July: Question 5

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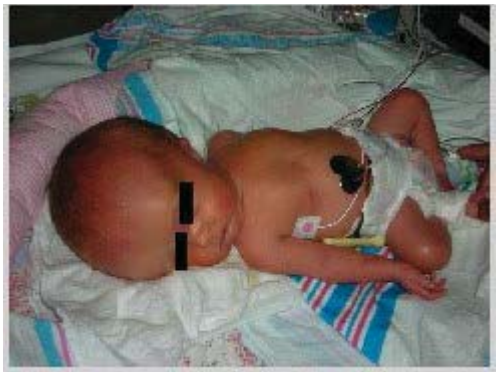
A 2,947-g female infant is born at 32 weeks' gestation. Physical examination reveals macrocephaly. Her head circumference measures 43.4 cm (>90th percentile). She does not have other dysmorphic physical features (Figure 1).

Figure 1: Infant with macrocephaly



Magnetic resonance imaging of her brain demonstrates agenesis of the corpus callosum, hydrocephalus, and a dilated fourth ventricle. The cerebellum is small, the cerebellar vermis is absent, and a large posterior fossa cyst is present (Figures 2 and 3).

Figure 2: Magnetic resonance image of brain demonstrating markedly enlarged lateral ventricles, absence of the corpus callosum, and a large posterior fossa cyst

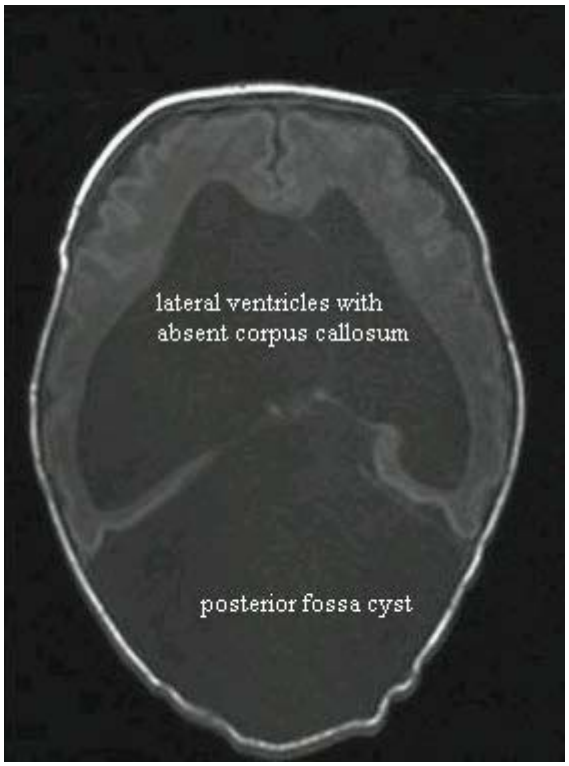
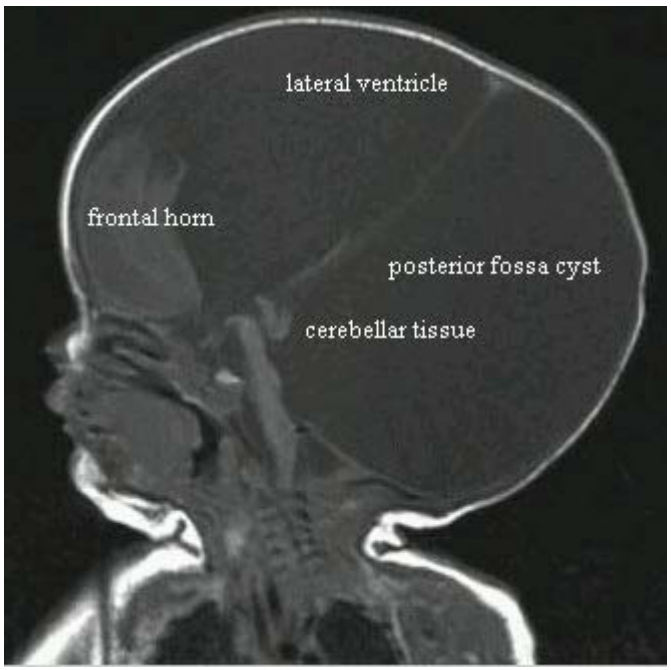


Figure 3: Magnetic resonance image of brain demonstrating markedly enlarged lateral ventricles, large posterior fossa cyst, and a small amount of cerebellar tissue



Of the following, the MOST likely cause of the hydrocephalus in this infant is:

1	aqueductal stenosis
2	arachnoidal cyst
3	Arnold-Chiari malformation
4	Dandy-Walker malformation
5	Joubert syndrome

You selected 3, the correct answer is 4.

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Hydrocephalus refers to ventricular enlargement, and is classified as obstructive (noncommunicating) or communicating. Cerebrospinal fluid (CSF) is secreted by the choroid plexus, and is generally absorbed by the arachnoid villi. Impaired CSF absorption leads to CSF accumulation and consequent ventricular enlargement or hydrocephalus. Obstruction of CSF flow can occur at any point in the pathway, including the foramen of Monro, aqueduct of Sylvius, fourth ventricle outlets (foramina of Luschka and Magendie), basal cisterns, and the arachnoid villi. In communicating hydrocephalus, CSF accumulates as a result of impaired absorption or obstruction within the subarachnoid space over the cerebral hemispheres.

The major causes of congenital hydrocephalus are aqueductal stenosis (33% of cases); Arnold-Chiari malformation with myelodysplasia (28%); "communicating" hydrocephalus (22%); Dandy-Walker malformation (7%); and other conditions, such as intrauterine infection, tumor, and hemorrhage (10%). Obstructive hydrocephalus is typically permanent, and a diversionary shunt is the preferred treatment.

The infant in the vignette has hydrocephalus and intracranial findings consistent with a diagnosis of Dandy-Walker malformation. Imaging of her brain demonstrates cystic dilation of the fourth ventricle, abnormal development of the cerebellar vermis, and hydrocephalus, the three major abnormalities comprising Dandy-Walker malformation (Figures 2 and 3). Enlargement of the posterior fossa and elevation of the tentorium are also distinguishing features of Dandy-Walker malformation. Associated CNS abnormalities, such as agenesis of the corpus callosum and neuronal migration defects, occur in as many as 70% of cases. Hydrocephalus with occipital prominence is a dominant clinical feature, but may not develop until later in the first year or rarely until adulthood. Management of the hydrocephalus involves shunting both the fourth ventricular cyst and the lateral ventricles. The long-term outcome of Dandy-Walker malformation is related to the severity of both the malformation and associated anomalies. Among infants with a postnatal diagnosis of Dandy-Walker malformation, the mortality is 10% and impaired cognitive development occurs in 25% to 75% of survivors. Dandy-Walker variant refers to the condition of vermian hypoplasia and cystic dilation of the fourth ventricle, without associated enlargement of the posterior fossa or hydrocephalus.

Aqueductal stenosis is the most common cause of congenital hydrocephalus, accounting for approximately one third of cases. Obstruction at the aqueduct of Sylvius results in marked enlargement of the third and proximal ventricles. The posterior fossa and cerebellar findings of Dandy-Walker malformation are not seen with aqueductal stenosis. Most cases of aqueductal stenosis are nonfamilial, but an X-linked variety associated with adducted thumbs, agenesis of the corpus callosum, and severe cognitive deficiencies has been associated with a mutation in the neural cell adhesion molecule L1CAM. In addition, aqueductal stenosis has been seen with the VACTERL association in an X-linked or autosomal recessive inheritance pattern.

Intracranial arachnoid cysts are benign, nongenetic developmental cysts that occur within the arachnoid membrane, and may obstruct CSF flow. Common locations for arachnoid cysts include the Sylvian fissure (66% of cases), the sellar region, and the posterior fossa. Arachnoid cysts of the posterior fossa can be difficult to distinguish from other cystic malformations of the posterior fossa, such as the Dandy-Walker malformation. However, the cerebellar abnormalities associated with Dandy-Walker malformation are not characteristic of arachnoid cysts.

Arnold-Chiari malformation (type II, occurring with myelodysplasia) is associated with obstructive hydrocephalus because of aqueductal compression and fourth ventricle outlet obstruction. The major features of the Chiari-II malformation include inferior displacement of the medulla and fourth ventricle, elongation and thinning of the upper medulla and lower pons, inferior displacement of the lower cerebellum through the foramen magnum, and a variety of bony defects of the occiput and upper cervical spine. Unlike Dandy-Walker malformation,

posterior fossa fluid collections are not characteristic of the Arnold-Chiari malformation.

Joubert syndrome is a disorder of autosomal recessive inheritance marked by agenesis or severe hypoplasia of the cerebellar vermis, and is also known as familial vermian agenesis. Clinical features include ataxia, hypotonia, oculomotor apraxia, dysregulation of breathing, and mental retardation. Although the fourth ventricle is enlarged with Joubert syndrome, the lack of both hydrocephalus and enlargement of the posterior fossa are features that distinguish this disorder from Dandy-Walker malformation.

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References:

Gleeson JG, Dobyns WB, Plawner L, Ashwal S. Congenital structural defects. In: Swaiman KF, Ashwal S, Ferriero DM, eds. *Pediatric Neurology, Principles & Practice*. 4th ed. Philadelphia, Pa: Mosby Elsevier; 2006:363-490

Pattisapu JV. Etiology and clinical course of hydrocephalus. *Neurosurg Clin North Am*. 2001;36:651-659

Volpe JJ. Neural tube formation and prosencephalic development. In: *Neurology of the Newborn.* 4th ed. Philadelphia, Pa: WB Saunders; 2001:3-44

American Board of Pediatrics Content Specification(s):

Be able to differentiate the familial/genetic features of neurology disorders associated with increased head circumference

Understand the etiology, familial/genetic features, and abnormalities associated with hydrocephalus

Understand the treatment of hydrocephalus

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July: Question 3

An infant with a birthweight of 3,200 g is born at 40 weeks' gestation. At 48 hours of age, you perform a neurologic examination.

Of the following, the finding MOST likely to represent nervous system dysfunction in this infant is:

1	absent patellar tendon reflex
2	ankle clonus of 5 to 8 beats
3	brisk pectoralis reflex
4	crossed adductor response
5	extensor plantar response

You selected 5, the correct answer is 1.

Assessment of deep tendon reflexes (DTRs) contributes to the neurologic examination of the newborn infant. In the absence of nervous system dysfunction, DTRs should always be elicitable in the term newborn. DTRs may be difficult to elicit in the triceps, variably obtainable at the biceps, and present at the patellar and Achilles tendons. In the newborn, the DTR easiest to elicit involves the pectoralis major. In the preterm infant, DTRs are elicitable by 27 weeks' gestation, and generally are less intense than in the term infant.

Deep tendon reflexes are qualified as absent, hypoactive, normal, or hyperactive. In the newborn infant, the response is often normally brisk or hyperactive. Absent or hypoactive DTRs occur with myopathies, neuropathies, and cerebellar disorders. Upper motor neuron lesions typically present with normal or increased DTRs. Detecting asymmetry in response is more useful than precisely qualifying the intensity of the reflex.

Ankle clonus is frequently elicited in the newborn, and it is enhanced during crying and hyperexcitable states. In the absence of other abnormal neurologic signs, up to 8 to 10 beats of symmetric clonus should be accepted as normal in the newborn infant. Newborn clonus is transient, and more than a few beats of clonus beyond 3 months of age suggests dysfunction of the corticospinal tracts.

Tapping the patellar tendon or the thigh adductors (medial aspect of the knee) may produce contraction in the opposite extremity, known as a crossed adductor response. Although a normal finding in the first months after birth, a crossed adductor response beyond 6 to 8 months of age suggests central nervous system dysfunction.

In the newborn, the plantar response is considered of limited value, because of competing reflexes and relative inconsistency of the response. In older infants, a positive Babinski reflex, extension of the great toe with fanning of the other toes, follows stimulation of the lateral aspect of the sole of the foot. However, in the newborn, the plantar response is normally extensor for the first month, and usually throughout the first year. Competing reflexes in the newborn stimulated by contact with the foot, such as with an examiner holding the foot to perform the plantar response, contribute to inconsistency in the finding. Nociceptive withdrawal and contact avoidance, stimulated by stroking of the dorsum of the foot, promote an extensor response. In contrast, plantar grasp and the positive supporting reflex, both elicited by pressure on the plantar aspect of the foot, promote flexion at the toes. A distinctly asymmetrical extensor plantar response, or persistence beyond infancy, suggests corticospinal tract impairment.

References:

Kuban KC, Skouteli HN, Urion DK, Lawhon GA. Deep tendon reflexes in premature infants. *Pediatr Neurol*. 1986;2:266-271

Swaiman KF. Neurologic examination of the term and preterm infant. In: Swaiman KF, Ashwal S, Ferriero DM, eds. *Pediatric Neurology, Principles & Practice*. 4th ed. Philadelphia, Pa: Mosby Elsevier; 2006:47-64

Volpe JJ. Neurological examination: normal and abnormal features. In: *Neurology of the Newborn.* 4th ed. Philadelphia, Pa: WB Saunders Co; 2001:103-133

American Board of Pediatrics Content Specification(s):

Recognize normal deep tendon reflexes (DTRs) in the newborn infant

Know that unsustained clonus is common in newborn infants

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