The exact cause of Wyburn-Mason syndrome is unknown. It is considered a developmental abnormality characterized by arteriovenous malformations. No specific genetic abnormality or hereditary tendencies have been identified. The specific, underlying mechanism(s) that cause the vascular malformations in Wyburn-Mason syndrome are not known. However, they are thought to result from abnormalities of blood vessel development during embryonic or fetal growth. Wyburn-Mason syndrome is an extremely rare disorder that appears to affect males and females in equal numbers. The incidence or prevalence rates of Wyburn-Mason syndrome in the general population are unknown. A diagnosis of Wyburn-Mason syndrome may be made based upon a thorough clinical evaluation, a detailed patient history, and identification of characteristic findings, especially ocular findings. Imaging studies such as a computed tomography (CT) scan or magnetic resonance imaging (MRI) may be performed to detect potentially dangerous central nervous system (CNS) malformations. During CT scanning, a computer and x-rays are used to create a film showing cross-sectional images of certain tissue structures. An MRI uses a magnetic field and radio waves to produce cross-sectional images of particular organs such as the brain.