Answers to neurology quiz

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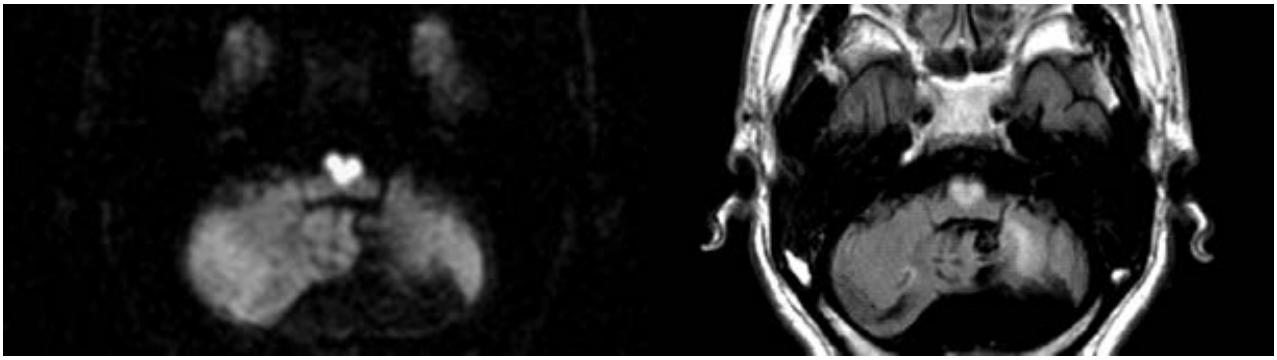
Answer 1

C) Bilateral medial medullary syndrome

Heart shaped sign



Bilateral medial medullary infarction (bilateral MMI) is an extremely rare cerebrovascular accident presenting with quadriplegia as the initial symptom and resulting in poor functional prognosis. Diagnosis of bilateral MMI has become possible based on brain MRI findings. In the early stage, bilateral MMI is sometimes misdiagnosed as Guillain-Barré syndrome. The medulla oblongata is divided into anterior-medial territory, anterior-lateral territory, lateral territory, and posterior territory, according to vascular supply. It is considered that blood is supplied to these areas by the vertebral artery and the anterior spinal artery, but it is often difficult to identify the occluded blood vessel because of the vastly complex network formed by these blood vessels. The “heart appearance” sign is considered to appear when the infarct occurs in the former two regions (anteriormedial territory, anterior-lateral territory). For an early diagnosis of bilateral MMI, it is essential to bear in mind that characteristic findings may be obtained by diffusion-weighted MRI.

Answer 2

a) Reversed Robin Hood syndrome

Reversed Robin Hood syndrome (RRHS) has recently been identified as one of the mechanisms of early neurologic deterioration in acute ischemic stroke (AIS) patients related to arterial blood flow steal from ischemic to nonaffected brain.

One of the mechanisms related to infarct expansion, leading to neurologic deterioration in the setting of acute cerebral ischemia, is an intracranial arterial blood flow steal phenomenon in patients with proximal arterial occlusions. Auto regulation of blood vessels is lost in the infarcted area and as result vasodilatation that occurs normally with hypercapnia does not occur. More specifically, with hypercapnia, flow velocities paradoxically decrease in the vessels supplying ischemic tissues at the time of velocity increase in the normal arteries, which are able to respond to the carbon dioxide stimulus with a more effective vasodilation. This results in an intracranial steal phenomenon. If this steal phenomenon leads to neurologic deterioration, the reversed Robin Hood syndrome (RRHS) is diagnosed. Robin Hood is known to have robbed the rich to give the poor, but as seen here when blood is robbed from the ischaemic region the term reversed Robin Hood syndrome is used.

Answer 3

c) Susac syndrome

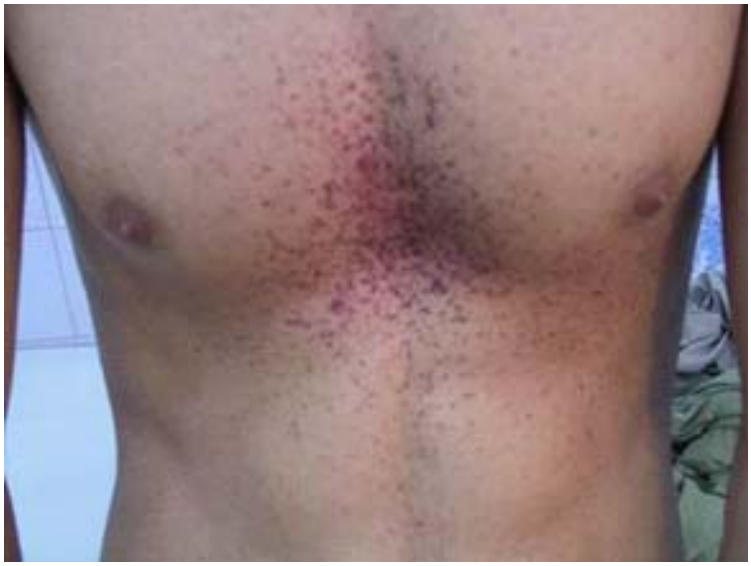
Susac syndrome (SS), also known as SICRET syndrome (small infarctions of cochlear, retinal and encephalic tissue), is a rare syndrome typically affecting young to middle-age women that is clinically characterized by the triad of acute or subacute encephalopathy, bilateral sensorineural hearing loss, and branch retinal arterial occlusions. Symptoms include episodes with headache, encephalopathic symptoms, focal neurologic deficits, sudden hearing loss for middle and low frequencies, and scintillating scotomata. Individuals with an encephalopathic course experience acute or subacute episodes of cognitive deficits, vigilance or mood changes, psychiatric symptoms (eg, psychosis or depression), fatigue, focal neurologic deficits, and, less frequently, seizures.

Characteristic MRI features are present even if all components of the clinical triad have not yet manifested. There tend to be multiple, small white matter lesions which have a predilection for the corpus callosum. Callosal lesions are considered almost pathognomonic and have many important characteristic features: typically involve the central fibers of the callosal body and splenium without abutting the callosal undersurface (with relative sparing of the periphery) – black holes in T1.

Answer 4

e) Autosomal recessive

Fabry's disease is a rare X linked recessive inherited lysosomal storage disorder. In young adult stroke Fabry's is the cause of stroke in about 5% of males and 3% of females labelled as 'cryptogenic stroke' [Fellgiebel A et al 2006]. Both males and females should be screened for this XLR inherited disease when investigating young adult cryptogenic stroke. Peripheral neuropathy: Patients complain of neuropathic pain with burning pains in the limbs "acroparaesthesia" which starts in adolescence or before aggravated by exercise. Skin: Cutaneous purplish angiokeratomas on the abdomen, umbilicus and genital areas Eyes: Corneal dystrophy. Corneal verticillata detectable by slit lamp examination. Cardiac: LV hypertrophy and conduction abnormalities. Hypertrophic cardiomyopathy may be seen. Renal: Progressive renal disease and eventual chronic kidney disease. CNS: small vessel stroke disease is significant.



Answer 5

b) CARASIL

CADASIL and CARASIL are hereditary small vessel diseases leading to vascular dementia. CADASIL commonly begins with migraine followed by minor strokes in mid-adulthood. Dominantly inherited CADASIL is caused by mutations (n > 230) in NOTCH3 gene, which encodes Notch3 receptor expressed in vascular smooth muscle cells (VSMC).

Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL), is disease of the arteries in the brain, which causes tissue loss in the subcortical region of the brain and the destruction of myelin in the CNS. CARASIL is characterized by symptoms such as gait disturbances, hair loss, low back pain, dementia, and stroke. In rare, recessively inherited CARASIL the clinical picture and white matter changes are similar as in CADASIL, but cognitive decline begins earlier. In addition, gait disturbance, low back pain and alopecia are characteristic features. Individuals with CARASIL may experience spondylosis and alopecia beginning in their teens, although alopecia is not seen in all patients.

T2 fluid-attenuated inversion recovery (FLAIR) hyperintensities involving the white matter of the anterior temporal poles (the O'Sullivan sign) seen in 90% of patients of CADASIL is not seen in CARASIL.

Answer 6

c) SMART syndrome

Stroke-like migraine attacks after radiation therapy (SMART) syndrome is a rare condition that involves complex migraines with focal neurologic findings in patients following cranial irradiation for central nervous system malignancies. Patients usually present years after radiation therapy (6-30 years in a case series) with seizures and subacute stroke-like episodes with symptoms such as hemiplegia, aphasia, and hemianopia. These episodes have been associated with headaches and are often preceded by a migraine-like aura. The hallmark of SMART syndrome is prominent unilateral gyral enhancement with mild mass effect, usually in an area included in the radiation ports. It is also observed as cortical thickening (hyperintense in T2 and FLAIR) with or without diffusion restriction. Typically, the condition is self-limiting and gradually resolves over the course of several weeks. Little is known about the mechanisms behind the disorder, making successful treatment challenging.

Differential diagnosis includes tumor recurrence, leptomeningeal carcinomatosis, infection, vascular disorders, mitochondrial encephalomyopathy with lactic acidosis and stroke, hemiplegic migraine, posterior reversible encephalopathy and post-ictal MRI changes.