Answers to picture quiz

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1. **Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes** – abbreviated to MELAS – is one of the family of mitochondrial cytopathies, which also include MERRF, and Leber's hereditary optic neuropathy. It was first characterized under this name in 1984. A feature of these diseases is that they are caused by defects in the mitochondrial genome which is inherited purely from the female parent. The disease can manifest in both sexes. Muscle biopsy of a person diagnosed with MELAS shows (a) Modified Gomori trichrome stain showing several ragged red fibers (arrowhead). (b) Cytochrome c oxidase stain showing Type-1 lightly stained and Type II fibers, darker fibers, and a few fibers with abnormal collections of mitochondria (arrowhead). Note cytochrome c oxidase negative fibers as usually seen in mitochondrial encephalopathy, lactic acidosis and stroke-like episodes (MELAS). (c) Succinate dehydrogenase staining showing a few ragged blue fibers and intense staining in the mitochondria of the blood vessels (arrow). (d) Electron microscopy showing abnormal collection of mitochondria with paracrystalline inclusions (arrowhead), osmiophilic inclusions (large arrowhead) and mitochondrial vacuoles (small arrowhead).

2. **CADASIL ("Cerebral Autosomal-Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy"**) is the most common form of hereditary stroke disorder, and is thought to be caused by mutations of the *Notch 3* gene on chromosome 19. The disease belongs to a family of disorders called the Leukodystrophies. The most common clinical manifestations are migraine headaches and transient ischemic attacks or strokes, which usually occur between 40 and 50 years of age, although MRI is able to detect signs of the disease years prior to clinical manifestation of disease. The definitive test is sequencing the whole Notch 3 gene, which can be done from a sample of blood. However, as this is quite expensive and CADASIL is a systemic arteriopathy, evidence of the mutation can be found in small and medium-size arteries. Therefore, skin biopsies are often used for the diagnosis.

3. **Cogan syndrome**

   Cogan syndrome or disease is a rare predominantly affecting young to middle aged adults. It is considered to be part of the spectrum of vasculitis involving small and medium size vessels in multiple organ systems. Ocular manifestations include interstitial keratitis, episcleritis, scleritis and retinal vasculitis. Hearing loss and vestibulo-auditory dysfunction, systemic vasculitis (aortitis, inflammatory bowel disease and pericarditis) and neurological manifestations such as neuropathy and mental changes are the usual manifestations. Stroke including cerebral venous thrombosis are rare complications of Cogan syndrome.

   The MRI venogram shows cerebral venous thrombosis. Limbal stromal opacity and episcleritis is evident in the eye.

4. **HEARNS disease** (Hereditary Endotheliopathy with Retinopathy Nephropathy and Stroke).

   HEARNS is an autosomal dominant, multisystem disease presenting with leukoencephalopathy, progressive visual loss, nephropathy and stroke. Initial presentation is progressive visual loss during the third and fourth decade followed by focal neurological deficits within 4 to 10 years.

   MRI shows an ischaemic stroke and the retinal photograph shows retinopathy.

5. **Susac syndrome**

   Susac syndrome is a triad of hearing loss, branched retinal artery occlusion and encephalopathy. A rare autoimmune condition that affects small vessels of the brain, retina and cochlea. This syndrome has a female predilection and usually presents between 20 and 40 years.

   MRI findings show infratentorial and supratentorial small multifocal white matter lesion with corpus callosum involvement. These lesions do not enhance with gadolinium. Cortical micro infarctions are seen.

   The MRI shows multiple small white matter lesions and micro infarcts. Fundal photograph shows branch retinal artery occlusion.

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