

ADVANCES IN TRANSLATIONAL RESEARCH FOR INHERITED EYE AND RETINAL DYSTROPHIES: BREAKTHROUGHS AND INNOVATIONS

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- **DOTT.SSA SIMONA ALIBRANDI**, UNIVERSITÀ DEGLI STUDI DI MESSINA E IEMEST
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- **DOTT. ANTONIO CRUPI**, UNIVERSITÀ DEGLI STUDI DI MESSINA E SCUOLA SUPERIORE SANT'ANNA DI PISA
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- **DOTT. ROCCO DI LORENZO**, PRESIDENTE DELL'ASSOCIAZIONE DEI RETINOPATICI ED IPOVEDENTI SICILIANI (ARIS)
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- **DOTT. FRANCESCO ROMANO**, UNIVERSITÀ DEGLI STUDI DI MILANO E HARVARD RETINAL IMAGING LAB MASS. EYE AND EAR, BOSTON (MA), USA
- **DOTT.SSA JANA SAJOVIC**, UNIVERSITY MEDICAL CENTRE LJUBLJANA, SLOVENIA
- **PROF. SERGIO ZACCARIA SCALINCI**, ALMA MATER STUDIORUM - UNIVERSITÀ DI BOLOGNA
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- **PROF.SSA ANTONINA SIDOTI**, UNIVERSITÀ DEGLI STUDI DI MESSINA E IEMEST DI PALERMO
- **DOTT.SSA ANNAMARIA TISI**, UNIVERSITÀ DEGLI STUDI DELL'AQUILA
- **PROF.SSA MARIA VADALÀ**, UNIVERSITÀ DEGLI STUDI DI PALERMO E IEMEST DI PALERMO

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Eye on Innovation: Transformative Digital Technologies

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Abstract

The digital revolution has created an unprecedented opportunity for advancements in eye care and ophthalmology. Within this domain, transformative digital technologies are emerging as pivotal tools in revolutionizing rare disease diagnosis and treatment development. From healthcare platforms, promising application of NGS (Next-Generation Sequencing) technologies and wearable healthcare devices, the integration of those digital solutions elevates patient care, while offering organizations various opportunities to devise novel eye care models. Furthermore, the increasingly widespread diffusion and adoption of the Internet of Things (IoT), blockchain technologies and artificial intelligence (AI) has enabled advanced data analytics. This advancement offers decision support for the diagnosis, monitoring, and treatment of eye diseases. As these technologies continue to evolve, they foster the creation of an inter-dependent ecosystem, especially in the landscape of rare diseases, where it is essential that digital health solutions harmoniously coexist alongside traditional practices. Therefore, in the rare disease space, with strong interconnections between community of patients, caregivers, patient associations, it is crucial to evaluate the efficacy and effectiveness of these technologies, ensuring their alignment with the overarching goal of meeting end-user needs and advancing the field of ophthalmology.

These technologies not only help improving patient outcomes, both at individual and population-level, but also significantly foster the implementation of a patient-centric approach. Consequently, organizations are called to leverage and update their digital and analytics capabilities. Nevertheless, a clear road map is needed to implement new technologies, capabilities and strategies, which are instrumental in reshaping companies' business models, thereby facilitating their transition towards digital technologies.

Keywords:

Management; Digital Transition; Green Transition.

Exploring lipid-mediated resolution of inflammation in age-related macular degeneration

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Abstract

Exacerbation of inflammatory processes is considered a major player in age-related macular degeneration (AMD), in which glial cells are actively involved. With the discovery of pro-resolving mediators biosynthesized from ω -3 (n-3) essential fatty acids, to date it is increasingly evident that the termination of inflammation is mediated by an active process, and that this is often altered in neuroinflammatory diseases. We hypothesized that resolution of inflammation may be altered in AMD as well. To confirm this hypothesis, we investigated the metabolism and signaling of Resolvin E 1 (RvE1), an eicosapentaenoic acid-derived lipid mediator involved in resolution of inflammation. The RvE1 receptor ChemR23 and the RvE1 metabolic enzymes (5-LOX and COX-2) were unchanged immediately after LD, but they were significantly up-regulated 7 days later. Instead, the RvE1 receptor BLT1 was not modulated by LD, neither was the RvE1 degradative enzyme 15-PGDH. Moreover, ChemR23, 5-LOX, COX-2 and BLT1 were found to be more expressed in the inner retina under all experimental conditions through confocal microscopy. Notably, amacrine cells highly expressed BLT1, suggesting a functional role of RvE1 in the retina. ChemR23 was found to be highly expressed in the activated microglia of the outer retina compared to the resting one.

Finally, ELISA assays also showed that LD rats displayed significantly higher circulating levels and reduced retinal levels of RvE1 compared to controls. Altogether, our data indicate that RvE1 metabolism and signaling are indeed modulated in the LD model, suggesting a potentially relevant role of pro-resolving lipid mediators in AMD.

Keywords:

Genetics; RvE1; RP; AMD.

Bridge research to industry: Open Innovation challenge

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Abstract

In the ever-evolving technological landscape, the gap between academic research and its industrial application has been a topic of enduring concern. The presentation titled "Bridge Research to Industry: Open Innovation Challenge" delves deep into this chasm, seeking pathways to cohesively unite the two realms. Open Innovation (OI) emerges as a potent paradigm, urging industries and researchers to venture beyond traditional boundaries and collaboratively foster innovation. Instead of guarding research as proprietary, OI promotes a shared knowledge ecosystem where external and internal ideas coalesce to drive economic growth and societal benefit. This presentation will underscore the successes achieved through this model, illustrating cases where academia-industry partnerships have fast-tracked technological advancements. Furthermore, we will explore the potential roadblocks in implementing OI and suggest strategic initiatives for its effective assimilation. Attendees will gain insights into how the global scientific community can pivot towards a more collaborative, open, and industrially-aligned future.

By promoting transparent knowledge exchange and cooperative endeavor, we envision a future where research seamlessly transitions from labs to real-world applications, benefiting both industry stakeholders and the wider society

Keywords:

Management; Digital Transition; Green Transition.

Blue-light Exposure induces Pro-Angiogenic Marker Expression in Human RPE Cell

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Abstract

The retinal pigment epithelium (RPE) is a monolayer of polarized neural-crista-derived pigmented epithelial cells separating photoreceptors from the choriocapillaris vascular bed. On the apical side, it interacts with the outer segments of the photoreceptors, while on the basolateral surface it makes contact with Bruch's membrane and choriocapillaris forming the blood-retinal barrier (BRB). Aberrant vascular remodeling occurs in age-related macular degeneration (AMD) or diabetic retinopathy, while choriocapillaris loss was observed in the late stage of Retinitis Pigmentosa (RP). Occasionally, choroidal neovascularization (CNV) occurs in RP patients. RP comprises a wide spectrum of inherited retinal dystrophies characterized by the progressive photoreceptor loss.

transcriptome analysis, we evaluated expression of angiogenesis related genes in RPE cells exposed to oxidative damage. Moreover, we evaluated involvement of these genes in RP onset. We found that two RP causative genes, AHR and ROM1, may be linked to CNV observed in RP, suggesting that this event could occur in AHR and ROM1 mutation carriers. Further confirmation of this hypothesis could lead to consider pro-angiogenic factors as therapeutic targets to prevent tardive photoreceptor loss and, the phenotype worsening.

Keywords:

Genetics; Angiogenesis, RP.

Excursus into AD-RP cross-link: could retinal and brain degeneration related diseases have a common humus?

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Abstract

In early stages, Alzheimer-Perusini's disease (AD) affected people often suffered from vision-related deficits; for instances, color vision, impaired contrast sensitivity, visual acuity, visual integration, visual field loss, macular thinning, visuomotor and visuospatial deficits. In parallel with the progression of the disease in the visual cortical areas, a glaucoma and age-related macular degeneration (AMD) related-cell death of the retina occurs. Probably retina is involved in neural trajectories, which also include hippocampus, where the great part of AD forms begins. Thinning of the retinal nerve fiber layer (RNFL), which results from the of retinal ganglion cells (RGCs) selective loss, has been proposed as a potential diagnostic test for AD, by means of electroretinography (ERG) and optical coherence tomography (OCT). AD-like APP processing occurs in the retina and other non-nervous eye tissues. In the vitreous and aqueous humor, it is possible to find Amyloid beta fragments (A β) in variable amounts as in the cerebrospinal fluid (CSF). A β is typically found in drusen too (one of the pathologic hallmarks of AMD, which interests retinal pigmented epithelium layer, RPE). Anyway, A β accumulates in all layers of the retina. Various research groups have demonstrated as A β induces tau hyperphosphorylation, a well-known pathologic mechanism followed by its accumulation as amorphous deposits and neurofibrillary tangles (NFTs) in ganglion cell layer, nerve fiber layer, photoreceptor layer, and the inner plexiform layer.

However, all neurodegenerative disease, in particular AD and retinal dystrophies, are now recognized as mixed proteinopathies. In various neurodegenerative disease affected brains and retinas, Prion protein (PrP) deposits has been found; while in Parkinson ones, α -synuclein accumulates was seen. Moreover, both in AD and retinal dystrophies, glial cells seem to play a fundamental role throughout the course of the illness. In particular, microglial ones are responsible for the phagocytosis of protein and other molecules accumulations; they constitute the guardian of immunoproteostasis. Retinal and brain microglia have been demonstrated to share the expression of several transcription factors. Most studies have also observed a certain degree of correspondence between retinal and brain morphotypes.

Keywords:

Genetics; Alzheimer disease, Retinal dystrophies.

University-Industry Technology Transfer

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Abstract

During the last decades, research on innovation management has largely recognized the importance of universities (and other public research centers) in the process of innovations generation, development and exploitation. However, while in the past the role of universities was mainly limited to the supply of skilled human resources and scientific knowledge (as primary input of an innovation development process within a linear model of innovation), in more recent years universities have started to assume a more proactive role, by also becoming competitors (not only partners) of companies in the broad market for technologies. This process has been the response to an increasing intensity of scientific knowledge in products and services, so that the traditional differentiation between university and industry has become more blurred. As a consequence, more and more universities have devoted internal resources and organizational competences for this end (for example, by setting up dedicated Technology Transfer Offices or Liaison Offices), have implemented programs to promote patent out-licensing and spin-offs creation, and have pursued business models of technology commercialization and valorization. The engagement of universities with society at large has thus expanded, and universities have become a key player of the complex innovation ecosystem.

Keywords:

Technological entrepreneurship; Technology licensing strategies; Intellectual property rights management; Management of technology transfer processes.

Non-coding RNAs in hereditary retinal dystrophies

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Abstract

Inherited retinal dystrophies (IRDs) represent a genetically heterogeneous group of retinal pathologies characterized by progressive photoreceptor degeneration leading to loss of vision. They are classified as rod-cone dystrophies (RCD) and cone-rod dystrophies (CRD), depending on the photoreceptor cell type which is primarily affected by the disease. Retinitis pigmentosa (RP) is the most common type of rod-cone dystrophy, with a prevalence of 1:3500 individuals worldwide. In the last decade, it has been shown that gene therapy is a promising approach to correct the genetic defects underlying IRDs. Although we have a profound understanding of etiopathogenesis of several IRDs and despite the recent advancements in gene therapy, there is still a lack of an effective treatment for this group of retinal disorders. Therefore, research is focusing on other therapeutic strategies as MicroRNAs. MicroRNAs (miRNAs) are small single-stranded non-coding RNA molecules that negatively regulate genes by silent post-transcriptional gene expressions. Evidence indicates that miRNAs play a key role in the development and maintenance of function within the eye, particularly the retina. Mutations in miRNAs or their target genes may contribute to a range of ocular disorders, including inherited retinal dystrophies. Nowadays, novel therapeutical approaches for IRDs based on miRNA modulation are being evaluated.

Keywords:

IRDs; miRNAs; RNA.

Unraveling the differences between Stargardt disease, Central areolar choroidal dystrophy and geographic atrophy: the SCOOP study

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Abstract

Macular dystrophies such as Stargardt disease (STGD) and central areolar choroidal dystrophy (CACD) represent important mimickers of dry age-related macular degeneration (dAMD). Difficulties in correctly identifying these cases can result in delayed diagnosis and, ultimately, in inappropriate inclusion in upcoming clinical trials. Our observational cross-sectional study aimed to investigate novel imaging and functional differences in these three conditions in order to facilitate their differential diagnosis, gain insight in their pathogenesis and optimize functional endpoints. We examined 18 eyes in each group, assessing best-corrected visual acuity, mesopic and two-color scotopic microperimetry, blue-light autofluorescence, high-resolution optical coherence tomography (Hi-Res OCT), macular OCT angiography (OCTA). Our microperimetry assessment revealed that patients with STGD and CACD experience a predominant cone dysfunction, especially in the central 3° of vision, unlike those with dAMD. Imaging assessment showed distinctive features: CACD group had a consistent autofluorescence pattern, while unique outer retinal and retinal pigment epithelium (RPE) alterations were identified on Hi-Res OCT.

Additionally, we observed more severe choriocapillary rarefaction in the atrophic areas of STGD and CACD patients. Understanding these differences sheds light on the pathogenesis of these macular conditions, emphasizing the need for tailored treatment approaches. Our study suggests that specific tests, such as mesopic microperimetry and Hi-Res OCT, can be crucial in diagnosing and managing these macular dystrophies. These insights pave the way for future clinical studies, especially for STGD and CACD patients, and hold promise for better therapeutic strategies.

Keywords:

IRDs; STGD; CACD; Hi-Res OCT.

Can electroretinography be used as a biomarker to monitor Stargardt disease?

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Abstract

Using electroretinography (ERG), retinal function is measured. Our study aimed to determine which ERG responses best correspond with disease progression in Stargardt disease (STGD1). 42 patients with STGD1 were included. Group 1 harbored two null mutations (8 patients, 3 male), while group 2 had other genotypes (34 patients, 10 male). Age at the time of exam, age at onset, visual acuity (VA) and ERG responses were evaluated. The correlation between ERG responses and age at the exam was compared using simple linear regression. Mann-Whitney U Test was used to compare the median values between the groups. The median age at onset was significantly earlier in group 1 (median 8 and 18.5 years, $p < 0.001$), while disease duration was similar between the two groups (median 12.5 and 12 years, $p = 0.320$). Group 1 had significantly worse VA (1.8 vs. 0.8, $p < 0.001$) and significantly lower all ERG responses ($p < 0.001$). Simple linear regression analysis revealed that in group 1, age significantly correlated with the DA 0.01 ERG b-wave ($\beta = -0.806$, $R^2 = 0.650$, $p = 0.016$) and DA 3.0 ERG a-wave ($\beta = -0.865$, $R^2 = 0.748$, $p = 0.006$), while in group 2, age significantly correlated with the S-cone ERG amplitude ($\beta = -0.065$, $R^2 = 0.446$, $p < 0.001$). Other ERG parameters did not show significant correlations with age in either group. To conclude, different ERG responses may be best suited to monitor disease progression in different STGD1 genotypes. The results are especially important for designing clinical trials that require sensitive and objective biomarkers to determine disease trajectory.

Keywords:

STGD; ERG; VA.

Don't forget your parents! Retinal ganglion cell lineage analysis on a cellular resolution

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Abstract

An essential challenge in regenerative neurobiology concerns replacing retinal ganglion cells (RGCs) and their axons, which are irreversibly lost or damaged in glaucoma and other optic neuropathies. Encouraging data suggests that RGC generation can be accomplished from induced pluripotent stem cells and even resident, closely related cell types by forcing them to reactivate essential developmental programs. However, before this can be achieved to target RGC replacement in mammals, acquiring knowledge on how such developmental programs can be activated and maintained in resident cells in the mature and damaged mammalian retina is required. Research in my group has contributed to the indispensable prerequisites for targeting RGC replacement: understanding the genesis of RGCs from retinal progenitor cells (RPCs) and the underlying genetics in the native retinal environment. We use completely transparent zebrafish embryos that grow rapidly outside their mother to study *in vivo* how RPCs activate RGC developmental programs as they divide, differentiate and mature, extending their axons into the brain. Our studies provided insights into the influence of extrinsic cues and modes of cell division in restricting the RGC commitment and differentiation of RPCs. We also began to gain insight into shared genetic signatures and lineage relationships amongst RGCs and other cell types, suggesting that they share epigenetic ancestry and developmental potential or neuronal plasticity.

With this knowledge and expertise, we generated a framework to advance understanding of resident cells' lineage and developmental potentials to target RGC genesis in the adult retina.

Keywords:

RGCs; RPCs; Retinal dystrophies; Zebrafish.

The Future of Retinal Molecular Genomics: a Convergence of Mitogenomics, G-Quadruplex, Non-coding RNAs and other Epigenetic Changes

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Abstract

The rapidly evolving field of retinal molecular genomics is witnessing a convergence of various epigenetic mechanisms that promise to revolutionize our understanding of retinal diseases and their treatments. Key among these are mitogenomics, G-quadruplex structures, non-coding RNAs, and other epigenetic modifications. Recent studies have highlighted the role of mitochondrial DNA (mitogenomics) in retinal health and disease, suggesting a direct link between mitochondrial dysfunction and retinal disorders. Concurrently, G-quadruplex structures, unique four-stranded DNA configurations, have been identified as potential therapeutic targets in retinal diseases due to their involvement in DNA replication, transcription, and genomic stability. Non-coding RNAs, particularly long non-coding RNAs (lncRNAs), have emerged as crucial regulators of gene expression in the retina. Their dysregulation has been associated with various retinal pathologies, emphasizing their potential as both biomarkers and therapeutic targets. Additionally, other epigenetic changes, including DNA methylation and histone modifications, are being explored for their roles in retinal development, function, and disease. The integration of these epigenetic mechanisms offers a holistic view of retinal genomics and provides novel avenues for the diagnosis, prognosis, and treatment of retinal diseases.

As we stand on the cusp of this genomic revolution, it is imperative to further our understanding through comprehensive research and collaborations, ensuring the translation of these findings into clinical applications.

Keywords:

Genetics; Omics sciences; Retinal dystrophies; G-Quadruplex; ncRNAs; Epigenomics.

Rehabilitation and associationism: that's also therapy

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Abstract

Global Burden of Disease estimates predict a three-fold increase to over 500 millions of visually impaired people worldwide by 2050, largely due to population ageing. While treatment options are improving, we will inevitably face an increasing burden of visual disability. We present two different ways to face visual impairment, from the point of view of health system and of patients; Vision Rehabilitation (VR) and Associationism. Our practical experience in VR is following the example of D.A.Re. registry and we present our project named PROVIT, Promoting Rehabilitation fOr the Visually Impaired in Italy. It has been participating to the selection for PNRR funding and it has already passed first selection. D.A.Re. registry first publication demonstrated that patients using electronic devices, especially smartphone and tablets, report better vision-related quality of life and IADL score. Aims of the project are to identify which patient characteristics and which devices are associated with better quality of life, to promote and implement a network of VR Centers in Sicily and Campania and to conduct qualitative or mixed-method research on barriers and facilitators for the successful implementation of a local VR service network. We then present the experience of patients' associationism in Italy targeting our interest on national and vision-related ones: their value as support to patient but also to clinicians and governance are examined. We also show our experience in some activities of narrative medicine.

Keywords:

VR; D.A.Re.; Retinal dystrophies.

Management of inherited retinal dystrophies: from diagnosis to genetics, our experience

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Abstract

Inherited retinal dystrophies are a heterogeneous group of genetically determined ocular pathologies, in which, due to the involvement of different genes and pathogenetic mechanisms, there is the progressive degeneration of the retinal photoreceptors/RPE complex. Two large categories can be recognized in the classification, depending on whether the rods (rod-cone dystrophies, incorrectly called peripheral forms) or the cones (cone-rod dystrophies) are initially and predominantly affected. The onset symptoms are different: the rod-cone forms are characterized by night blindness and concentric reduction of the visual field, while in the cone-rod forms there is an earlier central visual decline, congenital or acquired deficit of chromatic vision, photophobia. In some cases, retinal alterations constitute a sign in the context of syndromic diseases (i.e., which involve multiple areas of the organism) and in these cases a polyspecialistic evaluation of the patient is necessary (in particular otolaryngologist, neurologist, clinical geneticist). Hereditary retinal dystrophies can be transmitted with any mode of inheritance (autosomal recessive, autosomal dominant, X linked, mitochondrial). Among the main pathologies main peripheral forms are retinitis pigmentosa (typical and atypical forms) and choroideremia (also involving choriocapillaris), while the main central forms are Stargardt macular dystrophy, Best macular dystrophy, X-linked retinoschisis, progressive cone dystrophy.

The diagnosis is clinical and instrumental (OCT, autofluorescence and electroretinography) and in selected cases it can be completed with biomolecular diagnostic at the identification of the causing genetic defect. Starting from about 5 years ago the U.O.C. of Ophthalmology at Villa Sofia Hospital has been recognized as a Regional Reference Center for rare eye diseases.

Keywords:

IRDs; OCT; Retinal dystrophies.

The antioxidant properties of Nanoceria particles are useful to protect retinal morphology and function in an animal model of age-related macular degeneration (AMD)

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Abstract

As part of the central nervous system, the retina is particularly susceptible to alterations of its microenvironment, which can cause loss of vision. In fact, the retina is characterized by a state of physiological oxidative stress due to an elevated metabolism and high oxygen consumption and the maintenance of a balanced microenvironment is fundamental in order to allow the health of the retinal cells. AMD is a result of photoreceptor, retinal pigmented epithelium, Bruch's membrane and choriocapillaris alterations, which culminates in blood retinal barrier breakdown, activation of inflammatory events and retinal neurodegeneration. AMD can be considered a multifactorial disease and aging, cigarette smoke, high fat diet, light exposure, alcohol consumption, and specific genetic polymorphisms are considered the main risk factors. All these events share oxidative stress as a common feature that can be considered the driving force of all the risk factors. Based on the absence of effective therapies for the treatment of AMD, in recent years important improvements have been made surrounding nanomedicine, which represents a promising research field due to the unmatched properties of nanoparticles.

Cerium oxide nanoparticles (CeO₂-NPs), a pure antioxidant, have been tested in our animal model of AMD and we demonstrated that the main features of AMD can be counteracted. Specifically, we have demonstrated their ability to preserve retinal function, avoid the blood retinal barrier breakdown and debris accumulation, counteract neovascularization and microglial activation. Based on this evidence, it can be considered that CeO₂-NPs may represent a promising therapeutic approach for AMD.

Keywords:

Physiology; CeO₂-NPs; AMD.

The “State of Art” about Inherited Retinal Dystrophies Molecular Genetics

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Abstract

Inherited retinal dystrophies (IRD) are a group of diseases due to progressive loss or dysfunction of photoreceptors and characterized by a high genetic and clinical heterogeneity. To date, in fact, about 300 genes have been associated with IRDs for syndromic and non-syndromic forms to which we must add the high inter and intrafamily variability, variable expression and incomplete penetrance that characterize these diseases. IRDs can follow different patterns of inheritance including autosomal dominant, autosomal recessive, X-linked, mitochondrial mode, and some other less common, such as digenic inheritance. For these reasons, molecular diagnosis is very complex and in a significant number of patients it is not possible to identify causative mutations. The advent of the Next Generation Sequencing (NGS) has given an important acceleration to the identification of new genes and diagnosis. Thanks to different approaches such as custom panel designs, whole genome sequencing (WGS) and whole exome sequencing (WES) it is possible to improve the study of the molecular mechanisms of IRDs. These new sequencing techniques are essential to identify mutations in novel IRD-related genes, deep-intronic variants and modifier genes, able to influence the phenotype of single patient. In this way, it is possible to get to an early and accurate diagnosis that will allow patients to make accurate reproductive decisions and to be enrolled in future gene-based clinical trials.

Keywords:

Genetics; IRDs; WES; WGS.

Photobiomodulation and stem cells in retinal diseases

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Abstract

Age-related macular degeneration (AMD) is one of the most common causes of vision impairment worldwide. Among the two types, while there are several therapeutic strategies available for exudative macular degeneration, current clinical trials for the dry form offer no guarantee of cure or slowing of the disease. Our study leverages the widely accepted therapeutic potential of stem cells to promote the slowing of cell death and, consequently, the stabilization of the clinical picture in atrophic macular degeneration. One of the most promising therapeutic frontiers is the potential “cross-talk” between mesenchymal stem cells and retinal cells. Thanks to the molecular genetics support of the University of Messina and the Euro-Mediterranean Institute of Science and Technology (I.E.M.E.S.T) of Palermo, we implemented an innovative therapeutic protocol using mesenchymal secretome injected sub-sclerally and subsequently activated with Quantum Molecular Resonance Electrotherapy (QMR). This aims to preserve the morpho-functional integrity of the retina by regulating the inflammatory response, which is known to underlie clinical deterioration and progression of the disease towards the exudative form. The success of the results on cell-mediated retinal regeneration, obtained in preclinical studies, encourages clinical ones, and our protocol with Rexion-Eye has proven to be effective and safe in improving both subjective and objective ocular parameters; therefore, the treatment is considered innovative.

Keywords:

AMD; QME; Retinal regeneration.

New insights on GLO1-related Molecular Mechanisms from RNA-Seq on A2E-stressed RPE cells

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Abstract

One of the main risk factors in Retinitis Pigmentosa (RP) is the reactive oxygen species and advanced glycation products (AGEs) accumulation. These compounds can determine photoreceptor homeostasis alteration, autophagic impairment and other mechanisms related to oxidative stress. This scenario is amplified by the inflammation and damage to photoreceptors resulting in cell death. The role of GLO1 was investigated by comparing the expression profiles, obtained by RNA-seq, between untreated and treated RPE cells considering two time points (3 and 6 hours). The results of this analysis showed important expression differences and splicing events in 370 genes involved in the Glo1 pathway. From pathway clustered analysis it emerged that 23 of them are the main candidates to be associated with retinitis pigmentosa. These genes are involved in various molecular genetic mechanisms related to oxidative stress such as glyoxylate and dicarboxylate metabolism, glycolysis, axo-dendritic transport, lipoprotein activity and metabolism, SUMOylation and retrograde transport at the trans-Golgi network. These results could represent an important step towards clarification of new GLO1-related molecular mechanisms behind RP etiopathogenesis.

Keywords:

Genetics; IRDs; RP; GLO1; WES.

Visionary Strategies and Digital Marketing: The Potential of Digital Marketing in Raising Awareness of Hereditary Eye and Retinal Dystrophy

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Abstract

Hereditary eye and retinal dystrophy are a group of genetic diseases that can cause severe vision problems and even blindness. These diseases affect individuals of all ages and pose a significant challenge to patients, their families, and health professionals. However, digital marketing offers enormous potential to raise awareness and support this cause. In this talk, we will explore how digital marketing can be a valuable ally in the fight against inherited eye and retinal dystrophy. Thanks to the new digital technologies that the marketing function can implement, among many, the following opportunities can be identified: i) involving the community, ii) fundraising and sustainability, and iii) awareness campaigns. Involving the community: people affected by hereditary eye and retinal dystrophy, together with their families and health workers, constitute a valuable community. Digital marketing can help create online communities, such as forums and social media groups, where people can share their experiences, find support, and exchange useful information. These online spaces can become hubs for resource sharing and networking support. Fundraising and Sustainability: non-profit organizations engaged in research and support for hereditary eye and retinal dystrophy can leverage digital marketing to raise funds. Online campaigns on crowdfunding platforms, donation websites, and social media can help raise public awareness and obtain essential funds for research and assistance. In addition, digital marketing can help keep supporters informed about the progress and use of funds raised.

Awareness Campaigns: online awareness campaigns can have a significant impact on promoting awareness of hereditary eye and retinal dystrophy. The use of targeted social media ads, viral awareness campaigns, and dedicated hashtags can make the message reach a wide audience quickly and effectively. This increases the visibility of the cause and can lead to greater participation and support. Digital marketing offers numerous opportunities to raise awareness and support people affected by hereditary eye and retinal dystrophy. From the dissemination of information to the creation of online communities, from fundraising to the promotion of awareness campaigns, digital marketing can play a crucial role in the fight against these diseases. By working with digital marketing experts, organizations and interested individuals can amplify their voices and make a real difference in the lives of those facing these challenges. Harnessing the full potential of digital marketing to raise awareness and support for this important cause is critical.

Keywords:

Open Innovation; Product sustainability; Product personalization; FoPL.