



# Wyburn-Mason Syndrome



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#### LEARNING OBJECTIVES

1. Describe the anatomy and physiology of Wyburn-Mason syndrome
2. Discuss the diagnosis of Wyburn-Mason syndrome
3. Identify conditions associated with Wyburn-Mason syndrome



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Wyburn-Mason syndrome is a rare nonhereditary congenital disease characterized by abnormal arteriovenous anastomoses involving both the retina and midbrain. Similar arteriovenous malformations can also occur in the orbit, and less commonly in the face, skin, maxilla, or mandible.

This condition is classified as one of the phakomatoses, a group of disorders featuring congenital hamartomatous malformations affecting the eyes, skin, and central nervous system. Other diseases in this group include neurofibromatosis, tuberous sclerosis, von Hippel-Lindau disease, and Sturge-Weber syndrome. These disorders can produce significant visual and neurologic disturbances (Kerrison, 2005). The early ocular presentations discovered in some of these conditions may lead to identification of additional systemic implications (Chaudhuri & Kathikeyan, 2012).

#### Clinical Features

The prominent ocular feature of Wyburn-Mason syndrome is the presence of unilateral arteriovenous anastomoses of the retina. Arteriovenous anastomoses are congenital vascular

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malformations that arise due to disturbances in the embryonic maturation process of the retinal vascular system during early gestation. These anastomoses form a direct transition from arteries to veins without normal capillary connections (Wessing, 2007). There is no race or sex predilection for this condition. The appearance of the arteriovenous malformations in Wyburn-Mason syndrome may vary from a localized abnormality between an artery and a vein, to marked dilation and tortuosity of the retinal vessels. The malformations occur most often in the central or temporal retina and are readily apparent upon ophthalmoscopic examination. The retinal vascular abnormalities often remain stationary, but long-term observation has shown that remodeling of the malformations can occur in some cases (Ferry, 1994).

Larger arteriovenous malformations consisting of numerous convoluted and intertwined vessels are sometimes referred to as racemose hemangiomas. “Racemose” is a descriptive term meaning a cluster or bunch of grapes. This appearance has also been described as “like a bag of worms” (see Figure 1). The dilated vessels of a racemose hemangioma can reach a diameter of up to 10 times that of normal retinal vessels, extending through all layers of the retina and often protruding toward the vitreous body (Francois, 1972; Wessing, 2007; Ferry, 1994).

Large vessel diameter is believed to result in an abnormally high flow rate through the anastomosis. Both the arteries and veins in these malformations carry oxygenated blood and consequently appear similar in color, adding to the difficulty in distinguishing the arteries from the veins. Although markedly dilated, the vessels do not exhibit pulsations. Increased blood flow through the large-diameter vessels is believed to be the cause of damage to the surrounding capillary bed, causing avascular zones. Unlike other retinal vascular diseases with capillary dropout, these avascular zones do not appear to stimulate neovascularization (Wessing, 2007).

There seems to be a correlation between the severity of retinal changes and the prevalence of cerebral involvement. The larger the retinal arteriovenous malformation, the greater the chance it is associated with an arteriovenous communication in the central nervous system, face, or skin (Brown, 1999). Studies have shown that approximately 90% of patients with large racemose retinal malformations were found to have similar vascular abnormalities of the central nervous system (Ferry, 1994). Concurrent cerebral malformations and retinal anastomoses in Wyburn-Mason syndrome are ipsilateral in location.

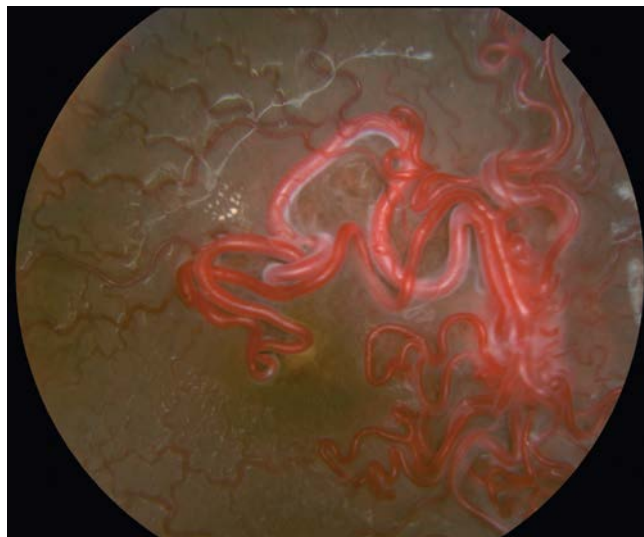


Figure 1. The classic racemose hemangioma found in Wyburn-Mason syndrome demonstrates dilated tortuous vessels that can be 10-12 times larger than normal. The vessels often appear similar in color, making it difficult to differentiate between arteries and veins.

Approximately 40% of patients with retinal and central nervous system anastomoses exhibit high-flow arteriovenous malformations of the maxillofacial or mandibular regions. Skin lesions have been found in about 25% of patients with this syndrome (Brown, 1999).

### Symptoms/Complications

Arteriovenous anastomoses represent a high-pressure hemodynamic system. Several of the complications and symptoms in Wyburn-Mason syndrome are presumably the result of hemodynamic stress. Visual symptoms can range from relatively normal to severely impaired. Vision loss can occur secondary to abnormal vessels in the macula, vitreous hemorrhage, retinal hemorrhage, macular hole formation, microvascular decompensation, or compressive nerve fiber layer loss (Harbour & Bonsall, 2002). Retinal vein occlusion may occur, leading to neovascular glaucoma. These occlusions are believed to result from partial thrombosis within the anastomosis or from enlargement of the malformation and direct compression of the central retinal vein (Kerrison, 2005).

Concurrent vascular abnormalities in the orbit can cause proptosis, dilated conjunctival vessels, papilledema, optic atrophy, and loss of vision. Neurological symptoms due to compression and bleeding in the midbrain include cranial nerve palsies, epileptic fits, strabismus, and visual field defects (Wessing, 2007). Homonymous hemianopia has been demonstrated in at least 30% of Wyburn-Mason syndrome patients as a result of vascular malformations of the optic track (Ferry, 1994). Visual

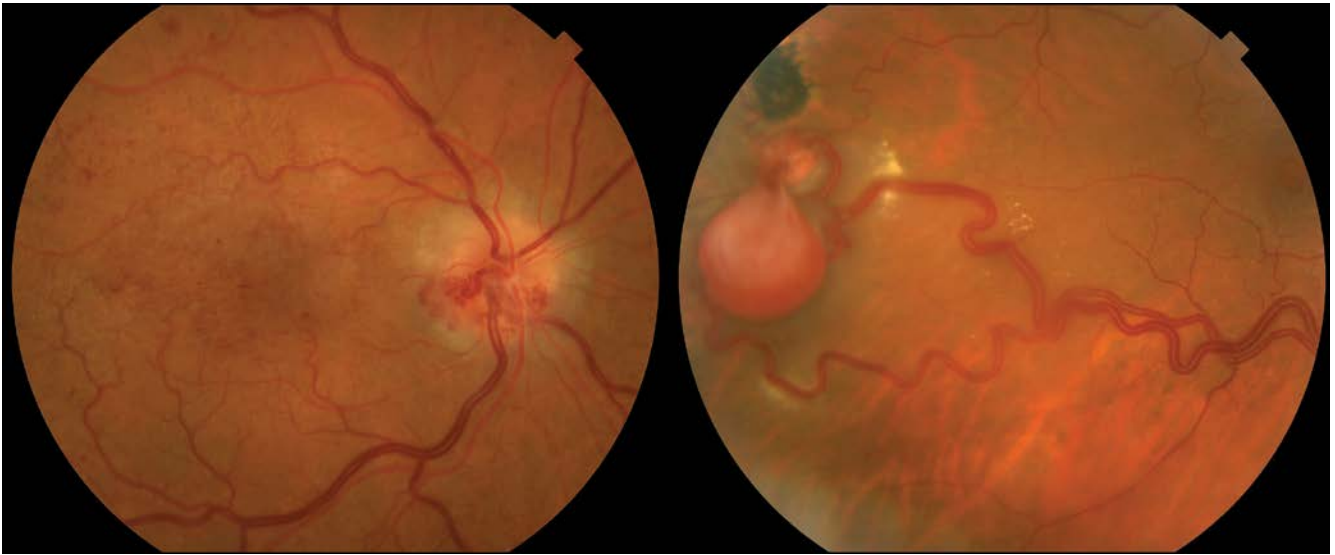


Figure 2. Other conditions that can cause dilation of vessels include central retinal vein occlusion (CRVO) and von Hippel-Lindau disease. Left: Resolving CRVO with dilated veins and collateral vessels on the optic nerve. Note that the arteries appear normal in caliber. Right: Dilated vessels feeding a capillary hemangioma in von Hippel-Lindau disease.

field testing, however, may not differentiate a neurological cause from a retinal one, as both may coexist (Chaudhuri & Karthikeyan, 2012).

Vascular malformations in the area of the maxilla, mandible, nasopharynx, buccal mucosa, or palate can cause epistaxis and hemorrhage (Brown, 1999; Wessing, 2007).

### Differential Diagnosis

Dilated and tortuous vessels may be present in a number of retinal conditions, but those found in Wyburn-Mason syndrome are usually distinctive in appearance. Central retinal vein occlusion can cause dramatic dilation and tortuosity of the retinal vasculature, but this is often accompanied by widespread hemorrhage and edema. Primary congenital vascular tortuosity

differs from Wyburn-Mason syndrome in that it is usually bilateral and the arteriovenous connection is via a normal capillary plexus (Wessing, 2007). Congenital retinal macrovessel is a rare finding of an aberrant large macular vessel with normal capillary communications.

The retinal anastomoses in Wyburn-Mason syndrome can resemble the dilated feeder vessels of retinal capillary hemangiomas found in von Hippel-Lindau disease (VHL), another one of the phakomatoses (see Figure 2). There are, however, several distinguishing differences between the arteriovenous malformations in these two diseases. Wyburn-Mason malformations are usually found in the posterior pole of the fundus, while VHL lesions can be located anywhere in the retina, including the far periphery. The arterial and venous components of VHL lesions are separated by the capillaries of the hemangioma. They are often multifocal and bilateral, and can cause lipid exudation or exudative retinal detachment. Leakage and exudation are not typical in Wyburn-Mason syndrome.

### Diagnostic Testing

Retinal arteriovenous anastomoses are typically discovered during routine ophthalmoscopic examination. Fundus photography may be used to document the location and extent of the dilated, tortuous vessels. Photo montage software or an ultrawide-field imaging device is often needed to document the full extent of the vascular lesion in a single image (see Figure 3). Fluorescein angiography can also be used to document the dilated, tortuous vessels as the characteristic filling pattern of

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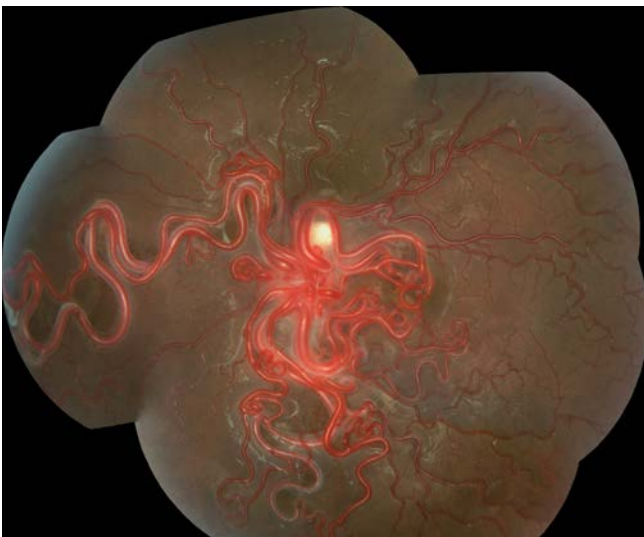


Figure 3. Photo montage of an extensive arteriovenous malformation in Wyburn-Mason syndrome.

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Figure 4. Fluorescein angiogram of an arteriovenous malformation (AVM) demonstrates very rapid filling of the AVM, while the nearby normal vein exhibits laminar flow and a more normal filling time.

the arteriovenous malformation. Early-phase fluorescein images will typically demonstrate a rapid transit through the vascular loops, with nearly simultaneous filling of the arterial and venous components of the malformation in the absence of a normal intervening capillary bed (see Figure 4). The flow rate through the anastomoses is relative to the diameter of the vessels; the larger the diameter, the higher the flow rate (Wessing, 2007). Typically there is no appreciable fluorescein leakage from the convoluted vessels.

Optical coherence tomography shows an irregular retinal surface with optical densities corresponding to the enlarged retinal vessels (Shields & Shields, 2008). Spectral domain optical coherence tomography (SD-OCT) typically shows a pattern of oval-shaped lesions that correspond to cross-sections of the enlarged, abnormal vessels (Fileta, Bennett, & Quillen, 2014) (see Figure 5). Increased thickness of the choroid and enlarged choroidal vessels have been reported in an eye with Wyburn-Mason syndrome using enhanced depth imaging (EDI) spectral domain OCT (Iwata et al., 2015).

Patients with Wyburn-Mason syndrome often have visual field abnormalities related to either retinal or intracranial abnormalities, with some 30% demonstrating homonymous hemianopia (Wessing, 2007).

### Management

Wyburn-Mason syndrome is typically nonprogressive, and currently there are no available treatment options for the primary vascular lesion. Visual acuity should remain stable, but regular

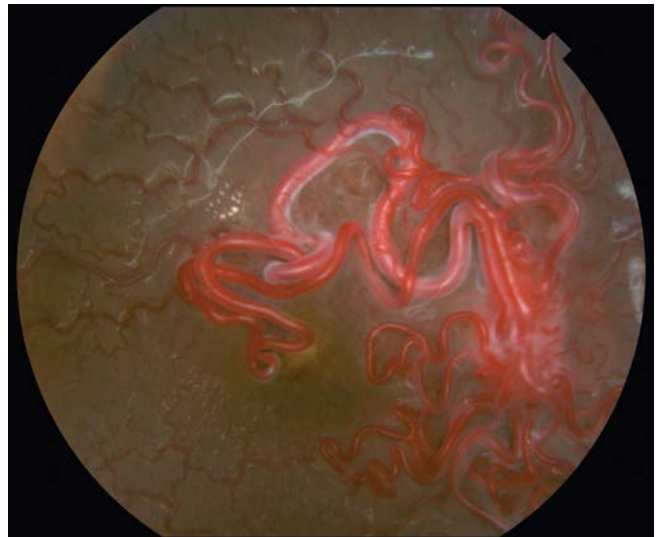


Figure 5. Spectral domain optical coherence tomography (SD-OCT) demonstrates a dramatic pattern of circular lesions corresponding to cross-sections of abnormal retinal vessels in Wyburn-Mason syndrome.

observation for secondary ocular complications is warranted. Standard therapy for complications such as retinal vein occlusion, neovascular glaucoma, or nonresolving vitreous hemorrhage may be recommended.

Patients identified with retinal arteriovenous malformations are typically referred for comprehensive neurological evaluation. Magnetic resonance imaging or computed tomography scans are often ordered to evaluate for concurrent central nervous system vascular malformations. Patients are advised that dental extractions should be approached with extreme caution because

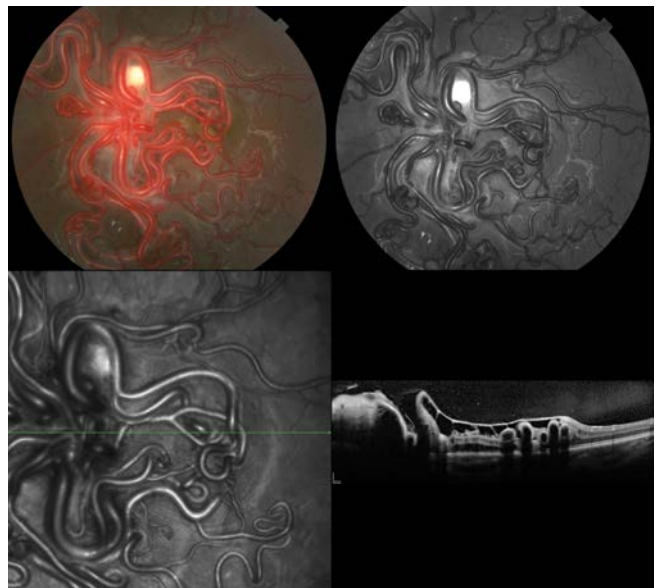


Figure 6. Clinical appearance of an arteriovenous malformation in a nine-year-old girl diagnosed with Wyburn-Mason syndrome.

of the possibility of severe bleeding from an arteriovenous communication located in the maxilla or mandible (Harbour & Bonsall, 2002).

### What's in a Name?

The history of initial identification and description of this syndrome is quite interesting and illustrates some of the pitfalls in using eponyms as a naming convention. Eponyms are conditions or findings that are named for a person, presumably the first person to discover or describe the finding. They are a long-standing tradition in science and medicine, and being awarded an eponym is considered an honor. But history shows that eponyms can be controversial or problematic and may lead to some confusion. For years there has been sparring in the literature over the use of eponyms in ophthalmology (Bennett, 2015). In addition to debate over who may have been first to identify a condition, eponyms often become outdated, or may not adequately describe the named condition (Cogan, 1978). Debate has also occurred over the perceived worthiness of the designee due to ethical considerations. Some authors recommend eliminating the use of eponyms entirely (Pulido & Matteson, 2010).

As one of the group of diseases known as the phakomatoses, Wyburn-Mason syndrome is a classic example of the use of an eponym to identify a rare condition. In fact, most of the phakomatoses are known by eponymous names, including von Recklinghausen's disease (neurofibromatosis), Bourneville's disease (tuberous sclerosis), von Hippel-Lindau disease, Sturge-Weber syndrome, and others. An association between

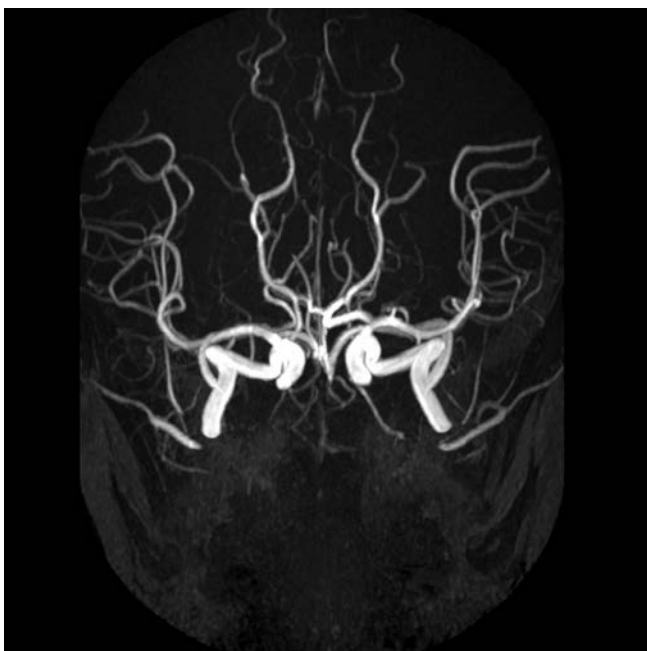


Figure 7. Magnetic resonance angiography showed no evidence of an arteriovenous malformation in the intracranial circulation.

arteriovenous malformations of the face, retina, and brain was first reported in the French medical literature in 1937 (Bonnet, Dechaume, & Blanc, 1937). In an extensive study six years later, British physician Roger Wyburn-Mason reviewed all previously reported cases and described nine additional examples (Wyburn-Mason, 1943). The association of retinal, facial, and cerebral vascular malformations became known as Bonnet-Dechaume-Blanc syndrome in France and continental Europe, and Wyburn-Mason syndrome in the English literature. Often these eponyms are used interchangeably, but sometimes Bonnet-Dechaume-Blanc syndrome is preferred for more severe cases with maxillofacial involvement.

In an attempt to eliminate confusion in the use of these eponyms, a new name that reflects the current understanding of this condition has been proposed: cerebrofacial arteriovenous metameric syndrome (CAMS) (Bhattacharya et al., 2001). To date, this name has not been universally adopted, and the condition is still most commonly referred to as Wyburn-Mason syndrome.

### Case Report

A nine-year-old girl was referred for a comprehensive eye exam by her school nurse after failing a vision screening in the left eye. Upon presentation, the patient had no visual complaints. Visual acuity measured 20/20 OD and 20/100 OS. Intraocular pressures were 18 and 16. Ocular motility and slit lamp examination were normal. No afferent pupillary defect was detected.

Dilated fundus examination was normal in the right eye. Examination of the left fundus revealed markedly dilated vascular loops consistent with a racemose hemangioma. The vascular loops extended from the optic nerve into the macula, as well as the nasal periphery (see Figure 6). SD-OCT demonstrated a dramatic pattern of circular lesions corresponding to cross-sections of abnormally dilated retinal vessels. Fluorescein angiogram was deferred at the parents' request.

The patient was referred for MRI/MRA to rule out intracranial arteriovenous malformations. Imaging was within normal limits (see Figure 7). Given the size of the retinal lesion, it was surprising that no other malformations were found in the central nervous system. The patient's parents were advised that there were no available treatment options for the retinal lesion, but that visual acuity should remain stable. Regular monitoring for secondary ocular complications was recommended. Although the arteriovenous malformation of the left retina fit the classic description of a racemose hemangioma, the absence of similar

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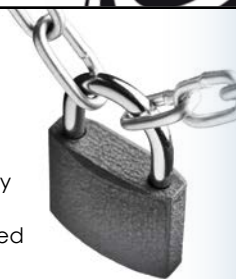
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orbital, facial, or cerebral lesions does not meet the true classification of Wyburn-Mason syndrome. Some authors however, believe that Wyburn-Mason syndrome may represent a continuous spectrum of disease without all the classic findings and that partial manifestations of the syndrome such as this case are possible (Madey, Lehman, Chaudry, & Vaillancourt, 2012).

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## POST-TEST QUESTIONS

- Wyburn-Mason syndrome is typically described as:
  - Rare, nonhereditary, congenital
  - Rare, hereditary, acquired
  - Common, nonhereditary, acquired
  - Common, hereditary, congenital
- Wyburn-Mason syndrome involves the following two anatomical structures:
  - Cornea and midbrain
  - Retina and midbrain
  - Retina and medulla
  - Cornea and medulla
- Wyburn-Mason syndrome malformations occur most often in the following area of the retina:
  - Ventral
  - Lateral
  - Medial
  - Central
- What is the major cause of complications associated with Wyburn-Mason syndrome?
  - High hemodynamic pressure
  - Increased intraocular pressure
  - Low hemodynamic pressure
  - Decreased intraocular pressure
- Which of the following tests usually identifies retinal arteriovenous anastomoses?
  - Photo montage
  - Fundus photography
  - Routine ophthalmic exam
  - Fluorescein angiography
- A common visual field abnormality related to Wyburn-Mason syndrome is:
  - Optic neuritis
  - Central scotoma
  - Optic neuropathy
  - Homonymous hemianopsia
- What additional tests are typically ordered for patients with retinal arteriovenous malformations?
  - MRI or CT scan
  - EKG or echocardiogram
  - EEG or evoked potentials
  - Ultrasound or angiography
- Approximately 40% of patients with retinal or central nervous system arteriovenous malformations also have them in the following anatomical location:
  - Face
  - Lungs
  - Spine
  - Heart
- The following name has been proposed to describe retinal arteriovenous malformations:
  - Von Hippel-Lindau syndrome
  - Roger Wyburn-Mason syndrome
  - Cerebrofacial arteriovenous metamerism syndrome
  - Bonnet-Dechaume-Blanc syndrome
- Treatments for Wyburn-Mason syndrome malformations include the following:
  - Angioplasty for occluded veins
  - Surgical removal of the malformation
  - Regular eye and neurological examinations
  - Palliative care consultation since there are no treatment options

ASORN members and other eye care professionals now have an online source for continuing education and training: EyeCareCE. The site is the collaborative effort of five organizations. Together, the International Joint Commission on Allied Health Personnel in Ophthalmology (JCAHPO), the American Society of Ophthalmic Registered Nurses (ASORN), the Association of Technical Personnel in Ophthalmology (ATPO), the Canadian Society of Ophthalmic Medical Personnel (CSOMP), the American Academy of Ophthalmic Executives (AAOE), the Contact Lens Society of America (CLSA), and the Ophthalmic Photographer's Society (OPS), teamed to produce a comprehensive online training resource for eye care professionals. ASORN is proud to participate in this online educational partnership.