Opinion

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Retinal Vasculopathy with Cerebral Leukoencephalopathy

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Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations (RVCL-S) is an autosomal dominant microvasculopathy of the brain, retina, and other organ systems. Retinal vasculopathy is a term for retina disorders that are associated with disease of the blood vessels in the eye. RVCL-S encompasses several previously described conditions including cerebroretinal vasculopathy (CRV), hereditary vascular retinopathy (HVR), hereditary systemic angiopathy (HAS), hereditary endotheliopathy, retinopathy, nephropathy and stroke (HERNS), and retinal vasculopathy with cerebral leukodystrophy (RVCL) RVCL-S is an autosomal dominant disorder caused by C-terminal frameshift mutations in the three prime repair exonuclease 1 (*TREX1*) gene, located on the short arm of chromosome.

The *TREX1* gene normally encodes for a DNA exonuclease that is involved in removing unnecessary nucleotides from DNA. Furthermore, the *TREX1* gene also plays a role in protein glycosylation. It is unclear which of these functions is important to lose in the development of RVCL-S. The causes of retinal vasculopathy are most commonly high blood pressure, ischemia, and neovascularization. These occur in such disorders as diabetes, macular degeneration, retinal occlusions and more. Leukoencephalopathy with vanishing white matter is a progressive disorder that mainly affects the brain and spinal cord (central nervous system). This disorder causes deterioration of the central nervous system's white matter, which consists of nerve fibers covered by myelin.

Importantly, these mutations in *TREX1* are distinct from those that cause Aicardi-Goutiéres syndrome and some cases of hereditary systemic lupus erythematosus. Affected tissue, such as cerebral white matter, demonstrates ischemia, necrosis, and dystrophic calcifications, with accompanying vasculopathy. This vasculopathy, affecting primarily small to medium sized vessels, manifests as fibrinoid vascular necrosis or thickened hyalinised vessels, notably without evidence of vasculitis. In the brain, the leptomeninges, extraparenchymal vasculature, and the cortical grey matter vessels are typically spared.

Diagnosing retinal vascular disorders requires a dilated eye examination performed by a retinal specialist, testing which includes fluorescein angiogram and optical coherence tomography, and a complete medical history. Treatment for retinal vasculopathy will depend on the cause, however, you commonly will benefit from injections of medication into the eye, that reduce swelling, and cause regression of abnormal blood vessels. A healthy lifestyle and regular medical examinations may help you prevent many complications of retinal vasculopathy.

There is no sure way to prevent all forms of this disorder. Treatment is usually successful in slowing or stopping the progression of retinal vessel disorders. Retinal vasculopathy with cerebral leukoencephalopathy and systemic manifestations (RVCL-S) is an autosomal dominant inherited angiopathy caused by mutations in the three-prime repair exonuclease 1 (*TREX1*) gene. RVCL-S is an under-recognized disorder. The main causes of morbidity and mortality are vascular retinopathy, focal neurological complaints including ischemic events and cognitive decline, and kidney failure. The retina has been increasingly investigated as a site of Alzheimer's disease (AD) manifestation for over a decade.

