Management of Ocular Complications associated with Dyskeratosis Congenita

Abstract: Dyskeratosis congenita is a rare disorder of shortened telomeres that can cause ocular manifestations. This report discusses a patient who was treated with vitrectomy for tractional retinal detachment and vitreous hemorrhage secondary to dyskeratosis congenita.

I. Case History

- Patient demographics: 4 year old Hispanic male
- Chief complaint: Doctor ordered 6 week follow up for pars plana vitrectomy OS following a tractional retinal detachment and vitreous hemorrhage secondary to dyskeratosis congenita and panretinal photocoagulation fill-in laser OD for ROP.
- Ocular history: retinopathy of prematurity (ROP) OU s/p laser, myopia OU, cortical cataract OD, infantile cataract OS, tractional retinal detachment with vitreous hemorrhage OS
- Medical history: premature at 28 weeks gestation (birth weight of 1 lb, 13 oz), dyskeratosis congenita, thrombocytopenia, microcephaly
- Ocular and systemic medications: none

II. Pertinent Findings

- Clinical:
  - Unaided distance visual acuities: OD counting fingers @ 3 feet, OS counting fingers @ 3 feet
  - Pupils: round and reactive to light OU, (-) RAPD OU
  - Extraocular motility: Full OU
  - Confrontation visual fields: unable OU
  - Intraocular pressure: OD, OS soft to palpation
  - Anterior segment: OD 2+ cortical cataract, OS infantile cataract; all other findings within normal limits OU
  - Posterior segment: OD large inferotemporal and temporal areas of retinal pigment epithelium (RPE) atrophy/scarring, PRP scars 360; OS PRP scarring, macular dragging secondary to previous traction, temporal peripheral fibrous ridge, PRP laser scars 5:00-2:30, resolution of the vitreous hemorrhage and residual pre-retinal hemorrhage noted inferotemporally, (-)exudates noted OU.
- Physical: microcephaly, mild reticulated skin pigmentation of neck, loose skin, short and frail stature
- Imaging:
  - Fundus photo documentation completed
  - Optical coherence tomography (OCT): OU loss of foveal contour, thickened intraretinal layers, disruption of inner/outer segment junction
  - Fluorescein angiography (FA): temporal leakage and avascular areas OU

III. Differential Diagnosis

- Primary: Revesz syndrome
- Other conditions causing tractional and/or exudative retinal detachment: include Hoyeraal-Hreidarsson syndrome, familial exudative vitreoretinopathy, retinopathy of prematurity (different retinal detachment appearance and scar tissue pattern), proliferative diabetic retinopathy, sickle cell retinopathy, toxocariasis, and trauma.

IV. Diagnosis and Discussion

- Diagnosis: Tractional retinal detachment and resolved vitreous hemorrhage OS 2’ dyskeratosis congenita, severe ROP OU
- Discussion
  - Pathophysiology
    - Dyskeratosis congenita (DC) is a rare, progressive congenital disorder caused by shortened telomeres and is associated with bone marrow failure, resulting in early mortality. Many symptoms resemble premature aging.
• The mechanism of retinal ischemia in DC is poorly understood. Vascular pruning and instability causes neovascularization, which in turn can lead to tractional retinal detachment and/or vitreous hemorrhage from contraction of fibrovascular proliferation.
  
  o Clinical characteristics
    • Include triad of reticulated skin pigmentation, nail dystrophy, and oral leukoplakia; peripheral cytopenia, bone marrow failure, pulmonary fibrosis, squamous cell carcinoma of head and neck and developmental delays.
    • Ocular signs: epiphora, blepharitis, madarosis, ectropion, entropion, trichiasis, retinal ischemia and neovascularization, fibrovascular proliferation, tractional and/or exudative retinopathy
  
  o Symptoms include flashes of light and/or floaters, veil or curtain over vision, decreased vision, loss of peripheral or central vision and watery eyes.
  
  o Diagnosis: clinical appearance based on fundus examination, OCT, FA, B-scan, bloodwork, genetic testing

V. Treatment and Management

• The patient was treated with vitrectomy OS and PRP fill-in laser for ROP OD.
  
  o At one week follow up, the vitreous hemorrhage had dispersed, tractional membranes were released, and the retina appeared flat OS with B scan.
  
  o At one month follow up, the vitreous hemorrhage had improved, and the retina appeared flat.
  
  o At 2.5 months follow up, resolving vitreous hemorrhage was noted, and the retina appeared flat. Optos fluorescein angiography revealed leakage and avascular areas OU. Additional laser treatment to prevent further neovascularization in the future may be needed. Patient scheduled for a one month follow-up appointment to monitor resolution and for Optos photos OU.

• Systemic condition
  
  o Bone marrow/stem cell transplant
  
  o Oxymetholone: increase red blood cell production
  
  o Filgrastim (Neupogen): stimulates granulocyte production

• Ocular treatment
  
  o If asymptomatic, the patient should be followed every 4-6 months with dilated fundus exam. For patients with complications, the optometrist should coordinate care with the retinal specialist for further surgical treatment such as pars plana vitrectomy, panretinal photocoagulation (PRP), anti-VEGF injection and scleral buckle.

• Prognosis is poor, as 80-90% of patients die from bone marrow failure, the leading cause of death, by 30 years of age. Ocular complications, such as tractional and/or exudative retinal detachment, can be sight threatening and must be treated promptly.

VI. Conclusion

• It is important for eye care professionals to be familiar with the ocular manifestations and complications associated with rare systemic disorders, especially in patients with multiple disease processes like in dyskeratosis congenita.

VII. References


