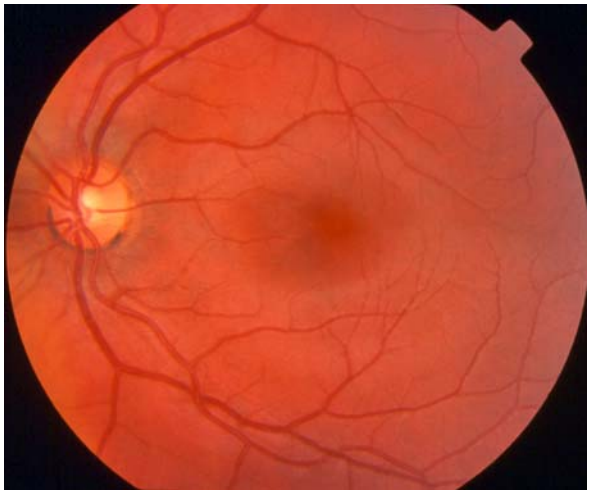
What You Should Know About Familial Exudative Vitreoretinopathy

By David J. Browning, MD, PhD

During development of the eye while a person is forming in the womb, the retina becomes vascularized. The retina is the neural lining of the back of the eye where light is transformed into a nerve signal, which travels to the brain.

Figure 1. Photo of Normal Retina



The process begins with blood vessel growth outward from the optic disk. The peripheral retina receives blood vessels last, and the temporal retina – that part closest to the patient's side – receives blood vessels after the nasal, superior, and inferior retina. Figure 1 shows the normal retina and its blood vessels so that you can visualize the process we are describing. In certain diseases, the orderly progression of blood vessel growth in the retina is interrupted and deranged. One rare and genetically determined cause for deranged retinal blood vessel growth is familial exudative vitreoretinopathy (FEVR). In this condition, the temporal retina fails to vascularize. Instead, the vessels stop advancing, often sprout abnormal endings which can leak fluid into the surrounding tissue or even bleed, and may produce scar tissue, which can then contract and cause retinal distortion and detachment. Figures 2 and 3 show examples of the retina of a patient with FEVR.

Figure 2. Retina with FEVR

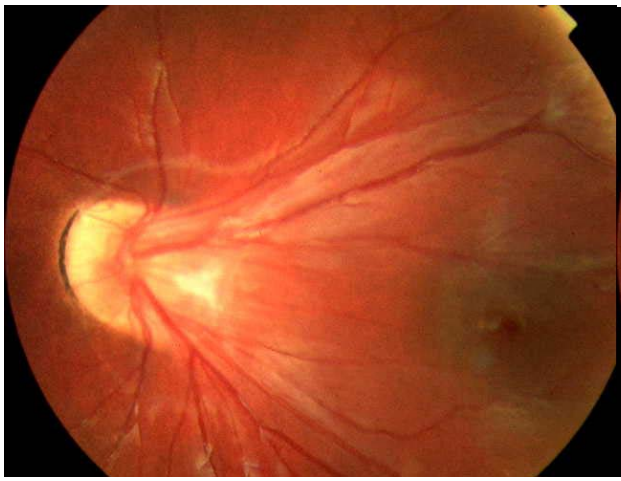
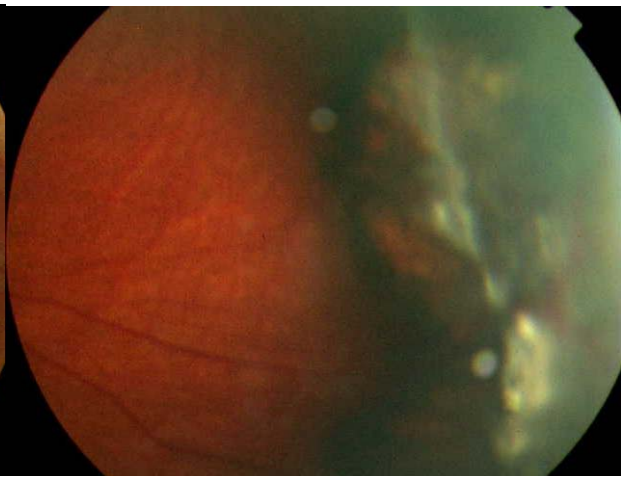


Figure 3. Temporal Retina of Same Eye



The Genetic Basis for Familial Exudative Vitreoretinopathy

Two forms of familial exudative vitreoretinopathy have been identified. Neither is common, but the more common of the two is caused by a mutation in the DNA of chromosome 11. The exact gene and its product enzyme have not been determined, but we know that a mutation in a single chromosome 11 will cause the disease – a characteristic of an autosomal dominant condition. This means that on average half of the offspring of a patient with FEVR will also be affected. It is always wise to screen as many family members of a patient with FEVR as possible, both to help potentially unsuspecting affected persons, and to allow for genetic counseling of the patient being treated.

A second, rarer form of the disease is caused by a DNA mutation on the X chromosome. Such patients will generally be males, since the male has but one X chromosome. The mother of an affected male carries one mutated chromosome, but has an intact second X chromosome, which prevents disease expression in her. On average half the sons of a female carrier will have the disease. All of the daughters of an affected male will be carriers of the disease, but will not show disease manifestations.

Possible Masqueraders

Certain other eye diseases can look like FEVR, but are distinct. Retinopathy of prematurity, caused by premature birth, low birth weight, and early exposure to elevated oxygen concentration, can cause eye findings similar to those in FEVR. Therefore, all

patients with FEVR should be questioned regarding birth history and neonatal exposure to oxygen.

Incontinentia pigmenti (IP) is a condition lethal to males but not to females. Retinal scarring, new vessels, and bleeding can occur in incontinentia pigmenti as in FEVR, but distinctions exist, including mental retardation, dental abnormalities, and skin lesions in IP but not in FEVR.

Treatment of FEVR

Patients with FEVR need to be monitored at regular intervals over time. If progressive leakage or bleeding develops, laser treatment of the retina having less circulation than normal may arrest the process. If scar tissue develops with retinal traction detachment, surgery to remove that scar tissue, called a vitrectomy, may help. Because the retina may be distorted in FEVR, patients may develop amblyopia, or “lazy eye”, in childhood, and may need patching of the good eye to reverse this neurologic process and allow the full visual potential to be reached.

FEVR may be unilateral or bilateral, and change can occur over years of follow-up. Diligent follow-up care is important in all patients with FEVR.

Final Comments

FEVR is a rare, serious, and potentially progressive genetic disease affecting the eye in isolation. Family members should be screened and affected patients closely followed over time. After reading this brochure, we encourage you to browse our website, including the Frequently Asked Questions section and the Forums, where patients may share their experiences with one another. If you have a focused question for which you cannot find an answer, we welcome you to ask Dr. Browning at: ask@theretinaexchange.com. Also, an excellent resource for medical literature is Pubmed, on the National Library of Medicine website, accessible at www.pubmed.com.