Question 1

Microcephaly is found in which of the following contexts EXCEPT:

Selected Answer: [None Given]
Correct Answer: A. mucopolysaccharidosis

Response Feedback:
Microcephaly refers to a head circumference less than 2 SD below the mean for age and sex and controlled for head shape, intrauterine growth retardation and/or low birth weight. Unlike macrocephaly, which may occur with an enlarged, normalized, or small brain, microcephaly occurs in parallel with microencephaly or small brain. Microencephaly may result from primary causes such as chromosomal abnormalities (e.g. Down syndrome) and neuronal proliferation/migration defects (e.g. lissencephaly) or from numerous secondary events such as maternal TORCH infections or metabolic/nutritional disorders. The neuronal storage diseases (e.g. the mucopolysaccharidoses) are more likely to cause macroencephaly or enlarged brain (Prayson 2005, pp. 95-97).

Question 2

Chiari II (Arnold-Chiari) is associated with which of the following:

Selected Answer: [None Given]
Correct Answer: C. displacement of cerebellar tissue below the foramen magnum

Response Feedback: Chiari type II malformation, also known as Arnold-Chiari malformation, comprises an open spinal neural tube defect, a small posterior fossa with hindbrain crowding with cerebellar tonsilar displacement below the level of the foramen magnum. Associated brainstem malformations frequently include fusion of inferior colliculi to give a beak-like appearance to the quadrigeminal plate and an indistinct pontomedullary junction with a rod-shaped pons. There is no cystic dilation of the fourth ventricle as this is feature of Dandy-Walker malformation. The open spinal neural tube defect is a myelomeningocele (or meningomyelocele) referring to a defect in spinal neural tube closure resulting in herniation of the spinal cord (myelo) and the meninges (meningo) through a vertebral defect - not a cranial neural tube defect. Spina bifida occulta is a closed spinal defect resulting from abnormal differentiation of tail bud tissues (Prayson 2005, pp. 98, 106-107). Agenesis of the olfactory tracts occurs in conditions involving failure of forebrain induction.

Question 3

All of the following are features of semilobar holoprosencephaly EXCEPT:

Selected Answer: [None Given]

Correct Answer: B. a single eye lying below a nasal anlagen

Response Feedback: Holoprosencephaly refers to a spectrum of malformations resulting from failure of forebrain induction and patterning. At the mild end of the spectrum is arrhinencephaly or absence/hypoplasia of the olfactory tracts and bulbs. A less mild form, termed lobar holoprosencephaly is characterized by an interhemispheric fissure which divides the whole forebrain except the most rostral/ventral aspect. At the severe end of the spectrum is lobar holoprosencephaly, characterized by a single telencephalic vesicle with a single or holoventricle and fused diencephalons. An intermediate form, semilobar holoprosencephaly, exhibits a partial interhemispheric fissure. In all forms, micrencephaly and arrhinencephaly are virtually always present. Associated craniofacial abnormalities tend to predict the severity of the underlying brain malformations with cyclopia underlying a nasal anlagen (proboscis) accompanying the complete alobar form, and cebocephaly (an elongated nose with a single nostril) accompanying the semilobar form (Prayson 2005, pp.110-112).

Question 4

Lissencephaly type I:

Selected Answer: [None Given]

Correct Answer: D. four layer cortex

Response Feedback: In classic, type I lissencephaly, the cortex consists of four abnormal layers instead of the normal six. Balloon cells, large cells with morphologic and immunohistochemical features of both neurons and glia, are present in the tubers of tuberous sclerosis as well as focal cortical dysplasia where localized somatic mutations in the TSC1 gene have occurred. The type II lissencephalies are
associated with mutations disrupting the pia-glia limitans allowing neurons to migrate past the pial surface into the leptomeninges producing a cobblestone appearance. These same mutations disrupt the sarcolemmal membrane of myocytes thereby causing muscular dystrophy as well (Prayson 2005, pp. 120-123).

**Question 5**

Which of the following is/are associated with callosal agenesis:

Selected Answer: [None Given]

Correct Answer: D. a, b, and c

Response Feedback: Agenesis of the corpus callosum, whether partial or complete, is a relatively common malformation found either in isolation or as part of various syndromes. In total agenesis, the hemispheric white matter fibers fail to either form or cross the midline. Remaining callosal fibers tend to abnormally project antero-posteriorly along the medial aspect of the lateral ventricles and are called Probst bundles. The cingulate gyrus is replaced by abnormally radiating gyri and the lateral ventricles have a vertical orientation, imparting a bat wing morphology (Prayson 2005, pp.125-126).

**Question 6**

Small/absent cerebellar vermian lobules, cerebellar heterotopia, and hypoplasia of the superior cerebellar peduncles:

Selected Answer: [None Given]

Correct Answer: C. Joubert’s syndrome

Response Feedback: Joubert’s syndrome is one of the forms of cerebellar hypoplasia. The cerebellar vermian lobules are small or absent, cerebellar cortical cell heterotopia are present, and the superior cerebellar peduncles are markedly hypoplastic. Lhermitte-Duclos disease, also known as dysplastic gangliocytoma of the cerebellum, comprises a unilateral hypertrophy of the cerebellar hemisphere with thickened folia containing displaced, enlarged dysplastic neurons. Rhombencephalosynapsis is a defect in midline cerebellar development so that the cerebellar hemispheres and dentate nuclei are fused across the midline. Supratentorial midline defects may accompany the cerebellar manifestations. Dandy-Walker malformation is characterized by cerebellar verm hypoplasia/agenesis, enlargement of the posterior fossa, and a cystically dilated fourth ventricle. Chiari type II malformation comprises an open spinal neural tube defect and a small posterior fossa with hindbrain crowding and cerebellar tonsilar displacement below the level of the foramen magnum (Prayson 2005, pp.128-133).

**Question 7**

Microcephaly, narrow superior temporal gyrus, cerebellar hypoplasia, Alzheimer’s disease:
Trisomies are frequently associated with malformations of the CNS. Trisomy 21 (Down's syndrome) is associated with gross abnormalities of the brain including microcephaly, brachycephaly, exposed insula with a narrow superior temporal gyrus, and hypoplasia of the cerebellum and brain stem. Individuals with Down syndrome are universally affected by the clinical and pathologic manifestations of Alzheimer's disease. Trisomy 13 (Patau syndrome) is associated with microcephaly and holoprosencephaly, which is frequently, associated with additional midline defects such as arhinencephaly and agenesis of the corpus callosum with basal ganglia fusion. Other findings may include myelomeningocele, hydrocephaly, and cerebellar heterotopias. Neonates with trisomy 18 (Edwards syndrome), like trisomy 13, tend to be microcephalic and may have abnormalities of the hippocampus, cerebellum and lateral geniculate nucleus (Prayson 2005, pp.157-158).

**Question 8**

Aqueductal stenosis is most frequently associated with:

Selected Answer: [None Given]

Correct Answer: A. hydrocephalus

Response Feedback: The cerebral aqueduct, also known as the aqueduct of Sylvius, is the narrowest portion of the ventricular system. Obstruction or even absence of the aqueduct may result from numerous causes including genetic, gliosis secondary to a variety of insults, vascular malformations, and pineal region neoplasms. Based upon its anatomic location, aqueductal stenosis can result in an obstructive hydrocephalus (Prayson 2005, p.134). It is not associated with defects of neural tube closure, holoprosencephaly (arrhinencephaly), or hydranencephaly which is a disruption resulting from massive intrauterine cerebral necrosis.

**Question 9**

Cystic dilation of the central canal of the spinal cord:

Selected Answer: [None Given]

Correct Answer: C. hydromyelia

Response Feedback: Hydromyelia refers to cystic dilation of the central canal of the spinal cord whereas syringomyelia is a glial lined cavity within the spinal cord parenchyma. Either may be seen in the setting of neural tube defects or disruptions such as trauma o tumors. Diplomyelia refers to duplication of spinal cord segments. This may also be seen in the context of a meningomyelocele (Prayson 2005, p.106).

**Question 10**

0 out of 1 points
Spina bifida occulta involves only the posterior vertebral elements and meninges:

Selected Answer: [None Given]  
Correct Answer: False

Response Feedback: Spina bifida occulta involves only the posterior vertebral elements. If meninges do herniate through a posterior vertebral defect, this is termed meningocele (Prayson 2005, p. 98).