AANP Teaching Rounds:

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Disclosures

• I have no relevant financial relationships to disclose
Learning Objectives

• Learning Objective #1: Outline the differential diagnosis of tumors of the ocular surface
• Learning Objective #2: Recognize the spectrum of ocular infections relevant to ophthalmic pathology
• Learning Objective #3: Recognize the morphologic features of the most common keratopathies and dystrophies
Case 1

• 60-year-old woman with past medical history of hypertension, GERD, Atrial fibrillation and DVT
• Autopsy: acute pulmonary hemorrhage, aortopulmonary fistula, and aortic dissection
• No clinical history of eye disease
Findings

- Fuchs Dystrophy
- Fuchs Adenoma
Case 1
Fuchs Endothelial Corneal Dystrophy

- Most common corneal dystrophy in the US
- Corneal edema in ~5th-6th decade of life
- Primary defect in corneal endothelium
- Relatively easy clinical and pathologic diagnosis
- PAS stain very useful in equivocal cases

Fuchs adenoma
- Benign tumor possibly developing from non-pigmented ciliary epithelium
- Age related
- Typically incidental at autopsy, but may rarely cause iris protrusion, shallowing of anterior chamber or glaucoma
Case 2

- 54-year-old man with visual loss
Masson Trichrome
Case 2
Granular Corneal Dystrophy

• Visual loss late in life
• May recur in grafts after transplantation
• Autosomal dominant inheritance
  • Transforming growth factor beta (TGFB1 p.R555W) mutation
• Avellino dystrophy variant: features of Granular (type I)+Lattice
• Other stromal dystrophies more aggressive
  – Lattice corneal dystrophy type I and II (confined and systemic amyloidosis)
  – Macular corneal dystrophy: most aggressive (autosomal recessive, ‘localized mucopolysaccharidosis’)
Case 3

- 71-year-old woman with corneal edema
Case 3
Gram Stains

GW

B&H
Case 3
Infectious pseudocrystalline keratopathy

• Indolent corneal infection
• Avirulent streptococcal strains
• Intrastromal opacities in the absence of inflammation
• Complication of corneal surgery, grafts and corticosteroids
• Treatment: aggressive antibiotic therapy or PKA
Case 3
Infectious keratitis

• Bacterial
• Mycobacteria
• Viral (Herpes simplex, Varicella zoster)
• Fungal (Candida, Asperigillus, Fusarium)
• Acanthamoeba
Case 4

• 6-month-old boy with intraocular mass
Characterisation of retinoblastomas without RB1 mutations: genomic, gene expression, and clinical studies

Case 4
Retinoblastoma with MYCN amplification

- Reported as occurring in a subset of RB1 wildtype retinoblastomas
- Young age of onset (infants)
- Aggressive histology
Case 5

• 63-year-old man
• Started experiencing tearing, swelling, pain and itching of left eye
• Progressive eye swelling over several months
• Left eye proptosis
Case 5
Flow cytometry

• “The majority of the B cells are phenotypically abnormal, accounting for 61% of total cells, and are small in size by forward light scatter.

• These show monoclonal expression of kappa light chain and also express CD19 and brighter than normal CD20 but lack CD5, CD10, CD200 and CD38.”
Case 5
Extranodal Marginal Zone B-cell Lymphoma

- Most common lymphoma of the ocular adnexa
- In contrast Large B-cell lymphoma most common primary intraocular type
- Flow cytometry very useful in diagnosis
- Indolent clinical course
Case 6

- 54-year-old man with lesion of the right eyelid
- Painful and itchy
- Appearance consistent with chalazion
Case 6
Sebaceous Carcinoma

• Usually affect elderly patients
• Predilection for the eyelids
• Presentation as a chalazion typical
• Diagnostic morphologic features usually present
• Minimal involvement, pagetoid spread more challenging to diagnose
Case 7

- 68-year-old woman
- Mass on the right nasal bulbar conjunctiva extending from the caruncle to the limbus 12 x 4.5 mm
BRAF p.V600K
VAF 78%
Case 7
Conjunctival melanoma

- Relatively rare compared to uveal melanoma
- Primary acquired melanosis (PAM) with atypia known precursor
- Lymphatic spread in advance cases
- Exenteration performed in cases with recurrence/orbital involvement
- Alterations in MAPK pathway components (BRAF, RAS, etc)
Case 7-
Follow-up

• Treated with I-125 plaque, cryo
• Orbital exentration 8 months later
• Right orbit-no evidence of recurrence 2 years later
Left eye-choroidal melanocytosis stable
Case 8

- 72-year-old man with complicated past medical history including sarcoidosis
- Sudden loss of vision in right eye
- Blindness
- Progressive decline, weakness, fatigue and mental fogginess
- Admitted for progressive hypoxemia, eventually shock and metabolic acidosis.
Autopsy

- Organizing pneumonia and hemophagocytic histiocytosis
Case 8: CMV retinitis
References

- Pacheco R. Conjunctival melanoma: outcomes based on tumour origin in 629 patients at a single ocular oncology centre. Eye (Lond) 2021.
Questions?
References

5. Pacheco R. Conjunctival melanoma: outcomes based on tumour origin in 629 patients at a single ocular oncology centre. Eye (Lond) 2021.