Study Questions

Please note that these questions are not part of your CME reporting process. They are provided here for your own educational use and for identification of any professional practice gaps. The required CME posttest is available online (see "Requesting Continuing Medical Education Credit"). Following the questions are answers with discussions. Although a concerted effort has been made to avoid ambiguity and redundancy in these questions, the authors recognize that differences of opinion may occur regarding the "best" answer. The discussions are provided to demonstrate the rationale used to derive the answer. They may also be helpful in confirming that your approach to the problem was correct or, if necessary, in fixing the principle in your memory. The Section 12 faculty thanks the Resident Self-Assessment Committee for developing these self-assessment questions and the discussions that follow.

1. What is the main glycosaminoglycan component of vitreous?
   a. chondroitin
   b. dermatan
   c. hyaluronic acid (hyaluronan)
   d. keratan

2. Within the retina, the density of rods is greatest at what degree of fixation?
   a. 90
   b. 45
   c. 12
   d. 0

3. What is the most common systemic condition associated with angioid streaks?
   a. oculocutaneous albinism
   b. Crohn disease
   c. pseudoxanthoma elasticum
   d. ulcerative colitis

4. What symptom following intravitreal injection would be unexpected and warrant immediate evaluation?
   a. progressive floaters with progressive blurring of vision
   b. sharp pain following the injection
   c. foreign body sensation
   d. small floaters that are noticed immediately after injection

5. A patient with long-standing poorly controlled diabetes presents with vision loss (20/200) in 1 eye. Clinical examination shows mild inferior vitreous hemorrhage, minimal macular edema outside the central macula, and a well-perfused optic nerve. Fluorescein angiography demonstrates an area of neovascularization elsewhere and a foveal avascular zone diameter of 1500 μm. What is the most likely cause of the poor vision?
   a. ischemic maculopathy
   b. capillary leakage
   c. traction (tractional) detachment
   d. bleeding from neovascularization

6. What traumatic extraocular condition is associated with a branch retinal artery occlusion?
   a. pelvic fracture
   b. femur fracture
   c. subarachnoid hemorrhage
   d. pancreatitis

7. What is the most significant risk factor for central retinal vein occlusion (CRVO)?
   a. hypertension
   b. older age
   c. hypercoagulable state
   d. glaucoma

8. What treatment has been proven to be effective for branch retinal vein occlusion–associated macular edema?
   a. panretinal photocoagulation
   b. verteporfin
   c. fluocinolone acetonide intravitreal implant
   d. ranibizumab

9. What is the most common cause of decreased visual acuity in patients who present with acute CRVO?
   a. macular ischemia
   b. macular edema
   c. vitreous hemorrhage
   d. neovascular glaucoma

10. A 5-year-old boy is found to have unilateral telangiectatic retinal vessels with an exudative retinal detachment. What histologic findings are characteristic of the likely diagnosis?
   a. "foamy" histiocytes and cholesterol crystals
   b. loss of capillary pericytes and thickening of the retinal capillary basement membrane
   c. dilated, thin-walled vascular channels between the retinal pigment epithelium (RPE) and outer aspect of Bruch membrane
   d. focal sclerosis of retinal vessels with an overlying pocket of liquefied vitreous

11. What is the mode of inheritance for von Hippel–Lindau syndrome?
    a. autosomal recessive
    b. autosomal dominant
    c. X-linked
    d. sporadic

12. What cause of leukocoria is associated with microphthalmos?
    a. persistent fetal vasculature
    b. infection with *Toxocara* species
    c. Coats disease
    d. retinoblastoma

13. How is threshold retinopathy of prematurity (ROP) defined?
    a. 3 contiguous or 10 cumulative clock-hours of extraretinal fibrovascular proliferation in zone II with any vascular engorgement
    b. at least 5 contiguous clock-hours of extraretinal neovascularization or 8 cumulative clock-hours of extraretinal neovascularization with plus disease as well as retinal vessels ending in zone I or II
    c. 6 clock-hours of any extraretinal fibrovascular proliferation in zone III with any vascular engorgement
    d. 3 contiguous or 8 cumulative clock-hours of extraretinal fibrovascular proliferation in zone III with any vascular engorgement

14. A 35-year-old otherwise healthy man presents with recent onset of unilateral slightly blurred vision. On evaluation, the right eye has a visual acuity of 20/25, there is serous subretinal fluid in the macula, and the choroid is thickened. The patient is minimally symptomatic. What is the most appropriate initial management?
    a. observation
    b. topical steroids
    c. photodynamic therapy
    d. laser photocoagulation therapy

15. A 24-year-old woman presents with acute-onset, paracentral "flashing lights" in 1 eye; nasal visual field loss by confrontation testing in both eyes; and normal visual acuity in both eyes. Vitreous cells are present posteriorly in both eyes. The fundus is otherwise unremarkable. Visual field, multifocal electroretinogram (mfERG), and optical coherence tomography (OCT) testing reveal scotomata corresponding to focally decreased mfERG responses and outer retinal layer disruption in both eyes. What is the most likely diagnosis?
    a. multiple evanescent white dot syndrome (MEWDS)
    b. acute zonal occult outer retinopathy (AZOOR)
    c. acute macular neuroretinopathy (AMN)
    d. intermediate uveitis

16. A 42-year-old man reports decreased vision in his right eye over the past 3 days. He does not report significant pain or discomfort, but on review of systems, the ophthalmologist discovers that he also has ulcers on the inside of his mouth. For which HLA haplotype would the patient most likely be positive?
    a. HLA-A29
    b. HLA-B27
    c. HLA-B51
    d. HLA-DR4

17. Once the diagnosis of sympathetic ophthalmia is established, what is the preferred initial management?
    a. intraocular injection of foscarnet to the sympathizing eye
    b. panretinal photocoagulation to the sympathizing eye
    c. enucleation of the exciting eye
    d. administration of oral corticosteroids

18. What bilateral white dot syndrome is characterized by acute vision loss that is typically followed by full or near-full vision recovery?
    a. acute posterior multifocal placoid pigment epitheliopathy (APMPPE)
    b. birdshot chorioretinopathy (vitiliginous chorioretinitis)
    c. MEWDS
    d. serpiginous choroiditis

19. What is the best oral induction treatment option for a patient with varicella-zoster virus–associated acute retinal necrosis?
    a. acyclovir 800 mg 5 times daily
    b. valacyclovir 1 g 3 times daily
    c. valacyclovir 2 g 3 times daily
    d. valganciclovir 900 mg 2 times daily

20. A 30-year-old woman presents with blurred vision and photopsias that started in her right eye and then affected her left eye as well. She had a fever, malaise, and headache 1 week ago. Her fundus examination reveals multiple yellow-white placoid lesions 1–2 disc areas in size. What is the most likely diagnosis?
    a. AZOOR
    b. APMPPE
    c. MEWDS
    d. serpiginous choroiditis

21. What is the most common pattern of color confusion associated with acquired color vision defects?
    a. blue-yellow
    b. red-yellow
    c. blue-green
    d. red-green

22. What is the inheritance pattern of choroideremia?
    a. autosomal recessive
    b. autosomal dominant
    c. mitochondrial DNA
    d. X-linked recessive

23. What ophthalmologic diagnostic test is characteristically expected to give a normal result in patients with Best disease?
    a. fluorescein angiography (FA)
    b. electroretinogram (ERG)
    c. electro-oculogram (EOG)
    d. OCT

24. What is the mode of inheritance of Best disease?
    a. autosomal recessive
    b. autosomal dominant
    c. X-linked recessive
    d. mitochondrial

25. What is the characteristic fundus appearance of dominant (familial) drusen?
    a. large soft drusen with irregular borders
    b. RPE hyperplasia
    c. drusen distribution beyond the vascular arcades and nasal to the optic nerve head
    d. clustering of lesions predominantly in the macula

26. Dermal erythema and bullae are a presenting feature of what retinal condition?
    a. X-linked retinitis pigmentosa
    b. severe form of enhanced S-cone syndrome (Goldmann-Favre syndrome)
    c. gyrate atrophy
    d. incontinentia pigmenti

27. Sorsby macular dystrophy results from mutations in which gene?
    a. *EFEMP1*
    b. *NYX*
    c. *CFH*
    d. *TIMP3*

28. Mutations in *CYP4V2* may lead to characteristic crystalline deposits in the retina with initially normal visual function but with enlarging regions of geographic-like paracentral atrophy. What disorder does this mutation cause?
    a. cystinosis
    b. oxalosis
    c. Bietti crystalline dystrophy
    d. macular telangiectasia type 2

29. A bull's-eye maculopathy may be present in what disease?
    a. Tay-Sachs disease
    b. cystinosis
    c. Fabry disease
    d. Batten disease

30. A male patient presents with photophobia, iris transillumination, hypopigmented fundi, and nystagmus. His skin and hair have normal pigmentation. What is the most likely inheritance pattern of this patient's condition?
    a. X-linked recessive
    b. autosomal dominant
    c. autosomal recessive
    d. X-linked dominant

31. A 60-year-old woman with unexplained peripheral neuropathy but no additional ophthalmic or medical history reports bilateral visually significant floaters. What is the most likely diagnosis?
    a. asteroid hyalosis
    b. cholesterolosis
    c. amyloidosis
    d. chronic vitreous hemorrhage

32. A 1-year-old boy presents with peripheral traction retinal detachment with temporal displacement of the maculae bilaterally. The child's father is similarly affected. There is no history of premature birth. What is the most likely diagnosis?
    a. familial exudative vitreoretinopathy
    b. congenital retinal telangiectasia
    c. juvenile retinoschisis
    d. incontinentia pigmenti

33. Where is the vitreous most firmly attached to intraocular tissue?
    a. optic nerve head
    b. macula
    c. retinal blood vessels
    d. vitreous base

34. Atrophic retinal holes are commonly associated with what condition?
    a. vitreous hemorrhage
    b. retinal detachment
    c. posterior vitreous detachment
    d. lattice degeneration

35. In an eye with retinoschisis, what examination findings are associated with an increased risk of progression to retinal detachment?
    a. demarcation line
    b. hyperopia
    c. inner and outer layer holes
    d. typical peripheral cystoid degeneration

36. What is a known risk factor for degenerative retinoschisis?
    a. prior retinal detachment
    b. hyperopia
    c. lattice degeneration
    d. collagen vascular disease

37. A stage 3 macular hole has at least what diameter?
    a. 200 μm
    b. 300 μm
    c. 400 μm
    d. 500 μm

38. What finding is most consistent with siderosis bulbi?
    a. dense vitritis
    b. sunflower cataract
    c. deposits in Descemet membrane
    d. peripheral retinal pigmentation

39. An ophthalmologist is surgically exploring a blunt globe injury secondary to racquetball trauma. The patient presented with 360° of hemorrhagic chemosis and an intraocular pressure of 3 mm Hg. Intraoperatively, a complete 360° peritomy is performed, and no globe rupture is identified. What is the most appropriate next step?
    a. closure of peritomy
    b. exploration under the rectus muscles
    c. pars plana vitrectomy
    d. placement of a scleral buckle

40. What is the most common ophthalmic manifestation of abusive head trauma?
    a. cranial nerve paresis
    b. bilateral intraocular hemorrhages
    c. papilledema
    d. exudative retinal detachment

41. A 68-year-old man reports a "missing spot" in his vision 1 week after cataract extraction by a first-year resident. Corrected distance visual acuity (also called *best-corrected visual acuity*) is 20/100. In addition to normal postoperative findings, the examination is significant only for subtle foveal hypopigmentation. Macular OCT demonstrates an outer retinal lucency. What is the most likely diagnosis?
    a. nonexudative age-related macular degeneration
    b. photic retinopathy
    c. macular hole
    d. cystoid macular edema

42. A 19-year-old man is struck in the left eye with a paintball. Visual acuity is 20/200 OS. Examination shows no afferent pupillary defect. Intraocular pressure is 18 mm Hg. There is no subconjunctival hemorrhage. The anterior chamber is formed. Fundus examination shows thickened, sheenlike retinal whitening in the macula. What is the most appropriate management for this patient?
    a. laser photocoagulation
    b. anterior chamber paracentesis
    c. high-dose intravenous corticosteroids
    d. observation

43. What imaging modality best helps determine the optimal time to surgically drain a suprachoroidal hemorrhage?
    a. B-scan ultrasonography
    b. FA
    c. OCT
    d. computed tomography

44. What surgical procedure is most indicated for a total retinal detachment with proliferative vitreoretinopathy and multiple posterior retinal breaks?
    a. vitrectomy
    b. indirect laser retinopexy
    c. pneumatic retinopexy with cryotherapy
    d. scleral buckle

45. Nine weeks after routine cataract surgery, a patient develops a steroid-responsive granulomatous uveitis/endophthalmitis, with a white plaque on her intraocular lens. What would be the most appropriate management of this condition?
    a. injection of intravitreal antibiotics
    b. observation
    c. pars plana vitrectomy and partial capsulectomy with injection of intravitreal antibiotic
    d. topical antibiotics and steroids

46. What agent used for retinal tamponade persists in the eye for approximately 2 months after pars plana vitrectomy?
    a. perfluoropropane gas
    b. silicone oil
    c. perfluorocarbon liquid
    d. sulfur hexafluoride gas

Answers

1. **c.** Hyaluronic acid (hyaluronan) is the main glycosaminoglycan found in vitreous. Other major components include water and collagen. Although dermatan, chondroitin, and keratan are glycosaminoglycans, they are not the principal ones found in the vitreous.
2. **c.** The rod photoreceptors have their greatest density (approximately 160,000 rods/mm^2) within the retina at around 12° of fixation. Rod density decreases toward the peripheral retina. Cone density is greatest (exceeding 140,000 cones/mm^2) in the fovea. Within the central fovea, only cones are observed. The fovea is defined as a concave central retinal depression seen on slit-lamp examination; it is approximately 1.5 mm in diameter.
3. **c.** Angioid streaks have been associated with a number of systemic disorders and with aging. The most common systemic association is with pseudoxanthoma elasticum (PXE), a predominantly autosomal recessive disorder caused by a mutation in *ABCC6*. PXE results in calcification and other mineralization of elastin, most visibly as yellow papules in the flexor regions of the skin ("plucked chicken" appearance). Angioid streaks are typically more prominent adjacent to or can extend from the optic nerve head, in contrast to Bruch membrane ruptures from trauma or high myopia ("lacquer cracks"). Other systemic diseases associated with angioid streaks include Paget disease of bone, β-thalassemia, sickle cell disease, and Ehlers-Danlos syndrome. Angioid streaks are not a feature of oculocutaneous albinism (OCA); patients with OCA may have depigmented skin spots but not papules. Crohn disease and ulcerative colitis are associated with uveitis but not angioid streaks.
4. **a.** Vitritis, or inflammation of the vitreous, is the first presenting feature of injection-related endophthalmitis. It is perceived by patients as progressive floaters and blurring of vision, often culminating in essentially no formed vision (eg, light perception only). Immediate evaluation is necessary to determine whether endophthalmitis is present. Sharp pain following the injection or foreign body sensation is likely caused by surface irritation or damage from the povidone-iodine solution, the manipulation of the conjunctiva, or the needle stick. Occasionally, patients may have severe pain that begins within 24 hours of the injection, which may be caused by corneal abrasions. Small floaters that are noticed immediately after injection are usually caused by small air bubbles in the injected liquid, a common occurrence. Less commonly, these floaters are due to silicone oil bubbles from the syringe lubricant. Air bubbles usually disappear after a few hours, whereas silicone oil bubbles persist but usually float out of view.
5. **a.** In patients with diabetic retinopathy, vision loss can be associated with capillary leakage, ischemic maculopathy (capillary occlusion), and sequelae from ischemia-induced neovascularization. Vitreous hemorrhage, macular edema, traction detachment, and macular ischemia can all cause vision loss. In this patient, macular ischemia (normal foveal avascular zone is approximately 500 μm in diameter) is the most likely cause because the vitreous hemorrhage is outside the visual axis (inferior) and the macular edema is noncentral.
6. **b.** A branch retinal artery occlusion (BRAO) is the result of embolization or thrombosis of the affected vessel. Common types of emboli include cholesterol emboli arising in the carotid arteries, platelet-fibrin emboli associated with large-vessel arteriosclerosis, and calcific emboli arising from diseased cardiac valves. Rare causes of a BRAO include emboli

resulting from cardiac myxoma, long-bone fractures (fat emboli), infective endocarditis (septic emboli), and intravenous drug use (talc emboli). Subarachnoid hemorrhage may be associated with intraocular hemorrhage or Terson syndrome. Traumatic pancreatitis is associated with Purtscher retinopathy, which is characterized by focal areas of retinal whitening that may superficially resemble a BRAO but are not a BRAO.

7. **b.** The most important risk factor for central retinal vein occlusion (CRVO) is age; 90% of affected patients are older than 50 years at the time of diagnosis. Other significant risk factors for development of CRVO include systemic hypertension, open-angle glaucoma, hypercoagulable states, diabetes mellitus, and hyperlipidemia.
8. **d.** The 2008 study Ranibizumab for Macular Edema Following Branch Retinal Vein Occlusion (BRAVO) found ranibizumab to be an effective treatment for macular edema associated with branch retinal vein occlusion (BRVO); monthly injection of either 0.5 mg or 0.3 mg of ranibizumab was superior to sham injection for improving visual acuity. Verteporfin is used in photodynamic therapy and is not an approved treatment for macular edema associated with BRVO. Fluocinolone acetonide intravitreal implant and panretinal photocoagulation are not used for treating macular edema associated with BRVO.
9. **b.** Patients with acute CRVO commonly present with reduced visual acuity secondary to macular edema. Neovascularization of the iris usually occurs 3 to 5 months after symptom onset. Peripheral ischemia with capillary dropout is seen in some severe cases of CRVO and can promote neovascularization, resulting in vitreous hemorrhage. Macular ischemia can occur as well but is a less likely cause of decreased acuity on presentation.
10. **a.** Coats disease is clinically evident within the first decade of life and is more common in boys. It is characterized by a unilateral retinal telangiectasia with mild to severe leakage and possible exudative retinal detachment. "Foamy" histiocytes and cholesterol crystals in the subretinal space are common histologic findings. Loss of capillary pericytes and thickening of the retinal capillary basement membrane may be seen in diabetic retinopathy. Dilated, thin-walled vascular channels between the retinal pigment epithelium (RPE) and the outer aspect of Bruch membrane are found in polypoidal choroidal vasculopathy. Focal sclerosis of retinal vessels with an overlying pocket of liquefied vitreous is found in lattice degeneration.
11. **b.** Von Hippel–Lindau syndrome is caused by a tumor suppressor gene mutation on the short arm of chromosome 3 (3p26–p25), the inheritance of which is autosomal dominant with incomplete penetrance and variable expression.
12. **a.** All of the conditions listed as answer choices (persistent fetal vasculature, Coats disease, retinoblastoma, and infection with *Toxocara* species) can cause unilateral leukocoria. However, only persistent fetal vasculature is associated with microphthalmos. It is also associated with elongated ciliary processes, cataract, retinal detachment, and angle-closure glaucoma.
13. **b.** The term *threshold retinopathy of prematurity (ROP)* was coined in the mid-1980s by investigators in the Cryotherapy for ROP (CRYO-ROP) study to define disease with equal chances of spontaneous regression or progression to an unfavorable outcome. Threshold disease is characterized by at least 5 contiguous clock-hours of extraretinal neovascularization or 8 cumulative clock-hours of extraretinal neovascularization with plus disease as well as retinal vessels ending in zone I or II.
14. **a.** The patient almost certainly has central serous chorioretinopathy (CSC). In most cases, initial observation, with the expectation of spontaneous resolution of the subretinal fluid, is the most appropriate management. Photodynamic therapy may be considered for CSC

when the patient is symptomatic or when the subretinal fluid does not resolve after a period of observation. Verteporfin photodynamic therapy (PDT) has been shown to decrease or eliminate subretinal fluid; it also decreases choroidal thickness and reduces choroidal vascular hyperpermeability. Laser photocoagulation therapy is no longer preferred for CSC. Moreover, unlike PDT, photocoagulation has no effect on choroidal thickness. Steroids are contraindicated in patients with CSC.

15. **b.** The findings are most consistent with those in acute zonal occult outer retinopathy (AZOOR), an idiopathic condition that typically affects young women with myopia. Acute onset of posterior photopsias may occur in many posterior inflammatory conditions, including multiple evanescent white dot syndrome (MEWDS) and multifocal choroiditis; however, the disruption of the outer retina observed on optical coherence tomography (OCT) differentiates AZOOR from both MEWDS and multifocal choroiditis. MEWDS is characterized by multiple small gray, white, or yellow-white dots at the level of the outer retina in and around the posterior pole and typically presents unilaterally. Acute macular neuroretinopathy is characterized clinically by reddish-brown teardrop or wedge-shaped lesions around the fovea, the tips of which point centrally; the lesions correspond in size and location to subjective paracentral scotomata. Intermediate uveitis is characterized by vitreous cells.

16. **c.** The clinical presentation is consistent with that of Behçet disease. Panuveitis with occlusive retinal vasculitis is the most common presentation. HLA-B51 is commonly associated with Behçet disease with ocular manifestations. However, the diagnosis of Behçet is clinical, and laboratory tests are of little value in confirming the diagnosis. HLA-A29 is more common in patients with birdshot chorioretinopathy, HLA-B27 is associated with acute anterior uveitis, and HLA-DR4 is weakly associated with Vogt-Koyanagi-Harada disease (or syndrome).

17. **d.** In the rare instances when sympathetic ophthalmia does follow either ocular injury or surgery, standard treatments such as corticosteroids almost always control the inflammation. Therefore, enucleation or evisceration of an injured eye to minimize risk of sympathetic ophthalmia is rarely practiced. Panretinal photocoagulation has no place in managing this form of uveitis. Foscarnet is an antiviral antibiotic that is used to treat cytomegalovirus retinitis or other herpes viral retinitis, such as that occurring in acute retinal necrosis or as a complication of iatrogenic or infectious immunosuppression.

18. **a.** Acute posterior multifocal placoid pigment epitheliopathy (APMPPE) is an uncommon, bilateral inflammatory disease characterized by acute-onset vision loss, usually followed by substantial or near-complete improvement weeks to months later. In the acute stage, fluorescein angiography (FA) of active lesions shows early blockage followed by progressive late leakage and staining. There is no definitive evidence that treatment with corticosteroids is beneficial in altering the outcome for APMPPE. Birdshot chorioretinopathy is chronic, progressive, and prone to recurrent episodes of inflammation. MEWDS is typically unilateral. Symptoms include decreased vision, scotomata, and sometimes photopsias. In most patients, the symptoms and fundus findings begin to improve in 2–6 weeks without treatment. Serpiginous choroiditis is an often vision-threatening, recurring inflammatory disease involving the outer retina, RPE, and inner choroid (the choriocapillaris). Persistent scotomata and decreased central vision are common symptoms.

19. **c.** While intravenous acyclovir 10 mg/kg every 8 hours for 10–14 days may be used for induction treatment in acute retinal necrosis, oral valacyclovir 2 g 3 times daily is considered to be an equally effective alternative. Oral acyclovir 800 mg 5 times daily or valacyclovir 1 g 3 times

daily is the appropriate maintenance dosage after induction has been completed. Oral valganciclovir is reserved for cytomegalovirus-associated intraocular infections.

20. **b.** Acute APMPPE is a bilateral inflammatory disease that typically occurs in otherwise healthy young adults, with men and women affected equally. Patients may have a viral prodrome. In rare cases, cerebrovasculitis occurs. The characteristic examination finding is multiple yellow-white placoid lesions that vary in size and involve the outer retina (RPE) and inner choroid (choriocapillaris). MEWDS can also have a viral prodrome but is usually unilateral. In some patients, a transient foveal granularity also develops and is highly suggestive of the condition. On initial presentation, eyes with AZOOR may have a normal-appearing fundus or show evidence of mild vitritis. Permanent visual field loss is often associated with late development of fundus changes. Depigmentation of large zones of RPE usually corresponds to scotomata; narrowed retinal vessels may be visible within these areas. In serpiginous choroiditis, lesions first appear at or near the optic nerve head and extend centrifugally in a serpentine pattern. With numerous recurrences, a serpiginous (pseudopodial) or geographic (maplike) pattern of chorioretinal scarring develops.
21. **a.** Acquired color vision defects are most frequently blue-yellow, or *tritan,* abnormalities; they affect males and females equally. Blue-yellow color defects often accompany an optic neuropathy but also can occur in a maculopathy. Hereditary color vision defects are most frequently red-green abnormalities. They are most often X-linked recessive and affect 5%–8% of males and 0.5% of females.
22. **d.** Choroideremia is an X-linked recessive degeneration of the choriocapillaris of the choroid, and of the overlying RPE and retina. Patients with choroideremia have nyctalopia and show progressive peripheral visual field loss over 3–5 decades. Most patients maintain good visual acuity until a central island of foveal vision is lost.
23. **b.** In Best disease, the electroretinogram (ERG) response is characteristically normal, and the electro-oculogram result is almost always abnormal, showing a severe loss of light response. OCT and FA results are abnormal in Best disease and reflect the changes in the macular lesion from vitelliform stage to atrophic appearance.
24. **b.** Best disease is an autosomal dominant maculopathy caused by mutations in the *BEST1 (VMD2)* gene. The encoded protein bestrophin localizes to the basolateral plasma membrane of the RPE and functions as a transmembrane ion channel.
25. **c.** Dominant (familial) drusen typically extend beyond the vascular arcades and nasal to the optic nerve head. A single missense mutation (Arg345Trp) on exon 10 in the *EFEMP1* gene of some patients with dominant drusen results in a fibrillin-like extracellular matrix protein expressed in the RPE and retina. Typically, the clinical findings first manifest bilaterally in the third to fourth decade of life. Although usually asymptomatic, choroidal neovascularization may develop in this patient population and lead to loss of vision. In contrast to drusen of age-related macular degeneration (AMD), the lesions of dominant drusen are typically small and round. Clumping and hyperplasia of the RPE and drusen clustering in the macula are features of AMD, not familial drusen.
26. **d.** Incontinentia pigmenti (Bloch-Sulzberger syndrome) is an X-linked dominant condition characterized by streaky skin lesions and abnormalities of the teeth and central nervous system. Ocular involvement includes pigmentary abnormalities as well as peripheral retinal nonperfusion and neovascularization that may cause traction (also called *tractional*) and cicatricial retinal detachment. In incontinentia pigmenti, erythema and bullae develop in the first few days of life.

X-linked retinitis pigmentosa is a retinal degeneration associated with vision loss and pigmentary retinopathy, but no dermatologic manifestations. The severe form of enhanced S-cone syndrome (also called *Goldmann-Favre syndrome*) is also a pigmentary retinopathy and is associated with pathognomonic ERG abnormalities and nyctalopia but no skin findings. Gyrate atrophy is an autosomal recessive congenital deficiency in ornithine aminotransferase that leads to large, geographic, peripheral paving-stone–like areas of atrophy of the RPE and choriocapillaris. It has no association with dermatologic changes.

27. **d.** Sorsby macular dystrophy (SMD) results from mutations in *TIMP3,* which plays an important role in the regulation of extracellular matrix turnover. Early signs of SMD are yellowish-gray drusenlike deposits or a confluent plaque of yellow material at the level of Bruch membrane within the macula and along the temporal arcades. The deposits progress over time to include the central macula and take on the appearance of geographic atrophy. Vision loss results from expansion of macular atrophy or from development of subfoveal choroidal neovascularization.

    Malattia Leventinese (also called *Doyne honeycomb dystrophy*) is caused by a mutation in *EFEMP1.* The drusen in this condition often show a distinctive pattern of radial extensions of small and intermediate-sized deposits emanating from the fovea. The clinical syndrome of cuticular (also called *basal laminar*) drusen features small clustered drusen and occurs in young adults. Although this syndrome is associated with mutation in complement factor H, it is not a monogenic disorder. Mutations in *NYX* are associated with congenital stationary night blindness.

28. **c.** The clinical description is most consistent with that of Bietti crystalline dystrophy, which is associated with mutations in *CYP4Y2.* Oxalosis is a rare inherited disorder of primary hyperoxaluria, which has been associated with mutations in 3 genes *(AGXT, GRHPR, DHDPSL).* Cystinosis is an abnormality of cystine accumulation that may result from genetic causes (mutation in *CTNS*) or exposure to some anesthetics. Typically, patients with oxalosis and cystinosis have renal insufficiency or failure resulting from amino acid crystallization in the kidneys. Characteristic findings in macular telangiectasia type 2 include a reduced foveolar reflex, loss of retinal transparency (retinal graying), superficial retinal crystalline deposits, and foveal atrophy, but the disease is not associated with *CYP4V2;* it is associated with serine metabolism abnormalities.

29. **d.** Batten disease, an example of a neuronal ceroid lipofuscinosis, may have retinal findings, including an atrophic bull's-eye maculopathy, optic nerve pallor, and attenuation of retinal vessels. The ophthalmic symptoms and signs may be the first, earliest manifestations of this disorder. The cherry-red spots seen in Tay-Sachs disease result from ganglioside accumulation within ganglion cells. Pigmentary abnormalities and fine retinal crystals are present in cystinosis (nephropathic type). Tortuous and dilated retinal vessels, as well as cornea verticillata and tortuous conjunctival vessels, are present in Fabry disease.

30. **a.** The clinical presentation described is consistent with ocular albinism, which is typically transmitted as an X-linked (recessive) trait. Albinism includes a group of different genetic abnormalities in which the synthesis of melanin is reduced or absent. A number of genes are associated with both oculocutaneous and ocular albinism. Oculocutaneous albinism is typically transmitted as an autosomal recessive trait.

31. **c.** Vitreous opacities may be one of the initial signs of the dominantly inherited form of hereditary familial amyloidosis. In addition to the vitreous, amyloid may be deposited in the retinal vasculature, the choroid, and the trabecular meshwork. Nonocular manifestations of

amyloidosis include upper- and lower-extremity polyneuropathy and central nervous system abnormalities. Congo red staining of vitreous samples can confirm the diagnosis. Asteroid hyalosis is typically unilateral and is rarely visually significant. Cholesterolosis, or *synchysis scintillans,* occurs in eyes with a history of intravitreal hemorrhage usually related to prior accidental or surgical ocular trauma. Although prior ophthalmic pathology leading to dehemoglobinized vitreous hemorrhage should always be considered, it is less likely in this scenario.

32. **a.** Unlike patients with ROP, individuals with familial exudative vitreoretinopathy (FEVR) are born full term. FEVR is characterized by failure of the temporal retina to vascularize. Retinal folds and peripheral fibrovascular proliferation, as well as traction and exudative retinal detachments, are often associated with FEVR. FEVR is usually inherited as an autosomal dominant trait, but X-linked transmission also occurs. Congenital retinal telangiectasia and juvenile retinoschisis are not typically associated with a traction retinal detachment. Incontinentia pigmenti is typically lethal in utero to males.

33. **d.** The vitreous is most tightly adherent at its base, where the vitreous remains firmly adherent to the anterior retina and pars plana epithelium. This attachment is so strong that it can lead to vitreous base avulsion after severe trauma. The vitreous is also firmly attached to the posterior lens capsule, retinal vessels, optic nerve, and fovea. With age, the vitreous contracts, which can result in a posterior vitreous detachment. With this contraction, the posterior cortical gel detaches toward the firmly attached vitreous base.

34. **d.** Atrophic retinal holes are commonly associated with lattice degeneration. They are infrequently the cause of rhegmatogenous retinal detachment. They represent atrophic rather than tractional retinal breaks, so they are not caused by posterior vitreous detachment, and they are not associated with vitreous hemorrhage.

35. **c.** The presence of inner and outer layer holes in retinoschisis is associated with an increased risk of retinal detachment. However, the presence of outer layer holes can lead to a localized retinal detachment that usually does not progress and seldom requires treatment. The presence of hyperopia and typical peripheral cystoid degeneration does not predispose to retinal detachment. Finally, the presence of a demarcation line in an eye with retinoschisis suggests that a full-thickness detachment is present or was formerly present and has spontaneously regressed; this is not likely to lead to a progressive retinal detachment.

36. **b.** Degenerative retinoschisis is commonly associated with hyperopia. Lattice degeneration is more common in myopic eyes. Collagen vascular disease and prior retinal detachment do not increase the risk of degenerative retinoschisis.

37. **c.** Stage 3 holes are full-thickness holes at least 400 μm in diameter. Stage 0 holes (pre–macular hole) represent vitreomacular adhesion (VMA). OCT examination shows that a stage 1A hole is a foveal "pseudocyst," or horizontal splitting (schisis), associated with vitreous traction on the foveal center. A stage 1B hole indicates a break in the outer fovea. Stage 2 holes are early full-thickness holes less than 400 μm in diameter. Stage 4 holes are full-thickness holes at least 400 μm in diameter in association with a complete posterior vitreous detachment.

38. **d.** The chalcosis and siderosis bulbi that result from retaining intraocular copper and iron, respectively, have distinct manifestations. Pure copper can cause an intense inflammatory response, whereas a copper content less than 85% can result in chronic chalcosis, with findings that include sunflower cataract and a Kayser-Fleischer–like ring of deposits in Descemet membrane. Intraocular iron deposits in neuroepithelial tissues produce findings that include retinal pigmentation. ERG can be used to assess retinal toxicity in siderosis bulbi.

39. **b.** Severe blunt trauma can cause scleral rupture. The most common sites of scleral rupture are at the corneal limbus and through areas of physiologic scleral thinning parallel to and under the rectus muscle insertions. Scleral rupture is associated with markedly decreased ocular ductions; boggy conjunctival chemosis with hemorrhage; deepened anterior chamber; severe vitreous hemorrhage; and usually hypotony, although intraocular pressure may also be normal or elevated. In this case, suspicion of occult scleral rupture is high, warranting further exploration, with disinsertion of 1 of more rectus muscles in order to identify the rupture site. Scleral lacerations or ruptures are typically closed with 7-0, 8-0, or 9-0 nonabsorbable suture. Some specialists recommend prophylactic vitrectomy and/or placement of a scleral buckle at the time of primary repair of some perforating ocular injuries, but the primary goal of most open-globe repairs is to achieve corneoscleral wound closure.

40. **b.** Retinal hemorrhages are the cardinal manifestation of abusive head trauma (AHT, formerly called *shaken baby syndrome*). The presenting sign of child abuse involves the eye in approximately 5% of cases. Ocular signs include retinal hemorrhages and cotton-wool spots, retinal folds, hemorrhagic schisis cavities, and pigmentary maculopathy. The retinal hemorrhages associated with AHT often have a hemispheric contour. Papilledema occurs in less than 10% of cases of AHT. Cranial nerve paresis and exudative retinal detachment are not commonly associated with AHT.

41. **b.** Nonexudative AMD, macular holes, and cystoid macular edema can decrease central visual acuity; however, in the setting of recent (and likely prolonged) ophthalmic surgery and outer retinal lucency, photic retinopathy secondary to exposure to the operating microscope light source is the most likely diagnosis. Although this retinopathy is more common following retinal surgery, it can occur with surgery times as short as 30 minutes; prolonged focal exposure can cause photochemical injury to the fovea, much like solar retinopathy. Patients typically present with central or paracentral scotomata, hypopigmented foveal lesions, and outer retinal cavitation. Most patients recover vision somewhat over a period of 3–6 months.

42. **d.** The clinical presentation described is consistent with commotio retinae, and the appropriate management is observation. High-dose intravenous steroids have no role in the management of commotio retinae. Anterior chamber paracentesis may be used in cases of acute retinal artery occlusion but has no role in commotio retinae. Laser photocoagulation may be used for a retinal break but is not used for commotio retinae.

43. **a.** When suprachoroidal hemorrhage occurs intraoperatively, immediate drainage is rarely effective, because of rapid coagulation of the blood. Waiting 7 to 14 days often allows the hemorrhage to liquefy. B-scan ultrasonography is useful to help identify clot liquefaction. It is also useful to determine whether appositional ("kissing") suprachoroidal hemorrhage is present and whether there is concurrent retinal detachment. In the presence of a large suprachoroidal hemorrhage, often no view is possible for OCT or FA. Computed tomography is not indicated for monitoring suprachoroidal hemorrhage.

44. **a.** For a total retinal detachment with proliferative vitreoretinopathy (PVR) and multiple posterior retinal breaks, pars plana vitrectomy with or without scleral buckle is indicated, along with membrane peeling to relieve traction, retinectomy (in case of incarceration or retinal shortening), flattening retina by using air or perfluorocarbon liquid, endolaser photocoagulation or cryopexy, and gas or silicone oil tamponade. Pneumatic retinopexy is contraindicated in the presence of PVR. Laser demarcation is not effective for a total retinal detachment. Scleral buckle alone is typically not effective in the presence of multiple posterior retinal breaks.