International Society of Ocular Oncology

ISOO VIRTUAL 2024

Jan 06, 2024 - Jan 07, 2024

Focussing On

Case Presentations with Interactive Discussion

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OVERVIEW - DAY 1

Saturday, January 6, 2024 (2:00 PM UTC to 4:00 PM UTC)
Session 1, Intraocular Tumors – Melanoma and other Uveal Tumors
Saturday, January 6, 2024 (4:00 PM UTC to 6:00 PM UTC)
Session 2, Intraocular Tumors – Retinoblastoma and other Retinal Tumors

- New York, USA (Eastern Time): 9:00 AM
- Chicago, USA (Central Time): 8:00 AM
- Denver, USA (Mountain Time): 7:00 AM
- Los Angeles, USA (Pacific Time): 6:00 AM
- London, UK (Greenwich Mean Time): 2:00 PM
- Berlin, Germany (Central European Time): 3:00 PM
- Paris, France (Central European Time): 3:00 PM
- Madrid, Spain (Central European Time): 3:00 PM
- Lagos, Nigeria (West Africa Time): 3:00 PM
- Cairo, Egypt (Eastern European Time): 4:00 PM
- Mumbai, India (Indian Standard Time): 7:30 PM
- Beijing, China (China Standard Time): 8:00 PM
- Tokyo, Japan (Japan Standard Time): 10:00 PM
- Sydney, Australia (Australian Eastern Standard Time): 12:00 AM (next day)

OVERVIEW - DAY 2

Sunday, January 7, 2024 (10:00 AM UTC to 12:00 Noon UTC)
Session 3, Ocular Surface and Eyelid Tumors
Sunday, January 7, 2024 (12:00 Noon UTC to 2:00 PM UTC)
Session 4, Orbital and Optic Nerve Tumors and Miscellaneous
Sunday, January 7, 2024 (02:00 PM UTC to 02:30 PM UTC)
Session 5, Special Session

- New York, USA (Eastern Time): 5:00 AM
- Chicago, USA (Central Time): 4:00 AM
- Denver, USA (Mountain Time): 3:00 AM
- Los Angeles, USA (Pacific Time): 2:00 AM
- London, UK (Greenwich Mean Time): 10:00 AM
- Berlin, Germany (Central European Time): 11:00 AM
- Paris, France (Central European Time): 11:00 AM
- Madrid, Spain (Central European Time): 11:00 AM
- Lagos, Nigeria (West Africa Time): 11:00 AM
- Cairo, Egypt (Eastern European Time): 1:00 PM
- Mumbai, India (Indian Standard Time): 3:30 PM
- Beijing, China (China Standard Time): 5:00 PM
- Tokyo, Japan (Japan Standard Time): 7:00 PM
- Sydney, Australia (Australian Eastern Standard Time): 8:00 PM

SESSION 1 - INTRAOCULAR TUMORS 1

SATURDAY, JANUARY 6, 2024 (02:00 PM TO 04:00 PM UTC)

Opening Remarks, Dan Gombos, ISOO President
Chairperson: Carol Shields, Jesse Berry
Moderator: Fairooz PM
Expert Panel: Bertil Damato, Bhavana Chawla, Carol Shields, Emine Kilic, Jesse Berry, Mandeep Sagoo, Martine Jager, Prithvi Mrutyunjaya

Sequence	Name	Title
1	Karim Al-Ghazzawi	Clinical Course and Local Resection of Mesoectodermal Leiomyoma of the Ciliary Body
2	Ahad Sedaghat	Proton Beam Radiation for Growing Choroidal Osteoma
3	Pranvera Emini	Bilateral Choroidal Osteoma in a Child with Börjeson-Forssman-Lehmann Syndrome
4	Elaine Binkley	Pigmented Choroidal Mass
5	Shreya Sirivolu	Diagnostic Aqueous Humor Proteome Predicts Metastatic Potential in Uveal Melanoma Patient with Tumor too Small to Biopsy
6	Buse Guneri Beser	Bilateral Choroidal Melanoma at Presentation in a Patient with Myotonic Dystrophy
7	Dan S Gombos	Asymmetric Response of an Intraocular/Orbital Uveal Melanoma Treated with Stereotactic Radiation Therapy
8	Vishal Raval	Peripheral Exudative Hemorrhagic Chorioretinopathy Simulating Choroidal Melanoma in a Patient with Nevus of Ota
9	Arun D Singh	RPE Adenoma Treated with Episcleral Brachytherapy
10	Almila Sarigul Sezenoz	Diagnosis of Choroidal Metastasis from Cutaneous Melanoma Based on BRAF Mutation on FNAB and Complete Response to Immunotherapy
11	Thomas M Catapano	Quite Rare, Not Fair

SESSION 2 - INTRAOCULAR TUMORS 2

SATURDAY, JANUARY 6, 2024 (04:00 PM TO 06:00 PM UTC)

Chairperson: Dan Gombos, Mahesh Shanmugam

Moderator: Rolika Bansal

Expert Panel: Carol Shields, Dan Gombos, Fairooz PM, Jyotirmay Biswas, Mahesh Shanmugam

Sequence	Name	Title
1	Abhijeet Beniwal	Histopathological Surprise in an Intraocular Mass
2	Hibba Quhill	Unusual Phenotypic Presentation of Ocular Erdheim Chester Disease
3	Ayushi Agarwal	A Rare Pseudoretinoblastoma in a Child
4	Ido Didi Fabian	An Incidental Rb1 Pathogenic Sequence Variant in a Fetus
5	Elaine Huang	Retinoblastoma with MDM4 Amplification Diagnosed from Cell-free DNA in Aqueous Humor
6	Hilary Racher	Retinoblastoma Discordant Monozygotic Twins and the Discovery of an Intron 15 Rb1 Variant
7	Chen-Ching Peng	Phenotypic Biomarkers of Aqueous Humor Extracellular Vesicles from a Retinoblastoma Eye
8	Aurora Rodriguez	Challenges in Familial Retinoblastoma
9	M Ashwin Reddy	Post-chemotherapy Macular Retinoblastoma: To Laser or not to Laser, That is the Question
10	Ruju Unadkat	Histopathological-Radiological Correlation of Optic Nerve Involvement in Retinoblastoma
11	Joana Providencia	Dual Retinitis with Cytomegalovirus and Herpes Simplex in a Paediatric Patient with Advanced Retinoblastoma

SESSION 3 - EYELID AND OCULAR SURFACE TUMORS

SUNDAY, JANUARY 7, 2024 (10:00 AM TO 12:00 NOON UTC)

Chairperson: Carol Karp, Vicktoria Vishnevskia-Dai

Moderator: Puneet Jain

Expert Panel: Carol Karp, Geetha Iyer, Krishha Tumuluri, Maria Pafkianaki, Sachin Salvi, Sima Das, Vicktoria Vishnevskia-Dai, Vijay Anand P Reddy

Sequence	Name	Title
1	Stephanie J Chiu	Eyelid Primitive Myxoid Mesenchymal Tumor of Infancy with BCOR Internal Tandem Duplication in A 4-year-old
2	Ritesh Verma	Endocrine Mucin Producing Sweat Gland Tumor of the Right Eyelid as a Presenting Feature of Breast Carcinoma in a Male Patient with Novel Mutation
3	Subhav Pershad	Sebaceous Cell Carcinoma: Done and Dusted
4	Delfitri Lutfi	Neoadjuvant Systemic Chemotherapy Can Successfully Reduce Extensive Eyelid Lymphoepithelioma-like Carcinoma: A Case Report
5	Rolika Bansal	A Raincheck to Extensive Squamous Cell Carcinoma
6	Ritu Arora	Anterior Segment OCTA in Monitoring Flat OSSN Resolution
7	Obaidur Rehman	Conjunctival Mucoepidermoid Papilloma with Atypia
8	Mrittika Sen	Apple of the Eye
9	Maria Manquez Hatta	Inflammatory Myofibroblastic Tumor of the Conjunctiva - Diagnosis Based on Molecular Studies
10	Irwin Leventer	Wham Bam Explosive PAM
11	Kelsey Roelofs	Management of Conjunctival Melanoma with Orbital Invasion: Alternatives to Exenteration

SESSION 4 - ORBITAL AND MISCELLANEOUS TUMORS

SUNDAY, JANUARY 7, 2024 (12:00 NOON TO 02:00 PM UTC)

Chairperson: Gangadhara Sundar, Hakan Demirci

Moderator: Mrittika Sen

Expert Panel: Gangadhara Sundar, Hakan Demirci, Hunter Yuen, Swathi Kaliki, Kaustubh Mulay, Miguel Materin, Sara Lally, Vikas Khetan

Sequence	Name	Title
1	Fairooz P Manjandavida	Epiphora - An Enigma
2	Yong Wei Wei Dayna	Disappearing Act - Magic of Neoadjuvant Chemotherapy in a Case of Lacrimal Sac Carcinoma
3	Manu Saini	A Massive Periorbital and Orbital Tumour in an Adult
4	Ruchi Goel	Well Differentiated Orbital Liposarcoma - A Diagnostic and Management Dilemma
5	Nirupama Kasturi	Congenital Neuroectodermal Tumor of the Orbit in a Newborn
6	Angela J Oh	Invasive Squamous Cell Carcinoma with Facial Nerve Palsy and Ophthalmoplegia
7	Anush Amiryan	A Case of Primary Orbital Melanoma
8	Deepthi Elizabeth Kurian	Iris Tumor in Neurofibromatosis: What Are the Chances?
9	Vicktoria Vishnevskia-Dai	Amelanotic Iris Lesion
10	Kushal U Agrawal	Vitritis in Acute Myeloid Leukemia Patient: What Could it Be?
11	Meenakshi Mahesh	The Tale of Two Children

SESSION 5 - SPECIAL SESSION

SUNDAY, JANUARY 7, 2024 (02:00 PM UTC TO 02:30 PM UTC)

Sequence	Name	Title
1	Katherine Paton	Ethics, Medicine, Organizations and Ocular Oncology: How is this relevant to you in 2024?
2	Fairooz P Manjandavida and Santosh G Hoinavar	A Curtain-raiser to ISOO 2024, Goa

SESSION 6 - ON-DEMAND ACCESS

ACCESS ON THE ISOO YOUTUBE CHANNEL FROM SATURDAY, JANUARY 6, 2024, 2:00 PM UTC

Sequence	Name	Title
D001	Arun D Singh	Primary Iris Leiomyoma
D002	Yagmur Seda Yesiltas	Necrotic Iris Melanocytoma with Secondary Glaucoma
D003	Kushal U Agrawal	Neoplastic Iris Condition Masquerading as Uveitis
D004	Miguel Hernandez-Emanuelli	The Bulge with A Nudge
D005	Saadia Razzaq Chaudhry	An Unusual Case of Ciliary Body Menaocytoma and Treatment.
D006	Dipti Gupta	A Wolf Under Sheep Skin
D007	Ravindra Kumar Saran	Ciliary Body with Rare Morphological Features
D008	Sarah B Pike	Potential of Aqueous Humor Liquid Biopsy in Diagnostically Challenging Intraocular Masses: Intraocular Ciliary Body Medulloepithelioma Masquerading as Atypical Retinoblastoma
D009	Akshay Agnihotri	Ciliary Body Melanoma with Extrascleral Extension Masquerading as a Conjunctival Nevus
D010	Sarinee Juntipwong	Acute Orbital Cellulitis and Focal Scleritis with Amelanotic Ciliochoroidal Lesion and Exudative Retinal Detachment Masqueraded Ciliochoroidal Melanoma with Scleral Invasion: A Case Report
D011	Neil Sheth	Atypical Ultrasound Biomicroscopy Appearance of a Ciliary Body Melanoma
D012	Xiaolu Yang	Local Resection Via Partial Lamellar Sclerouvectomy for Ciliary Body Tumors
D013	Shagun korla	Choroidal Melanoma
D014	Vincenzina Mazzeo	Peculiar Uveal Melanomas

SESSION 6 - ON-DEMAND ACCESS

ACCESS ON THE ISOO YOUTUBE CHANNEL FROM SATURDAY, JANUARY 6, 2024, 2:00 PM UTC

Sequence	Name	Title
D015	Helya Aghazadeh	An Atypical Presentation of Diffuse Choroidal Melanoma
D016	Hartej Singh	Stop! Eyes Have Got a Pulse
D017	Zack Oakey	De Novo Choroidal Melanoma
D018	Catriona Downie	Malignant Melanoma of the Choroid Presenting as Orbital Cellulitis and Apparent Response to Intravenous Antibiotics
D019	Prapti Praful Chheda	Retinal Capillary Hemangioblastoma: Feeder Vessel Ligation Technique
D020	Athar Shadmani	The Unexpected while Expecting - An Exceptional Journey
D021	Hung-Da Chou	Retinal Vascular Proliferation with Fibrotic Regression in Von Hippel-Lindau Disease: A Case Report and Literature Review
D023	Kushal U Agrawal	Coats Plus Syndrome with New Observation of Drusenoid Retinal Pigment Epithelial Detachments in a Teenager
D024	Subhav Pershad	Plaque Brachytherapy - A Port Wine Aficionado
D025	Georgios Blatsios	Neoadjuvant Proton Beam Radiation Therapy Followed by Endoresection of a Large Uveal Glial Tumour in a Young Patient.
D026	Akshay Agnihotri	A Cotton Wool Spot Like Lesion Unveils Underlying Tuberous Sclerosis
D027	Cristina Rastoaca	Agressive Giant Retinal Astrocytoma
D028	Shebin Salim	A Rare Case of Retinal Pigment Epithelial Adenocarcinoma in a Painful Blind Eye

SESSION 6 - ON-DEMAND ACCESS

ACCESS ON THE ISOO YOUTUBE CHANNEL FROM SATURDAY, JANUARY 6, 2024, 2:00 PM UTC

Sequence	Name	Title
D029	Prajakta S Thakur	Usual Presentation of Unusual Masquerade, Choroidal Lymphoma - A Case Report
D030	Vishal Raval	An Unusual Delayed Treatment Response to Intravitreal Methotrexate for Primary Vitreoretinal Lymphoma
D031	Hidayet Sener	Triple Whammy - A Heart Touching Story
D032	Stefan Klein	Intraocular Manifestation of Nocardia Asteroides
D033	Rolika Bansal	Behind the Curtain's Mystic Fold, Lies the Glowing Future Untold
D034	Patrick Murtagh	Cutaneous Melanoma Metastatic to the Vitreous Cavity
D035	Ridham Nanda	Leukemic Infiltrates in Blast Crisis of Chronic Myeloid Leukemia
D036	Saloni Desai	Relapse of ALL Masquerading as Ocular Involvement
D037	Hridya H	Bilateral Retinoblastoma in a Child with Simpson Golabi Behmel Syndrome Phenotype - A Possible Non-fortuitous Association
D038	Ritesh Verma	Enigma of Intraocular Mass Lesion in A 22 Year Old Male
D039	Shabana Choudhary	It is not Merely a Retinal Tumour but the Journey of Tears, Fears, Hope, and Faith
D040	Deepthi Elizabeth Kurian	Conquering Retinoblastoma: The Homestretch
D041	Dan S Gombos	Very Late Recurrence of Retinoblastoma following Primary Treatment with Intra-arterial Chemotherapy
D042	Chhavi Gupta	Double Plaque for Double Trouble

SESSION 6 - ON-DEMAND ACCESS

ACCESS ON THE ISOO YOUTUBE CHANNEL FROM SATURDAY, JANUARY 6, 2024, 2:00 PM UTC

Sequence	Name	Title
D043	Hansell Soto Ramos	Xeroderma Pigmentosum with Ocular Involvement
D044	Sonal P Yadav	Globe Salvage in Invasive Squamous Cell Carcinoma of the Orbit: Is it Possible?
D045	Bijnya Birajita Panda	Radiation Therapy in Ocular Surface Squamous Cell Carcinoma
D046	Alua Aubakirova	A Case of Conjunctival Melanoma Simulating Iris Prolapse
D047	Chhavi Gupta	An Unusual Presentation of Multifocal Conjunctival Melanoma Arising from a Nevus
D048	Efstathios Detorakis	Elevated Melanotic Ocular Surface Lesion: A Reverse Masquerade Condition
D049	Komal Bakal	A Sheep in Wolves Clothing - The Story of an Eyelid Tumor
D050	Devika Chauhan	Salivary Gland-like Duct Carcinoma of the Lacrimal Drainage System: An Exceedingly Rare Entity
D051	Christine Anggun Putri	To Exenterate or Not: Lacrimal Sac Squamous Cell Carcinoma with Orbital Involvement
D052	Ankita Aishwarya	Can I Be Ready for Marriage?
D053	Maria Manquez Hatta	A Rare Cystic Orbit Tumor to Keep in Mind
D054	Yong Wei Wei Dayna	Residual Solitary Fibrous Tumour of the Orbit
D055	Mrittika Sen	A Solitary Affair
D056	Arun D Singh	GLI-1 Amplified Spindle Cell Tumor of the Intraconal Orbit: Novel Surgical Management

SESSION 6 - ON-DEMAND ACCESS

ACCESS ON THE ISOO YOUTUBE CHANNEL FROM SATURDAY, JANUARY 6, 2024, 2:00 PM UTC

Sequence	Name	Title
D057	Yagmur Seda Yesiltas	En-Bloc Optic Nerve Resection/Biopsy
D058	Hector Gabriel M Solano	Navigating the Ocular Odyssey: A Tale of Glioma, Resilience, and Specialized Care
D059	Obaidur Rehman	Bilateral Angiolymphoid Hyperplasia with Eosinophilia in an Elderly Male
D060	Nikhil Kumar	An Unusual Case of Orbital Metastasis from Anorectal Malignant Melanoma

SESSION 1

INTRAOCULAR TUMORS 1

Clinical Course and Local Resection of Mesoectodermal Leiomyoma of the Ciliary Body

Presenting Author: Karim Al-Ghazzawi, *University Hospital Essen, Germany*, karim.al-ghazzawi@uk-essen.de

Co-authors: Ilias Diamantis, Tobias Kiefer, Miltiadis Fiorentzis, Nikolaos E. Bechrakis

Abstract

Introduction: Intraocular leiomyomas tend to involve the ciliary body but may extend or arise posteriorly in the choroid or anteriorly in the iris. Mesoectodermal leiomyoma is a rare tumor of neural origin. Its clinical diagnosis is difficult, and it's usually mistaken for an amelanotic uveal melanoma, Secondary complications include cataract and exudative retinal detachment.

Case Report: A 42year-old man was found to have a symptomatic drop of visual acuity due to increasing lens opacity on the left eye. On examination a pink ciliary body lesion with infiltration chamber angle was found. Slit lamp examination revealed atypical cataract and a 2.5 X 3 mm round, translucent, amelanotic, tan-pink stromal lesion. There were fine intrinsic vessels with surrounding conjunctival feeder vessels. The lesion was located at 6:00, approximately 2 mm from the pupillary margin. Ultrasound biomicroscopy showed a dense iris stromal mass of 5.3 X 6.1 X 3 mm with infiltration to the ciliary body, pressing on the capsular bag and lens. Immunohistochemical workup was positive for: smooth muscle actin and Caldesmon however negative for S100, Pancytokeratin and Melan-A, confirming diagnosis of intraocular mesoectodermal leiomyoma. Postoperatively within 4 years tumor growth was documented and the tumor was excised via transscleral resection in hypotension and hypothermia. Postoperatively he developed localized rhegmatogenous retinal detachment, which was treated successfully with pars plana vitrectomy and SF6 Gas tamponade. Final visual acuity was 25/25 with an IOP of 16 mm Hg.

Conclusion and Clinical Implication: Although rare, the diagnosis of primary ciliary body leiomyoma should be considered when evaluating an amelanotic circumscribed uveal tumor. Immunohistochemistry is necessary to confirm the diagnosis. Incisional biopsy might be diagnostic however extra-scleral growth can occur and excision of the tumor by adequate surgery is imminent to finally control tumor growth.

Financial Interest: *None*

S1C1

Proton Beam Radiation for Growing Choroidal Osteoma

Presenting Author: Ahad Sedaghat, *Cole Eye Institute, Cleveland Clinic, Cleveland, Ohio, USA*, sedagha@ccf.org

Co-authors: Arun D. Singh

Abstract

Purpose: To present a case of growing choroidal osteoma treated with proton beam radiation.

Case Report: A 21-year female was referred for evaluation of an amelanotic choroidal lesion in her left eye with an initial visual acuity of 20/20 in both eyes.On fundus exam a distinct well-defined, yellow-orange colored lesion, measuring 3.5x3.5x1 mm was identified supratemporal to the optic disc.The lesion appeared as an echodense mass on B-scan with both high and low gain, diagnostic of choroidal osteoma. Over 10 months, visual acuity decreased to 20/30, with evidence of growth, reaching dimensions of 4.5x4x1 mm, accompanied by the development of subretinal fluid. Three cycles of photodynamic therapy (PDT) administered over 22 months effectively resolved the subretinal fluid, restoring visual acuity to 20/20. in addition, choroidal neovascularization (CNV) was treated with intravitreal anti-vascular endothelial growth factor (VEGF) injections. Over next 7 months, the tumor grew towards the fovea, measuring 5x4.5x1 mm with foveal involvement and decreased visual acuity to 20/125. Patient was treated with proton beam radiation (Total dose: 20 Cobalt Gray Equivalent in 4 fractions).in the subsequent 30-month follow-up period, the tumor size remained unchanged (absence of growth) and visual acuity improved to 20/40.

Conclusion: proton beam radiation was effective to stop tumor growth with favorable response.

Clinical Implication: Low-dose proton beam radiation represents a potential intervention to stop tumor growth in cases of choroidal osteoma that exhibit progressive growth.

Financial Interest: *None*

S1C2

Bilateral Choroidal Osteoma in a Child with Börjeson-Forssman-Lehmann Syndrome

Presenting Author: Pranvera Emini, *Universitätsklinikum Essen, Essen, Germany*, pranvera.emini@uk-essen.de

Co-authors: Tobias Kiefer, Anna Gruber, Mathis Steindor, Nikolaos E. Bechrakis

Abstract

Introduction: The Börjeson–Forssman–Lehmann syndrome is a rare recessive disorder caused by mutations in the PHF6 gene on the X chromosome gene locus q26.2, leading to intellectual disability, epilepsy, obesity, hypogonadism and various facial and physical abnormalities as well as behavioral and developmental problems. The syndrome affects primarily males, but rarely it can also develop in females.

Case Report: An 8-year-old girl with an initially unknown syndromic disease was referred to the Pediatric Department of our University Hospital. The girl exhibited several multiorgan abnormalities such as global mental and oral developmental disorder, macrocephalus, hypermelanosis of Ito, muscle hypotonia or horizontal nystagmus. Computed tomography (CT) -scan revealed bilateral calcification of the posterior pole of both eyes, which led to the referral to the Department of Ophthalmology. Fundoscopy revealed bilateral yellowish prominences of the choroid located at the posterior pole. in context with the calcification in the CT a diagnosis of bilateral choroidal osteoma was established. Few days later, a Börjeson–Forssman–Lehmann syndrome was genetically confirmed. A connection between this syndromic disease and choroidal osteomas have not been reported in the literature so far.

Conclusion and Clinical Implication: To the best of our knowledge choroidal osteomas have not been reported in association with the Börjeson–Forssman–Lehmann syndrome.

Financial Interest: *None*

S1C3

Pigmented Choroidal Mass

Presenting Author: Elaine Binkley, *University of Iowa, Iowa City, USA*, elaine.binkley@gmail.com

Co-authors: Lola P Lozano, Chau Pham, Jennifer Menke, Nasreen Syed, H. Culver Boldt

Abstract

Introduction/Purpose: To describe a rare case of pigmented juxtapapillary choroidal schwannoma with extraocular extension mimicking choroidal melanoma.

Case Report: A 79-year-old Black male was referred for evaluation of a juxtapapillary, pigmented choroidal mass that had been noted on a routine diabetic eye exam. He was found to have a 12x11x3.9 mm dome-shaped, pigmented choroidal mass abutting the optic nerve. Echographically, the lesion was medium reflective without vascularity. There was a 3.8mm area on echography posterior to the globe that was suspicious for extraocular extension. After an extensive discussion of options including biopsy of the lesion, treatment with radiation, and enucleation, the patient elected to undergo enucleation of the left eye. Intraoperatively, there was noted to be an off-white area of extraocular tumor adherent to the superior optic nerve sheath. Pathology showed scattered large areas of Antoni A pattern tissue consistent with schwannoma involving the juxtapapillary choroid and episcleral space.

Conclusion: Choroidal schwannoma may have extraocular extension and can mimic choroidal melanoma.

Clinical Implication: While rare, the diagnosis of schwannoma should be considered for choroidal lesions, particularly in populations of patients in which choroidal melanoma is less common.

Financial Interest: *None*

S1C4

Diagnostic Aqueous Humor Proteome Predicts Metastatic Potential in Uveal Melanoma Patient with Tumor too Small to Biopsy

Presenting Author: Shreya Sirivolu, *Keck School of Medicine, University of Southern California, Los Angeles, USA,* sirivolu@usc.edu

Co-authors: Chen-Ching Peng, Sarah Pike, Mary E. Kim, Liya Xu, Jesse L. Berry

Abstract

Introduction/Purpose: Gene expression profiling (GEP) is clinically validated to stratify metastatic risk by assigning uveal melanoma patients to two highly prognostic molecular classes: class 1 (low metastatic risk) and class 2 (high metastatic risk). However, GEP requires intraocular tumor biopsy, which is limited by small tumor size, resulting in the inability to predict the metastatic risk in these patients. in this study, we seek to determine the aqueous humor (AH) proteome related to the advanced GEP class 2 using twenty diagnostic AH liquid biopsy specimens. By analyzing differential protein expression, we aim to determine the metastatic risk of a patient with a tumor too small to biopsy.

Case Report: The patient is a 71-year-old male diagnosed with choroidal melanoma in the left eye. The tumor was classified as AJCC Stage I (T1a). At diagnosis, B-scan ultrasonography demonstrated a dome-shaped choroidal lesion that measured 1.65 mm in height and 9.86 mm in largest basal diameter. This patient was treated with Iodine-125 plaque brachytherapy for one week. Given the small size of the tumor, biopsy was not able to be performed.

Conclusion: GEP classes could be differentiated by 45 AH differentially expressed proteins (DEPs), which were detected even when the tumor was too small to biopsy in three cases (posterior tumors with <2.5mm height). 31 DEPs were upregulated [fold-change (FC) >2, $P < 0.01$] and 14 were down-regulated [FC < 0.5, $P < 0.01$] in GEP class 2 compared to GEP class 1. Further analysis showed that IL1R and SPRY2 are potential upstream regulators for the 8/45 DEPs that contribute to metastasis-related pathways. Based on its 45 DEP-based classification, this case was presumed to be likely GEP class 1 with low metastatic risk.

Clinical Implication: Analyzing the proteome in the AH offers a new opportunity to determine uveal melanoma metastatic potential, with notable clinical utility for small-sized tumors that cannot be biopsied.

Financial Interest: *None*

S1C5

Bilateral Choroidal Melanoma at Presentation in a Patient with Myotonic Dystrophy

Presenting Author: Buse Guneri Beser, *University of Michigan, Kellogg Eye Center, Ann Arbor, Michigan, USA*, gbuse@med.umich.edu

Co-authors: Hakan Demirci, James Hayman

Abstract

Introduction/Purpose: Myotonic dystrophy is the most common muscular dystrophy in adulthood, with an estimated prevalence of 1/8000. in a recent meta-analysis of cancer risk in a cohort of 2779 patients with myotonic dystrophy, it was concluded that elevated risk was observed for cancers of the thyroid, endometrium, ovary, cutaneous melanoma, colon/rectum, and testis. To our knowledge, there are eleven case reports with myotonic dystrophy and uveal melanoma in the literature. But none had bilateral involvement at presentation.

Case Report: A 38-year-old male, with a history of myotonic dystrophy and rhabdomyosarcoma of the bladder status post removal thirty-seven years ago, presented with bilateral choroidal lesions. Fundus examination showed a large mushroom-shaped pigmented choroidal lesion, measuring 16x19x8.9mm, located superotemporally with associated exudative retinal detachment in the right eye and a dome shaped pigmented choroidal lesion, measuring 13x12x4.1mm, on the macula with overlying orange pigment and subretinal fluid in the left eye. Ultrasonography exhibited bilateral acoustically solid lesions with intrinsic vascularity on B-scan and medium internal reflectivity on A-scan. Fine needle aspiration biopsy of the right eye confirmed the diagnosis of choroidal melanoma. He underwent bilateral iodine-125 plaque radiotherapies at the same time. His genetic expression profile exhibited Class1B molecular signature in the right eye and Class1A in the left. Two years after the plaque surgeries, his vision was 20/200 OD and 20/125 OS. There was no sign of radiation retinopathy or systemic metastasis.

Conclusion: There might be a possible association between myotonic dystrophy and uveal melanoma.

Clinical Implication: To our knowledge, this is the first case report of primary bilateral choroidal melanoma at presentation in a patient with myotonic dystrophy. Annual ophthalmologic examination is important in patients with myotonic dystrophy.

Financial Interest: *None*

S1C6

Asymmetric Response of an Intraocular/Orbital Uveal Melanoma Treated with Stereotactic Radiation Therapy

Presenting Author: Dan S Gombos, MD *Anderson Cancer Center, Houston, Texas USA*, dgombos@mdanderson.org

Co-authors: William Morrison, Spana Patel

Abstract

Introduction/Purpose: Advanced uveal melanomas with large extraocular extension can be challenging to treat with globe salvaging modalities.

Case Report: We present a gentleman presenting with a uveal melanoma associated with a large extraocular extension. After refusing modified enucleation the intra and extraocular components were treated with stereotactic radiotherapy. Despite unform dosing the response between the two regions was asymmetric; neither recurred locally. The eye developed neovascular glaucoma but was successfully retained.

Conclusion: Stereotactic radiation therapy may be a reasonable treatment option in advanced cases of uveal melanoma presenting with orbital involvement. The Implications of asymmetric response to therapy are unclear but may nonetheless be associated with adequate local control.

Clinical Implication: Stereotactic radiation is a treatment option for patients presenting with intra & extraocular uveal melanoma.

Financial Interest: *None*

S1C7

Peripheral Exudative Hemorrhagic Chorioretinopathy Simulating Choroidal Melanoma in a Patient with Nevus of Ota

Presenting Author: Vishal Raval, *L V Prasad Eye Institute, Hyderabad, India*, drvishalraval@gmail.com

Co-authors: Saumya Jakati

Abstract

Introduction/Purpose: Nevus of Ota is a risk factor for primary uveal melanoma. Intraocular hemorrhage, although rare, is a manifestation of uveal melanoma. Peripheral Exudative Hemorrhagic Chorioretinopathy (PEHCR) is the 2nd most common mimicker of choroidal melanoma after choroidal nevus. Herein, we describe a patient with Nevus of Ota referred for melanoma in the contralateral eye, who was found on examination to have PEHCR.

Case Report: A 66-year-old male presented with complaints of diminution of vision in the left eye for 5 days. On examination, the patient had hyperpigmentation on the right side of his face, including the forehead, brow, upper lid and superotemporal conjunctiva and iris. The left eye had a conjunctival nevus approximately 1 mm in size in the nasal aspect. Right eye vision was 20/20 and left eye was hand movements. The posterior segment of the right eye revealed choroidal melanocytosis. The left eye had dense vitreous hemorrhage with no view of the retina. B scan revealed multiple dot-like and membranous echoes suggestive of vitreous hemorrhage and multi-lobulated subretinal echoes with a high index of suspicion for choroidal melanoma. The patient underwent 25G pars plana vitrectomy with a drainage retinotomy to obtain a biopsy sample. Thick loculi of blood came out. The histopathology report revealed hemorrhagic aspirate, and no evidence of tumor cells. We concluded this was a breakthrough hemorrhage due to PEHCR simulating choroidal melanoma in a patient with Nevus of Ota.

Conclusion: Nevus of Ota is a risk factor for choroidal melanoma. Since patients with PEHCR are commonly diagnosed as having posterior uveal melanoma, multimodal imaging and in rare instances biopsy should be performed to confirm the diagnosis.

Clinical Implication: PEHCR can resemble choroidal melanoma and remains a diagnostic dilemma. As such, one should be mindful of masquerades for choroidal melanoma especially in patient with Nevus of Ota.

Financial Interest: *None*

S1C8

RPE Adenoma Treated with Episcleral Brachytherapy

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Abstract

Purpose: To explore the safety and efficacy of episcleral brachytherapy as a primary management option for eyes with retinal pigment epithelial (RPE) adenoma.

Case Report: A 40 year old woman noted to pigmented fundus mass on a routine examination (10 x 9 x 3.4 mm). The tumor had dark uniform color with absence of drusen, orange pigmentation. The details of overlying retina were obscured. Prominent lipid exudation along the base of the tumor was also observed. The retinal vessels seem to lead into the tumor but the feeder vessels were not dilated. Fluorescein angiography confirmed that intrinsic vasculature of the tumor was derived from of retinal vasculature. B-scan ultrasonography revealed a dome shaped lesion that was located anterior to the choroid with high internal reflectivity on A-scan. A clinical diagnosis of RPE adenoma was made and patient observed every 3 months. At 6 month visit, the tumor was noted to have enlarged. Transvitreal FNAB was performed without significant complications. Cytology specimen revealed bland cuboidal cells with granules of pigment suggestive of RPE cells rather than choroidal melanoma. 3 years after episcleral brachytherapy (Iodine-125; 85 Gy apical dose), the tumor was regressed with surrounding chorio retinal atrophy.

Conclusion: Diagnosis of RPE adenoma can be confirmed by cytological assessment following diagnostic FNAB.

Clinical Implication: Episcleral brachytherapy is an effective management option for RPE adenoma capable of inducing long-term tumour regression while maintaining normal visual acuity.

Financial Interest: *None*

S1C9

Diagnosis of Choroidal Metastasis from Cutaneous Melanoma Based on BRAF Mutation on FNAB and Complete Response to Immunotherapy

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Co-authors: Selin Orge, Victor Elner, Hakan Demirci

Abstract

Introduction/Purpose: Choroidal metastasis from cutaneous melanoma poses a diagnostic challenge due to its rarity and the difficulty in differentiation from primary choroidal melanoma. Here we report a unique case of identifying BRAF mutation on fine-needle aspiration biopsy (FNAB) for diagnosis of choroidal metastasis from cutaneous melanoma and hence complete response with immune checkpoint inhibitors.

Case Report: A 51-year-old male, with a history of cutaneous melanoma three years ago and papillary thyroid carcinoma one year ago, presented with a choroidal lesion in the right eye. He did not have any known systemic metastasis at presentation. Fundus examination showed a partially pigmented choroidal lesion extending to the macula, with associated exudative retinal detachment. Ultrasonography showed an acoustically solid lesion with intrinsic vascularity on B-scan and mid to low internal reflectivity on A-scan. FNAB showed melanoma, and genetic testing revealed a positive BRAF V600E mutation. Nivolumab and ipilimumab therapy were started. After ten cycles, he had a 20/50 visual acuity and choroidal lesion was regressed to a flat scar tissue, with resolved exudative detachment.

Conclusion: While cutaneous and choroidal melanomas have similar embryonic origin and histological features, they differ in genetic characteristics. The presence of BRAF V600E mutation on FNAB confirmed the diagnosis in this case. Immunotherapy, either alone or in combination with other therapies, has altered the management of metastatic cutaneous melanoma. To the best of our knowledge, this is the first report showing the effectiveness of immunotherapy alone treatment in the eye.

Clinical Implication: Our findings suggest immune checkpoint inhibitors could be a safe treatment option for patients with choroidal metastasis from cutaneous melanoma to preserve the eye and vision.

Financial Interest: *None*

S1C10

Quite Rare, Not Fair

Presenting Author: Thomas M Catapano, *Wills Eye Hospital, Philadelphia, USA*, thomasc@shields.md

Co-authors: Carol L. Shields

Abstract

Purpose: To assess differential diagnosis in a case of vitreoretinal metastasis from lung adenocarcinoma as primary.

Case Report: A 76-year-old Caucasian male was referred for decrease in vision in the left eye over six months with a suspected diagnosis of vitritis and retinitis. Systemic work-up was negative for inflammatory or infective etiology. He is a former smoker with a history of left lung adenocarcinoma stage IIB (4 years back) which was treated with surgery and chemotherapy. On examination, best corrected visual acuity was 20/30 in the right eye (OD) and hand motion in the left eye (OS). Fundus examination OD was within normal limits and OS showed diffuse vitreous haze and retinal exudates at the macula and the temporal retina. Diagnostic pars plana vitrectomy OS revealed atypical epithelial cells, prominent nuclei, intracytoplasmic mucin, and mitotic figures confirming the diagnosis of vitreoretinal metastasis from lung carcinoma as primary. Although vitreous haze reduced following vitrectomy, the retinal lesions remain unchanged. He underwent external beam radiation therapy with a total dose of 4140 cGy in 23 fractions of 180 cGy each to posterior globe OS. After two months of radiation the vitreoretinal disease showed regression with no evidence of recurrence and improvement in the visual acuity OS to counting fingers at 4 feet.

Conclusion: Metastasis to the retina and vitreous remains exceedingly rare and may be overlooked in favor of more common entities, such as inflammation or infection.

Clinical Implication: Patients with aggressive systemic cancers should be encouraged of the potentially life-saving benefits of routine medical visits, including regular visits to the ophthalmologist.

Financial Interest: *None*

S1C11

SESSION 2

INTRAOCULAR TUMORS 2

Histopathological Surprise in an Intraocular Mass

Presenting Author: Abhijeet Beniwal, *RP Centre, AIIMS, New Delhi, India*, abhijeetbeni1993@gmail.com

Co-authors: Bhavna Chawla, Seema Kashyap

Abstract

Purpose: To report an unusual and rare presentation of an intraocular mass seen in a young adult

Case Report: A 25-year-old male presented with diminution of vision in the left eye for six months. On examination, visual acuity was 6/6 OD and perception of light with inaccurate projection of rays OS. Clinical examination of the left eye revealed a shallow anterior chamber, posterior synechiae, and cataractous lens. The fundus could not be visualized due to vitreous hemorrhage. The right eye was unremarkable. Ultrasonography of left eye revealed an intraocular mass lesion in the posterior segment with retinal detachment. Contrast-enhanced MRI of orbits confirmed the mass. The optic nerves were normal. Imaging features were inconclusive and a malignancy could not be ruled out. Given the poor visual prognosis, and inability to rule out malignancy, an enucleation was performed with informed consent. Histopathology revealed an astrocytic neoplasm with piloid processes, occasional rosenthal fibres, and eosinophilic granular bodies, suggestive of pilocytic astrocytoma.On IHC, it was found to be GFAP (Glial Fibrillary Acidic Protein) and Vimentin positive.

Conclusion: Astrocytoma originates in astrocytes, a kind of glial cells. Pilocytic astrocytoma is a grade I tumor, more commonly seen in the first two decades. It is the most common childhood brain tumor. Astrocytes are present in the optic nerve. However, optic nerve pilocytic astrocytoma is extremely rare. Further, in this case, the tumor was intraocular in location and the optic nerve was unremarkable. Hence, the diagnosis of pilocytic astrocytoma came as a histopathological surprise.

Clinical Implication: We report an unusual case of a pilocytic astrocytoma presenting as an intraocular mass in a young adult. The clinical and imaging features were inconclusive given the rarity of this lesion and final diagnosis could be established on histopathology and immunohistochemistry.

Financial Interest: *None*

S2C1

Unusual Phenotypic Presentation of Ocular Erdheim Chester Disease

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Co-authors: Hardeep Singh Mudhar

Abstract

Introduction/Purpose: Erdheim Chester Disease (ECD) is a rare systemic non-Langerhans histiocytic disease that most commonly affects the long bones and visceral organs. The disease can be fatal with a 5-year survival of 68%. Ocular involvement occurs in about a fifth of patients, commonly with intraconal orbital mass. We present a very rare case of intraocular ECD, with an unusual phenotype.

Case Report: 64-year-old male with a background of stable ECD affecting lung, skin and bone, managed with weekly peginterferon and filgrastim. He had no past ocular history, other than bilateral xanthelasma. He presented with incidental findings of symmetrical macular lesions, on a background of chronic low grade alternating anterior uveitis for 8 months, managed with topical steroids. Ocular oncologist examination revealed visual acuity of 6/12, pinhole 6/9 OU, subtle bilateral orange subconjunctival infiltrative lesions in the fornices, unremarkable anterior segments and bilateral macular choroidal amelanotic calcified mass lesions with no associated sub-retinal fluid or exudate. Multimodal imaging including MRI orbits will be presented, which did not detect orbital involvement. The patient underwent subconjunctival incisional biopsy as chorioretinal biopsy risked vision loss in a visually asymptomatic patient. Histology of the specimen will be presented the findings were consistent with ocular ECD.

Conclusion: Intraocular involvement is extremely rare and can be the presenting feature of this multisystem disease. To our knowledge, this is only the second case to describe calcific choroidal lesions which resemble osteoma, and the first to describe this change in conjunction with subconjunctival disease.

Clinical Implication: Ophthalmologists need to be aware of the manifestations of ECD as early diagnosis can improve survival and visual prognosis. Identification of extraocular sites of disease can prevent morbidity associated with intraocular biopsy, as in our case.

Financial Interest: *None*

S2C2

A Rare Pseudoretinoblastoma in a Child

Presenting Author: Ayushi Agarwal, *Operation Eyesight Universal Institute for Eye Cancer, L V Prasad Eye Institute, Hyderabad, India,* ayushiagarwal.2110@gmail.com

Co-authors: Dilip K Mishra, Swathi Kaliki

Abstract

Purpose: To present an exceedingly rare case of Retinal Pigment Epithelium (RPE) adenoma simulating as retinoblastoma.

Case report: A 12-year-old boy presented with reduced vision and white reflex in right eye (RE) since childhood. RE visual acuity was light perception and a giant amelanotic yellowish-white lesion was noted behind the crystalline lens with overlying dilated, tortuous vessels. RE ultrasonography revealed hyperechoic lesion in the vitreous cavity with low to moderate internal reflectivity and hyperechoic dot opacities with high internal reflectivity within the lesion suggestive of intraocular mass with calcifications. T1W Magnetic resonance imaging revealed hyperintense right intraocular lesion, which was hypointense on T2W-MRI. A provisional diagnosis of RE retinoblastoma was established and enucleation with implant was performed. Gross examination showed a gray-white homogenous mass, filling the entire vitreous cavity with detached retina. Histopathological evaluation revealed a nodular tumor attached from the RPE. These cells had round nuclei exhibiting mild pleomorphism and abundant vacuolated cytoplasm, with intercellular septations visible in Periodic Acid Schiff stain. Ciliary body was free of the tumor. Ki-67 index was <2%. Immunohistochemistry revealed positivity for S-100 and pan-cytokeratin. Synaptophysin, CK-7, CK-20, Epithelial membrane antigen, HMB-45, TTF-1, and Melan-A were not expressed. Based on clinicopathological correlation, a definitive diagnosis of primary RPE adenoma of the RE was confirmed.

Conclusion: This is the youngest reported case of RPE adenoma, which mimicked a retinoblastoma, owing to the younger age at presentation, amelanotic nature of the lesion, and imaging findings suggestive of intraocular mass with calcification.

Clinical Implication: This report highlights the rarity of RPE adenoma simulating as retinoblastoma in a child and the significance of considering it amongst the differential diagnoses of leukocoria.

Financial Interest: *None*

S2C3

An Incidental Rb1 Pathogenic Sequence Variant in a Fetus

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Co-authors: Mattan Arazi, Eitan Friedman

Abstract

Purpose: To report the management and outcome of an incidental RB1 pathogenic sequence variant (SV) detected on unrelated genetic testing in a fetus.

Case Report: A 37-year-old woman at 29 weeks of gestation, underwent amniocentesis due to a suspected polycystic kidney (PCKD) detected on routine fetal ultrasonography. While no genetic abnormality associated with PCKD was identified on whole genome sequencing (WES), an incidental finding of a heterozygous c.501-2A<G SV in the RB1 gene was noted. The SV, which had not been previously reported, was deemed pathogenic by the genetic lab. There was no family history of retinoblastoma, and ophthalmic evaluation of both parents showed no suspected retinomas. Further WES of the parents confirmed that the father harbored the same genetic SV as the fetus, and RNA sequencing demonstrated a disruption in the splice site, but showed that the reading frame remained intact, downgrading the SV to be classified as a Variant of Undetermined Significance. Fetal MRI performed at 38 weeks gestation did not demonstrate any intraocular or intracranial abnormalities. The parents proceeded with the pregnancy, and the child was born at 39+6 weeks with no complications. Examination following delivery demonstrated no evidence of intraocular tumors. The child, currently 10-months-old, continues to be periodically evaluated and has not been found to have any pathogenic ocular abnormalities.

Conclusion: An incidental RB1 pathogenic SV detected in a fetus poses clinical and ethical dilemmas, both to the clinician and family. Further clinical and genetic investigations are instrumental in order to reach the best outcome.

Clinical Implication: As genetic analyses increase, incidental, potentially pathogenic SVs may become more common, including in the RB1 gene. Further investigations, including WES and RNA analysis are crucial to disclose the real nature of the SV, obviating in selected cases the need for early delivery or pregnancy termination.

Financial Interest: *None*

S2C4

Retinoblastoma with MDM4 Amplification Diagnosed from Cell-free DNA in Aqueous Humor

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Co-authors: Heidy Paniagua, Liya Xu, Jesse Berry

Abstract

Introduction: Retinoblastoma (RB) poses molecular diagnostic challenges due to risk of extraocular cancer spread in invasive tissue biopsies. Aqueous humor (AH) is a minimally invasive high-yield liquid biopsy source of tumor information to monitor treatment response and identify primary oncogene amplifications such as MDM4. We report clinical features and outcome of one MDM4-amplified RB case identified genetically by AH sampling.

Case Report: A 35-month-old male with 2-3 week history of leukocoria in the left eye was diagnosed with unilateral RB (Group D/AJCC Stage cT2B). There was a c.1333-5_1333del RB1 mutation in the AH with no evidence of RB1 germline mutation. 6 cycles of systemic chemotherapy and 15 intravitreal chemotherapy injections of melphalan (25-50mcg) and topotecan (25-50mcg) were administered. AH samples from 17 timepoints throughout treatment were subject to cell-free DNA extraction and whole-genome sequencing. While there was no evidence of 6p gain, a negative prognostic biomarker, there was focal amplification of MDM4 on 1q32. Tumor fraction (TFx), the proportion of tumor-derived cell-free DNA, remained elevated (>25%) during melphalan monotherapy but significantly decreased after 6 combined topotecan and melphalan injections. Continued topotecan monotherapy led to a final AH TFx < 1.6%, indicating disease regression.

Conclusion: Using AH as a liquid biopsy enables detection of molecular biomarkers. Rarely, amplification of MDM4 is seen as in this case with aggressive tumor behavior. Monitoring TFx showed initial chemotherapy resistance with TFx > 25%. Using intravitreal topotecan (initially with melphalan) increased treatment effectiveness, correlating with decreases in TFx. At a clinical follow up of >30 months, this eye is now salvaged.

Clinical Implication: Mutational analysis from AH can inform treatment plans in RB. Focal MDM4 amplification may be relevant for a subset of the RB population and correlate with an aggressive phenotype.

Financial Interest: *None*

S2C5

Retinoblastoma Discordant Monozygotic Twins and the Discovery of an Intron 15 Rb1 Variant

Presenting Author: Hilary Racher, *Impact Genetics, Ontario, Canada*, racherh@dynacare.ca

Co-authors: Mario Zanolli, Diego Ossandon, Jaime Jessen

Abstract

Introduction/Purpose: in most families with heritable RB, all individuals who inherit the pathogenic variant develop bilateral tumors. Monozygotic twins harboring germline RB1 variants would generally display concordant phenotypes. Furthermore, 100% of patients presenting with bilateral RB carry a constitutional RB1 variant putting them at risk for life-long RB related cancers. Many large multigene hereditary cancer next generation sequencing (NGS) panels do not interrogate intronic regions known to harbor RB1 variants.

Case Report: Discordant monozygotic twins, one presented with bilateral RB at 11 months, while his identical brother was unaffected. The affected twins blood sample was initially tested via a large multi-gene NGS panel which was negative. Subsequently a second blood sample was sent to Impact Genetics, who identified a c.1421+18_1421+38del RB1 intron 15 deletion. This deletion has been previously identified in the literature in which all 3 carriers presented with bilateral RB, and by Impact Genetics in a unilateral RB patient. Testing performed on the unaffected twin showed he carried the same intron 15 deletion. Given the twin remains unaffected, confirmed via MRI at 17 months, suggests that this RB1 deletion is likely a reduced penetrance variant. Parents could also be carriers of the RB1 variant posing a risk of up to 50% for future pregnancies.

Conclusion: While all bilateral RB patients are assumed to carry a constitutional RB1 pathogenic variant, it is still recommended as standard of care to confirm the genetic diagnosis.

Clinical Implication: Recognizing the potential reduced penetrance of a RB1 variant warrants testing of all at-risk relatives even if unaffected with RB, as risk remains for other RB related cancers. Labs performing RB1 analysis should ensure they include known regions of the RB1 gene that harbor disease-causing variants.

Financial Interest: *Hilary Racher and Jaime Jessen are salaried employees of Impact Genetics*

S2C6

Phenotypic Biomarkers of Aqueous Humor Extracellular Vesicles from a Retinoblastoma Eye

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Co-authors: Annie Amacker, Bibiana Reiser, Liya Xu, Jesse Berry

Abstract

Purpose: Retinoblastoma (RB) is the most common pediatric intraocular cancer, yet tumor biopsies are contraindicated. This challenge emphasizes the need of non-tissue biomarkers. Liquid biopsy aqueous humor (AH) provides great potential for intraocular tumor information. Extracellular vesicles (EVs) have emerged as promising biomarkers. Our group has previously proposed that CD63/81+ EVs are found in AH and associated with retinoblastoma, but specific tumor-derived EV surface markers have not been identified. in this study, we employed Macsplex, a high-dimensional magnetic bead-based immunoassay, to further explore RB derived EV.

Case Report: The patient is a 64-month-old male diagnosed with unilateral RB, International Intraocular Retinoblastoma Classification (IIRC) Group E, clinical stage cT3b. Spherical vitreous seeds were identified at diagnosis. Seeding was present in four quadrants. This patient did not have high risk pathology.

Conclusion: Macsplex was used to analyze the expression profiles of 39 surface markers on EVs by using specific capture beads. Tetraspanin positive EVs bound to the beads were then incubated with APC conjugated tetraspanin antibodies. The resulting mean fluorescence intensity (MFI) from APC could be measured by flow cytometry. The MFI signals from each beads indicated the relative expression levels of the specific surface antigens. Analysis of the RB AH demonstrated a strong CD133 signal which is a known cancer stem cell marker. By using single tetraspanin or triple tetraspanin antibodies for detection, the Macsplex results revealed a higher coexpression of CD63 and CD81 with CD133 than with CD9.

Clinical Implication: This is the first study identifying the expression of CD133, a cancer stem cell marker, on retinoblastoma derived EVs in AH. CD133 was found to be associated with CD63/81+ EVs. Macsplex technology illustrates an incredible clinical potential of AH EVs for characterizing retinoblastoma phenotype in vivo.

Financial Interest: *None*

S2C7

Challenges in Familial Retinoblastoma

Presenting Author: Aurora Rodriguez, *Creighton University, Phoenix, Arizona, USA*, aurorarodriguez@creighton.edu

Co-authors: Eric Hansen, Todd Abruzzo, Luis Goncalves, Aparna Ramasubramanian

Abstract

Introduction/ Purpose: Familial retinoblastoma has an autosomal dominant inheritance pattern and 90% penetrance. The inherited dysfunctional RB1 allele predisposes the child to a higher risk of developing retinoblastoma as well as other secondary cancers.

Case Report: A family had three generations of retinoblastoma. Mother had bilateral retinoblastoma and osteosarcoma; maternal grandmother had bilateral retinoblastoma, nasopharyngeal carcinoma, and thyroid carcinoma. Mother and grandmother had external beam radiation therapy. The oldest child (currently 4) was diagnosed with bilateral retinoblastoma at 6 weeks of age. The child had bilateral macular tumors and was controlled with systemic chemotherapy, intra-arterial chemotherapy in both eyes, intravitreal chemotherapy in the left eye, and chemoplaque in the left eye. For the families next child, prenatal ultrasounds were commenced at 30 weeks, tumors were identified, the child was delivered at 37 weeks and the tumors were treated. For their third child the family decided to undergo preimplantation genetic testing facilitated by in-vitro fertilization, embryo selection based on genetic analysis, and an embryo has been placed on long-term preservation.

Conclusion: Families with mutation of the retinoblastoma gene (RB1) interested in having children should consider early retinoblastoma screening and diagnosis because prompt implementation of treatment has been associated with better vision and treatment outcomes in the child.

Clinical Implication: Individuals with a family history of retinoblastoma are strongly advised to obtain genetic testing for a retinoblastoma gene mutation. Early retinoblastoma screening can be done during the prenatal and immediate postnatal period. Prenatal screening options include preimplantation, early pregnancy, and late pregnancy screening.

Financial Interest: *None*

S2C8

Post-chemotherapy Macular Retinoblastoma: To Laser or not to Laser, That is the Question

Presenting Author: M Ashwin Reddy, *Royal London Hospital, London, UK*, mashwinreddy@hotmail.com

Co-authors: Guy S Negretti, Mandeep S Sagoo

Abstract

Introduction/Purpose: The role of laser/TTT (transpupillary thermotherapy) for patients with macular retinoblastoma requiring six cycles of systemic chemotherapy will be discussed with an illustrative case and literature review.

Case Report: A 13-month-old girl presented with unilateral unifocal Group C (cT2a) retinoblastoma. After 6 cycles of systemic chemotherapy (Carboplatin, Etoposide and Vincristine) she had Type III regression of her macular tumour. She was found to have mosaicism of a pathogenic variant of the RB1 gene. No further treatment was given and she was stable after 13 years.

Conclusion: This child did not relapse after chemotherapy alone. A Cochrane review has shown that there is weak evidence to treat with focal therapy post-chemotherapy in such cases. Other relevant studies will be presented with respect to vision.

Clinical Implication: Patients who have post-chemotherapy laser/TTT for macular tumours relapse at the same rate as those who are treated solely with chemotherapy. Whether adjuvant laser/TTT is given or not will depend on several factors 1)guaranteed follow-up to detect relapse 2) status of other eye 3) anxiety and experience of treater.

Financial Interest: *None*

S2C9

Histopathological-Radiological Correlation of Optic Nerve Involvement in Retinoblastoma

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Co-authors: Kaustubh Mulay, Ravi Varma, Santosh Honavar

Abstract

Purpose: To analyze correlation between magnetic resonance imaging (MRI) and histopathology for optic nerve extension in a case of retinoblastoma.

Case report: A 4-year-old girl presented to us with unilateral retinoblastoma International Classification of Retinoblastoma (ICRB) group E with clinical high-risk factors, MRI didn't show any optic nerve extension. Primary enucleation with implant was performed for her followed by histo-pathological evaluation. On histopathology, retrolaminar optic nerve extension (ONE) up to 5mm from sclera was noted, which warranted a need for adjuvant chemotherapy. This intriguing case analysis made us inquest further and to our surprise we found cases with similar clinical case presentation but results of correlation between MRI and histopathology for optic nerve extension were inconsistent and contradictory to our previous case findings.

Conclusion: MRI doesn't always predict optic nerve extension in retinoblastoma. Histopathological evaluation is always a gold standard in evaluating ONE and for deciding further management protocol.

Clinical Implication: While the role of MRI in retinoblastoma is invaluable, its utility in predicting ONE is modest.

Financial Interest: *None*

S2C10

Dual Retinitis with Cytomegalovirus and Herpes Simplex in a Paediatric Patient with Advanced Retinoblastoma

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Co-authors: I. Figueiredo, Madalena Monteiro, Cristina Fonseca, Guilherme Castela

Abstract

Introduction: Herpesvirus retinitis, specifically cytomegalovirus (CMV) and herpes simplex virus (HSV) are greatly prevalent viruses that can cause opportunistic infections in patients with suppressed immune system. We report a case of a non-necrotizing retinitis caused by dual infection by CMV and HSV1 in a paediatric patient under immunosuppressive treatment for advanced stage retinoblastoma.

Case report: A 4-year-old boy with past ocular history of enucleation of left eye for locally advanced retinoblastoma, currently on adjuvant systemic immunosuppressive chemotherapy complicated by bone marrow aplasia, was routinely observed under anaesthesia for periodic disease follow-up. Right eye fundus evaluation showed areas of focal haemorrhagic retinitis with no retinal necrosis on mid and far periphery. Aqueous Polymerase Chain Reaction (PCR) was requested and revealed dual positivity for CMV and HSV1. The patient was successfully managed with intravitreal foscarnet injections, intravenous ganciclovir and foscarnet. The retinitis lesions regressed gradually, leaving behind areas of chorioretinal scarring at two-month follow up. The patient is currently stable on prophylactic dose oral foscarnet.

Conclusion: in paediatric patients undergoing immunosuppressive chemotherapy, infectious cause must always be suspected and ruled out. Although very rare, dual viral infection can cause atypical retinitis in predisposed individuals. PCR from ocular fluids should be performed earliest in cases of poor response to therapy or atypical clinical presentation.

Clinical Implication: Awareness of opportunistic infections in immunossupressed retinoblastoma patients

Financial Interest: *None*

S2C11

SESSION 3

EYELID AND OCULAR SURFACE TUMORS

Eyelid Primitive Myxoid Mesenchymal Tumor of Infancy with BCOR Internal Tandem Duplication in A 4-year-old

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Co-authors: Bashar M Bata, Dan Yeomanson, Sachin M Salvi

Abstract

Introduction/Purpose: We present an interesting case of primitive myxoid mesenchymal tumor of infancy (PMMTI) with BCOR internal tandem duplication (BCOR-ITD) in a 4-year-old child.

Case Report: A 4-year-old previously well boy presented with a 6-week history of rapidly progressive right lower lid swelling. There was no family history of cancer. His vision was unaffected. Due to suspicion of rhabdomyosarcoma, he underwent an MRI head/orbit and core needle biopsy under a general anaesthetic. The MRI showed a 2.5x2x1.2 cm encapsulated mass involving the right lower lid without intraorbital extension. The biopsy was non-confirmatory, so he underwent excision. Histopathology was consistent with a primitive myxoid mesenchymal tumour of infancy with BCOR-ITD. Staging showed no metastases. Following MDT discussion, adjuvant radiotherapy was not given due to his age. Six months later, he presented with recurrence at the core needle biopsy site. He underwent further resection, but 9 months later, recurrent nodules were noted. He underwent further surgical excision with adjuvant proton beam therapy (PBT). He tolerated this fairly well with expected skin and ocular surface toxicity. Two years later, he is doing well without any recurrence of the tumour or metastases. There are adnexal changes in keeping with post-radiation therapy, which are currently managed conservatively.

Conclusion: PMMTI is a rare sarcoma with less than 25 reported cases, preferentially affecting infants. This case occurred in a slightly older child. Complete surgical excision is often effective. Recurrence at the deeper margin of surgical excision necessitated proton beam therapy, which has so far induced remission.

Clinical Implication: To our knowledge, this is the first reported case of adnexal PMMTI. Due to initial suspicion of rhabdomyosarcoma, biopsy was performed but resulted in local recurrence at the biopsy site. Surgical excision of recurrence with adjuvant PBT appears curative.

Financial Interest: *None*

S3C1

Endocrine Mucin Producing Sweat Gland Tumor of the Right Eyelid as a Presenting Feature of Breast Carcinoma in a Male Patient with Novel Mutation

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Co-authors: Aminder Singh, Kunal Dhall, Yesha Gupta

Abstract

Introduction: Endocrine mucin producing sweat gland carcinoma (EMPSGC) is a rare indolent tumor with predilection for periocular skin of elderly women. Though recurrences may occur, metastases have not been reported so far. We present a case of EMPSGC of the eyelid as a presenting feature of invasive carcinoma of the breast in a 71-year-old male patient.

Case Report: A 71-year-old male presented with a right eyelid mass from past 6 months. The lesion was bilobed, granular with large surface vessels suggestive of malignant pathology. A wide excision biopsy with intraoperative frozen section margin control and reconstruction was done. Histopathology and immunohistochemistry were suggestive of EMPSGC. On systemic exam, the patient had a breast mass 2x3x1cm which he did not reveal at presentation. A Trucut biopsy was performed and the histopathology was similar to the eyelid tumor. On PET scan, there was avid uptake in the breast mass, axillary and mediastinal lymph nodes and multiple nodular lesions in lungs were noted. The patient then underwent germline BRCA1/2 mutation test which revealed a variance of uncertain significance (possibly novel). The patient was then started on hormonal therapy and the tumor showed significant regression on successive PET scans.

Conclusion: EMPSGC has been reported in the literature but this is the first case with eyelid metastasis from the breast. We will discuss the management of the eyelid lesion and compare the histopathology and IHC of the primary and metastatic tumor.

Clinical Implication: Breast carcinoma is rare in males and breast examination is not performed routinely in males. EMPSGC is also a rare eyelid tumor affecting periocular skin of elderly women. Detection of metastatic breast carcinoma from eyelid tumor in a male patient makes this case a unique with ample learning points.

Financial Interest: *None*

S3C2

Sebaceous Cell Carcinoma: Done and Dusted

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Co-authors: Kaustubh Mulay, Santosh G Honavar

Abstract

Introduction: Sebaceous gland carcinoma (SGCa), is the most common eyelid malignancy in India. It is largely misdiagnosed owing to its ability to masquerade, with nodular variant simulating a chalazion & intraepithelial (pagetoid) variant simulating unilateral blepharoconjunctivitis.in this case, we highlight the step-wise approach to a case of SGCa and its management respecting the oncological principles.

Case report: a 71-year-old male presented with complaints of ulceration of the upper lid right eye since 20 days, history of multiple episodes of red eye since 5 years and no contributory systemic history. On evaluation, visual acuity was 6/6 both eyes and a 8x3x2mm ulcerated lesion involving the lid margin, obliteration of meibomian gland orifices,congestion & thickening of the upper tarsal conjunctiva suggestive of surface inflammation.There was no evidence of regional lymphadenopathy. Clinically, a diagnosis of pagetoid variant of SGCa accompanied with surface inflammation was made. Following a short course of topical steroids for inflammation, a conjunctival map biopsy from 17 sites ruled out intra-epithelial malignancy. Excision biopsy with a 4mm clinically clear margin with intra-operative frozen section for margin control was performed followed by lid reconstruction by modified reverse Hughe's procedure. Histopathology confirmed the clinical diagnosis. On 1-year follow up visit, no evidence of local recurrence was seen with an acceptable lid height, no ectropion/entropion & no lagophthalmos.

Conclusion: A comprehensive ocular exam with keen observation of differential laminar involvement, regional exam & systemic evaluation to rule out metastasis are keystone in an oncological workup.

Clinical Implication: Pagetoid variant of SGCa, may have an overlying inflammatory component which needs to be addressed prior to an optimal assessment of the extent of involvement.An intraoperative margin control confirmation by frozen section followed by appropriate reconstruction of the lid is the standard of care to minimise risk of local recurrence and regional spread of SGCa.

Financial Interest: *None*

S3C3

Neoadjuvant Systemic Chemotherapy Can Successfully Reduce Extensive Eyelid Lymphoepithelioma-like Carcinoma: A Case Report

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Abstract

Introduction: To report the efficacy of neoadjuvant systemic chemotherapy in the management of extensive eyelid sebaceous gland carcinoma (SGC) to avoid invasive surgery.

Case Report: A 71-year-old woman complained of progressive edematous tumor in the right upper eyelid for four months. Right eye examination found diffuse eyelid thickening tumor extended from upper eyelid to anterior orbital superior. On examination, no palpable lymph nodes were found. Incisional biopsy histopathology revealed sebaceous gland carcinoma. We consulted the patient with a hematologist and gave her two cycles of neoadjuvant chemotherapy with paclitaxel and carboplatin for three weeks interval. After two cycles of chemotherapy, the tumor gave a good response and was limited in 2/3 lateral of the right upper eyelid. We performed wide eyelid excision biopsy under frozen section control and Cuttler-Beard reconstruction procedure for better cosmesis.

Conclusion: Neoadjuvant chemotherapy is a promising and safe treatment strategy for patients with locally advanced eyelid SGC providing adequate tumor volume reduction with the additional benefit of regional tumor control during surgery. This combination treatment gives significant benefits of organ preservation, and patient cosmesis also downstages the disease and lowers the risk of distant metastasis.

Clinical Implication: in the beginning, complete treatment for this patient required extensive surgery, orbital exenteration, or more than 75% loss of eyelid tissue which can cause a cosmetic issue. Neoadjuvant chemotherapy gives a good response with more than 75% reduction in tumor size, so we can perform tumor-free margin excision surgery with reconstruction to achieve the better function of the eye.

Financial Interest: *None*

S3C4

A Raincheck to Extensive Squamous Cell Carcinoma

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Abstract

Introduction: Diffuse extensive squamous cell carcinoma is often inoperable due to its extent and surgical resection may lead to possible post-operative disfigurement or might require exenteration (based on extent). Herein, we demonstrate the efficacy of check-point inhibitors in reducing tumor extent and bringing them to operable state.

Case report: A 55-year-old Caucasian female was referred with an eyelid and conjunctival lesion persisting for 6 months. On examination, the visual acuity was 20/20 in both eyes. The left eye showed a massive lesion involving the left lower eyelid medially, inferior fornix and medial bulbar conjunctiva with feeder vessel, intrinsic vascularity and overlying leukoplakia. After biopsy-based diagnosis of squamous cell carcinoma, the patient was started on cemiplimab which showed a variable response over the 3-weekly 6 cycles of cemiplimab, a programmed death receptor (PD-1) check-point inhibitor. Considering a possible increase in the lesion size at the end of 6 cycles, surgical excision was carried out which surprisingly showed excellent response to cemiplimab with pathologically proven 20% viable tumor which was completely excised. The patient is scheduled to receive cemiplimab regularly for the next one year as maintenance therapy to prevent tumor recurrence.

Conclusion: Check-point inhibitors provide a promising role in treating extensive lesions, hence making them operable and preventing mutilating surgeries like exenteration.

Clinical Implication: Better aesthetic outcomes and longevity can be achieved by the advent of check-point inhibitors in extensive squamous cell carcinoma, thus providing a rain check to tumor recurrence and bringing them to operable size.

Financial Interest: *None*

S3C5

Anterior Segment OCTA in Monitoring Flat OSSN Resolution

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Abstract

Purpose: To monitor OSSN resolution with AS-OCTA

Case Report: A 58-year-old male presented with decreased vision associated with prominent vascular lesion in the inferior nasal conjunctiva of right eye. There was associated whorl like superficial corneal opacification overlying the pupillary area. Minimal elevation was also present from 3-5 O'clock limbus.Imprint cytology from superior and inferior margin of the limbal lesion suggested features consistent with high grade OSSN. He was started on topical 1 million unit Interferon alpha 2b four times a day. The corneal lesion disappeared completely at four weeks, however nasal elevated vascularity persisted. AS-OCTA images obtained using swept source OCT system (PLEX Elite 9000;Carl Zeiss Meditec) equipped with 10 D optical adaptor lens with a 3x3 mm scan pattern revealed dense crisscross vasculature, and the B scan showed deep flow. Topical interferon alpha 2 b was continued and patient was followed up monthly. Clinically the vascular lesion started regressing at 2 months and completely disappeared at six months after initial presentation. Monthly AS-OCTA follow up revealed marked reduction and disappearing of zigzag vascular pattern at conjunctival epithelium at limbus, especially in the deeper layers of conjunctival epithelium. No deep flow was picked up at six months. ASOCTA picture has remained same at further 6 months. Interferon alpha 2b was stopped at four months after initial presentation.

Conclusion: AS-OCTA is a useful adjunct to slit lamp biomicroscopy to evaluate the clinical resolution of OSSN.

Clinical Implication: AS-OCTA can be a useful tool to monitor the regression of OSSN with vascularisation especially when topical chemo/immunotherapy is used for the management.

Financial Interest: *None*

S3C6

Conjunctival Mucoepidermoid Papilloma with Atypia

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Abstract

Introduction: Mucoepidermoid papillomas are rare benign conjunctival lesions. These lesions are inverted papillomas clinically and histopathologically. We report a rare case of conjunctival mucoepidermoid papilloma with atypia.

Case Report: A 39-year-old female complained of a pink mass beneath the right upper eyelid for the past 6 months. The swelling had progressively increased in size and a single episode of bleeding was reported. A pinkish pedunculated mass was noted in right upper fornix, extending to the bulbar conjunctiva. The fleshy mass had well-defined margins with a papillary surface and hair-pin loop vessels on the surface. There was no feeder vessel or keratin on the surface. On palpation, consistency was soft and the lesion was freely mobile over the conjunctival surface. Anterior segment OCT showed a dome-shaped lesion with corrugated surface and intrinsic spaces, but without any scleral involvement. An excision biopsy by no touch technique and double freeze thaw cryotherapy at the margins was performed. Histopathology revealed papillary fronds with mucinous areas, with mild cellular atypia and an intact basement membrane. A diagnosis of mucoepidermoid variant of papilloma with mild atypia was made. The patient received topical interferon alpha-2b eyedrops postoperatively. At the last follow-up, the patient is doing well with early forniceal symblepharon and no signs of recurrence.

Conclusion: Conjunctival papillomas usually have a squamous epithelium and rarely, may be mucoepidermoid variants, which are inverted in nature. Atypia may be seldom noted. We report a mucoepidermoid papilloma, which was not inverted clinically and histopathologically.

Clinical Implication: Conjunctival papillomas may be associated with human papilloma virus and share features with Ocular surface squamous neoplasia. High clinical suspicion with histopathological confirmation is necessary in these uncommon cases. Recurrence is common, if not managed properly.

Financial Interest: *None*

S3C7

Apple of the Eye

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Abstract

Introduction/Purpose: To report a rare case of a caruncular ocular surface squamous neoplasia (OSSN) with amyloidosis.

Case Report: A 44-year-old male patient presented with a caruncular mass in the right eye. His visual acuity was 6/6 in both eyes. The patient gave a history of a lacrimal surgery done at 10 years of age. There was no proptosis or limitation of movements. An excision biopsy with double freeze thaw cryotherapy to the base was done. Histopathology showed intraepithelial dysplasia. The stroma showed acellular eosinophilic material that showed positive staining with Congo Red and apple green birefringence under polarized light.

Conclusion: The patient was diagnosed with ocular surface squamous neoplasia with dysplasia and amyloidosis. Evaluation for systemic involvement was advised.

Clinical Implication: Amyloidosis of the eye can be localized or secondary to systemic involvement. Cases of localized amyloidosis in patients with oral or vulvar epithelial dysplasia have been reported but this appears to be the first case of OSSN with amyloidosis.

Financial Interest: *None*

S3C8

Inflammatory Myofibroblastic Tumor of the Conjunctiva - Diagnosis Based on Molecular Studies

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Co-authors: Pablo Zoroquiain, Pablo Vigorena Salomon, Eugenio V

Abstract

Purpose: To report a very rare type of conjunctiva tumor, its differential diagnosis and test that providred definitive diagnosis

Case Report: A 34-year-old white female with no eye prior history, consult due to subconjunctival red lesion, supero nasal area in OD, in January 2022. No pain associated. Scleritis diagnosis was made. She started on topical steroid. After 1 month with no improvement, she asked for second opinion. Steroids were stopped. Images were done, those included anterior segment OCT, UBM. Surgery was performed in Sept 2022. Surgical findings revealed a 4mm lesion unattached to sclera, intimately attached to medial rectus muscle insertion. Only 90% of the tumor was excised to avoid sectioning medial rectus muscle. Path report was spindle cell tumor. Differential diagnosis included: Spindle cell carcinoma, Spindle cell melanoma, Nerve sheath tumor, Leiomyoma, Solitary fibrous tumor, Fibromatosis, Nodular fascitis and Inflammatory myofibroblastic tumor. Histopathological analysis showed a low-grade spindle cell neoplasm with a scant inflammatory component. Immunohistochemical analysis revealed myofibroblastic differentiation. Next-generation sequencing analysis demonstrated the TMP3-ALK fusion.

Conclusion: The final integrated diagnosis was an inflammatory myofibroblastic tumor. This very rare tumor is a neoplasm of low metastatic potential composed of spindle myofibroblastic cells accompanied by an inflammatory infiltrate. Localization in the eye is rare. Most cases arereported in Orbit and lacrimal gland. There are only 4 cases in conjunctiva- subconjunctival (none with molecular studies). To our knowledge this case would be the first one supported with molecular studies.

Clinical Implication: Inflammatory myofibroblastic tumors should be in the differential diagnosis of subconjunctival lesions. Diagnosis is based on morphology, but molecular studies are desirable. Locally aggressive, but unlikely metastatic.

Financial Interest: *None*

S3C9

Wham Bam, Explosive PAM

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Co-authors: Carol L. Shields

Abstract

Introduction: Primary acquired melanosis (PAM) is an acquired pigmentation of the conjunctival epithelium that has malignant potential when involving more than two clock hours and/or with histo-pathologically proven moderate to severe atypia. Herein, we present a case of extensive conjunctiva melanoma arising from primary acquired melanosis.

Case Report: A 68-year-old Asian male presented elsewhere with a right upper eyelid nodule, progressively increasing over 4 months. Transdermal excision biopsy elsewhere showed extensive malignant melanoma with involvement of the tarsus, muller's muscle and forniceal conjunctiva. On presentation to us, the right eye showed extensive primary acquired melanosis (superior fornix, medial fornix, and 80% of the inferior fornix) and a 60x50x40mm right upper eyelid mass. Excision biopsy was performed, and pathology results indicated invasive melanoma associated with primary acquired melanosis with severe atypia with positive margins. Biomarker testing was positive for v-Raf murine sarcoma viral oncogene homolog B (BRAF) and Kirsten rat sarcoma virus (KRAS). Positron emission tomography scan showed no evidence of residual or metastatic disease. The sentinel lymph node was negative and PD-1 inhibitor immunotherapy was prescribed under the care of a medical oncologist. Close follow-up has been advised with a potential need for exenteration in the future.

Conclusion: Primary acquired melanosis with severe atypia has a high degree of malignant potential. When left untreated, it can progress to fulminant and invasive melanoma, putting life at risk. This should prove a cautionary tale for close monitoring of primary acquired melanosis with prompt referral to a specialist.

Clinical Implication: Despite its appearance as innocent conjunctival pigmentation, primary acquired melanosis can become quite severe and life threatening. Prompt assessment and biopsy by a specialist is critical for further management in light of its malignant potential.

Financial Interest: *None*

S3C10

Management of Conjunctival Melanoma with Orbital Invasion: Alternatives to Exenteration

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Co-authors: Antoni Ribas

Abstract

Introduction: Locally advanced conjunctival melanoma has traditionally been managed with exenteration; however, advances in targeted and immunotherapies may provide opportunities for globe sparing treatment.

Case Report: An 84-year-old man presented with a 7-week history of right upper eyelid fullness. An incisional biopsy had been performed at an outside hospital, confirming the diagnosis of melanoma. Upon presentation to our service, he had a large, predominantly amelanotic superior forniceal conjunctival tumour. Extraocular motility was limited to 40 degrees in adduction. MRI orbits confirmed anterior orbital extension of the tumour and PET-CT was negative for nodal or distant metastatic disease (AJCC 8th edition T3c). Next-generation DNA sequencing identified a BRAFV600E mutation. With the goal of avoiding exenteration, pembrolizumab monotherapy was initiated. After the first two infusions there has been a reduction in tumour size.

Conclusion: Conjunctival melanoma carries a similar mutational profile to cutaneous melanoma. There have been a handful of cases of locally advanced conjunctival melanoma successfully treated with immunotherapy reported in the literature, though many were recurrent tumors, often with evidence of distant metastatic disease at presentation.

Clinical Implication: Extrapolation of data from cutaneous melanoma research suggests locally advanced BRAF mutant conjunctival tumors could be treated with either combination (nivolumab plus ipilimumab) or single-agent immunotherapy (pembrolizumab or nivolumab), depending on how well systemic toxicities are likely to be tolerated. As immunotherapy may be less effective if given after progression on a BRAF inhibitor, combined BRAF/MEK inhibition tends to be a second line choice. Targeted therapy with BRAF/MEK inhibition is not indicated in BRAF wild-type tumors, and in these cases, next generation sequencing to investigate for other actionable mutations is valuable.

Financial Interest: *None*

S3C11

SESSION 4

ORBITAL AND MISCELLANEOUS TUMORS

Epiphora - an Enigma

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Co-authors: Smitha K S, Jayaprasad Bhaskar, Kaustubh Mulay

Abstract

Introduction/Purpose: Epiphora is a common symptom that you deal with in your daily practice as an ophthalmologist. Aetiology varies from ocular surface disorders to nasolacrimal duct obstruction. Nasolacrimal duct obstruction is indeed a very common condition presenting in an oculoplasty practice that can be primary obstruction or secondary. Rarely tumors in and around the lacrimal sac can mimic dacryocystitis. Meticulous clinical evaluation is warranted to differentiate inflammation from neoplastic aetiology. The eyes sees only what the mind knows. Here is a case series of neoplasia presenting as epiphora.

Case Report: Four cases who presented with epiphora of which 2 patients had undergone dacryocystorhinostomy (DCR) for diagnosed chronic dacryocystitis. All thefour patients were referred for DCR in our oculoplasty clinic of which 2 were for failed DCR. Clinical evaluation showed pointers towards neoplasia and was further investigated. Three patients were proven to be oculoadnexal lymphoma and one patient was solitary fibrous tumor. The cases will be presented in detail.

Conclusion: Not all epiphora is dacryocystitis. Neoplasia can be lurking in any part of the eye

Clinical Implication: The case series highlights the need for thorough evaluation in the presence of medial canthal swelling or puffiness. Dacryocystorhinostomy in epiphora should be planned after meticulous work up.

Financial Interest: *None*

S4C1

Disappearing Act - Magic of Neoadjuvant Chemotherapy in a Case of Lacrimal Sac Carcinoma

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Co-authors: Gangadhara Sundar

Abstract

Introduction: To describe a case of complete pathologic remission with chemoreduction for squamous cell carcinoma (SCC) of the lacrimal drainage system (LDS).

Case Report: A 62-year-old chinese male with left epiphora presented with an ulcerated 3cm medial canthal mass involving the eyelid margins and lower canaliculus, displacing the globe superotemporally. Nasoendoscopy revealed fullness of left lateral nasal wall with no fluorescein. CT and MRI scans showed a large enhancing hypodense mass in the left lacrimal sac extending to the ethmoid sinus, eroding medial orbital wall with globe displacement. Histopathology of transnasal incisional biopsy suggested moderately-differentiated keratinizing SCC. Final Staging was T4N0M0. While he was referred for orbital exenteration, neoadjuvant chemotherapy (5FU, Cisplatin and Docetaxel) was commenced which resulted in remarkable clinical and radiologic reduction of tumour. Decision made for local wide excision with frozen section of margins prior to exenteration. No malignant tumour was seen histologically, including both ends of LDS. Thus a limited medial maxillectomy and reconstruction was done, with orbit and globe preservation followed by adjuvant chemotherapy and radiation with good response.

Conlcusion: While neoadjuvant chemoreduction is mainstream for intraocular tumors such as retinoblastoma, it has only now become more applicable for malignant orbital and LDS tumours. This case highlights complete pathologic remission following neoadjuvant chemotherapy for a T4 LDS SCC that was planned for orbital exenteration.

Clinical Implication: LDS SCC is an aggressive malignancy. Standard of care for cases with extensive sinonasal and orbital extension had been radical excision with organ/vision sacrifice and adjuvant radiotherapy. We present a case that wad listed for orbital exenteration, with complete pathologic response following neoadjuvant chemotherapy, preserving organ and vision. The introduction of targeted and immunotherapy may provide more therapeutic options in years to come.

Financial Interest: *None*

S4C2

A Massive Periorbital and Orbital Tumour in an Adult

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Co-authors: Usha Singh, Pankaj Gupta, Khushdeep Abhaypal

Abstract

Introduction: Malignant peripheral nerve sheath tumours is an extremely rare orbital tumour and are associated with a highly aggressive course of recurrence, metastases. Herein we report a patient with MPNST treated with orbital exenteration and radiation

Case Report: A 58-year-old female presented with a one-year history of progressively increasing proptosis and vision loss attributed to a sizable tumour in her left eye. Upon physical examination, a pinkish, firm, irregular, bosselated, ulcerated mass measuring 8.5 x 10 cm was identified, extending above the orbital rims and obscuring the eyelid and globe of the left eye. Systemic examination yielded unremarkable findings. A contrast-enhanced computed tomography (CECT) scan delineated a heterogeneous mass abutting the left eyeball in the superolateral and inferior quadrants of orbit, involving the superior, lateral, and inferior rectus muscles. Notably, no intracranial involvement, extension in adjacent sinus, or bony erosions were observed. Consequently, the patient underwent lid-sparing orbital exenteration. Microscopic examination revealed pleomorphic spindle tumour cells with inconspicuous nucleoli and extensive areas of necrosis, characteristic of a malignant peripheral nerve sheath tumour. Tumour cells tested negative for smooth muscle actin (SMA). Importantly, retro orbital fat and the optic nerve were devoid of tumour cells. Following surgery, the patient underwent irradiation therapy, and there has been no evidence of tumour recurrence six months postoperatively. Determining the precise cell of origin remains challenging, but the predominant superolateral orbital location suggests possible involvement of the ophthalmic division.

Conclusion and Clinical Implication: Malignant peripheral nerve sheath tumour is a highly aggressive tumour. Orbital exenteration often does not prevent recurrence and extension due to posterior perineural spread, therefore mandate radiotherapy.

Financial Interest: *None*

S4C3

Well Differentiated Orbital Liposarcoma - A Diagnostic and Management Dilemma

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Abstract

Purpose: To report the challenges faced during the 5 years in the management of case of well differentiated orbital liposarcoma.

Case Report: A 21 years old female presented with painless progressive abaxial proptosis of left eye. Her visual acuity was 20/20 and 20/30. The left eye showed 15mm proptosis and 2mm medial displacement. The mass was visible temporally with dilated and tortuous vessels on its surface. There was limitation of abduction, negative Valsalva, absence of bruit and unremarkable systemic examination. On MRI, an ill-defined marginated lobulated lesion 5.3X 4.3X 3.5 cm, hyperintense on T1 and T2 and complete suppression on fat suppression. The blood and systemic investigations were unremarkable. Incision biopsy showed few atypical cells, positive for MDM2 and S-100, amidst mature adipose tissue with myxoid areas suggestive of well differentiated orbital liposarcoma. She underwent debulking with recurrence at 3 months. A repeat debulking was followed by recurrence at 5 months. She was administered 3 cycles of injection ifosfamide, adriamycin and doxorubicin, leading to worsening of proptosis and development of lagophthalmos. PET scan showed the limitation of disease to the orbit. Options of exenteration and repeat debulking were discussed with the patient. Given the of visual acuity of 20/30, extensive debulking was undertaken. No further recurrences were seen on PET scan or clinically 10 months post third debulking.

Conclusion: Primary exenteration can be replaced by repeated debulking with close follow-up in well differentiated orbital liposarcoma with good visual acuity.

Clinical Implication: Orbital liposarcoma is a rare entity with non-specific radiologic features.. Due its pathological similarity to benign adipose tissue, immunohistochemistry plays a crucial role in its diagnosis. It should be considered in any recurrent orbital mass with non-specific fibrofatty changes.

Financial Interest: *None*

S4C4

Congenital Neuroectodermal Tumor of the Orbit in a Newborn

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Abstract

Purpose: Primitive neuroectodermal tumors arise from the progenitor cells of the neural crest, most commonly in the central nervous system. However, in recent years there has been an increased incidence of PNETs in peripheral locations, most commonly in the thoracopulmonary region. Orbital involvement is extremely rare and has not been previously described in the Indian population. We report a rare case of a congenital malignant tumor, diagnosed as a primary orbital primitive neuroectodermal tumor on histopathological examination.

Case Report: A 5-day-old male neonate, born out of a second-degree consanguineous marriage, presented with a protruding mass measuring 3x3 cm in the inferonasal part of the right orbit with otherwise normal systemic examination and no evidence of local spread or metastasis. Contrast-enhanced magnetic resonance imaging (MRI) of the orbits revealed a well-defined homogeneously intense lesion with clear margins seen in the inferomedial aspect of the right orbit and extending posteriorly to the globe. However, there was no involvement of the optic nerve or brain parenchyma. Histopathological examination of an incisional biopsy showed findings consistent with PNET, including Immunohistochemistry. The child underwent primary chemo reduction using Vincristine, Doxorubicin, Cyclophosphamide, Ifosfamide, and Etoposide (VDC/IE) regimen for six cycles followed by surgical excision using the trans-caruncular approach. A complete response to chemotherapy was demonstrated on the HPE of the excised mass. The child has since completed chemotherapy, and strabismic correction and is on follow up at 2 years with amblyopia therapy.

Conclusion: Multidisciplinary approach with adjuvant chemotherapy and surgery is needed for the management of these cases.

Financial Interest: *None*

S4C5

Invasive Squamous Cell Carcinoma with Facial Nerve Palsy and Ophthalmoplegia

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Co-authors: Wanxing Chai-Ho, Daniel B. Rootman, Kelsey A. Roelofs

Abstract

Introduction: To present a case of invasive perineural squamous cell carcinoma with facial nerve palsy

Case Report: A 58-year-old male with a one year of progressive left sided cranial neuropathies presented to our service with acute left proptosis and ptosis. He also noted left facial weakness, numbness, and binocular diplopia. Medical history was notable multiple recurrent cutaneous squamous cell carcinomas (SCC) involving the face treated several years prior. Examination of the left side was notable for complete ptosis with no levator excursion, inferior conjunctival chemosis, and complete limitation of extraocular motility. There was decreased V1-V2 sensation to 20% with a complete left facial nerve paralysis. MRI showed an enhancing left superior apical mass involving the superior rectus and extending to the orbital apex and cavernous sinus. There was asymmetric enhancement of multiple cranial nerves (left CN 3-7). An orbital biopsy was performed. Pathology revealed moderately-differentiated infiltrating SCC. Given the extensive degree of perineural spread, the patient was started on carboplatin, paclitaxel and cemiplimab.

Conclusion: Perineural invasion of head and neck SCC can result in multiple cranial neuropathies. For cutaneous skin cancers of the face, palsies of the trigeminal and facial nerves are most frequently involved and may be the presenting symptom.

Clinical Implication: Cases of perineural spread often present in the setting of recurrent cutaneous disease,1 and may not have a concurrent, clinically apparent skin lesion. Thus, there must be a high index of suspicion in patients with a history of prior cutaneous malignancies. While surgery and post-operative radiotherapy have traditionally been the mainstay for management of perineural spread,1 cases with extensive involvement, such as this one, necessitate alternate approaches to management. Outcomes are improved in earlier stage disease, highlighting the need for prompt diagnosis.

Financial Interest: *None*

S4C6

A Case of Primary Orbital Melanoma

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Co-authors: Svetlana Saakyan, Nina Panina

Abstract

Introduction: Primary orbital melanoma (POM) is exceedingly rare, with approximately 50 cases reported to date. We have observed 5 cases with a histologically verified diagnosis of PMO in last 16 years. Here we present a case of the patient with the most malignant course of PMO.

Case Report: A 57-year-old Caucasian male who had a primary oculodermal melanosis (nevus of Ota) from birth, presented with rapid proptosis, double vision and periodic pain in the orbit. On examination, there was edema of lids, bluish black pigmentation in the periorbital region and conjunctiva. Ocular motility was restricted in all directions, proptosis OD/OS=26/20mm with globe downward displacement, partial blepharoptosis and there was no eye reposition. Palpation in the upper-outer part under the superior orbital wall revealed a painful, soft-elastic mass with a smooth surface, fused to the surrounding tissues and extending deep into the orbit. CT scan demonstrated a 34 mm maximum diameter, well-circumscribed lesion in the superior orbit, the external and superior rectus muscles were somewhat thickened. The adjacent bone was normal. During the surgery intensive pigmentation of skin, soft tissue, and orbital fat was detected. Histopathological examination revealed a mixed cell type melanoma (80% epithelioid type) with a moderate mitotic count of 7/ 40 HPF. Immunohistochemistry examination confirmed the diagnosis of POM. Follow-up external beam radiotherapy was planned, but not performed because the patient was subjected to 5 courses of chemotherapy for metastases of the chest organs detected on CT in a month after surgery. Local recurrence of POM was detected in 6 months after surgery. Patient died from the consequences of metastatic disease in 24 months.

Conclusion: The presented case clearly demonstrates the need for monitoring patients with oculodermal melanosis. Such patients are at risk of developing POM. POM has a very poor, it is associated with the histological type of tumor.

Financial Interest: *None*

S4C7

Iris Tumor in Neurofibromatosis: What Are the Chances?

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Co-authors: Miguel Hernandez-Emanuelli, Carol Shields

Abstract

Introduction: Neurofibromatosis (NF) is a neurocristopathy caused by inactivation of a tumor suppressor gene predisposing patients to various tumors arising from embryonic neural crest cells. It is one of the largest genes to code with over 500 identified mutations. We present a case of iris tumor in a patient with NF and explore the relative risks of malignancies in NF.

Case Report: A 60-year-old Caucasian female was referred for an iris lesion in the left eye (OS). She has a history of optic nerve glioma of the right eye (OD) diagnosed 40 years ago and managed conservatively. Family history was significant for neurofibromatosis in her mother. General physical examination revealed cutaneous freckling and facial sub-cutaneous neurofibromas. On ocular examination, visual acuity was hand movements OD and 20/60 OS. Anterior segment showed Lisch nodules in both eyes (OU) and a large, pigmented iris mass OS, inferotemporally 10 millimetres in largest diameter. Fundus examination revealed optic disc pallor OD and choroidal abnormalities OU, evident on infrared optical coherence tomography (OCT) images. Anterior segment OCT and ultrasound biomicroscopy OS showed the lesion to be arising from the iris stroma. Based on clinical features and family history, she was diagnosed to have NF type 1. Fine needle aspiration biopsy OS confirmed iris melanoma.

Conclusion: Although causative association is debatable, patients with NF need monitoring for uveal melanoma. Iris melanoma in patients with NF is rare, with only four previously reported cases.

Clinical Implication: Mutations associated with uveal melanomas are BAP1, EIF1AX, GNA11, GNAQ and SF3B1. Although, somatic NF mutations are found in 5 to 10 % of all human sporadic cancers, an association between NF mutation and uveal melanoma has not been established. However, since NF gene mutations affect cell proliferation via the ‘RAS’ pathway, it is prudent to monitor these patients for uveal melanomas.

Financial Interest: *None*

S4C8

Amelanotic Iris Lesion

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Co-authors: Rachel Shemesh

Abstract

Case Report: A 58-year-old female patient presented with a resently asymptomatic multi-lobular amelanotic Iris lesion. Multimodel imaging reveald on UBM scan a round multi-lobular hypoechogenic iris lesion reaching the basement membrane with iris-lens touch but no ciliary process or Ciliary body involvemen. The basement membrane was intact on anterior segment OCT. Based on the resent growth and the multimodal imaging amelanotic melanoma was suspected.Biopsy of the lesion demonstrated a spindle cell lesion composed of poorly defined fascicles. The FISH study showed monosomy of chromosomes 3, 6, and 8, consistent with a low-grade spindled melanoma. The patient was treated with margins free local lesion exsion.At 18 months follow-up, the VA remained stable, and the IOP was normal. A small descemet scar and a nucleus sclerosis cataract were seen. Systemic work up was negative for secondary lesions.

Financial Interest: *None*

S4C9

Vitritis in Acute Myeloid Leukemia Patient: What Could it Be?

Presenting Author: Kushal U Agrawal, *1. Gujarat Cancer Research Institute, Ahmedabad, India, 2. K.D. Hospital, Ahmedabad, India,* kushal@shields.md

Co-authors: Chetna Bakshi, Supriya Dutta

Abstract

Purpose: To report a novel case of acute myeloid leukemia (AML) on remission who presented as vitritis and diagnosed to have relapse inside the vitreous cavity. To report advantages of doing vitreous biopsy and diagnostic vitrectomy only under air.

Case Report: A 47 year old female was a known case of acute myeloid leukemia (AML-M5) presented with diminution of vision in the right eye (OD) since two weeks. Examination revealed pigmented cells in the anterior vitreous both eyes with vitritis OD. Systemic evaluation was non contributory and patient was in remission phase of AML. She was further treated with vitreous biopsy and diagnostic vitrectomy under air OD. A Cytology examination of vitreous biopsy revealed infiltration with myeloblasts confirming the diagnosis of extramedullary relapse of AML inside vitreous. She was further treated with intrathecal chemotherapy considering it as a central nervous system (CNS) relapse of AML. She did not have any further relapse till three years of her last follow up.

Conclusion: AML can relapse in vitreous and treatment on the line of CNS relapse is warranted in such cases. Clinical Implication: This case highlights a rare location of extramedullary (vitreous) relapse of AML which initially presented as vitritis and needed treatment on the line of CNS relapse. AML relapse inside vitreous presenting as vitritis has not been reported as per literature search. Moreover, this case also emphasizes added advantage of doing diagnostic vitreous biopsy and vitrectomy only under air to get undiluted vitreous fluid which could have also helped in confirming the diagnosis.

Financial Interest: *None*

S4C10

The Tale of Two Children

Presenting Author: Meenakshi Mahesh, *Sankara Eye Hospital, Bangalore, Karnataka, India*, meenakshimahesh123@gmail.com

Co-authors: P. Mahesh Shanmugam

Abstract

Introduction: Retinal manifestations of Leukemia are known,but some cases can pose as a diagnostic dilemma with confusing presentations,and warrant multiple modalities of investigation to come to a diagnosis.

Case Report: Two children on ALL remission presented with sudden diminution of vision in 1 eye. An 18-year-old boy who was diagnosed elsewhere to have Retinochoroiditis with a negative anterior chamber tap. He had been treated with IVMP for Optic Neuritis in the same eye. Fundus examination revealed temporal disc pallor,subretinal yellowish lesions and deposits superior to the disc. With a suspicion of leukemic infiltration vs fungal granuloma,we planned a vitrectomy with vitreous and retinal biopsy. The specimen revealed active fungal elements. The second case was a 12-year-old girl with counting fingers vision and appearance of a central black spot in the left eye since 3 days.Her systemic status and blood counts were stable. Fundus examination showed cells and seeds in the vitreous cavity with a creamy white elevated lesion over the posterior pole and disc, with detachment. The Oncologist team suspected CMV Retinitis with a slim chance of Infiltration. Vitrectomy with biopsy of the lesion was done. Unfortunately, the cytology report was suggestive of large lymphoid cells with blast component, confirming leukemic infiltration.

Conclusion: This presentation shows some of the myriad presentations of Leukemia, along with the techniques of biopsy used to clinch the diagnosis. The staging of the disease changes if a diagnosis of leukemic infiltrate is established. Most patients will need bone marrow transplant.Hence early diagnosis is of primordial importance.

Clinical Implication: Leukemic infiltrates can mimic multiple conditions, most commonly, infections. Factors such as systemic stability of the disease and normal blood counts can be confusing,and cloud the diagnosis. A strong suspicion can help in timely diagnosis and correct treatment in these cases.

Financial Interest: *None*

S4C11

SESSION 5

SPECIAL SESSION

Ethics, Medicine, Organizations & Ocular Oncology: How is this relevant to you in 2023?

Presenting Author: Katherine Paton, *Department of Ophthalmology and Visual Sciences, The University of British Columbia, Vancouver, BC, Canada*, katherinepaton@me.com

Abstract

Purpose: To enable those practicing Ocular Oncology to discuss the evolution of the components of medical and organizational ethics across international boundaries, that affect our field, and illuminate what use a Code of Ethics could be to Ocular Oncology.

Methods: Review of available academic literature on medical, ophthalmological, oncological and professional organizational domains, as well as review of existing journal and literature coverage of contentious issues in the past decade was conducted looking for themes and common strands. Care was taken to include information from countries and cultures beyond the Americas and Europe. Academic dialogue with ethics experts was used to aid analysis.

Results: While medical ethics with respect to individual patient interactions are standard for many medical practitioners, ethics beyond the individual patient – doctor relationship is less familiar, and organizational ethics rarely discussed. A matrix of the background of identified ethics approaches was created, and common components were identified. Separately, a “recent ethics issues” list was developed. Existing ophthalmology clinical and research organizations codes, and medical association codes were evaluated for guidance on the components of a code, the identification of issues, and mechanisms for resolving issues.

Conclusions: A firm understanding of the development, components and utility of ethics codes can help provide for the best functioning of organizations of medical professionals with international collaborations. This is relevant to Ocular Oncology in 2023. These results will form the basis of a survey, which in turn will inform the creation of a Code of Ethics for our community.

SESSION 6

ON-DEMAND ACCESS

(ACCESS ON THE ISOO YOUTUBE CHANNEL)

Primary Iris Leiomyoma

Presenting Author: Arun D Singh, *Cole Eye Institute/ Cleveland Clinic, Cleveland, USA*,singha@ccf.org

Co-authors: Gabrielle A. Yeaney

Abstract

Purpose: To report clinical, histopathological, immunohistochemical, and electron microscopic features of iris leiomyoma.

Case Report: A 58-year-old white man was found to have an asymptomatic iris lesion in the right eye on routine examination. Slit lamp examination, revealed a 2 x 2 x 1.2 mm round, translucent, tan-pink stromal lesion (without melanocytic pigment). Fine intrinsic and surrounding feeders vessels confirmed by anterior segment fluorescein angiography). The lesion was located at 5:30, pproximately 2 mm from the pupillary margin. Anterior segment optical coherence tomography revealed a dome-shaped mass. Ultrasound biomicroscopy showed an iris stromal mass with intact pigment epithelial layer. Clinical differential diagnosis included amelanotic melanoma or nevus, inflammatory lesion, hemangioma, and metastasis. The lesion was excised via a sector iridectomy with pupilloplasty. Postoperative assessment at 1 week showed return to baseline vision and normal intraocular pressures. Light microscopy (H&E stained sections) showed a round mass in the anterior iris composed of fascicles of spindle cells with ovoid nuclei and moderate fibrillar eosinophilic cytoplasm. The mass was discrete from the iris stroma. On immunohistochemistry the lesional cells were positive for both smooth muscle actin and desmin and negative for h-caldesmon, vascular markers (CD31, CD34), neural crest or melanoma markers (S-100, HMB45, SOX10, MelanA). Electron microscopy showed distinct cell borders, folded nuclei, cytoplasmic myofilaments with focal fusiform densities and macropinocytotic vesicles, diagnostic of smooth muscle. Of note, tumor cells did not have melanosomes.

Conclusion: Although rare, the diagnosis of primary iris leiomyoma should be considered when evaluating an amelanotic circumscribed iris stromal tumor.

Clinical Implication: Immunohistochemistry is necessary to confirm the diagnosis and electron microscopy is a helpful addition.

Financial Interest: *None*

D001

Necrotic Iris Melanocytoma with Secondary Glaucoma

Presenting Author: Yağmur Seda Yeşiltaş, *Gülhane Training and Research Hospital, Ankara, Turkey,* yesiltas.seda@gmail.com

Co-authors: Arun D Singh

Abstract

Purpose: To report a rare case of necrotic iris melanocytoma with pigment dispersion glaucoma that resolved after resection of primary tumor and glaucoma surgery.

Case Report: A 54-year-old female was referred to Ocular Oncology Service, Cole Eye Institute because of a pigmented iris lesion with uncontrolled glaucoma in the left eye. The intraocular pressure (IOP) was 54 mm Hg despite anti-glaucoma medications. Her visual acuity (VA) was 20/100. A slit lamp examination showed corneal edema and a darkly pigmented small iridociliary lesion (UBM : 4 x 3 x 2 mm) with diffuse pigment shedding onto anterior iris surface and diffuse circumferential pigmentation of the trabecular meshwork with wide-open angle (gonioscopy). Diagnostic fine needle aspiration biopsy (FNAB) of the main tumor and anterior chamber lavage were performed. Cytology revealed polygonal cells with dense intracytoplasmic pigment obscuring nuclear details diagnostic of a melanocytoma. During follow-up, as the tumor continued to cause new pigment shedding with high IOP, excision of the primary tumor via iridocyclectomy was performed. At postoperative 1 month, the IOP was 13 mm Hg with maximal medical therapy for glaucoma requiring glaucoma shunt surgery. At 6 months, the IOP remained below 13 mm Hg and the VA was 20/50 without recurrence of pigment shedding.

Conclusion: Necrotic iris melanocytoma, though rare, can lead to pigment dispersion glaucoma. Surgical excision of the primary tumor, coupled with glaucoma surgery, proved effective in controlling intraocular pressure and preserving vision in this case.

Clinical Implication: A small pigmented iridociliary tumor causing extensive pigment dispersion and secondary glaucoma should raise a suspicion of melanocytoma (to be differentiated from melanoma). Prior to any therapeutic intervention (radiation, excision or enucleation), a diagnostic FNAB can reliably establish the diagnosis of melanocytoma.

Financial Interest: *None*

D002

Neoplastic Iris Condition Masquerading as Uveitis

Presenting Author: Kushal U Agrawal, *Wills Eye Hospital, Jefferson University, Philadelphia, USA*, kushal@shields.md

Co-authors: Carol L. Shields, Konica Singla

Abstract

Purpose: To describe a case of iris mass who presented with features of hypertensive uveitis and heterochromia. To describe role of fine needle aspiration biopsy (FNAB) in such cases for the definitive diagnosis and further management.

Case Report: A 64 year old female was referred with initial diagnosis of hypertensive uveitis with heterochromia. On examination there were keratic precipitates and diffuse thickening of iris with neovascularization. Fine needle aspiration biopsy (FNAB) proved large B-cell lymphoma. The patient was treated with systemic immunochemotherapy and methotrexate leading to complete resolution of iris mass.

Conclusion: FNAB in patients with iris mass could be beneficial in the definitive diagnosis and further management. Iris Lymphoma is a relatively rare disease and can present as a masquerade disease.

Clinical Implication: Patient with iris lymphoma needs a multimodal approach of management with FNAB, cytology and if needed systemic immunochemotherapy or external beam radiation.

Financial Interest: *None*

D003

The Bulge with A Nudge

Presenting Author: Miguel Hernandez-Emanuelli, *Wills Eye Hospital, Jefferson University, Philadelphia, USA*, miguelh@shields.md

Co-authors: Rolika Bansal, Carol L. Shields

Abstract

Introduction: Malignant melanoma of the iris constitutes a small proportion of all the uveal melanomas. Herein, we describe a case of iris melanoma in a young girl, diagnosed with ultrasound biomicroscopy and managed with plaque brachytherapy.

Case Report: A 17-year-old Caucasian female presented with a history of a stable asymptomatic iris cyst since 10-years which was followed by an episode of sudden decrease in vision due to hyphema for she was treated conservatively elsewhere and referred to us for further evaluation. On presentation, her best corrected visual acuity was 20/25 in both eyes. Anterior segment examination of the right eye revealed an iris lesion at 8:00 meridian, 6 x 6 x 3mm in size, tan-colored, with intrinsic vascularity and a resolving hyphema inferiorly. Gonioscopy confirmed no angle involvement. Ultrasound biomicroscopy revealed an iris lesion with a small anterior and larger posterior component with cavities and no ciliary body involvement. With a suspicion of melanoma, fine-needle aspiration biopsy confirmed the diagnosis of iris malignant melanoma arising from a nevus and iodine-125 plaque radiotherapy was performed. Cytogenetics confirmed The Cancer Genome Atlas Group A/B (low grade) and at a follow-up of 1-year-4-months the lesion has regressed well to a thickness of 1.3mm, visual acuity of 20/25, with no systemic involvement.

Conclusion: Iris cysts present a diagnostic dilemma due to the atypical features, especially in younger age group and must be screened well by imaging to rule out the possibility of melanoma.

Clinical Implication: Cavitations occur rarely in intra-ocular melanomas and are known to be most commonly associated with ciliary body melanomas. A rare finding of cavitations in iris lesions hints towards the diagnosis of iris melanoma.

Financial Interest: *None*

D004

An Unusual Case of Ciliary Body Menaocytoma and Treatment.

Presenting Author: Saadia Razzaq Chaudhry, *Royal Hallamshire Hosiptal, Sheffield Teaching Hospitals NHS Trust, Sheffield, UK,* saadia_razzaq@yahoo.com

Co-authors: Sachin M Salvi

Abstract

Introduction: We present an unusual case of 72 year old lady with growing ciliary body (CB) melanocytoma who initially underwent open flap biopsy but had subsequent extraocular extension (EOE) at biopsy site. A further shave biopsy of EOE and plaque brachytherapy was planned to treat any possible conversion into melanoma. On the day of operation she had complete erosion of the scleral-flap which prompted an out of box solution on the day.

Case Report: A 72 year old lady referred for mass in left eye seen during cataract surgery; better visualised on gonioscopy. Bscan showed CB mass measuring 4x6mm, pigmented with no scleral extension. MDT decision was to monitor. Few months later the mass increased in size hence an open flap biopsy was performed with cryotherapy. Histology confirmed CB melanocytoma. Patient was closely followed up; after a year the lesion started growing and there was EOE through the flap site. Shave excision of EOE and Ru106 plaque brachytherapy was planned as it was deemed there was a possible conversion of melanocytoma to melanoma. At surgery complete erosion of flap tissue was noted. She underwent shave excision of EOE. As there was no tutopatch ordered at the time, decision was made to suture multilayer Amniotic membrane graft over the scleral defect. Ru106 plaque was applied over the AMG. Second histology again confirmed melanocytoma with no malignant cells noted. Patient has done very well post op as the multi-layered AMG has held well even with high IOP from secondary glaucoma and Brachytherapy. We will present the photo diary of the patients treatment journey.

Conclusion: Growing melanocytoma can be treated with Ru106 plaque brachytherapy with good results. Multilayered AMG can be useful as a patch material and takes well even with high IOP and brachytherapy postoperatively.

Clinical Implication: Growing melanocytomas can be treated with brachytherapy. in cases where tutopatch not available AMG can be good option as evident in this case.

Financial Interest: *None*

D005

A Wolf Under Sheep Skin

Presenting Author: Dipti Gupta, *Govind Ballabh Pant Institute of Post Graduate Medical Education & Research, New Delhi, India*, diptig2@gmail.com

Co-authors: Ravindra K. Saran

Abstract

Introduction: Intraocular malignant tumors including primary and metastatic tumors, are mainly found in Retina and uvea, and very few cases originate from the sclera and optic nerve. Intraocular malignant tumors include primary malignant and metastatic tumors, and the most common sites of malignancies are uvea and retina. Choroidal melanoma and retinoblastoma are the most common primary intraocular malignant tumors in adults and children respectively. Medulloepithelioma are rare introrbital tumors, presenting most commonly in children, but only occasionally in adults. We hereby present a case of Medulloepithelioma presenting in an adult female.

Case Report: A 22-year pregnant female presented to the eye opd with an episcleral bleb since 2 months which was increasing in size since last 4 days. She had a previous history of non-penetrating trauma to same eye 4 months back & of cataract surgery 1 year back. We received aspirate fluid from the episcleral bleb followed by scleral nodule biopsy specimen in the pathology department of GIPMER, New Delhi. Relevant IHC was applied to the paraffin sections. Further eye exenteration specimen was also received in the pathology department. H&E sections & IHC was applied to the sections.

Conclusion: Medulloepithelioma most commonly occurs in children between 4 and 5 years but can also be seen in adults as well. Variability in the immunohistochemical markers such as HMB-45, S100 & synaptophysin & positivity with CK & CK 8 may lead to erroneous diagnosis if not kept in mind.

Clinical Implication: Since the tumor can hide for months to years in the ciliary body region & produces secondary features of glaucoma & cataract, leading to surgery & later discovery of tumor. Ophthalmologists must exercise due caution in children presenting with unilateral secondary glaucoma, cataract & iris neovascularisation & must advise relevant imaging investigations.

Financial Interest: *None*

D006

Ciliary Body with Rare Morphological Features

Presenting Author: Ravindra Kumar Saran, *Govind Ballabh Pant Institute of Post Graduate Medical Education & Research, New Delhi, India*, ravindraksaran@hotmail.com

Co-authors: Ravindra Kumar Saran, Praveen Mongre, Ashish Jain

Abstract

Introduction: A rare ciliary body benign tumour mimicking as uveal / ciliary body melanoma is discussed here.

Case Report: A 43-year-male presented with painful blind left eye for duration two to three months. P/H/O shown to ophthalmologist 3-4 yrs back, asked to get it operated for enucleation. Patient ran away. B-scan U/A Doom shaped moderately low reflective choroidal mass displacing cataractous lens. Basal dimension 17.9mm & Tumour Hight 14.2mm. No Extrasclaral extension. MRI: Left eye- 11x21x17 mm heterogenous signal lesion involving iris- ciliary body. Hypointense on T2W and hypointense on T1W. Clinical Diagnosis of Primary Uveal-Ciliary body melanoma. Enucleation of left eye done. On slicing well defined tumour in antero medial aspect of globe. identified. HistoPathology on HE stained section, diffuse sheets of monomorphic cells with abundant eosinophilic cytoplasm with central round monomorphic nuclei noted. No atypia, mitosis, necrosis or melanin pigment noted. Tumour cells are also negative for S-100, HMB45 and melan A, thus ruling out malignant epithelioid melanoma. Tumour cells are positive for SMA, h-Caldesmon and Calretinin. Favouring smooth muscle differentiation. IHC negative for GFAP, CK, EMA, Synaptophysin, Chromogranin. Positivity was also noted for NSE, CD56 and NeuN resulting in confusion.

Conclusion: Rarely ciliary body leiomyoma can clinically mimics to uveal or ciliary body melanoma. Preoperative diagnosis may help ophthalmologist for taking steps to save eye from enucleation.

Clinical Implication: Leiomyoma of ciliary body- mesoectodermal origin, is a benign tumour. Relative conservative surgery for biopsy and decision after biopsy report could be important steps for possible preservation of eye. Local excision of iris leiomyoma in the form of iridectomy or iridocyclectomy. For ciliary body and posterior leiomyoma trans-scleral resection via partial lamellar sclerouvectomy or endoresection via pars plana vitrectomy is recommended.

Financial Interest: *None*

D007

Potential of Aqueous Humor Liquid Biopsy in Diagnostically Challenging Intraocular Masses: Intraocular Ciliary Body Medulloepithelioma Masquerading as Atypical Retinoblastoma

Presenting Author: Sarah B Pike, *Children's Hospital Los Angeles, Keck School of Medicine of USC, Los Angeles, USA*,sbpike@usc.edu

Co-authors: Rahul Iyengar, Chen-Ching Peng, Patricia Chevez-Barrios, Rachana Shah, Liya Xu, Jesse L. Berry

Abstract

Introduction/Purpose: Intraocular ciliary body medulloepithelioma (CBME) is a rare congenital tumor often mistaken for retinoblastoma (RB) due to clinicopathologic similarities, requiring histology for definitive diagnosis. For both tumors, direct biopsy is strictly contraindicated. Aqueous humor (AH) is a source of tumor analytes and serves as a surrogate liquid biopsy for RB; analysis of AH cell-free DNA (cfDNA) in advanced RB has established the highly recurrent somatic copy number alterations (SCNAs) 1q2p6p gain and 13q16q loss. At this time, the AH has not been explored as a liquid biopsy source of CBME tumor information.

Case Report: A 5-year-old male was diagnosed with a mass in the left eye (OS) by clinical examination and imaging. The patient presented with a painful, phthisic left eye. Ocular B-scan revealed a lesion with calcifications and retinal detachment. Orbital MRI suggested left globe hypercellularity. An infiltrative lesion involving the ciliary body was seen on examination under anesthesia. Differential diagnosis was broad, with concern for atypical advanced RB; enucleation was pursued. AH liquid biopsy was collected at enucleation. Whole-genome sequencing of AH cfDNA determined SCNA profile. Tumor was assayed for oncogenic mutations and SCNAs. The SCNA profile of AH cfDNA showed loss-of-copy of chromosomes 4, 6, and 9. Tumor was negative for clinically significant mutations and SCNAs. Histopathology ultimately diagnosed malignant teratoid CBME.

Conclusions: The SCNA profile of AH from the affected eye was distinct from typical advanced RB suggesting an alternative diagnosis, which was ultimately confirmed by negative genetic testing and final histopathology. The loss-of-copy of chromosomes 4, 6, and 9 found in the AH in this case is consistent with the limited CBME tumor cytogenetic literature.

Clinical Implication: Our results highlight the potential of AH liquid biopsy analysis to rule out rare RB mimicking lesions.

Financial Interest: *None*

D008

Ciliary Body Melanoma with Extrascleral Extension Masquerading as a Conjunctival Nevus

Presenting Author: Akshay Agnihotri, *UCSD Shiley Eye Institute, San Diego, California, USA*, akshay06020@gmail.com

Co-authors: Nathan Scott

Abstract

Introduction: Uveal melanoma is the most common intraocular malignancy in adults and can manifest with a multifaceted initial clinical presentation based on its location and growth. Like our case, there are few case reports highlighting extrascleral extension as the primary presentation leading to a diagnosis.

Case report: A 74-year-old woman with a history of breast cancer, now in remission, presented for evaluation of a conjunctival lesion of the right eye. Per her history, the lesion was noted over 10 years in the past by her optometrist. She reported no noticeable growth or changes to the lesion. The conjunctiva of the right eye was notable for a small elevated pigmented lesion with sentinel vessels. Fundus photographs and autofluorescence as well as optical coherence tomography (OCT) were normal bilaterally. An anterior segment OCT was performed and revealed a small elevated subepithelial lesion with shadowing versus possible scleral extension.

Conclusion: The UBM of the right eye revealed an intraocular lesion measuring 2.96 mm in depth with low acoustic internal reflectivity and cystic cavities.Upon transillumination of the extrascleral lesion, a 10 mm shadow appeared in the 4 o'clock position. Diagnosis confirmation involved a B-scan ultrasound revealing a 9.5 x 8 x 3mm mass. The diagnosis was a ciliary body melanoma with extrascleral extension masquerading as a conjunctival lesion. This lesion was likely to have gone undetected for years given that it was not visible on fundus examination without depression and did not affect the patient's vision.

Clinical Implication: Although the most common melanocytic lesion of the conjunctiva is a conjunctival nevus, melanoma must be considered in the differential of any pigmented conjunctival lesion. Given the clinical findings above, we feel it is imperative to first obtain a UBM to confirm diagnosis.

Financial Interest: *None*

D009

Acute Orbital Cellulitis and Focal Scleritis with Amelanotic Ciliochoroidal Lesion and Exudative Retinal Detachment Masqueraded Ciliochoroidal Melanoma with Scleral Invasion: A Case Report

Presenting Author: Sarinee Juntipwong, *Kellogg eye center, University of Michigan, Ann Abor, MI, USA,* jesarinee@gmail.com

Co-authors: Kubra Serbest Ceylanoglu, Tassapol Singalavanija, Almila Sarigul Sezenoz, Victor M. Elner, Bernadete Ayres, Hakan Demirci

Abstract

Introduction: This case is atypical presentation of ciliochoroidal melanoma in a young patient which was masqueraded by orbital cellulitis and focal scleritis.

Case Report: A 26-year-old female presented right orbital cellulitis with focal scleritis supero-temporally. She had a history of leaves hitting the right eye during gardening couple days prior. Visual acuity was 20/80 in her right eye. Fundus examination showed amelanotic ciliochoroidal lesion supero-temporally, measuring 18x17x4.0mm with exudative retinal detachment. Ultrasonography showed multilobulated solid ciliochoroidal lesion on B-scan and low to medium reflectivity on A-scan. Biopsy for periocular tissue and fine needle biopsy (FNAB) of ciliochoroidal lesion showed inflammation. There was a small focus of gram-positive cocci in the periocular tissue. After 7 days of oral antibiotic, her orbital cellulitis was improved. Serum Blastomyces antibody came back weakly positive. The patient was treated empirically for ocular Blastomyces, given fluconazole and then itraconazole. Ciliochoroidal mass initially shrunk from its original thickness of 4mm to 2.6mm in 4 weeks. But later it increased to 4mm and then 5.4mm in 6 and 12 weeks follow up, respectively. The 2nd FNAB was performed, which was undiagnostic. Enucleation of the right eye was performed and showed ciliochoroidal melanoma, with full thickness scleral invasion but no extrascleral excision and granulomatous inflammation/scarring on episcleral surface. Gene expression profiling revealed Class 1A, PRAME positive, GNAQ and SF3B1 positive tumor. There was no sign of systemic metastasis after 1-year follow-up.

Conclusion: Ciliochoroidal melanoma can be masqueraded by atypical presentation such as orbital cellulitis and focal scleritis.

Clinical Implication: When the clinical presentation does not correlate with diagnosis, biopsy of the lesion and close follow-up is important.

Financial Interest: *None*

D010

Atypical Ultrasound Biomicroscopy Appearance of a Ciliary Body Melanoma

Presenting Author: Neil Sheth, *Illinois Eye and Ear Infirmary, Chicago, USA,* neilshethmdmba@gmail.com

Co-authors: Michael Heiferman

Abstract

Purpose: Uveal melanoma (UM) is the most common intraocular malignancy in adults and has a poor prognosis.

Case Report: A 70-year-old male with a past medical history of hypertension, hyperlipidemia, prediabetes, and chronic sinusitis was referred for an asymptomatic intraocular tumor in the right eye. Seven years prior to presentation, the mass was incidentally noted on an MRI evaluating the patient's sinusitis. The patient was informed about the tumor and recommended evaluation but did not seek further care. Upon presentation, the patient's uncorrected vision was 20/50 and 20/40 due to cataracts with normal intraocular pressure. Anterior segment examination was without neovascularization, a scleral sentinel vessel, angle involvement or extraocular extension. The dilated fundus examination revealed an elevated melanocytic tumor 12 mm from the optic nerve head without vascularity, hemorrhage, inflammation or overlying pigment. Ultrasound biomicroscopy (UBM) revealed a heterogenous mass measuring 3.89 x 9.06 x 11 mm with discrete hypoechoic and hyperechoic foci. Fluorescein angiography was notable for blockage with an adjacent area of leakage. Fundus autofluorescence revealed hypoautofluorescence. Ocular coherence tomography demonstrated minimal drusen. MRI showed an interval increase in lesion size with enhancement but no extraocular extension. A subsequent full body PET/CT noted increased activity at the prostate. Incisional transscleral choroidal biopsy revealed atypical melanocytes, predominately spindle-type, consistent with a diagnosis of UM. Iodine-125 plaque brachytherapy was performed and the biopsy tissue was sent for further evaluation with gene expression profiling.

Conclusion: The atypical appearance on UBM led to the decision for incisional biopsy, which confirmed a diagnosis of ciliary body melanoma. Systemic workup was concerning for prostate cancer.

Clinical Implication: Biopsy of choroidal tumors is useful for cases that present atypically.

Financial Interest: *None*

D011

Local Resection Via Partial Lamellar Sclerouvectomy for Ciliary Body Tumors

Presenting Author: Xiaolu Yang, *Shanghai General Hospital, Shanghai, China*, dryangxiaolu@163.com

Co-authors: Kun Liu, Ruonan Wang

Abstract

Introduction: Ciliary body tumor is extremely rare and treatment is challenging. The aim of this study is to present our experience in treating this rare entity and to evaluate the surgical outcomes and complications of local resection via partial lamellar sclerouvectomy in four cases of ciliary body tumors in China.

Case Report: Four patients with ciliary body tumors who underwent partial lamellar sclerouvectomy (PLSU) in a single-center setting (Shanghai General Hospital, China) between October 2019 and April 2023 were enrolled. Tumor features, histopathologic findings, complications, visual acuity, and surgical outcomes were reviewed.

Conclusion: Four patients with a mean age of 31.8 years (range: 20-48 years) were included in this study. The histopathological diagnosis was adenoma of non-pigmented ciliary epithelium in 2 cases, schwannoma in 1 case, and multiple ciliary body pigment epithelial cysts in 1 case. The mean largest tumor base diameter was 6.0 mm (range: 2.0 - 10.0) and the mean tumor thickness was 3.5 mm (range: 2.0 - 5.0). Preoperative complications included cataract in 3 (75%) eyes, lens dislocation in 2 (50%), and secondary glaucoma in 1 (25%). Temporary ocular hypotonia was observed in one case and no other postoperative complications were observed. At a mean follow-up of 20.8 months (range: 5 - 47) , the best corrected visual acuity increased in 3 (75%) eyes and was stable in 1 (25%) eye. Tumor recurrence was absent in all eyes. All patients were alive at the end of follow-up.

Clinical Implication: Local tumor resection via PLSU is useful in the treatment of ciliary body tumors, especially for tumors with indeterminate features. It is a challenging technique, especially for large tumors, but is an ideal treatment because the eyeball is preserved, accurate histopathologic diagnosis is possible, and useful vision may be expected. Surgery-related complications were manageable with adequate preoperative assessment and careful operation during surgery.

Financial Interest: *None*

D012

Choroidal Melanoma

Presenting Author: Shagun Korla, *Maharishi Markandeshwar Medical College and Hospital, Solan, Himachal Pradesh, India,* shagunkorla@gmail.com

Co-authors: Shagun Korla, Ravinder Kumar Gupta

Abstract

Introduction: The choroid melanomas occur more frequently in comparison to iris and ciliary body melanoma and is the most common occurring intraocular malignancy amongst the adults. It is a rare malignancy with incidence of 6 cases per million population. Clinical presentation depends on the location and size of the choroidal melanoma.

Case Report: A 86-year-old, male Presented to Ophthalmology OPD with loss of vision in right eye associated with pain and right sided headache since 3 months.The ophthalmic examination showed no perception of light and projection of rays inaccurate in right eye. The intraocular pressure in right eye recorded was 42 mmHg. Slit lamp examination revealed circumciliary congestion with flat anterior chamber both centrally and peripherally. Pupil was slightly dilated and non reacting with cataractous crystalline lens. B-scan ultrasound of right eye showed a Collar button shaped lesion with solid consistency and low to medium internal reflectivity. There was presence of serous retinal detachment extending from margins of tumour. CEMRI showed a well defined altered signal intensity lesion with broad base towards wall, involving lateral half of posterior chamber of right globe in sub retinal location with right sided retinal detachment with sub retinal and vitreous haemorrhage. Right Optic nerve was unremarkable. There was no evidence of liver and lung metastasis. Modality of management involved enucleation of right eye.

Conclusion: Symptomatology is extremely variable with a choroid mass. Enucleation tends to be the method usually preferred for medium and large ocular melanomas.

Clinical Implication: Prognosis depends on several factors including, age of the patient, tumor size, histological characteristics and the presence of metastasis. Metastases, general health status and age should be considered in the treatment decision.

Financial Interest: *None*

D013

Peculiar Uveal Melanomas

Presenting Author: Vincenzina Mazzeo, *Solo Private Practice*, vmazzeosimonini@gmail.com

Abstract

Introduction: The adjective 'peculiar' may be given tumoral cases that also are rare Uveal Melanoma types. One case was presented at the ISOO Conference in 2019 under the tile 'The spontaneously presumed Small Pigmented Choroidal lesion shift into a T0 Choroidal Melanoma. A four-stage story'. The peculiar cases considered are the cavitary, amelanotic, bilateral and multiple Uveal Melanoma (UM).

Purpose: To describe some peculiar case

Case Report: A part UM, metastatic tumours and retinoblastoma may also be cavitary. Three cases of cavitary lesions will be described: one Choroidal Melanoma (CM) and two Cilio-Choroidal mass were seen by the presenter, and one is reported in a case of multiple tumours in the same eye. The tumour colour relates to the clinical aspect and so to the immediate diagnosis, recently it is considered a prognostic factor too by studying the fundus images and evaluating the pathology slides colour i.e. low pigment, high malignancy. Two cases of UM in patients with Oculocutaneous Albinism are also described. 26 cases multicentric UM were described from 1953 to 2022, 4 cases associated to Ocular Melanocytosis. The personal case described was also amelanotic in the three masses. The two news were found after five years. One of the multicentric masses showed no BAP1mutation, while another one a different gene expression profile in the two masses.

Conclusion: Bilateral cases are extremely rare, 15 papers were published from 1994 to 2023. One population study on 8915 pts demonstrate no difference in survivaland not poorer prognosis (Scott JF, Vyas R, Galvin J et Al. Clin Exp Ophtalmol. 2018; 46;502-10)

Financial Interest: *None*

D014

An Atypical Presentation of Diffuse Choroidal Melanoma

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Co-authors: Aghazadeh H, Negretti GS

Abstract

Introduction/Purpose: We report a case of uveal melanoma with diffuse infiltration of the ciliary body and choroid presenting initially as scleritis, followed by a presentation more in keeping with an eccentric disciform lesion than melanoma.

Case Report: A 68-year-old male had been treated for anterior scleritis elsewhere for 2 years prior to presentation with a vitreous haemorrhage (VH) and multi-lobulated superior ciliary body lesion with overlying haemorrhage, surrounding exudate, inferior retinal detachment and choroidal effusions. The appearance of the lesion was more suggestive of an eccentric disciform lesion or nodular scleritis than melanoma. There was no reduction in size with a trial of intravenous methylprednisolone. Two vitrectomies were performed but biopsy was impossible due to bleeding from the lesion. The lesion grew in elevation by 1.1mm during a 5-month period of observation. Transcleral biopsy eventually confirmed a diagnosis of uveal melanoma. Proton beam radiation was initially planned but enucleation was subsequently recommended due to ongoing VH and significant lesion growth. Pathology showed a large diffuse choroidal melanoma with 2 focal nodules and no loss of BAP1. Cytogenetics showed gains of 6p and 8q but normal chromosome 3.

Conclusion: This case emphasizes how challenging the diagnosis of uveal melanoma can be, particularly in the presence of vitreous haemorrhage, and how uveal melanoma can masquerade as benign conditions such as scleritis/eccentric disciform lesions for several years prior to the correct diagnosis being made.

Clinical Implication: Although rare, uveal melanoma can cause pain and in the presence of a sentinel vessel could be confused with scleritis. Eccentric disciform lesions are the most common masquerade for uveal melanoma. in the presence of VH, distinguishing between uveal melanoma and these masquerades becomes more difficult clinically and a biopsy may be needed.

Financial Interest: *None*

D015

Stop! Eyes Have Got a Pulse

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Co-author: Carol L. Shields

Abstract

Purpose: To emphasize the importance of radiological investigations for the evaluation of intraocular masses, in cases with obscured view of the posterior segment.

Case report: A 53-year-old male presented with a history of pain in the left eye for 2 weeks. His ophthalmologist noted elevated intraocular pressure and opted for conservative management along with a referral for further evaluation. At presentation, his vision was light perception in the right eye (OD) and no light perception in the left eye (OS). On fundus examination, peripheral bony spicules were noted in OD, consistent with his history of retinitis pigmentosa, and the view into OS was obscured due to dense cataract and vitreous hemorrhage. Ultrasonography of OS revealed a mushroom-shaped mass with a thickness of 15.9mm, “V-shaped” retinal detachment and spontaneous venous pulsations indicating towards a diagnosis of choroidal melanoma. The diagnosis was confirmed by magnetic resonance imaging, which revealed a choroidal mass with surrounding sub-retinal hemorrhage. He was advised systemic evaluation and was treated with enucleation of OS which confirmed the histopathological diagnosis of mixed-cell type choroidal melanoma and associated retinitis pigmentosa.

Conclusion: Obscured view of the posterior segment, especially in the setting of subretinal hemorrhage, offers a distinct diagnostic challenge in determining the prognostic trajectory of a choroidal mass. Spontaneous venous pulsations on ultrasonography help in differentiating a congealed sub-retinal hemorrhage from a more concerning vascularized tumor of the choroid. Furthermore, magnetic resonance imaging helps in confirming the clinical diagnosis.

Clinical Implication: Mushroom configuration, “V-shaped” retinal detachment and spontaneous venous pulsations on ultrasonography are critical findings which can aid in the diagnosis of choroidal melanoma, despite a poor view of the posterior segment.

Financial Interest: *None*

D016

De Novo Choroidal Melanoma

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Abstract

Introduction: Small choroidal melanocytic tumors may be a melanoma or nevus. Assigning diagnosis can be aided by features outside of the ophthalmoscopic examination including documented de novo onset. While a known phenomenon, very few reports exist describing these tumors, lack demonstrative imaging, or are restricted to single cases prohibiting any analysis that may provide insights into special features that many be unique relative to what one may consider common melanomas.

Case Report: We reviewed cases of de novo small choroidal melanocytic tumors presenting to the Cleveland Clinic and used an epidemiologic framework to recognizes them as melanoma. This is a retrospective case series including 5 patients from April 5, 2012 and July 22, 2022 with a mean follow-up of 5.7 years. All had de novo onset and were treated with plaque brachytherapy. The mean time between the last photo available for review and diagnosis of melanoma was 1466 days (40-3002) or 4.0 years (0.1-8.2). Tumors were small in 80% of cases (COMS) and medium in one case (20%). The mean largest basal diameter was 8 mm (4.5-16) and the mean apex height was 2.3 mm (1-6.6). All tumors except for the one medium were flat in appearance. All tumors demonstrated orange pigmentation and lacked drusen.

Conclusion and Clinical Implication: De novo choroidal melanoma can be mistaken for nevus without the aid and context of prior fundus photographs. There are no unique clinical characteristics specific to de novo melanoma except lack of detectable presence on a previous photograph. The approximate date of their first appearance can be estimated post hoc given growth rates known ex ante. Choroidal melanoma may arise de novo and may represent a unique class of choroidal melanoma.

Financial Interest: *None*

D017

Malignant Melanoma of the Choroid Presenting as Orbital Cellulitis and Apparent Response to Intravenous Antibiotics

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Co-author: Amit Arora, Mandeep Sagoo

Abstract

Introduction: We present a case of uveal melanoma presenting as orbital cellulitis which improved following intravenous antibiotic therapy. Interestingly, the tumour was minimally necrotic on pathological examination.

Case Report: A 51-year-old woman presented with 2 days of headaches, swollen lids & reduced vision. Visual acuity in the right eye was 6/12, left 6/5. Examination found 1mm right sided proptosis, chemosis & conjunctival hyperaemia. Dilated examination revealed a nasal pigmented lesion with overlying retinal haemorrhages & vitritis. B-scan found a moderate echogeneticy lesion 15mm x 9.1mm x 2.7mm, with associated suprachoroidal fluid & distension of sub-tenon's space. MRI demonstrated streaky enhancement of retrobulbar fat adjacent to an en-plaque curvilinear T2 hyperintensity along the supranasal aspect of the choroido-retinal layer. She was diagnosed with a subretinal abscess & treated with IV linezolid & voriconazole for 3 days followed by oral therapy. Pain & swelling improved on day 2. Repeat ultrasound on day 5 found a reduced elevation of 2.2mm, & clinically the haemorrhages & vitritis were improved. The patient was lost to oncology follow up. On review 1 year later the lesion was found to have increased to 20.5mm x 11.4mm x 4.9mm elevation. She chose to proceed with enucleation. Pathology found spindle cell melanoma with minimal focal necrosis, modest melanin pigmentation & patchy intratumoral lymphocytic infiltrate.

Conclusion: Cellulitis is an atypical but known presentation of uveal melanoma, however reduction in melanoma thickness following intravenous antibiotic therapy has not been previously documented. It is also unusual for a tumour with minimal necrosis to present with cellulitis.

Clinical Implication: This case illustrates the importance of considering uveal melanoma as a consideration in a cellulitic presentation despite apparent response to antibiotic therapy. It also demonstrates a tumour with minimal necrosis can present with a cellulitic picture.

Financial Interest: *None*

D018

Retinal Capillary Hemangioblastoma: Feeder Vessel Ligation Technique

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Co-author: Vishal Raval

Abstract

Introduciton: To present the surgical technique of feeder vessel ligation in the case of retinal capillary hemangioblastoma

Case Report: A 42-year-old woman was diagnosed with a solitary large retinal capillary haemangioma associated with localized tractional retinal detachment and massive exudation in the macular area. The patient's workup was negative for Von Hippel-Lindau syndrome. in this case, we demonstrate a unique surgical technique of 25G chandelier assisted bimanual vitrectomy highlighting feeder vessel ligation, followed by en bloc transscleral endoresection of the tumor with clinicopathological correlation. Surgical pearls during the various steps of surgery are highlighted in the video.

Conclusion: Surgical treatment for large and complex retinal capillary haemangioma is challenging. Bimanual feeder vessel ligation and endoresection of tumor can be an effective treatment in such cases, achieving long term anatomical success.

Clinical Implication: In summary, the clinical Implications of this surgical technique lie in its potential to advance treatment options, promote personalized care, enhance precision, and serve as an educational resource for the ocular oncology community.

Financial Interest: *None*

D019

The Unexpected while Expecting - An Exceptional Journey

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Abstract

Introduction: Hemangioblastomas are known to involve the cerebellum, spinal cord and central nervous system. Rarely, they occur at the retina or optic nerve, either alone or as a manifestation of von Hippel Lindau disease. The influence of pregnancy on disease progression in these cases remains uncertain. Here we are presenting a case of retinal hemangioblastoma diagnosed in a pregnant woman without an association of von Hippel Lindau disease and it's further management.

Case Report: At 20-weeks of gestation, a 29-year-old Caucasian female was referred with decreased vision in the right eye. On examination, an orange-coloured retinal lesion was seen inferiorly with surrounding exudates and cystoid macular edema which was causing deterioration of her vision. However, due to her ongoing pregnancy she denied the proposed treatment and interestingly over the course of her pregnancy, her vision improved spontaneously with resolution of cystoid macular edema. She was negative for von Hippel Lindau disease. Post-pregnancy, the lesion was treated with photodynamic therapy to secure vision. However, she developed cystoid macular edema again along with exudative retinal detachment which were managed well with anti-vascular endothelial growth factor injections leading to complete tumor control and a visual acuity of 20/25.

Conclusion: Asymptomatic retinal hemangioblastomas may turn symptomatic during pregnancy and it is recommended to treat them if associated with retinal exudative response.

Clinical Implication: Photodynamic therapy is an ideal mode of treatment for retinal hemangioblastomas with excellent visual outcomes and tumor control.

Financial Interest: *None*

D020

Retinal Vascular Proliferation with Fibrotic Regression in Von Hippel-Lindau Disease: A Case Report and Literature Review

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Abstract

Introduction: Retinal vascular proliferation in Von Hippel-Lindau disease is uncommon. We present a case of a sizable macular fibrovascular lesion in a young VHL patient. We also provide a literature summary, discussing clinical presentation and outcomes.

Case Report: A 12-year-old male patient came to our department with symptoms of obstructive hydrocephalus secondary to a left cerebellar hemangioblastoma. Simultaneously, the patient reported experiencing blurred vision for 1 to 2 years, with a visual acuity of 20/200 in the right eye and 20/100 in the left eye. Fundoscopic examination revealed a broad epiretinal fibrovascular membrane extending from the optic disc to the macula, causing significant traction in the right eye. Optical coherence tomography of the right macula demonstrated retinal distortion due to traction from the neovascular tissue. Genetic testing identified a pathogenic missense mutation (c.223A>G) within the VHL gene. Over a 3-year follow-up, the fibrovascular membrane persisted, with regressed vasculature on its surface. Despite undergoing pars plana vitrectomy, the patient's visual acuity remained at 20/200 one year postoperatively, accompanied by the development of cataracts.

Conclusions: For rare cases of retinal vascular proliferation in Von Hippel-Lindau disease, diligent consideration of treatment and outcomes remains imperative.

Clinical Implication: Our literature review underscores the recommendation for pars plana vitrectomy, given its potential to enhance macular anatomy and improve visual acuity.

Financial Interest: *None*

D021

Coats Plus Syndrome with New Observation of Drusenoid Retinal Pigment Epithelial Detachments in a Teenager

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Co-author: Carol L. Shields, Niko Kalafatis

Abstract

Purpose: To describe a case of Coats Plus Syndrome (CPS), a vision and life threatening disease belonging to a family of diseases known as the Telomere Biology Disorders.

Case Report: A 15-year-old girl with a history of small for gestational age, short stature, microcephaly, thinning/greying of scalp hair, skin hyperpigmentation, nail ridging, and multiple pathological fractures presented with bilateral Coats-like retinopathy. On presentation, visual acuity was 20/30 in the right eye (OD) and 20/25 in the left eye (OS). Anterior segment examination was normal in both eyes (OU). Fundoscopy OD revealed retinal telangiectasia, subtle exudation inferior to telangiectatic vessel, and vascular sclerosis temporally, whereas fundoscopy OS showed clinically normal vascularity Fluorescein angiography (FA) revealed localized temporal non-perfusion with related telangiectasia and minimal leakage OD, minimal temporal non-perfusion without leakage OS, and pinpoint staining in the retinal pigment epithelial detachments (PEDs) OU. We discovered a new observation of multiple peripheral pinpoint retinal PEDs. Further genetic testing revealed CTC1 gene mutation and she was diagnosed with Coats plus syndrome with features of dyskeratosis congenita, a telomere biology disorder. Patient received laser photocoagulation treatment in OD and observation OS and was referred to paediatrician for further systemic management.

Conclusion: Coats Plus Syndrome is a vision and life threatening disease and needs a multimodal approach of management.

Clinical Implication: Patients with bilateral Coats-like retinopathy and associated systemic features suggestive of CPS should be evaluated through genetic testing to diagnose this disease and treat vision and life threatening manifestations as early as possible. Moreover, multiple pinpoint PEDs that could be related to an accelerated aging process with telomere dysfunction.

Financial Interest: *None*

D023

Plaque Brachytherapy - A Port Wine Aficionado

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Abstract

Introduction: Choroidal haemangioma, is a congenital hamartomatous lesion. It may circumscribed or diffuse. Diffuse choroidal haemangioma patients are known to exhibit features of Sturge-Weber syndrome. Most commonly presents to an ocular oncology clinic in the adulthood. Diagnosis is essentially clinical acoupled with an ultrasound B scan for tumour measurements and multi-modal imaging.

Case Report: A 12-year-boy presented to us with complaint of pinkish discolouration of left side of the face since birth. He gave history of prior ophthalmic consult for red eye and was opined to have choroidal haemangioma of left eye as a part of spectrum of Sturge-Weber syndrome. External examination revealed the port wine stain of left hemiface with no evidence of lip hypertrophy. Ocular evaluation revealed,VA of 6/6 RE, PL+, inaccurate PR LE; grade 1 RAPD, raised IOP, diffuse injection of the ocular surface & dilated tortuous episcleral, scleral vessels. Fundoscopic exam showed a diffuse, pinkish-red mass with exudative retinal detachment. Ultrasound B-scan revealed a 5.68 x 12.28 x 19.42mm lesion in the largest dimension. A contrast-enhanced MRI of the orbits & brain ruled out cerebellar involvement. He was diagnosed to have Sturge-Weber syndrome with diffuse choroidal haemangioma & secondary glaucoma. He underwent episcleral plaque brachytherapy with a notched BARC 2022 plaque with 4 doses of intravitreal anti VEGF injections LE &showed an excellent response with respect to eye and vision salvage.

Conclusion: Episcleral plaque brachytherapy coupled with intravitreal anti-VEGF is an excellent modality of treatment in cases of diffuse choroidal haemangioma which addresses the pathology &minimises the risk of radiation retinopathy respectively.

Clinical Implication: Choroidal haemangioma, most commonly involves the posterior pole and thereby warrants intervention to mitigate the risk of amblyopia and alleviation of symptoms arising from the secondary effects of choroidal lesion

Financial Interest: *None*

D024

Neoadjuvant Proton Beam Radiation Therapy Followed by Endoresection of a Large Uveal Glial Tumour in a Young Patient.

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Co-author: Georgios Blatsios, Peter Meyer, Gertud Haas

Abstract

Introduction: A rare case of a large uveal glial tumour in a young patient.

Case Report: A 17-year-old male patient presented with progressive vision loss to counting fingers and visual field defects on his left eye. Clinical examination revealed a large uveal amelanotic tumour mass in the inferotemporal quadrant with serous retinal detachment involving the macula. The tumour was treated with proton beam radiation therapy (PBRT) followed by endoresection, to avoid toxic tumour syndrome, due to its large mass. Histology revealed a low malignancy glial tumour. Visual acuity recovered significantly and no signs of metastasis were found.

Conclusion: Proton Beam Irradiation Therapy and endoresection are a valuable treatment of large uveal glial tumours.

Clinical Implication: Glial uveal tumours are extremely rare and although often of relatively low malignancy, they can have a devastating potential on vision. Irradiation (such as PBRT) and potential endoresection of large uveal glial tumours can be beneficial and vision saving. Differential diagnosis should always include these entities in atypical uveal tumour cases.

Financial Interest: *None*

D025

A Cotton Wool Spot Like Lesion Unveils Underlying Tuberous Sclerosis

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Abstract

Introduction: An ophthalmologist is often the first person to give an insight into the diagnosis of many undetected systemic syndromes. We present one such case where a retinal lesion unveiled the diagnosis of an underlying systemic syndrome.

Case Report: A 27-year-old male patient attended our hospital for routine check-up. His best corrected visual acuity was 20/20 in right eye (RE) and 20/80 in left eye (LE). On refraction, he was found to have emmetropia in RE and high myopia with anisometropic amblyopia in LE. On fundus examination, an isolated cotton wool spot was noted in his left eye for which he was referred to the retina clinic. On careful examination in the retina clinic, the cotton wool spot like lesion had atypical features with diffuse margins and an approximate size of a little more than half a disc diameter with no other associated changes in background retina. Optical coherence tomography (OCT) was done whichshowed the presence of a hamartomatous lesion in the nerve fiber layer characteristic of astrocytic hamartoma seen in Tuberous sclerosis. Magnetic resonance imaging (MRI) of brain was then done which clearly revealed the presence of multiple cortical tubers. The patient was also referred to a dermatologist, who identified the skin lesions as angiofibromas of tuberous sclerosis.

Conclusion: The patient was counseled regarding the disease and was informed about the warning symptoms of possible complications along with the need for regular follow up with a neurologist.

Clinical Implication: This case classically depicts the importance of careful ophthalmic examination in identifying syndromes like tuberous sclerosis, an early diagnosis of which can be crucial for better management and long-term prognosis of the patients.

Financial Interest: *None*

D026

Agressive Giant Retinal Astrocytoma

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Abstract

Purpose: To report on the clinical and histopathological findings of a refractory aggressive retinal astrocytoma in tuberous sclerosis, which showed progressive growth after two intra-arterial chemotherapy sessions.

Case Report: A 15-year-old girl diagnosed with tuberous sclerosis (TSC) since infancy was referred to our clinic with blurred vision in her left eye since 1 year. The patient presented with a large papillary astrocytoma in the left eye. Visual acuity was 20/500 and the intraocular pressure was 18 mmHg. Firstly, photodynamic therapy (PDT) was offered, but was not possible due to regional drug shortage. On follow-up, the tumor showed enlargement with progressive exudative retinal detachment and vitreous hemorrhage, such that PDT was no longer feasible. As the patient and family refused enucleation, intra-arterial chemotherapy (IAC) was offered as an eye salvage attempt. However, two courses of IAC with combined chemotherapy agents showed no improvement but further growth of the tumor. Therefore, secondary enucleation was performed. We present the ocular findings, the clinical course and the histopathologic findings.

Conclusion and Clinical Implication: Although retinal astrocytic lesions in tuberous sclerosis are generally stationary, they can sometimes grow inexorably and cause serious ocular complications such as exudative retinal detachment with visual loss. Patients with TSC should have regular ophthalmological examinations, even if asymptomatic, in order to be able to offer alternative therapy in a timely manner. We herewith, report a treatment attempt by IAC as treatment attempt for aggressive giant retinal astrocytoma, which was however in this case unsuccessful.

Financial Interest: *None*

D027

A Rare Case of Retinal Pigment Epithelial Adenocarcinoma in a Painful Blind Eye

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Co-author: Marian Pauly

Abstract

Introduction: Retinal pigment epithelial (RPE) adenocarcinoma are rare tumors which has been rarely if ever diagnosed prior to enucleation.

Case Report: A 68-year-old lady presented with right eye pain of 3 months duration. She gave history of no perception of light in the same eye since child hood, the aetiology for the same was not clear . On examination right eye showed nil perception of light and showed early features of pthysis bulbi. Since there was no view of fundus a B scan ultrasonography (USG) was done which showed features suggestive of possible old retinal detachment with subretinal echoes and ocular coat calcification with no evidence of any mass lesion. She was diagnosed as right painful blind eye and started on topical steroid eye drops . Her next follow up visit was 2 years later with persistent pain in right eye. A USG B scan was repeated which showed an area of high echogenicity suggestive of calcification, the retina and choroid could not be delineated properly .Since her ocular pain was persisting she was taken up for an evisceration + ball implant. Intra-operatively a whitish mass lesion was noted along with scleral thickening and adherent uveal tissue. Hence proceeded with an enucleation. Histopathology was suggestive of adenocarcinoma involving sclera and choroid with possibilities of either a primary retinal pigment epithelium adenocarcinoma or a metastatic lesion. A detailed systemic work up was done, which ruled out any primaries or metastasis. Hence the final diagnosis was primary RPE adenocarcinoma. Her last follow up visit was 3 years following the enucleation and she was doing well without any recurrence.

Conclusion: In the management of a blind painful eye, it is extremely important to rule out an intraocular malignancy.

Clinical Implication: In long standing painful blind eye with unknown aetiology always go for primary enucleation.

Financial Interest: *None*

D028

Usual Presentation of Unusual Masquerade, Choroidal Lymphoma - A Case Report

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Abstract

Introduction: Diagnosis and management of intraocular lymphoma are challenging as they often mimic other intraocular diseases. There is a paucity of literature relating to choroidal lymphoma (CL) in particular. We intend to report this rare case of CL to explore the specific clinical manifestations and enable early diagnosis.

Case Report: A 61-year-old Caucasian male was referred to Yeovil Hospital, UK in 2019 with increasing soft drusen-like lesions in the right eye. His vision was -0.1 LogMAR in both eyes with quiet anterior and posterior chambers. Fundus examination in the right eye showed multifocal, yellowish-creamy circular, hyperautofloroscent choroidal lesions. OCT macula was normal. So, he was observed without intervention. During the year review, the Fundus examination was stable. However, the OCT macula showed increased choroidal thickness. Therefore, referred to the ocular oncology for suspected CL. Unfortunately, he was lost to follow-up and presented with a drop in right-eye vision. Fundus examination showed multiple hypopigmented choroidal spots with pronounced choroidal folds, confirmed on OCT scan which showed choroidal thickening and undulating folds of the choroidal surface, like a "sea storm" (seasick appearance), suggestive of CL. MRI brain and PET scan ruled out lymphoma at any other site. A biopsy of the choroidal lesions confirmed low-grade B-cell lymphoma. Hence, a primary CL was confirmed and referred for Right eye External Beam Radiotherapy.

Conclusion: Establishing the diagnosis of CL can be challenging and is often delayed. A definitive diagnosis of malignant lymphoma requires histopathological confirmation. A thorough systemic examination and evaluation with appropriate imaging is crucial to rule out systemic lymphoma.

Clinical Implication: The possibility of CL should be considered early in the diagnostic process based on the specific signs seen on fundus examination and OCT. OCT often gives more conclusive evidence early on.

Financial Interest: *None*

D029

An Unusual Delayed Treatment Response to Intravitreal Methotrexate for Primary Vitreoretinal Lymphoma

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Co-author: Saumya Jakati

Abstract

Introduction: Primary vitreoretinal lymphoma (PVRL) can masquerade as posterior uveitis. PVRL is a condition that can easily be misdiagnosed. Vitreous and/or subretinal biopsy of subretinal infiltrates is imperative for clinching the diagnosis of PVRL. Intraocular methotrexate (MTX) is the most widely used treatment option for PVRL. Response to treatment is typically observed within 2 weeks of initiating therapy. However, in rare scenarios, resistance to MTX is observed.

Case Report: A 66-year-old woman presented with complaints of blurred vision in her right eye for three months. The patient was diagnosed with toxoplasma retinochoroiditis and was already being treated with oral sulfamethoxazole-trimethoprim and oral prednisolone for two weeks. The patient had vitritis with yellowish subretinal nodular lesions. Optical coherence tomography (OCT) showed multiple subretinal hyperreflective lesions. To confirm the diagnosis, a 25G pars plana vitrectomy-based subretinal biopsy was performed using a novel beveled soft tip cannula. The patient was diagnosed with PVRL and received three intravitreal injections of MTX (400 micrograms/0.1 cc) over 10 days. However, no response to treatment was observed, and the patient developed corneal epithelial toxicity due to MTX. Further injections were withheld, and the patient returned for review a month later. Surprisingly, there was a marked improvement in her condition, and PVRL lesions were regressing. Treatment with intravitreal MTX was restarted.

Conclusion: This case highlights an unusual, delayed response to intravitreal MTX for PVRL treatment.

Clinical Implication: We have described a novel technique using a modified beveled soft tip cannula to increase the yield of biopsy. Various plausible explanations for the delayed response to intravitreal MTX are highlighted in this study.

Financial Interest: *None*

D030

Triple Whammy - A Heart Touching Story

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Abstract

Introduction: Choroidal sarcoidosis is a rare manifestation of systemic sarcoidosis, affecting approximately 1% of patients with differentials including neoplastic etiology based on clinical presentation of lesions. Here we have described the management of such a case with a complex past history of systemic sarcoidosis and renal cell carcinoma.

Case Report: A 66-year-old Caucasian male with a complex medical history of pulmonary and cardiac sarcoidosis, systemic hypercalcemia, and renal cell carcinoma, presented with vision loss in the left eye to 20/40. Fundus examination revealed multifocal confluent yellowish-white lesions with vermiform margins in the peripapillary area with hypoautofluorescence. Optic coherence tomography showed retinal pigment epithelial irregularity and sub-retinal fluid with hyperreflective deposits. Ultrasonography showed dome-shaped multifocal lesions with moderate acoustic solidity, base of 12mm and maximum thickness of 4mm. Fluorescein angiography showed hyperfluorescence, whereas indocyanine green angiography displayed hypofluorescence. Systemic evaluation ruled out metastasis based on positive emission tomography and a final diagnosis of choroidal sarcoid granuloma was made. Systemic corticosteroids were initiated which led to complete resolution of the choroidal lesions with complete vision salvage.

Conclusion: Choroidal sarcoidosis often needs to be differentiated from choroidal metastasis in cases with complex history. Corticosteroid therapy leads to excellent outcomes with vision salvage, and complete resolution of these lesions.

Clinical Implication: With the differentials of etiology ranging from an underlying systemic manifestation to neoplastic disease, a systematic evaluation is mandatory for achieving the final diagnosis, as the treatment differs in both the conditions.

Financial Interest: *None*

D031

Intraocular Manifestation of Nocardia Asteroides

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Abstract

Introduction: Nocardia asteroides is a grampositive , filamentous and aerobic bacterium. Its commonly found in soil and can cause a range of infections in humans, particularly in individuals with a suppressed or weak immune system. Here, it oftentimes causes pulmonary, skin and soft tissue infections. Even under adequate antibiotic therapy the mortality rate is up to 47%.

Case Report: A 70-year old male was referred to our Department with the high suspicion for a choroidal metastasis in his right eye. He was treated for a renal cell carcinoma and prostate carcinoma Visual acuity on the right eye was 20/30 and on the left of 20/20. The initial IOP was normal. The anterior segments of both eyes was unremarkable, whilst on fundoscopy a 2x8x12mm amelanotic lesion was present on the nasal mid periphery on the right eye. Based on these results, we initially recommended a staging for prostate and renal cell carcinoma. Two weeks later, the patient showed a rapidly declined VA on the right eye down to hand movement, fundus examination showed dense vitreous opacities. We decided to perform a vitrectomy and biopsy and referred the patient to the general oncology department. Two Months later, the patient presented with corneal perforation associated with endophtalmitis of the right eye. Enucleation was performed which resulted in Nocardia asteroides complex infiltration. Thereafter the antibiotic treatment with imipenem and amikacin resulted in resolution of a small lesion, which simultaneously developed on the left eye. Despite this, the general health of the patient deteriorated over the following months and the patient succumbed due to septic shock.

Conclusion: Nocardiosis is an important differential diagnosis to choroidal amelanotic lesions such as metastasis, amelanotic uveal melanoma and intraocular lymphoma. Despite antibiotic treatment, as much as 50% of immunocompromised patients succumb due to their general condition associated with systemic nocardia manifestation.

Financial Interest: *None*

D032

Behind the Curtain's Mystic Fold, Lies the Glowing Future Untold

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Co-author: Hidayet Sener, Miguel Hernandez-Emanuelli, Irwin Leventer, Carol Shields

Abstract

Introduction: Choroidal metastasis are rare manifestations with 8% occurring on autopsy. The primary lies in breast in 33% and lung in 25% and the lesions present to the ophthalmologists first as multi-focal, post-equatorial choroidal lesions and are often misdiagnosed. Even though they are associated with disseminated disease and poor prognosis, the advent of check-point inhibitors holds a promising future for these patients.

Case Report: A 56-year-old Caucasian female was referred with complete retinal detachment in the right eye (with visual acuity limited to light perception), thus obscuring the fundus view. Left eye showed multiple creamy-yellow choroidal lesions (with visual acuity of 20/50). She had a history of basal cell carcinoma over the lip 2-years ago and a recent diagnosis of breast carcinoma. With the initiation of a novel check-point inhibitor, abemaciclib as oral chemotherapy, the patient showed excellent response with vision improvement of 20/50 in right eye and 20/20 in left eye with complete regression of the choroidal lesions. The wonder drug led to the miraculous improvement in her vision, lifting off the curtain and unfolding the glowing future of these patients.

Conclusion: Choroidal metastasis requires accurate timely diagnosis and it's presence leads to early detection of a possible systemic primary carcinoma. Timely initiation of treatment in these cases leads to resolution of symptoms and a longer lifespan.

Clinical Implication: A clear understanding of choroidal metastasis for the ophthalmologists helps in early detection and treatment initiation for the primary lesion. The check-point inhibitors play a promising role and hold a bright future for treatment in these patients.

Financial Interest: *None*

D033

Cutaneous Melanoma Metastatic to the Vitreous Cavity

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Co-author: Noel Horgan

Abstract

Introduction/Purpose: Cutaneous melanoma metastatic to the vitreous is a very rare clinical entity. We present a case which was seen in our outpatients prior to the commencement of immune checkpoint inhibitors. We aim to make our audience aware of its differential diagnosis, treatment and potential complications.

Case Report: A 31-year-old female was reviewed in the ophthalmology clinic for screening of a tumour primary after local excision of a pigmented lesion on her left shoulder (Malignant Melanoma pTA4, BRAF V600 mutant). She was 20/20 OU and asymptomatic from an ocular point of view. Examination of the posterior segment revealed a sheet like veil of pigmented cells in the sub hyaloid space. Vitrectomy was undertaken which revealed that the cells were positive for Melan-A and SOX10. Subsequent systemic imaging was revealed ovarian, brain lesions and iliac nodes suspicious for metastatic melanoma metastases. Patient was commenced on combination immunotherapy (Ipilimumab and Nivolumab)

Conclusion: Cutaneous melanoma metastatic to the vitreous appears to be more common in the era of immune checkpoint inhibition. With these medications, patients are evading life-threatening systemic metastases and are therefore living longer. Treatment was originally with enucleation or external beam radiotherapy but recently there has been a move toward intravitreal melphalan due to its safety and potential efficacy profile.

Clinical Implication: Systemic treatment with immune checkpoint inhibitors may unmask vitreous disease, either by allowing the patient to live longer and therefore allow distant sites to develop metastases or by immune activation of immune-privileged sites. Clinical suspicion for vitreous deposits from metastatic disease should be high and, in many cases, a vitreous biopsy in warranted. The jury is still out on the most appropriate treatment protocol, but intravitreal melphalan is proving to be a valid option.

Financial Interest: *None*

D034

Leukemic Infiltrates in Blast Crisis of Chronic Myeloid Leukemia

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Abstract

Introduction: Chronic Myeloid Leukemia is a myeloproliferative disorder with triphasic presentation- chronic, accelerated and blast phase. Clinically significant ocular involvement (mainly posterior segment) can occur in upto 50 % of leukemia patients at the time of diagnosis.

Case Report: A young male in his late twenties complained of progressive painless diminution of vision in his right eye noticed for 2 weeks. The patient had a loss of vision in the left eye for 5 years secondary to penetrating ocular trauma. His best corrected visual acuity in the right eye was 20/100 with no apparent abnormality in an anterior segment on slit lamp examination. Fundus examination of the right eye revealed dilated tortuous retinal veins, and multiple retinal infiltrates with interspersed hemorrhages more confluent in mid periphery. Baseline laboratory investigations revealed a strikingly raised total leucocyte count - 253000/mm3 with peripheral blood film showing numerous blast cells, and the cytogenetic study demonstrated- abnormal Breakpoint cluster-Abelson 1 (Bcr-Abl 1) fusion gene (Philadelphia chromosome), which confirmed the diagnosis of blast crisis of Chronic Myeloid Leukemia (CML). The patient was hospitalized and treated with systemic chemotherapy (imatinib mesylate) for CML; clinical improvement was noted in the right eye on follow-up, with a reduction in the size of retinal infiltrates, vascular tortuosity, and improvement in vision to 20/50.

Conclusion: The presence of large retinal infiltrates along with dilated tortuous vessels could be a sign of leukemic retinopathy. Our patient responded well to systemic tyrosine kinase inhibitor with a reduction in retinal infiltrates and improvement in vision.

Clinical Implication: It is advisable to rule out masquerade in patients with massive retinal infiltrates and unexplained vascular tortuosity. Ocular involvement could be the first presentation in patients with Chronic Myeloid Leukemia.

Financial Interest: *None*

D035

Relapse of ALL Masquerading as Ocular Involvement

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Co-authors: Saloni Desai, Sashwanthi Mohan, Jyotirmay Biswas

Abstract

Introduction/Purpose: To report a rare case of ocular involvement in a child with a relapse of Acute Lymphoblastic leukemia (ALL).

Case Report: We report an unusual case of a 7-year-old female, a known case of ALL diagnosed 2 years ago, who came to our out patient service with complaints of diminished vision in the right eye for 2 months. She had a history of chemotherapy in the past and was on maintenance treatment when she presented to us. MRI Brain was done recently which was within normal limits. The visual acuity on Snellen's distant vision chart was 6/7.5 and for near it was N6. The anterior segment examination showed a grayish-white hypopyon along with whitish fluffy diffuse infiltrates over the iris. The lens was clear and had pigments on the anterior lens capsule. The left eye slit lamp examination and fundus were within normal limits. Right eye ultrasound was done in which there were few low reflective dot vitreous echoes and the retina was attached. Anterior segment Optical coherence tomography was done which showed thickening and undulations of the iris. Ultrasound Biomicroscopy (UBM) confirmed the thickening of the iris and ciliary body along with the presence of thick pars plana membranes. The peripheral blood smear was sent which was negative for malignant cells. Thereafter the patient was taken up for a right eye aqueous chamber tap under general anaesthesia and the aqueous was sent for cytological analysis. The H&A smear was highly cellular and showed numerous large lymphoid cells with a high nucelo-cytoplasmic ratio which was suggestive of Blast cells. This concluded the relapse of ALL, presenting as pseudohypopyon with iris and ciliary body infiltration. The child was immediately referred back to the oncologist for re-initiation of chemotherapy.

Conclusion: Relapse of ALL can masquerade rarely as hypopyon and it signifies CNS involvement with poor prognosis.

Clinical Implication: Patients with ALL need routine ocular examination.

Financial Interest: *None*

D036

Bilateral Retinoblastoma in a Child with Simpson Golabi Behmel Syndrome Phenotype - A Possible Non-fortuitous Association

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Abstract

Introduction: Bilateral retinoblastoma (RB) usually results from germline mutations in the RB1 gene. Simpson-Golabi-Behmel syndrome (SGBS) is caused by mutations in a gene encoding Glypican 3 (GPC3). GPC3 gene mutations prevent glypican 3 from inhibiting the Sonic Hedgehog (SHH) signalling pathway. Dysregulation of SHH signalling is one of the possible candidates of an additional pathological alteration needed for RB development.

Case Report: A 1-year-old boy, was evaluated for seizure disorder, was incidentally found to have bilateral intraocular mass and was referred as a case of suspected bilateral RB. The boy is the first and only child of healthy unrelated parents. in view of abnormal gestational USG, amniocentesis was done and FISH was negative for any abnormality of chromosomes 13, 18, 21. The child was born on term by vaginal delivery. Child has coarse facial features, prominent forehead, hypertelorism, epicanthal folds, broad nasal bridge, upturned nostrils, narrow bow shaped upper lip, macrostomia, macroglossia, accessory nipple. Clinical Geneticist's opinion was sought and a possibility of SGBS was given. He had Group B RB in right eye and Group E RB in left eye with secondary angle closure glaucoma. MRI showed no extrascleral extension/ optic nerve/ CNS involvement. CSF cytology showed 70% atypical cells. He was started on palliative chemotherapy with standard VEC.

Conclusion: This is the first reported case of RB in a child with SGBS phenotype. However, molecular results are awaited. High SHH signalling are associated with advanced disease as seen in this case.

Clinical Implication: Management of bilateral RB with CSF positivity in a syndromic child is always challenging. It never stops with addressing RB alone, it extends to addressing other issues associated with SGBS, genetic counselling, social, financial and emotional support for the parents. in future, therapeutic targeting of pathways like SHH in RB could make some difference.

Financial Interest: *None*

D037

Enigma of Intraocular Mass Lesion in A 22 Year Old Male

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Co-authors: Amita S Verma, Aminder Singh

Abstract

Introduction: Retinoblastoma (RB) is the most common intraocular tumor in children <5 years of age. Its occurrence in adults is rare and a diagnostic challenge. Herein, we describe a case of adult-onset RB with clinical, radiological, and pathological findings.

Case Report: A 22-year-old boy presented with blurring of vision in right eye from past 1 month. On examination, the visual acuity was 6/24, N36 in the right eye and 6/6, N6 in the left eye. On fundus examination of the right eye, there was a mass lesion in the temporal half associated with multiple subretinal and vitreous seeds. Widefield imaging, USG B scan, optical coherence tomography and contrast enhanced MRI were obtained and the diagnosis of Group D retinoblastoma was established. Patient underwent enucleation of the right eye. Histopathology and IHC confirmed the diagnosis of retinoblastoma with no high-risk features. All the relevant images will be shown and discussed in the presentation.

Conclusion: The diagnosis of RB in an adult patient is challenging and a misdiagnosis could lead to wrong or delayed treatment. A widefield fundus image and the clinical, radiological, and pathological evidence in this case are invaluable in making a correct diagnosis of adult-onset RB.

Clinical Implication: The cause of adult-onset RB is still unknown. It has been speculated to occur due to the reactivation of a spontaneously regressed RB also known as retinocytoma or through the persistence of embryonic retinal cells with the RB1 gene mutation. The reliability of adopting ICRB to predict outcomes in patients with adult-onset RB is debatable and requires further research.

Financial Interest: *None*

D038

It is not Merely a Retinal Tumour but the Journey of Tears, Fears, Hope, and Faith

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Co-authors: Shabana Choudhary, Aisha Azam, Andleeb Zahare, Samreen Jamal, Asim Amjad, Asad Aslam Khan

Abstract

Introduction: A late presentation of retinoblastoma in low-income countries is frequently mentioned in the literature but a late-onset retinoblastoma as a masquerading syndrome is rare.

Purpose: To report a case of late-onset retinoblastoma mimicking anterior uveitis and the perplexity of the management in our setup.

Case Report: A 9-year-old girl, presented in 2019 with a complaint of headaches and blurred vision in both eyes. On ocular examination, there was hypopyon in the right eye. The fundus examination revealed vitritis in the right eye. On the B scan, there were no signs of intraocular calcification. After EUA and MRI, the final diagnosis of Group E retinoblastoma (IIRC) was made. Parents refused enucleation. The patient survived cardiac arrest during the 5th chemo cycle.

Conclusion: A patient's vision and life were saved through multidisciplinary teams' efforts, but concerns remain about late metastasis or secondary tumors, along with ongoing psychosocial and economic Implications.

Clinical Implication: Recent global data shows a surge in late-onset retinoblastoma cases, highlighting the need to identify reasons to reconsider age ranges and prevent misdiagnosis. The focus on vision and globe salvage in treatment protocols raises the question of updating classification systems (IIRB & IIRC) and treatment guidelines, considering visual acuity in decision-making. e.g., anterior segment involvement is labelled as group E Can we label a patient with BCVA of 6/12 presented as a masquerading syndrome as group E retinoblastoma? Is it worth saving the eye given the potential risk of endangering a child with late metastasis or secondary tumors? In low-income countries, it is crucial to comprehend the psychosocial Implications and economic burdens before deciding on a turbulent path.

Financial Interest: *None*

D039

Conquering Retinoblastoma: The Homestretch

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Abstract

Introduction: Midline intracranial primitive neuroendocrine tumors (PNET) can be seen in 3 - 5% of children with germline retinoblastoma (RB) together called trilateral RB (TRB). Three quarters of PNET occur in the pineal gland, the rest being non-pineal, commonly suprasellar and larger. in the pre-chemotherapy era non-pineal TRB was fatal. Currently with high-dose chemotherapy and stem cell rescue the survival in both pineal and non-pineal tumors have increased to above 50%. We present a case of non-pineal TRB treated with chemotherapy, neurosurgery and stem cell rescue and explore the available evidence-based management guidelines.

Case Report: A 7-month-old, Hispanic girl, diagnosed with delayed motor milestones and poor eye tracking was referred to us for evaluation. General physical examination revealed lethargy. Anterior segment was normal in both eyes (OU) with no leukocoria. Fundus examination showed bilateral endophytic, multifocal RB, ICRB (International classification of retinoblastoma) group B. Magnetic resonance imaging (MRI) of the brain and spine showed a large suprasellar mass 46 millimetres in largest diameter, with normal spine. Genetic analysis showed germline mutation with 13q deletion. Cerebrospinal fluid was negative for malignant cells. She was treated with intensive multimodality therapy including 4 cycles of chemotherapy according to the Children's Oncology group trial for trilateral RB (ARET0321), followed by neurosurgery, high-dose chemotherapy and autologous stem cell rescue. Child continues to be stable 4 months following treatment, with good resolution of PNET and regressed RB, but on supportive care for diabetes insipidus.

Conclusion: Neuro-imaging at the time of diagnosis of bilateral RB is imperative, especially in children with delayed milestones.

Clinical Implication: Due to its rarity, there exists little level one evidence for the treatment of TRB. Centres of excellence must collaboration for robust protocols.

Financial Interest: *None*

D040

Very Late Recurrence of Retinoblastoma following Primary Treatment with Intra-arterial Chemotherapy

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Co-authors: Stephen Chen, Cindy Herzog, Brandon Z Brown, Emmanuel Chang

Abstract

Introduction/Purpose: Intra-arterial chemotherapy (IAC) is a standard technique used to treated advanced and recurrent intraocular retinoblastoma. Unlike other modalities less is known about its risk for very late recurrence.

Case Report: We present a female teenager successfully treated as an infant for bilateral Group D retinoblastoma with multi-agent IAC. Over a decade later, on routine follow up, one eye developed an asymptomatic recurrence. The eye was successfully salvaged with repeat IAC therapy. We present the subsequent therapy associated photos & angiograms in this heavily pre-treated eye.

Conclusion: Like other modalities, treatment of intraocular retinoblastoma with IAC can be associated with late recurrence. Its long-term impact on the orbital vasculature is less understood.

Clinical Implication: All eyes successfully treated with IAC should be serially examined for the risk of very late recurrence.

Financial Interest: *None*

D041

Double Plaque for Double Trouble

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Co-authors: Sima Das

Abstract

Introduction/Purpose: Radioactive episcleral plaque is a treatment method for selected retinoblastomas (Rb) where other therapeutic methods of treatment have failed.

Case Report: A 4-year-old female with bilateral retinoblastoma (OD Group D multifocal with diffuse vitreous seeding and OS group E Rb with neovascular glaucoma) received systemic chemotherapy and focal therapy. Left eye was non salvageable and was enucleated whereas, right eye had multiple retinal recurrences, even after focal treatment and intravitreal chemotherapy (IVitC). Ruthenium plaque brachytherapy was performed for residual disease. After few months, patient had another recurrence over the treated tumor for which intra-arterial chemotherapy was tried. Extremely difficult cannulation and intraoperative spasm made further cycles impossible. Minimal response was noted for which further IVitC were performed. Sudden vision drop due to melphalan toxicity with multiple new pre-retinal seeds were seen. Six months later, a new tumor recurrence was noted over a different site. Second plaque was then performed after tumour board approval. The tumour regressed on follow up but subsequently developed radiation retinopathy changes. Fundus fluorescein angiography was performed periodically and anti VEGF injections were given. Patient is maintaining a stable functional vision for now.

Conclusion: Plaque radiotherapy is successful in effectively treating selective recalcitrant cases providing adequate tumour control and improving patient survival.

Clinical Implication: Rb remains the most commonly seen malignancy occurring in pediatric age group with delayed presentation in these parts of the world. Repeat plaque therapy may be needed in selectively difficult cases, such as our case with a sole precious eye. Proper counselling, correct dosimetry calculations, co-ordination with physicist, monitoring of radiation related changes and timely anti-VEGF can prevent complications in such cases.

Financial Interest: *None*

D042

Xeroderma Pigmentosum with Ocular Involvement

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Co-authors: Alena Calvo, Yanisis HernÃ¡ndez-Ortiz

Abstract

Purpose: to showcase the complexity of management in xeroderma pigmentosum with ocular involvement.

Case series: Case 1 is a 7-year-old female with typical XP cutaneous xerosis, visual acuity of 20/30 both eyes and a 10 mm corneal leucoma in the nasal periphery of left cornea. Main treatment was observation and eye lubricants. Case 2 is a 6-year-old male with personal and family history of XP and localized squamous cell carcinoma in the left shoulder. Visual acuity 20/40 both eyes and slit lamp microscopy shows bilateral conjunctival hyperemia with localized epithelial thickening and vascular changes. Both corneas showed sectoral haziness towards the temporal limbus. Main treatment was topical fluorouracil as prevention. Case 3 is an 8-year-old female with XP and history of multiple surgeries in the face, head, and neck region due to carcinomas. Visual acuity is 20/70 both eyes and upon evaluation, an irregular and multilobulated lesion in the right lower lid margin is noticed. The ocular surface in both eyes have gelatinous, vascularized, and diffuse lesions with corneal involvement. Main treatment was topical fluorouracil and steroids, eye lubricants, and systemic metronomic therapy. Neither of our cases were surgically treated.

Conclusion: management of xeroderma pigmentosum is very complex, not every case qualifies for primary surgical treatment and those with ocular involvement must be evaluated by a pediatric oncology to determine if a systemic metronomic treatment might be useful.

Clinical Implication: if a patient with XP has ocular involvement the mortality rate will be higher compared to those without ocular involvement. Thus, the clinician must be very cautious when selecting the objective of the treatment to avoid iatrogenesis. in selected cases, systemic metronomic therapy can help control the ocular surface and eyelid tumors so to avoid unnecessary surgical intervention.

Financial Interest: *None*

D043

Globe Salvage in Invasive Squamous Cell Carcinoma of the Orbit: Is it Possible?

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Co-authors: Anirudha Puntambekar, Sanjay MH, Rahul Deshpande

Abstract

Introduction: Invasive squamous cell carcinoma (SCC) of orbit is an aggressive malignancy and is considered an indication for orbital exenteration. Primary SCC of orbit is considered rare while, secondary orbital involvement from an advanced conjunctival SCC is known.

Purpose: To present a case of invasive orbital SCC likely originating form superior forniceal conjunctiva managed with multimodal treatment including neoadjuvant chemotherapy, wide local excision and radiotherapy resulting in globe salvage.

Case Report: A 68 years old lady was referred for recent onset ptosis of her left eyelid. On detailed clinical evaluation, she was found to have a firm irregular mass lesion occupying left superior-lateral orbital quadrant with partial restriction in elevation and abduction. CT scan revealed 4.5x3.5x1cm fairly defined mass in the extraconal compartment of left superior-lateral orbital quadrant extending from anterior to mid-orbit. LPS-SR complex as well as lacrimal gland was not separable from the mass. Histopathology from incision biopsy showed presence of moderately differentiated squamous cell carcinoma. Whole body PET CT scan ruled out regional or distant metastasis. Neoadjuvant chemotherapy with paclitaxel and carboplatin was administered to consolidate the lesion prior to surgical excision Histopathology confirmed diagnosis of invasive SCC with evidence of lymphovascular invasion. Adjuvant intensity modulated radiotherapy (60Gy) with rapid arc technology was then administered to the orbit. At a 3-years of follow up, she is doing well with no loco-regional recurrence or distant metastasis.

Conclusion/Clinical Implications: Management of orbital invasive SCC is challenging. Multimodal treatment with chemotherapy, surgery and radiotherapy can be viable alternative to orbital exenteration.

Financial Interest: *None*

D044

Radiation Therapy in Ocular Surface Squamous Cell Carcinoma

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Co-authors: Debasish Sadangi

Abstract

Introduction: Definitive radiation therapy can be an effective treatment option for conjunctival squamous cell carcinoma (SCC), particularly for lesions that are not amenable to surgical excision or for patients who are not suitable surgical candidates.

Case Report: We describe the case of a young male with a large orbital mass of size 5X5cm probably arising out of the ocular surface covering whole globe The patient underwent conformal beam radiotherapy, specifically external beam radiotherapy (EBRT), targeting the left orbit. The treatment involved the use of 6 mV photon beams, with a total dose of 30 Gy administered over a span of two weeks, consisting of ten fractions. EBRT was divided into 3 Gy/fraction as phase 1 and boost dose of 10 Gy in 5 fractions. The patient responded well to radiotherapy with adequate reduction in tumor mass, globe salvage as well as vision salvage. The patient has been consistently attending follow-up appointments and there has been no recurrence of the condition for the past 2 years.

Conclusion: Radiation therapy is a safer alternative for selected cases of squamous cell carcinoma with very few side effects and can save the eye from enucleation or exenteration.

Clinical Implication: The advantage of radiotherapy as highlighted in our case could potentially offer a less intrusive treatment option for selective cases of conjunctival squamous cell carcinoma.

Financial Interest: *None*

D045

A Case of Conjunctival Melanoma Simulating Iris Prolapse

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Co-authors: Dariga Abdrakhimova, Idris Rassulov

Abstract

Introduction: Conjunctival melanoma is a rare but sight and life threatening malignancy with an associated mortality of up to 30% (Kastelan S., 2016). Bulbar melanoma can grow rapidly and spread on the cornea.

Case Report: Patient K., 70 years old, 2 months ago, redness appeared in the right eye. Three days later a formation appeared and has been growing. At the age of 12, there was a trauma to the right eye (kicked by a horse), after that, the patient noticed the opacity of the eye. Visual acuity of the right eye was the hand movement, left eye was 0.9. Objectively: Right eye is injected. in the inner quadrant of the cornea there is a large prominent brown tumor on a stalk. in the upper half of the cornea there is a flat pink formation with infiltrative growth. Deeper structures and fundus was not visible. Ultrasound examination of the right eye shows a round hypoechogenic homogeneous tumor on the cornea with own vessels, the prominence 8.27 mm, diameter 7.92 mm. Considering the rapid growth of the tumor, its penetration into the cornea, and the presence of its own vessels, enucleation of the right eye was performed. Histologically confirmed conjunctival melanoma. The patient was referred to general oncologist for further treatment.

Conclusion: The history of trauma of the right eye and corneal opacity did not exclude the possibility of corneal perforation in the area of opacity with iris prolapse. However, oncological concerns, the possibility of malignant tumor development, and the large size of the tumor indicated to enucleation, after which malignant melanoma was confirmed.

Clinical Implication: Elderly age, large tumor size, rapid growth and the presence of its own blood flow in the tumor should raise suspicion of a malignant process and require a radical surgery.

Financial Interest: *None*

D046

An Unusual Presentation of Multifocal Conjunctival Melanoma Arising from a Nevus

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Co-authors: Sima Das

Abstract

Introduction/Purpose: To describe an unusual case of a recurring multifocal conjunctival nevi that eventually transformed into a melanoma.

Case report: A 22-year-old male presented with history of recurrent brownish masses in right eye since childhood. They had been excised twice previously elsewhere and labelled as a junctional nevus on histopathology. On examination, multilobulated fleshy blackish - brown masses were seen in the temporal bulbar (local recurrence) and nasal palpebral conjunctiva (new site). Ultrasound bio-microscopy revealed scleral involvement. Excision biopsy was performed for both sites and histopathology was suggestive of an atypical melanocytic lesion.

Conclusion: Conjunctival melanoma is an uncommon and potentially devastating periocular malignancy. Local recurrence, multicentric involvement, development of a new lesion on non-bulbar conjunctival location (particularly medial) after prior surgical treatment was alarming in this case as all these are risk factors for regional spread and metastasis. Multifocal conjunctival melanoma without primary acquired melanosis (PAM) is rare and also associated with increased risk of mortality. On discussion with the tumor board, a decision for adjuvant plaque brachytherapy was made. Six months follow up showed no recurrence or radiation complications with additional cosmetic outcome.

Clinical Implication: This case highlights the infiltrative, aggressive and notorious behavior of a less commonly seen multicentric conjunctival melanoma arising from a nevus, without any PAM. Wide conservative surgeries followed by customized brachytherapy have appeared as viable strategies for such lesions. This multidisciplinary ophthalmic malignancy requires close co-ordination between ocular oncologist, pathologist, physicist and radiotherapist.

Financial Interest: *None*

D047

Elevated Melanotic Ocular Surface Lesion: A Reverse Masquerade Condition

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Abstract

Introduction/Purpose: Ocular surface melanotic lesions are a frequent cause of concern among both physicians and patients. We present a case of elevated melanotic lesion of the corneoscleral limbus of recent appearance, which was referred for management as a melanotic tumor but proved to be non-neoplastic.

Case Report: A 20-year old male patient was referred to our center for management of an elevated melanotic tumor of the corneoscleral limbus (OS), of recent origin. Visual acuity and intraocular pressure measurements were normal (20/20cc OU and 12mmHg, OU, respectively). Anterior segment biomicroscopy was significant for an elevated melanotic lesion at 7o'clock position of the left corneoscleral limbus, measuring 2x3mm in width and about 2mm in height. The lesion appeared rather lobulated with sharp borders, covered with epithelium and a central area of epithelial thinning. Interestingly there was no associated topical hyperemia and gonioscopy revealed topical peripheral iris synechiae without pigment dispersion. Dilated fundoscopy and a CT scan of the orbits were also non-contributory. Detailed history taking revealed that the patient had been hospitalized, including some days in intensive care unit, prior to the appearance of the lesion for head injury associated with a car crash and fractured windshield. Accordingly, it was suggested that the lesion represented an anterior staphyloma related to subclinical ocular perforation in association with that injury and this hypothesis was confirmed by the fact that the lesion remained stable during a 6-month follow-up.

Conclusion: Ocular surface elevated melanotic lesions, if stable and indolent, may represent post-traumatic staphylomas even if the initial history of the case is non-contributory.

Clinical Implication: In such cases, biopsy should be contra-indicated due to the risk of iatrogenic ocular injury.

Financial Interest: *None*

D048

A Sheep in Wolves Clothing - The Story of an Eyelid Tumor

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Co-author: Swathi Kaliki

Abstract

Purpose: To discuss a case of benign eyelid lesion, closely mimicking malignancy

Case summary: A 44-year-old gentleman came with complaints of swelling in left eye since 3 months. He has a previous history of chalazion surgery performed twice, 4 years ago. Incision biopsy of the current swelling was done elsewhere which was suggestive of inflammatory lesion. On examination, he had a left eye upper lid mass about 30x30mm with surface ulceration, crusting and bleeding, infiltrating the entire upper lid. Lower lid was also distorted and there was no view of the globe. He also had an enlarged parotid gland. Whole body PET scan showed FDG uptake in the left eyelid lesion with enhancing left intraparotid and cervical nodes. A provisional diagnosis of LE upper lid Sebacceous gland carcinoma was made and incision biopsy was planned. The lesion was very vascular and had excessive bleeding during the surgery. To our surprise, HPE showed presence of granulation tissue with inflammation, there was no evidence of atypia. A repeat deeper biopsy along with parotid gland biopsy was taken which was again suggestive of inflammation and IHC confirmed inflammatory myofibroblastic tumor. Patient was Administered IVMP 1gm/day for 3 days, and he showed significant improvement within 4-5 days . It was then followed with tapering oral steroids.

Conclusion: Although rare, aggressive nature of the tumor can be seen in benign lesions.

Clinical Implication: Incision biopsy from deeper aspect of the tumor is gold standard for diagnosis and further management.

Financial Interest: *None*

D049

Salivary Gland-like Duct Carcinoma of the Lacrimal Drainage System: An Exceedingly Rare Entity

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Co-author: Ravindra Kumar Saran, Parveen Mongre

Abstract

Introduction: Lacrimal drainage system (LDS) neoplasms are extraordinarily rare. They typically show up with non-specific symptoms such as epiphora, or the presence of a mass and are therefore mistakenly diagnosed as inflammatory and benign conditions. Due to their aggressive behavior, they usually require prompt diagnosis and urgent treatment modalities.

Case Report: We report an exceedingly rare case of salivary gland-like duct carcinoma of the LDS with less than 5 cases described in literature to date. The patient was a 77-year-old male who presented with a swelling in the medial canthus of right eye which was gradually progressive and associated with redness and epiphora. Interestingly, on immunohistochemistry, the tumor cells were positive for Androgen receptor and expressed Her-2-Neu.

Conclusion: It is of utmost importance to correctly and promptly diagnose such high-grade carcinomas. Histopathology and immunohistochemistry are invaluable for accurate diagnosis. Since AR evaluation may have significant therapeutic and diagnostic ramifications, we recommend it be included in the immunohistochemical analysis of high-grade carcinomas of the LDS that morphologically mimic salivary gland duct carcinoma.

Clinical Implication: Surgery with adjuvant radiotherapy is being used widely; however, the exact course of treatment depends on the patient's preferences, overall health, and the extent and severity of the disease. A multidisciplinary team approach should be considered for the management of this rare and aggressive tumor.

Financial Interest: *None*

D050

To Exenterate or Not: Lacrimal Sac Squamous Cell Carcinoma with Orbital Involvement

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Co-author: Sachin Salvi

Abstract

Introduction: Treatment of lacrimal sac squamous cell carcinoma (LSSCC) with orbital involvement typically involves en bloc excision of the entire lacrimal drainage system (LDS) along with medial maxillectomy, anterior ethmoidectomy and orbital exenteration, often accompanied by adjuvant radiotherapy. We present a case of LSSCC with extensive medial anterior orbital involvement successfully managed with a globe and sight-preserving approach.

Case Report: A 46-year-old female patient presented with right-sided epiphora and enlarging medial canthal mass. CT scan revealed a large mass involving the right lacrimal sac, measuring over 3cm in dimension, extending into the anterior orbit medially and inferomedially, while abutting the globe. The patient preferred a globe-preserving approach. Staging scans were normal. Patient was discussed at multidisciplinary meeting. She underwent surgery in which: A. Soft tissue en bloc excision of the lacrimal sac tumour along with the entire LDS and B. Removal of adjacent bone including anterior and mid-ethmoidectomy and medial maxillectomy and C. Reconstruction with paramedian forehead flap was performed. Histopathology confirmed poorly differentiated invasive LSSCC with evidence of perineural invasion and probable vascular invasion. Surgical margins were clear. Due to the possibility of residual microscopic disease, she underwent adjuvant radiotherapy. She has excellent cosmetic and functional results with no signs of recurrence 24 months postoperatively.

Conclusion: Our case highlights the short-term success of globe-preserving approach. A globe-preserving approach should be considered for managing LSSCC with even extensive orbital extension if there is no globe involvement.

Clinical Implication: The option of a globe-preserving approach for managing LSSCC with orbital involvement should be offered to selected patients. Careful counselling on the risk of local recurrence and a slightly increased risk of metastasis is essential.

Financial Interest: *None*

D051

Can I Be Ready for Marriage?

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Abstract

Introduction/Purpose: This case report details a unique solution for managing a dermoid cyst near the left eye in a 28-year-old male with uncontrolled type 1 diabetes, who was facing surgery constraints due to an impending marriage. The use of foam sclerotherapy with sodium tetradecyl sulfate (STS) is explored as an alternative treatment, aiming for minimal scarring and a swift recovery.

Case Report: The patient presented with a growing dermoid cyst near the left eye, complicating surgical options due to uncontrolled type 1 diabetes and an upcoming marriage. To address these challenges, the patient underwent left eye intralesional foam sclerotherapy using STS with a 2:1 ratio of aspirate to injected sclerosant. A total of 3 ml of cyst content was aspirated, followed by the meticulous injection of 1.5 ml of foam created from 2 ml of STS and 1 ml of filtered air. Cytopathology confirmed the lesion as a dermoid cyst. At the 6-month follow-up, the patient showed no residual lesion, scar, or aesthetic blemish.

Conclusion: Foam sclerotherapy, a non-surgical approach, proved to be a game-changer in managing a dermoid cyst in a patient deemed medically unfit for surgery due to uncontrolled diabetes. The technique collapsed the cyst without leaving significant scarring, offering an alternative to traditional surgical excision. The 13.4-month follow-up reaffirmed the efficacy of foam sclerotherapy in achieving a favorable outcome.

Clinical Implication: While surgical excision remains the standard treatment for dermoid cysts, foam sclerotherapy emerges as a viable option, especially for medically unfit patients. Further research is warranted to better understand the efficacy of these approaches in this specific patient population. The presented case provides valuable insights into the successful use of foam sclerotherapy, offering clinicians an additional tool for tailored treatment plans.

Financial Interest: *None*

D052

A Rare Cystic Orbit Tumor to Keep in Mind

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Abstract

Purpose: To present a rare case of Orbit tumor and the pathology studies done to obtain final diagnosis.

Case Report: A 36-year-old Hispanic male with a history of a conjunctival cystic nodule in temporal conjunctiva OS since 2018. He had biopsy elsewhere. No photo was done before surgery. After a couple of months, he started with proptosis OS. Systemic steroid was started for months. There was no improvement. He was referred to our team in 01/2021. There was a remarkable exoftalmo OS with severe periorbital edema and complete ptosis OS. Motililty was reduced almost 100% into levo -version in OS. MRI revealed cystic tumor attached to lateral rectus muscle. Capsule enhanced with contrast. During surgery the tumor was gelatinous in consistency, something that was recorded. Histopathological analysis revealed a hypocellular tumor composed of small to medium cells with elongated shapes within a myxoid stroma. The tumor was highly vascularized, with vessels arranged in an arcuate pattern, and in some areas, resembling a chicken wire disposition. The tumor infiltrates the skeletal muscle, dissecting the bundles. No atypical mitotic figures were observed. S100 was negative, making the diagnosis of a peripheral nerve tumor with myxoid changes less likely. The absence of DDIT3 gene rearrangement by FISH ruled out myxoid liposarcoma. All morphological, immunohistochemical, and molecular analyses supported a diagnosis of low-grade myxofibrosarcoma. EBRT was done. After 18 months, the patient is doing well. Vision in OS is 20/25. No Proptosis/periorbital change. Motility was recovered 100%

Conclusion: This neoplastic process represents a low grade neoplasm with a possible local slowly growing infiltration without metastatic potential. To our knowledge this is one of the few cases reported in literature.

Clinical Implication: To consider Low grade myxofibrosarcoma within the differential diagnosis of cystic tumor attached to orbit muscle

Financial Interest: *None*

D053

Residual Solitary Fibrous Tumour of the Orbit

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Co-author: Gangadhara Sundar

Abstract

Purpose: To describe the dilemma in management and prognosis of residual orbital solitary fibrous tumour (SFT).

Case Report: A 32-year old Chinese female presented with recent onset pain with a right eye prominence for 2 years. BCVA was 6/9 and 6/6 respectively. Eyes were orthophoric, with full motility and no RAPD. There was a 6mm axial non-pulsatile proptosis with epibulbar dilated vessels and resistance to retropulsion. Orbital MRI showed a large intraconal well-defined pear-shaped enhancing retrobulbar mass extending from orbital apex to the globe. She underwent lateral orbitotomy and osteotomy with attempted total excision. As the posterior aspect was tightly adherent to the apex, subtotal resection was performed with intent of preserving vision and apical orbital structures. Recovery was good with preservation of vision and ocular motility. Histopathology showed a spindle cell lesion separated by collagen bundles, without evidence of malignancy(no mitotic activity, low Ki-67). Immunochemistry was positive for STAT6, negative for S100 and desmin, confirming diagnosis of SFT. Postoperative imaging revealed a residual orbital apex lesion for which she was offered repeat surgery or radiation which she declined. Follow up over 5-yrs showed stable findings clinically and radiologically.

Conclusion: SFT should be considered in a gradually enlarging painless benign orbital tumour and ideally be completely excised. While desirable, en-bloc resection of tumours adherent to the orbital apex is difficult without visual consequences. Incomplete excision of SFTs may lead to recurrent growth with possible malignant transformation and metastasis. However, based on Demicco's risk assessment model, in our patient it appeared to be a low-risk tumour and Grade 1 (WHO 2016 CNS tumour classification).

Clincial Implication: It is essential to counsel patients pre and postoperatively on residual lesions if present, its prognosis, need for long term clinical and radiological follow up and threshold for additional interventions.

Financial Interest: *None*

D054

A Solitary Affair

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Abstract

Introduction/Purpose: To report a case of recurrent orbital solitary fibrous tumour presenting as a subconjunctival mass treated with orbitotomy and adjuvant radiotherapy.

Case Report: A 25-year-old male presented with a subconjunctival mass, ptosis and proptosis in the left eye. The mass was yellow-pink and located in the subtenon space. A computed tomography revealed a larger orbital component. A transconjunctival excision biopsy was performed. Histopathology showed stellate-spindle shaped cells in a myxoid matrix with CD34, vimentin, CD99 and BCL2 positivity on immunohistochemistry. A diagnosis of myxoid solitary fibrous tumour was made. The lesion recurred after 5 months and after a repeat surgical debulking with sparing of the lateral rectus, lacrimal gland and optic nerve, he was treated with adjuvant external beam radiotherapy.

Conclusion: The case reports a rare presentation of an orbital solitary fibrous tumour as an ocular surface mass.

Clinical Implication: Recurrent SFTs are more aggressive with local infiltration. Meticulous debulking with adjuvant radiotherapy achieves local control with good functional outcome.

Financial Interest: *None*

D055

GLI-1 Amplified Spindle Cell Tumor of the Intraconal Orbit: Novel Surgical Management

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Abstract

Purpose: To present combined open transorbital and endoscopic, endonasal approach for resection of posterior intraconal orbital tumors (GLI-1 amplified spindle cell tumor).

Case Report: A 54-year-old woman presented with proptosis, eye pain, and ocular motility restriction in the left eye. MR imaging demonstrated a tumor occupying the superomedial intraconal orbit that was distinct from the extraocular muscles, optic nerve, and globe. She had an open biopsy done at an outside hospital which demonstrated a GLI-1 alerted spindle cell tumor with unclear malignant potential and was offered left orbital exenteration. She presented at our institutes for a second opinion as this her only functional eye. The patient underwent a combined endonasal and open, transorbital approach for gross total tumor resection. Histopathological analysis demonstrated a spindle and epithelioid cell mesenchymal tumor with diffuse nuclear GLI1 expression. PCR-based, next generation sarcoma fusion panel was negative for GLI-1 fusions including GLI-1:: ACTB fusions. Post-operatively the patient had progressive vision improvement and she tolerated adjuvant radiation therapy (60Gy in 30 fractions) without complications.

Conclusion: Combined endoscopic endonasal and open trans-orbital approach is safe, effective approach to achieve gross total resection in tumors with low malignant potential.

Clinical Implication: A rare case of GLI-1 amplified spindle cell epithelioid neoplasm of the intraconal orbit managed with gross total surgical resection is presented. Given the molecular features, adjuvant radiation therapy and close surveillance for local, regional, and distal disease is advised.

Financial Interest: *None*

D056

En-Bloc Optic Nerve Resection/Biopsy

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Co-author: Arun D. Singh

Abstract

Purpose: To report novel surgical technique for en bloc optic nerve resection/biopsy based on medial transconjunctival approach facilitated by globe rotation.

Case Report: The surgical procedure involved the medial transconjunctival approach to the optic nerve (ON) with disinsertion of the medial rectus. Lateral globe rotation was achieved through medial rectus disinsertion, enabling direct ON visualization. Double-fold 2-0 silk traction suture was passed around the ON using a muscle hook. With traction applied at the ON insertion, the globe was further rotated laterally bringing the full thickness orbital ON into the surgical view. The distal portion of the ON was transected about 9 mm from the sclera by monopolar cautery (blended-cutting mode). The cut end of ON segment was grasped and lifted out of the orbit, further rotating the globe, and exposing its scleral attachment. The proximal ON remnant was then resected en bloc from scleral attachment. The medial rectus was reattached, and the conjunctiva closed with absorbable sutures.

Conclusion: The described en bloc optic nerve resection/biopsy technique via a medial transconjunctival approach with globe rotation provides improved access, exposure, and direct visualization of the ON compared to existing methods. Globe rotation achieved in three steps: first, medial rectus disinsertion allowing, second, lateral globe rotation which permits direct traction of the ON leading to the possibility of additional lateral rotation. The third and final rotation includes cutting the distal end of the ON facilitating extreme rotation of the globe. This final step presents the optic nerve with its proximal scleral attachment into the surgical field.

Clinical Implication: Enhanced view, exposure, and direct access to the full thickness retrobulbar ON via medial transconjunctival approach facilitated by globe rotation described herein, offers novel technique for en bloc optic nerve resection/biopsy.

Financial Interest: *None*

D057

Navigating the Ocular Odyssey: A Tale of Glioma, Resilience, and Specialized Care

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Abstract

Introduction: Optic nerve glioma, globally affecting 1.5 cases per million, predominantly occurs in children (3-5% of pediatric intracranial tumors). Early diagnosis challenges highlight the importance of therapeutic strategies.

Case Presentation: A 23-year-old with a decade-long history sought medical attention after a fall, revealing optic nerve glioma on MRI. Despite enucleation and chemo recommendations, patient explored alternative therapie from 2015 to 2021 with no improvement, resulting in visual loss. This year, patient consulted us in Eye Cancer Institute in Monterrey, Mexico. Clinical examination revealed proptosis, lagophthalmos, restricted ocular motility, and corneal opacity in right eye, hindering the assessment of anterior segment and fundus. Immediate MRI and a multidisciplinary approach were initiated due to corneal exposure, absent vision, and complications. Subsequently, enucleation with neurosurgical approach was performed, encompassing intracranial portion and placing an orbital implant. After diagnosing a right optic nerve glioma, patient underwent chemo and radiotherapy. Further investigation excluded neurofibromatosis type 1, categorizing it as sporadic. Long-term follow-up includes visual fields and MRI scans, ensuring monitoring for recurrence and contralateral involvement providing comprehensive ongoing care.

Conclusion: The patient received an accurate diagnosis 11 years ago; however, fear and uncertainty led them to explore alternative treatments. Through establishing a support network and a comprehensive approach, it was possible to provide a quality treatment.

Clinical Implication: Management of optic nerve glioma necessitates a precise treatment approach tailored to its complexity. Equally crucial is building patient confidence, recognizing the impact of uncertainty on treatment decisions. Strong communication and detailed information foster trust, enabling effective personalized care and emotional support throughout the journey.

Financial Interest: *None*

D058

Bilateral Angiolymphoid Hyperplasia with Eosinophilia in an Elderly Male

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Co-author: Namita Kumari, Sima Das, Chhavi Gupta

Abstract

Purpose: Angiolymphoid hyperplasia with eosinophilia (AHLE) is a rare proliferative disorder affecting the head and neck, more commonly diagnosed in females. We describe bilateral presentation of AHLE in an adult male.

Case Report: A 70-year-old asthmatic male presented with progressive diminution of vision and painful swelling of both eyes for past 1 month. Examination showed bilateral proptosis with periorbital edema, conjunctival chemosis and lagophthalmos. Ocular motility restriction was noted in all gazes and firm palpable masses were present in superotemporal side of both orbits. Exposure keratopathy with corneal ulceration were noted in the left eye. Corneal scraping examination showed a diagnosis of fungal keratitis. Orbital imaging revealed well-circumscribed homogenous mass, isodense to muscle in lacrimal gland region on both sides, along with lateral rectus thickening. Lacrimal gland biopsy showed mixed inflammatory cells, reactive lymphoid aggregates with blood vessels and fibrosis. On systemic evaluation, absolute eosinophilia and hilar lymphadenopathy were present but the autoimmune profile was normal. The patient received pulse intravenous steroid therapy, topical and systemic anti-fungals, with supportive treatment, leading to improvement in symptoms and improvement in ocular motility. At the last follow-up of 6 months, the patient has complete resolution of proptosis, has full and free ocular motility in all gazes and a healed corneal scar in the left eye.

Conclusion: AHLE shares common features with Kimura disease and may be difficult to distinguish from it. We present a rare case of bilateral AHLE with asthma, lymphadenopathy and peripheral eosinophilia.

Clinical Implication: Clinicopathological correlation and detailed systemic evaluation are must in rare presentations of uncommon disorders. Timely diagnosis and management with pulse steroid therapy, even in presence of fungal keratitis led to satisfactory outcomes

Financial Interest: *None*

D059

An Unusual Case of Orbital Metastasis from Anorectal Malignant Melanoma

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Co-author: Lata Singh, Seema Sen, Rachna Meel, Seema Kashyap

Abstract

Introduction: Uveal melanoma (UM) is a highly malignant intraocular tumor in adults with poor prognosis. Colorectal cancer is the third most common cancer and the second most leading cause of mortality due to distant metastasis. This is an unusual case of anorectal malignant melanoma (ARMM) presented with proptosis in left eye.

Case Report: A 50-year-old Asian female presented with abaxial proptosis of the left eyeball, underwent anterior orbitotomy. A densely pigmented mass was evident intraoperatively, and histopathological examination revealed malignant melanoma, spindle cell type with areas of necrosis and haemorrhage. Immunohistochemical staining was strongly positive for HMB-45 and S-100. PET-CT revealed heterogeneously enhanced lobulated soft tissue mass measuring 3.1 x 3.8 x 5.3 cm in the lumen of distal rectum extending across the anorectal junction into the lumen of proximal-mid anal canal suggesting a primary anorectal mass. Similar metastatic lesions were found in the hepatic lobes, lungs, lymph nodes along with lytic lesions in D5 vertebrae and right femoral head. Patient underwent colonoscopy whereupon a large friable proliferative growth arising from anal verge with a wide base, nodular surface and black pigmentation was noted and a biopsy taken. Histopathological examination of the anal mass confirmed the diagnosis of ARMM. The patient was duly counselled and referred to the oncology department for palliative chemo-radiotherapy. However, the patient died 15 months after the diagnosis.

Conclusion: We have presented an extremely rare case of orbital metastasis from ARMM, which to our knowledge is rare in clinical practice and has not been documented earlier.

Clinical Implication: This case report highlights the rare occurrence of malignant melanoma. Clinicians should be aware of the possibility of ocular melanomas manifesting in atypical ways, prompting thorough ophthalmic examinations and PET-CT evaluation for early detection.

Financial Interest: *None*

D060

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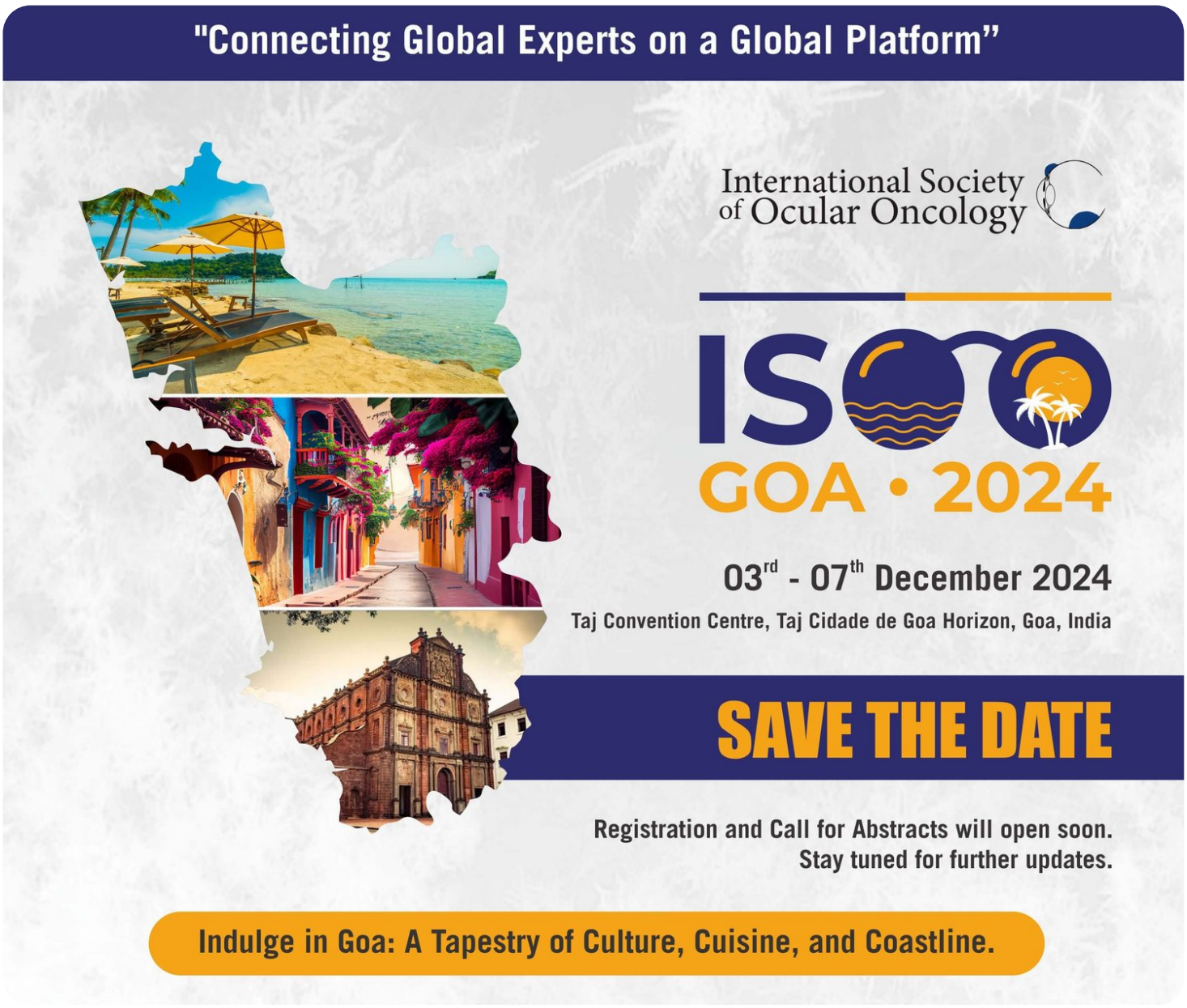

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