Chapter 3
Vascular Malformations of the Retina and Posterior Segment of the Eye

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3.1 Macular Telangiectasia

Macular telangiectasia, also known as idiopathic juxtafoveal telangiectasia, is a vascular abnormality that affects the capillaries in the posterior pole. There are two types of macular telangiectasia: type 1, which is a congenital unilateral vascular anomaly and may be a part of the spectrum of Coats disease, and type 2, which is typically bilateral with onset in middle or older age. Macular telangiectasia is a rare vascular condition, and the incidence of macular telangiectasia type 2 ranges from 0.0045 to 0.1 % [1, 2]. Visual disturbances are typically mild, usually consisting of mild metamorphopsia or a scotoma [3, 4]. Visual acuity may decline with progression of the disease, but visual acuity less than 20/200 is rare [5, 6]. The earliest sign is a loss of temporal retinal transparency, or a grayish discoloration, followed by dilation of temporal parafoveal capillaries [5, 6]. Abnormal capillaries can be noted to make a right-angle turn diving deeper into the retina. Later on, retinal pigment epithelium migration and hyperplasia along abnormal capillaries can be noted as well as retinal atrophy [5, 6]. Perifoveal crystalline deposits may be seen (Fig. 3.1). The diagnosis can be made with fundus autofluorescence imaging, which reveals a loss of the hypofluorescent center of the macula due to the depletion of macular pigment. This change is one of the earliest signs and is diagnostic of the disease [7]. Fluorescein angiography shows telangiectatic capillaries, beginning temporal to the macula, with late staining [6]. Optical coherence tomography shows an early enlargement of the temporal side of the foveal pit followed by

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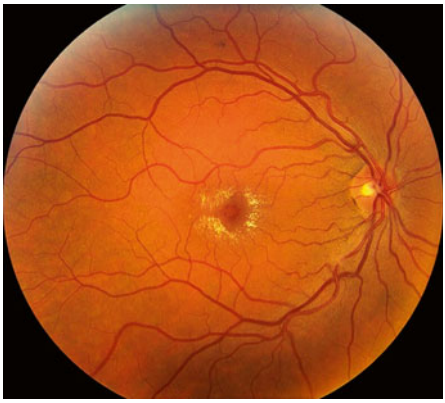

Fig. 3.1 Perifoveal crystalline deposits in macular telangiectasia type 2

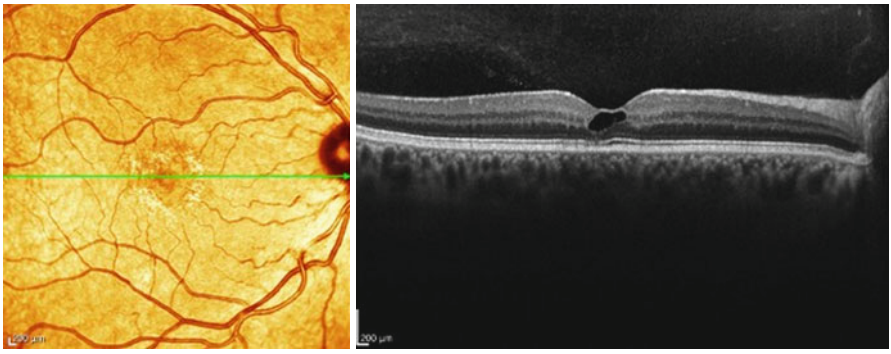

Fig. 3.2 Optical coherence tomography of the macula in macular telangiectasia type 2 showing pseudocystic spaces in the fovea caused by loss of tissue

disruption in the outer retinal and photoreceptor layers (Fig. 3.2). The development of retinal cavities and hyporeflective spaces at times leaving only the overlying internal limiting membrane, without corresponding leakage on fluorescein angiography, occurs with disease progression due to loss of retinal layers [7]. The most feared complication is the development of retinal neovascularization. Currently, there is no effective treatment of the disease. In cases of retinal neovascularization, the mainstay of treatment is intravitreal injections of anti-vascular endothelial growth factor (anti-VEGF). The visual prognosis is good with 50 % maintaining visual acuity 20/32 or better [8].

3.2 Retinal Arterial Macroaneurysm

Retinal arterial macroaneurysms (RAMA) are fusiform outpouchings or dilatations of retinal arterioles, commonly at retinal bifurcations or arteriovenous crossings. Acquired RAMAs are generally associated with atherosclerosis and

hypertension and tend to occur in the sixth and seventh decade of life [9–12]. The location is usually in the posterior pole within the first three orders of arterial bifurcation, and 90 % are unilateral (Fig. 3.3). RAMAs can be associated with lipid exudation and hemorrhage in the subretinal, intraretinal, and preretinal space and can result in visual decline when involving the macula [13]. Fluorescein angiography demonstrates early arterial filling of the RAMA with a variable amount of late leakage (Fig. 3.4). If hemorrhage is present, the RAMA and surrounding leakage may be obscured on fluorescein angiography. Optical coherence tomography can delineate subretinal hemorrhage and intraretinal fluid and exudation. Treatment is not always necessary, because RAMAs can thrombose and spontaneously involute with clearance of associated lipid exudation [9]. When the macula is involved, treatment options include laser photocoagulation of the RAMA, pneumatic displacement for submacular hemorrhage, and vitrectomy for vitreous hemorrhage [14–16].

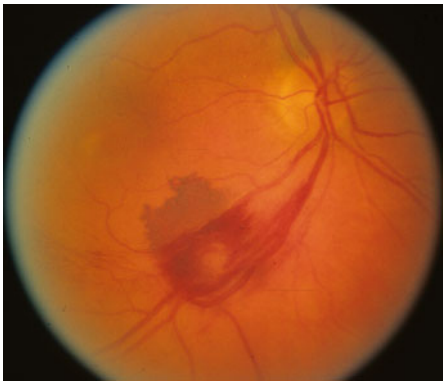

Fig. 3.3 Retinal arterial macroaneurysm surrounded by retinal hemorrhage (Image courtesy of Richard A. Lewis, M.D.)

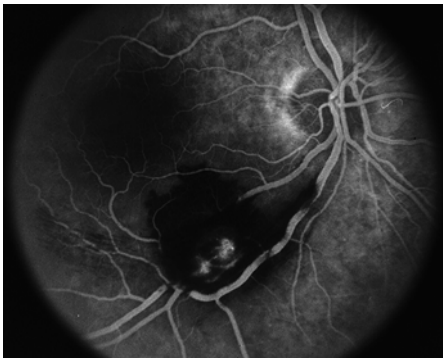

Fig. 3.4 Fluorescein angiography showing blockage owing to retinal hemorrhage and hyperfluorescence with late leakage from the aneurysm itself (Image courtesy of Richard A. Lewis, M.D.)

3.3 Coats Disease

Coats disease was originally described in 1908 by George Coats as an idiopathic condition with telangiectatic vessels and associated lipid exudation [17]. It affects males three times more frequently than females and is unilateral in 80–95 % of cases [18–20]. The average age of diagnosis is 5 years of age. Children most frequently present with decreased visual acuity, and in some cases, strabismus, leukocoria, heterochromia, nystagmus, or asymptomatically [19]. On fundus examination, telangiectatic vessels, subretinal lipid exudation, exudative detachments, retinal hemorrhage, retinal macrocyst, and optic disc neovascularization can be present (Fig. 3.5). Coats disease is typically slowly progressive. A staging system proposed by Shields et al. is summarized in Table 3.1 [19]. Fluorescein angiography reveals telangiectatic vessels and aneurysms with early and progressive leakage, peripheral retinal nonperfusion, and areas of capillary dropout. Coats disease must be differentiated from retinoblastoma, in which children can also present with leukocoria and exudative retinal detachments. Ultrasound sonography, computed tomography, and magnetic resonance imaging can aid with differentiating the two entities. Treatment of Coats disease consists of laser photocoagulation to leaking telangiectatic vessels in early stages of the disease [21–23]. Cryotherapy can also be used for peripheral lesions and lesions

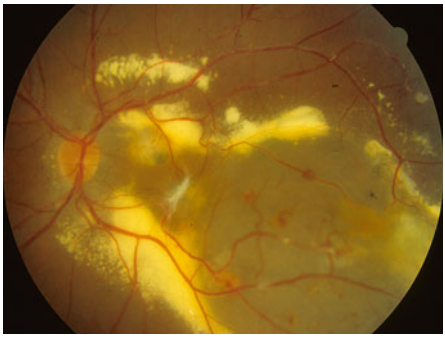

Fig. 3.5 Coats disease with telangiectatic vessels in the temporal macula leading to an exudative retinal detachment (Image courtesy of Richard A. Lewis, M.D.)

Table 3.1 Staging of Coats disease

Stage	Ocular findings
1	Retinal telangiectasias without exudation
2a	Retinal telangiectasias with extrafoveal exudation
2b	Retinal telangiectasias with subfoveal exudation
3a1	Subtotal exudative detachment, extrafoveal
3a2	Subtotal exudative detachment, involving the fovea
3b	Total exudative detachment
4	Exudative detachment with glaucoma
5	End stage

with a poor response to laser photocoagulation [24]. Treatment is often performed under anesthesia with multiple treatment sessions over the course of several months [25]. Intravitreal triamcinolone acetonide and anti-VEGF alone or in combination have also been reported to be effective in treating subretinal fluid and macular edema [26–32]. In severe cases, vitrectomy and subretinal drainage may be necessary [33–36]. Despite treatment, visual prognosis is poor due to damage to the macula. In one series, 47 % of cases had visual acuity ranging from hand motions to no light perception and another 29 % ranging from 20/200 to counting fingers [25].

3.4 Familial Exudative Vitreo-Retinopathy

Familial exudative vitreo-retinopathy (FEVR) is a bilateral ocular disease due to a genetic mutation that results in incomplete formation of the retinal vasculature. It is inherited in an autosomal dominant pattern, although X-linked transmission has been reported. The retinal findings can vary in severity, with earlier onset associated with more severe disease. On examination, ocular findings range from straightening of the retinal vessels and peripheral retinal nonperfusion to more severe forms with peripheral neovascularization, lipid exudation and subsequent exudative retinal detachments, and tractional retinal detachment as neovascular membranes contract. Treatment involves laser photocoagulation to peripheral avascular retina and vitrectomy for management of tractional retinal detachment.

3.5 Norrie Disease

Similar to FEVR, Norrie disease is due to a genetic mutation that results in incomplete development of the retinal vasculature. It is inherited in an X-linked recessive manner and can be associated with hearing loss and mental retardation. Retinal findings include peripheral retinal neovascularization, lipid exudation, and tractional retinal detachment. Treatment involves laser photocoagulation to peripheral avascular retina and vitrectomy for management of tractional retinal detachment.

3.6 Retinal Cavernous Hemangioma

Retinal cavernous hemangioma is a rare vascular hamartoma consisting of clumps of saccular aneurysms in the inner retina or on the optic nerve head (Fig. 3.6). It is typically unilateral and 1–2 disc diameters in size. The size typically does not change over time. There is no clearly visible feeding vessel, and

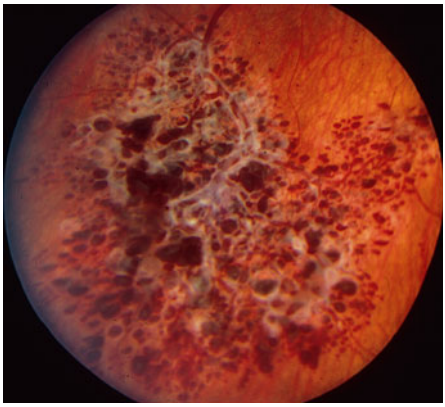

Fig. 3.6 Retinal cavernous hemangioma with grapelike clusters of malformed vessels (Image courtesy of Richard A. Lewis, M.D.)

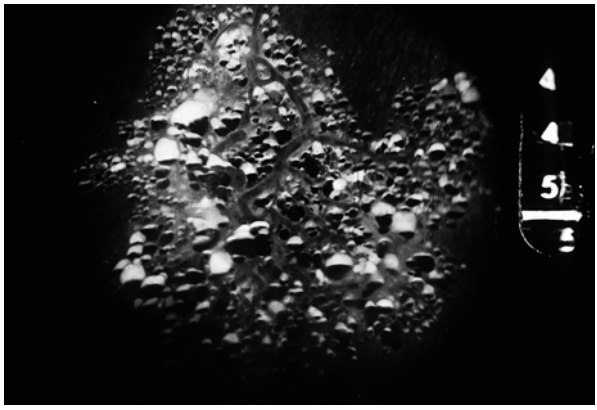

Fig. 3.7 Fluorescein angiography of the cavernous hemangioma showing the laying of dye in the saccular malformations (Image courtesy of Richard A. Lewis, M.D.)

the lesion is usually located over a vein. The aneurysms are usually filled with dark blood, giving the appearance of a "cluster of grapes." Retinal cavernous hemangioma is typically asymptomatic but has been reported to cause mild visual disturbance if located in the macula or if vitreous hemorrhage occurs [14]. Lipid exudation is rare due to the slow circulation through the aneurysms. Red blood cells can layer inferiorly within the aneurysm resulting in a plasma-erythrocyte separation. In some cases, retinal cavernous hemangiomas can be associated with cutaneous and central nervous system angiomatous lesions. Fluorescein angiography demonstrates an incomplete and slow filling of the aneurysms, with fluorescein often accumulating in the superior portion of the aneurysm while inferior accumulated red blood cells cause blockage (Fig. 3.7). Retinal cavernous hemangiomas typically do not require treatment. If vitreous hemorrhage occurs, the hemangioma can be treated with laser photocoagulation or cryotherapy.

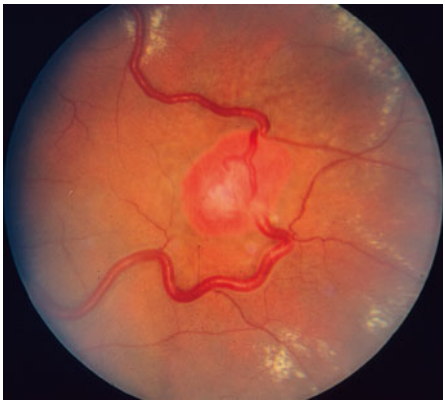

Fig. 3.8 Retinal capillary hemangioma in von Hippel-Lindau disease showing dilated feeder vessels and surrounding lipid exudate (Image courtesy of Richard A. Lewis, M.D.)

3.7 Retinal Capillary Hemangioma

Retinal capillary hemangioma is a vascular tumor that occurs as an isolated ocular lesion or part of a systemic condition known as von Hippel-Lindau disease, an autosomal dominant disorder with multiple benign or malignant lesions that occur in the retina, central nervous system, and visceral organs. Retinal capillary hemangiomas begin as small lesions, typically in the peripheral retina, that progressively enlarge over time with dilation of blood vessels to and from the lesion (Fig. 3.8). Lipid exudation from the lesion can result in an exudative retinal detachment. Late fibrosis of the lesion can cause a tractional retinal detachment [37]. Decreased vision occurs secondary to exudation in the macula and development of exudative and tractional retinal detachment. The diagnosis is made clinically, but ancillary testing can aid in diagnosis. Fluorescein angiography shows early leakage from the hemangioma that may persist or decrease with time. Ocular coherence tomography reveals the extent and amount of exudation. Treatment includes laser photocoagulation to smaller lesions and cryotherapy for larger, peripheral lesions [38–41]. Exudative and tractional retinal detachment can be managed with vitrectomy [42]. Photodynamic therapy, radiation therapy, and systemic anti-angiogenic agents have been used with limited success [43–49].

3.8 Retinal Arteriovenous Malformation (Racemose Hemangioma)

Retinal arteriovenous malformation is also known as racemose hemangioma. This is a vascular malformation in which direct anastomoses occur between the retinal arterial and venous circulation with dilation of retinal vessels (Fig. 3.9). It is nonhereditary and is associated with ipsilateral vascular malformations of the midbrain in Wyburn-Mason syndrome. There is typically no lipid exudation and no leakage on fluorescein angiography. The Archer classification scheme separates the vascular

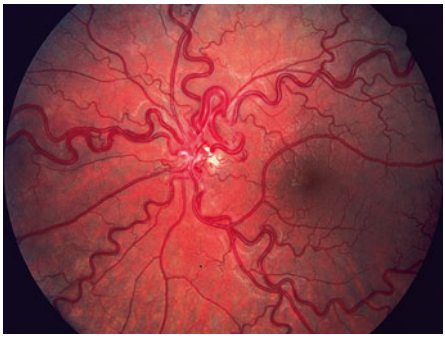

Fig. 3.9 Dilated and tortuous retinal vessels in the setting of Wyburn-Mason syndrome (Image courtesy of Richard A. Lewis, M.D.)

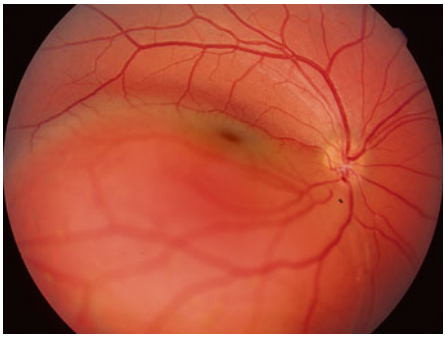

Fig. 3.10 Large choroidal hemangioma in a Sturge-Weber patient resulting in a serous retinal detachment affecting the macula (Image courtesy of Richard A. Lewis, M.D.)

malformations into 3 groups. Group 1 consists of an abnormal arteriovenous communication with an intervening capillary plexus and is typically asymptomatic. Group 2 is defined as a direct arteriovenous communication without intervening capillary vessels and with few visual symptoms. Group 3, the most severe form, consists of a complex arteriovenous communication with dilated and tortuous vessels commonly with associated visual loss [50]. Approximately 30 % of cases with retinal findings will have lesions in the central nervous system [51]. Racemose hemangioma does not require treatment.

3.9 Encephalofacial Hemangiomatosis

Encephalofacial hemangiomatosis (Sturge-Weber syndrome) is a nonhereditary disorder characterized by congenital hamartomas of the eye, brain, and skin. The most common central nervous system finding is a diffuse leptomeningeal hemangioma ipsilateral to the facial hemangioma or nevus flammeus (port-wine stain) [52]. Ocular findings include dilated epibulbar vessels, glaucoma, tortuous retinal vessels, retinal pigmentary changes, and diffuse choroidal hemangioma that results in a "tomato-catsup" fundus appearance. Diffuse choroidal hemangioma can lead to a total retinal detachment and secondary neovascular glaucoma (Fig. 3.10) [14]. On

B-scan ultrasonography, the choroidal lesion demonstrates high echogenicity. Choroidal hemangiomas that are minimally elevated can be observed. Refractive changes should be monitored closely to prevent amblyopia. Large tumors with elevation and retinal detachment can be treated with oral propranolol, photodynamic therapy, or external beam radiation [53].

3.10 Summary

Vascular malformations of the eye and retina are important to be recognized. Ocular findings can aid with the diagnosis and management of systemic disease. These disorders often require close monitoring to prevent or detect complications, and prompt treatment is required in some cases to preserve or improve vision.

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