiration.

Subgaleal hemorrhage describes bleeding into the aponeurosis covering the scalp, which lies between the subcutaneous tissue and skull bone periosteum. Blood in this aponeurosis may spread beneath the entire scalp and subcutaneous tissues of the neck and present as a firm, fluctuant mass that increases in size after birth. A large amount of blood loss and consumptive coagulopathy may occur, which can result in respiratory distress, hypotension, shock, and disseminated intravascular coagulation. Progression can be rapid and result in death unless recognized and treated early. Hypotension and shock are due to hypovolemia rather than heart failure unless the shock is advanced; high-output cardiac failure is not present. Hyperbilirubinemia may complicate the recovery phase. Right atrial oxygenation is not increased, and neurologic signs become apparent only if associated with intracranial bleeding or advanced stages of shock.

Transient tachypnea of the newborn most often presents immediately after birth rather than during subsequent hours and days. Cardiomegaly, heart murmur, and increased vascular markings on chest radiographs may suggest congestive heart failure, although blood pressure and pulse pressure are typically normal.bounding pulses usually are not found with transient tachypnea. Right atrial oxygenation, as with meconium aspiration, is not increased significantly above venous levels.

Transposition of great arteries is a cyanotic congenital heart lesion. The usual presentation is cyanosis. High-output congestive heart failure is unusual. If coarctation of the aorta accompanies transposition of great arteries, full pulses may be present in the upper extremities and neck, similar to that found with high-output congestive heart failure. However, cyanosis and lack of elevated right atrial oxygenation reported for the infant in the vignette argue against transposition.

References:


Content Specifications:

Understand the clinical features and evaluation of arteriovenous malformations
Understand the management, complications of management, and outcome of arteriovenous malformations

Understand the diagnosis, clinical and radiographic features of extracranial hemorrhage, including cephalohematoma and subgaleal hemorrhage