Fabiana Mallone

ESPERIENZA LAVORATIVA

ATTUALE

Specialista in Oftalmologia

20

Medico frequentatore - Servizio Speciale di Cornea e Superficie Oculare

Azienda ospedaliera Universitaria Policlinico Umberto I, Roma

19

Tirocinio di alta specializzazione presso il Dipartimento di Chirurgia Vitreoretinica del Royal Eye Hospital di Manchester, UK. Supervisori Prof.N.Patton e F.D.Scala.

15 - 19

Specializzanda in Oftalmologia

Universitaria 'Sapienza', Roma

18

Missione Umanitaria in Togo, Africa

15

Iscrizione all'Albo dei Medici-Chirurghi dell'ordine Provinciale di Roma dal 19/03/2015 con numero: 61490

12-13

Borsa di Collaborazione (150 ore lavorative), Dipartimento di Igiene e Sanità Pubblica, Università 'Sapienza' di Roma

ISTRUZIONE E FORMAZIONE

20

ESASO eCampus - Masterclass 1 Pearls on Medical Retina, Masterclass 2 Pearls on Anterior Segment & Glaucoma, Masterclass 3 Live Surgery

20

Speaker, Alcon Vitreous Club – Alcon vitreoretinal surgery course, Milano.

19

Tirocinio di alta specializzazione presso il Dipartimento di Chirurgia Vitreoretinica del Royal Eye Hospital di Manchester, UK. Supervisori Prof.N.Patton e F.D.Scala.

19

Partecipazione a V Meeting Scientifico Annuale BEECS (British Emergency Eye Care Society). Centre for Life, Newcastle upon Tyne

15 – 19

Diploma di Specializzazione in Oftalmologia conseguito in data 20/11/2019. Tesi sperimentale:'Le neurotrofine nelle membrane epiretiniche: ERM idiopatiche Vs ERM secondarie', Supervisori Prof.M.Gharbiya and Prof. A.Lambiase. Votazione 70/70 con lode.

Università 'Sapienza', Roma

19

Speaker, Congresso: 'Case Reports 2019: La gestione dei casi complessi 2019', responsabile Prof. T. Rossi. Fondazione Santa Lucia, Roma

19

SOU - Corso teorico e pratico di Eco Oftalmologia ed OCT. Organizzatori: Prof.N.Delle Noci, Prof.T.Avitabile. Foggia/Chieti/Roma

18

Corso di chirurgia oculare 'SarEye Itinerario nella professione. L'Ora della Chirurgia' (2018)', Roma.

18

Vitreoretinal Surgery Esaso Simulator Course. Lugano, Switzerland.

18

Vitreoretinal Surgery Esaso Module. Lugano, Switzerland.

18

XIV Corso SOU, Chieti 2018

15

American Heart Association Basic Life Support - Defibrillation (BLSD). Certificazione di competenza in rianimazione cardiopolmonare.

14

Esame di Stato per l'abilitazione alla Professione di Medico Chirurgo (DM 270), Sessione di Novembre 2014

Azienda Ospedaliera Universitaria Policlinico Umberto I 'Sapienza', Roma

11 - 14

Percorso di Eccellenza, supervisore Prof.ssa M.Gharbiya. Partecipazione a 120 ore di lezioni frontali e 480 ore suddivise tra internato elettivo e il progetto di ricerca 'Gli anticorpi monoclonali in ambito oftalmologico: nuovo approccio terapeutico'

Università 'Sapienza', Roma

08 - 14

Laurea in Medicina e Chirurgia conseguita il 24/07/2014.Tesi sperimentale 'L'assottigliamento dello spessore coroideale e dello strato delle fibre nervose retiniche nella malattia di Alzheimer'.Supervisore Prof. M.Gharbiya.Votazione 110/110 con lode.

Università 'Sapienza' di Roma

12

Basic Life Support (BLS) Course Rescue Doctor Diploma e Volontario

Croce Rossa Italiana

08

‘Bocconi Talent Scout Program’

Università Bocconi, Milano

07 - 08

'Awarding of Excellence’

Liceo Scientifico Innocenzo XII, Anzio (Rm)

03 - 08

Diploma di maturità scientifica. Votazione 100/100.

Liceo Scientifico Innocenzo XII, Anzio (Rm)

08

First Certificate in English University of Cambridge (Council of Europe level B2)

07

Trinity College London Exam, Grade 10

07

European Computer Driving Licence (ECDL)

06

University College of Dublin (UCD) – Upper English Course.

ATTIVITA’ DI RICERCA

18

Principal Investigator del progetto di ricerca: ’Neurotrophins in Idiopatic Epiretinal (iERM) Membranes at the time of Membrane Peeling: molecular characterization of the two types of iERMs’. Numero protocollo: AR11816436687015

Tutor Prof. Alessandro Lambiase, ‘Sapienza’ University Umberto I Hospital, Rome

19

Co-Investigator del progetto: ‘Long-term outcomes of intravitreal injections for choroidal neovascularization in patients with pathologic myopia: analysis of prognostic factors.’

Tutor Prof. Alessandro Lambiase, ‘Sapienza’ University Umberto I Hospital, Rome

18

Co-Investigator del progetto: ‘Neurofibromatosis Type I: morpho- functional evaluation at chorioretinal level’.

Tutor Prof. Alessandro Lambiase, 'Sapienza' University Umberto I Hospital, Rome

17

Co-Investigator del progetto: 'Studio prospettico, multicentrico, sul ruolo delle neurotrofine come biomarkers di progressione e di risposta alla terapia in pazienti con degenerazione maculare senile e glaucoma.'Cod.20152EKS4Y.

Tutor Prof. Alessandro Lambiase, 'Sapienza' University Umberto I Hospital, Rome

20

Co-investigator del progetto: 'Caratterizzazione del pathway dell'IL-8 d e del suo recettore CXCR1 nella superficie oculare di pazienti affetti da glaucoma sottoposti a chirurgia filtrante: possibile biomarker predittivo dell'outcome chirurgico'.

Tutor Prof. Alessandro Lambiase, 'Sapienza' University Umberto I Hospital, Rome

18-20

Reviewer per l'International Journal Ophthalmology (IJO)

PUBBLICAZIONI

Mallone F, Sacchetti M, Lambiase A, Moramarco A. Molecular Insights and Emerging Strategies for Treatment of Metastatic Uveal Melanoma. Cancers (Basel). 2020 Sep 25;12(10):2761. doi: 10.3390/ cancers12102761. PMID: 32992823; PMCID: PMC7600598.

2020

Abstract

Uveal melanoma (UM) is the most common intraocular cancer. In recent decades, major advances have been achieved in the diagnosis and prognosis of UM allowing for tailored treatments. However, nearly 50% of patients still develop metastatic disease with survival rates of less than 1 year. There is currently no standard of adjuvant and metastatic treatment in UM, and available therapies are ineffective resulting from cutaneous melanoma protocols. Advances and novel treatment options including liver-directed therapies, immunotherapy, and targeted-therapy have been investigated in UM-dedicated clinical trials on single compounds or combinational therapies, with promising results. Therapies aimed at prolonging or targeting metastatic tumor dormancy provided encouraging results in other cancers, and need to be explored in UM. In this review, the latest progress in the diagnosis, prognosis, and treatment of UM in adjuvant and metastatic settings are discussed. In addition, novel insights into tumor genetics, biology and immunology, and the mechanisms underlying metastatic dormancy are discussed. As evident from the numerous studies discussed in this review, the increasing knowledge of this disease and the promising results from testing of novel individualized therapies could offer future perspectives for translating in clinical use.

Keywords: adjuvant therapy; combined therapy; immunotherapy; liver-directed- therapies; metastatic dormancy; metastatic therapy; metastatic uveal melanoma (mUM); prognostication; targeted-therapy; uveal melanoma (UM).

Mallone F, Lucchino L, Franzone F, Marenco M, Carlesimo SC, Moramarco A. High-dose vitamin B supplementation for persistent visual deficit in multiple sclerosis: a pilot study. Drug Discov Ther. 2020;14(3):122-128. doi: 10.5582/ddt.2020.03031. PMID: 32669520.

2020

Abstract

The aim of this study is to investigate the potential neuroprotective effect of high- doses vitamins B1, B6 and B12 in patients with relapsing-remitting multiple sclerosis (RRMS) and persistent visual

loss after acute optic neuritis (AON). Sixteen patients (20 eyes) diagnosed with RRMS and visual permanent disability following AON were enrolled for the present open, pilot study. Each patient was treated with oral high-doses 300 mg of vitamin B1, 450 mg of vitamin B6 and 1,500 mcg of vitamin B12, as add-on treatment to concomitant disease-modifying therapies (DMTs) for consecutive 90 days. Outcome measures were to determine changes from baseline to month three in visual acuity (VA) and visual field (VF) testing, with correlations with clinical parameters. Logistical regression was performed to evaluate predictors of final VA. A statistically significant improvement was registered in visual acuity ($p = 0.002$) and foveal sensitivity threshold (FT) ($p = 0.006$) at follow-up compared to baseline. A similar trend was demonstrated for mean deviation (MD) ($p < 0.0001$), and pattern standard deviation (PSD) ($p < 0.0001$). Age at the time of inclusion was positively correlated with latency time (rho = 0.47, $p = 0.03$), while showing a negative correlation with visual acuity (rho= - 0.45, $p = 0.04$) and foveal sensitivity threshold (rho = - 0.6, $p = 0.005$) at follow up. A statistically significant correlation was demonstrated between foveal sensitivity threshold and visual acuity at baseline (rho = 0.79, $p < 0.0001$). In a linear regression model, the main predictor of visual acuity at follow up was the foveal sensitivity threshold ($B = 1.39$; $p < 0.0001$). Supplemental high-dose vitamins B1, B6 and B12 resulted as effective therapy to improve visual function parameters in MS-related visual persistent disability.

Keywords: B Vitamins group; neuroprotection; visual function.

Mallone F, Marcelli M, Monsellato R, Franzone F, Gharbiya M, Lambiase A. Self-sealing posterior scleral perforation in airgun ocular trauma, surgical tip: a case report. BMC Ophthalmol. 2020 Apr 22;20(1):164. doi: 10.1186/s12886-020-01435-8. PMID: 32321467; PMCID: PMC7178978.

2020

Abstract

Background: Intraorbital metallic foreign bodies have varied clinical presentations. Here, we report the unusual case of intraoperative evidence of spontaneously healed posterior scleral perforation in a severe ballistic trauma without previous instrumental signs of penetrating wound and complete visual restoration after surgery.

Case presentation: The patient was hit by several lead hunting pellets in the chest, abdomen, limbs, face and orbit. Computed Tomography (CT) images revealed the presence of a pellet within the orbitary cavity, close to the optic nerve, with no signs of penetrating ocular wound. While performing vitrectomy for severe vitreous hemorrhage, a point of strong adherence between a old hemorrhage and retinal surface was identified and managed conservatively, as it was attributed to trauma related-impact area. So, lead foreign body took an unusual trajectory impacting the globe and finally lodging back in the deep orbitary cavity, in absence of significant ocular injury and with visual prognosis preservation.

Conclusions: Our findings provide further information on orbital injuries from airguns, a theme of growing popularity and concern. Intraoperative recognition of hardly removable old hemorrhagic clot as self-blockage site of posterior scleral penetrating trauma, allowed for surgical stabilization and minimal solicitation of the area to avoid inadvertent perforation.

Keywords: Airgun; Case report; Ocular trauma; Pellet; Vitreous hemorrhage.

Moramarco A, Mallone F, Pirraglia MP, Bruscolini A, Giustolisi R, La Cava M, Lambiase A. Clinical Features of Ocular Syphilis: a Retrospective Clinical Study in an Italian Referral Centre. Semin Ophthalmol. 2020 Jan 2;35(1):50-55. doi: 10.1080/08820538.2020.1723651. Epub 2020 Feb 8. PMID: 32036734.

2020

Abstract

Purpose: To describe the clinical characteristics and visual prognosis of ocular involvement in syphilis. Design: A retrospective cohort study. Methods: We studied the charts of 24 patients who visited our Ophthalmological Centre in Rome, Italy. All patients with serological evidence of syphilitic infection were included. Results: Ocular involvement was the first manifestation of syphilitic disease in 96% and Human Immunodeficiency Virus (HIV) seropositivity was found in 29% of the cases. The most frequent ocular manifestation was posterior uveitis. Vitreous involvement was frequent. Patients with papillitis at onset showed better visual outcome with antisyphilitic treatment. Posterior uveitis at onset and HIV seropositivity were negative prognostic factors for visual outcome. HIV-positive patients showed more severe and frequent bilateral course of ocular involvement in syphilis. Conclusions: The ophthalmologist should suspect syphilis in patien ts with uveitis or optic neuropathy associated with high-risk sexual behaviour and/or HIV, or in patients with

posterior placoid chorioretinitis, necrotising retinitis, or interstitial keratitis.

Keywords: HIV; infectious uveitis; posterior uveitis; syphilis.

Moramarco A, Himmelblau E, Miraglia E, Mallone F, Roberti V, Franzone F, Iacovino C, Giustini S, Lambiase A. Ocular manifestations in Gorlin- Goltz syndrome. Orphanet J Rare Dis. 2019 Sep 18;14(1):218. doi: 10.1186/s13023-019-1190-6. PMID: 31533758; PMCID: PMC6749644.

2019

Abstract

Background: Gorlin-Goltz syndrome, also known as nevoid basal cell carcinoma syndrome, is a rare genetic disorder that is transmitted in an autosomal dominant manner with complete penetrance and variable expressivity. It is caused in 85% of the cases with a known etiology by pathogenic variants in the PTCH1 gene, and is characterized by a wide range of developmental abnormalities and a predisposition to multiple neoplasms. The manifestations are multiple and systemic and consist of basal cell carcinomas in various regions, odontogenic keratocistic tumors and skeletal anomalies, to name the most frequent. Despite the scarce medical literature on the topic, ocular involvement in this syndrome is frequent and at the level of various ocular structures. Our study focuses on the visual apparatus and its annexes in subjects with this syndrome, in order to better understand how this syndrome affects the ocular system, and to evaluate with greater accuracy and precision the nature of these manifestations in this group of patients.

Results: Our study confirms the presence of the commonly cited ocular findings in the general literature regarding the syndrome [hypertelorism (45.5%), congenital cataract (18%), nystagmus (9%), colobomas (9%)] and highlights strabismus (63% of the patients), epiretinal membranes (36%) and myelinated optic nerve fiber layers (36%) as the most frequent ophthalmological findings in this group of patients.

Conclusions: The presence of characteristic and frequent ocular signs in the Gorlin- Goltz syndrome could help with the diagnostic process in subjects suspected of having the syndrome who do not yet have a diagnosis. The ophthalmologist has a role as part of a multidisciplinary team in managing these patients. The ophthalmological follow-up that these patients require, can allow, if necessary, a timely therapy that could improve the visual prognosis of such patients.

Keywords: Gorlin syndrome; Gorlin-Goltz syndrome; Myelinated optical nerve fiber layers; Nevoid basal cell carcinoma syndrome; Ocular anomalies; Odontogenic keratocyst.

Bruscolini A, La Cava M, Mallone F, Marcelli M, Ralli M, Sagnelli P, Greco A, Lambiase A. Controversies in the management of neuromyelitis optica spectrum disorder. Expert Rev Neurother. 2019 Nov;19(11): 1127-1133. doi: 10.1080/14737175.2019.1648210. Epub 2019 Aug 12. PMID: 31339052.

2019

Abstract

Introduction: Neuromyelitis optica spectrum disorders (NMOSD) are autoimmune diseases of the central nervous system mainly involving the optic nerves and spinal cord. Many advances have been made in understanding the immunopathology of NMOSD and related clinical classification, nevertheless, open issues in management and effective therapeutic approaches still remain. Areas covered: In this article, the authors reviewed and discussed the scientific evidence in pathogenesis and pharmacological therapy of NMOSD addressing the more recent advances in new biological treatment option and therapeutic strategy that may help to improve management of this condition. Expert opinion: Despite current immunopathogenic evidence, NMOSD management represents a challenge due to the poor-validated diagnostic, prognostic and therapeutic biomarkers. A tailored approach is mandatory to improve the management of the different disease clinical settings.

Keywords: Neuromyelitis optica spectrum disorders (NMOSD); aquaporin-4 immunoglobulin G (AQP4-IgG); diagnostic criteria; emerging treatment; immunopathogenesis; management; myelin oligodendrocyte glycoprotein immunoglobulin G (MOG-IgG); standard treatment.

Moramarco A, Miraglia E, Mallone F, Roberti V, Iacovino C, Bruscolini A, Giustolisi R, Giustini S. Retinal microvascular abnormalities in neurofibromatosis type 1. Br J Ophthalmol. 2019 Nov;103(11):

1590-1594. doi: 10.1136/bjophthalmol-2018-313002. Epub 2019 Jan 31. PMID: 30705042.

2019

Abstract

Purpose: The aim of this study was to provide a classification of the different retinal vascular arrangements in neurofibromatosis 1 (NF1), with appropriate qualitative and quantitative information.

Methods: This study was conducted on 334 consecutive patients with NF1 and 106 sex-matched and age-matched healthy control subjects. Each patient underwent a comprehensive ophthalmological examination inclusive of near-infrared reflectance retinography by using the spectral domain Optical coherence tomography (OCT), a complete dermatological examination and 1.5T MRI scan of the brain to assess the presence of optic nerve gliomas. To evaluate the predictability and the diagnostic accuracy of our identified retinal microvascular arrangements, we calculated the diagnostic indicators for each pattern of pathology, with corresponding 95% CI. In addition, we evaluated the association between the microvascular arrangements and each National Institutes of Health diagnostic criteria.

Results: Microvascular abnormalities were detected in 105 of 334 NF1 patients (31.4%), the simple vascular tortuosity was recognised in 78 of 105 cases (74.3%) and whether the corkscrew pattern and the moyamoya-like type showed a frequency of 42.8% (45 of 105 cases) and 15.2% (16 of 105 cases), respectively. We found a statistically significant correlation between the presence of retinal microvascular abnormalities and the patient age (p=0.02) and between the simple vascular tortuosity, the patient age and the presence of neurofibromas (p=0.002 and p=0.05, respectively).

Conclusions: We identified microvascular alterations in 31.4% of patients and a statistically significant association with patient age. Moreover, the most frequent type of microvascular alterations, the simple vascular tortuosity, resulted positively associated with age and with the presence of neurofibromas.

Keywords: NF1; NIR-OCT; microvascular abnormalities; moya-moya disease; ophthalmoscopy; retina.

Moramarco A, Giustini S, Nofroni I, Mallone F, Miraglia E, Iacovino C, Calvieri S, Lambiase A. Near-infrared imaging: an in vivo, non-invasive diagnostic tool in neurofibromatosis type 1. Graefes Arch Clin Exp Ophthalmol. 2018 Feb;256(2):307-311. doi: 10.1007/s00417-017-3870-z. Epub 2017 Dec 30. PMID: 29290016.

2018

Abstract

Purpose: Only a few reports in the literature have investigated the presence of ocular abnormalities in neurofibromatosis type 1 (NF-1) patients. The aim of this study was to evaluate the prevalence of ocular abnormalities in a large population of NF1 patients, focusing on the choroidal changes.

Methods: This study was conducted on 160 consecutive patients with NF1 and 106 sex- and age-matched healthy subjects (control). Each patient underwent a complete ophthalmological examination inclusive of best-corrected visual acuity, intraocular pressure measurement, slit-lamp biomicroscopy, indirect fundus biomicroscopy, and near-infrared reflectance (NIR) retinography by using the spectral domain OCT. Moreover, all patients underwent complete dermatological exam and 1.5-Tesla MRI scan of the brain to assess the presence of optic nerve gliomas. Results: Choroidal abnormalities were detected in 97% of patients, with a positive predictive value of 100% and a negative predictive value of 96.4%. Interestingly, a small number of patients (4/160; 2.5%) showed Lisch nodules without choroidal abnormalities, whereas a larger number of patents (22/160; 13.75%) presented choroidal lesions in absence of Lisch nodules. None of the patients showed the absence of both choroidal lesions and Lisch nodules. The number of choroidal lesions increased with age (r = 0.364, p = 0.0001) and with the severity of pathology (r = 0.23, p = 0.003). Any statistically significant correlation between choroidal lesions, visual acuity, and intraocular pressure was observed.

Conclusions: NIR imaging represents an in vivo, non-invasive, sensitive and reproducible exam to detect choroidal nodules in NF-1 patients, suggesting that choroidal changes may represent an additional diagnostic criterion for NF1.

Keywords: Choroid; Lisch nodules; NIR-Oct; Neurofibromatosis type 1; Ophthalmoscopy.

Gharbiya M, Giustolisi R, Marchiori J, Bruscolini A, Mallone F, Fameli V, Nebbioso M, Abdolrahimzadeh S. Comparison of Short-Term Choroidal Thickness and Retinal Morphological Changes after Intravitreal Anti- VEGF

Therapy with Ranibizumab or Aflibercept in Treatment-Naive Eyes. Curr Eye Res. 2018 Mar;43(3):391-396. doi:10.1080/02713683.2017.1405045. Epub 2017 Nov 22. PMID: 29166140.

2018

Abstract

Purpose: To evaluate choroidal thickness (CT) and retinal morphological changes in eyes with neovascular age-related macular degeneration (nAMD) following ranibizumab or aflibercept intravitreal treatment.

Materials and methods: This was a prospective, observational, comparative study where 76 eyes of 76 consecutive patients with treatment-naiven AMD were consecutively enrolled and randomized to ranibizumab 0.5 mg or aflibercept 2 mg injections. Spectral-domain optical coherence tomography images of the choroid were obtained by enhanced depth imaging modality. CT measurements were made of the subfoveal choroid, and at 500 µm from the center of the fovea in the superior, inferior, temporal, and nasal quadrants. Central subfield retinal thickness, intraretinal fluid, subretinal fluid, and pigment epithelium detachment were evaluated. Patients were followed up for 3 months.

Results: Compared with baseline, CT decreased over time in both the ranibizumab and aflibercept group (P=0.04 and 0.001, respectively). At each location, the decrease in CT was significantly more prominent in aflibercept with respect to ranibizumab-treated eyes (P < 0.05). Among the different choroidal neovascularization subtypes, type 3 lesions showed the greatest CT decrease after anti-vascular endothelial growth factor injections (P = 0.003). Choroidal thinning was significantly greater in type 3 lesions treated with aflibercept compared with ranibizumab (F = 13.6, P = 0.002). Post-treatment incidence of dry macula was higher in aflibercept- versus ranibizumab-treated eyes (50% vs. 76%, P = 0.03).

Conclusions: CT reduction is greater in aflibercept-treated eyes, and type 3 lesions show the greatest thickness decrease. The post-treatment frequency of dry macula, evaluated by qualitative parameters, is higher in aflibercept-treated eyes, but is not correlated with CT change.

Keywords: Age-related macular degeneration; aflibercept; anti-VEGF; choroidal thickness; dry macula; optical coherence tomography; ranibizumab.

Trebbastoni A, Marcelli M, Mallone F, D'Antonio F, Imbriano L, Campanelli A, de Lena C, Gharbiya M. Attenuation of Choroidal Thickness in Patients With Alzheimer Disease: Evidence From an Italian Prospective Study. Alzheimer Dis Assoc Disord. 2017 Apr-Jun;31(2): 128-134. doi: 10.1097/WAD.0000000000000176. PMID: 27875364.

2017

Abstract

Introduction: To compare the 12-month choroidal thickness (CT) change between Alzheimer disease (AD) patients and normal subjects.

Methods: In this prospective, observational study, 39 patients with a diagnosis of mild to moderate AD and 39 age-matched control subjects were included. All the subjects underwent neuropsychological (Mini Mental State Examination, Alzheimer disease Assessment Scale-Cognitive Subscale, and the Clinical Dementia Rating Scale) and ophthalmological evaluation, including spectral domain optical coherence tomography, at baseline and after 12 months. CT was measured manually using the caliper tool of the optical coherence tomography device.

Results: After 12 months, AD patients had a greater reduction of CT than controls (P≤0.05, adjusted for baseline CT, age, sex, axial length, and smoking).

Discussion: CT in patients with AD showed a rate of thinning greater than what could be expected during the natural course of aging.

Moramarco A, Lambiase A, Mallone F, Miraglia E, Giustini S. A characteristic type of retinal microvascular abnormalities in a patient with Neurofibromatosis type 1. Clin Ter. 2019 Jan-Feb;170(1):e4-e9. doi: 10.7417/CT.2019.2101. PMID: 30789191.

2019

Abstract

This study aims to describe a typical retinal microvascular abnormality in patients with neurofibromatosis type 1 (NF-1). A 64-year-old man with diagnosis of NF-1 was

evaluated by complete ophthalmological examination, including fluorescein angiography and spectral Domain OCT in Near-Infrared (NIR-OCT) modality. Slit lamp exam showed the presence of more than 10 Lisch nodules for each eye.

Ophthalmic examinations and NIR-OCT scans showed the presence of retinal tortuous vessels ending in a 'puff of smoke' arrangement. The clinical significance as diagnostic and prognostic factor of this novel type of retinal microvascular abnormality in NF-1 should be further investigated. Keywords: Microvascular abnormalities; Moya-Moya disease; NIR-OCT; Neurofibromatosis; Retina.

Miraglia E, Iacovino C, Corbo G, Mallone F, Moramarco A, Giustini S. Lisch Nodules in Schwannomatosis: A New Manifestation. Acta Scientific Ophthalmology. 2020 Dec.; Volume 3 Issue 12. (Article in course of publication).

2020

Abstract

Schwannomatosis is a syndrome characterized by presence of schwannomas in the absence of bilateral vestibular schwannomas and meningiomas. Schwannomas interest frequently peripheral nerves (90%) and spinal nerves (75%).
Schwannomatosis are generally sporadic; in 15 - 25% are familiar. The genes involved are SMARCB1 (40-50% of familial) and LZTR1. The reported phenotype continues to expand and evolve. We report the case of a patient with Schwannomatosis and Lisch nodules, typical manifestation of NF1.

Keywords: Lisch Nodules; Schwannomatosis; Schwannomas

COMPETENZE LINGUISTICHE

LINGUA MADRE: Italiano

Inglese

Ascolto	Lettura	Produzione orale	Interazione orale	Scrittura
C1	**C1**	**C1**	**C1**	**C1**

COMPETENZE DIGITALI

European Computer Driving Licence (ECDL)

Autorizzo il trattamento dei miei dati personali ai sensi del Decreto Legislativo 30 giugno 2003, n. 196 "Codice in materia di protezione dei dati personali" e del GDPR (Regolamento UE 2016/679).

Roma, 23/11/2020