Global Eye Genetics Consortium

Newsletter#01

About Us

The Global Eye Genetics Consortium (GEGC) was originally established as Asian Eye Genetics Consortium (AEGC) in 2014 and expanded in 2018 to promote and enhance collaboration on eye genetic research with developing countries of Asia, Africa and Central & South America.

GEGC now has more than 200 members of scientists and ophthalmologists from 30 counties. The new GEGC Phenotype-Genotype Database "GenEye" is now open to collect phenotype-genotype information. GEGC is actively organizing sessions in local and international ophthalmology meetings around the globe.

Goals of Global Eye Genetics Consortium

Goals

- Share genetic information to isolate common genetic variants associated with genetic eye diseases
- Establish cost effective genetic analysis and accurate diagnosis for grouping of genetic eye diseases
- Develop research-oriented database to collect and catalog genetic eye diseases at global scale
- Support and foster global collaboration for the advancement of genetic eye research
- Collaborate with other international or regional organizations with similar goals
- Organize regional congresses and other educational and scientific activities to promote goals of the consortium

Executive Members

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Molecular and Cellular Biology Division, National Institute of Sensory Organs National Hospital Organization Tokyo Medical Center, Tokyo, Japan

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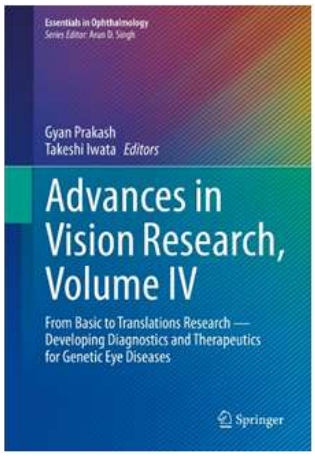
Pediatric Ophthamology, Strabismus, Neuro-ophthalmology and Ocular Genetics, Dr Shroff's Charity Eye Hospital, New Delhi, India

From Our President's Desk

As a president of GEGC, it is my great pleasure to announce the launch of GEGC Newsletter in 2024. This launch was coordinated to celebrate the 10th year anniversary of GEGC, which started at 2014 ARVO in Orlando, Florida, USA. The aim of this consortium is to establish global network to tackle genetic eye diseases by international collaborations and sharing of phenotype-genotype information. Since 2014, we have organized over 30 scientific meetings and sessions, conducted young scientist visiting programs, established genetic labs, and research collaborations followed by publications. I am happy to see that GEGC members has now increased to 220 from 38 countries. This year GEGC will have the first scientific session in Africa and South America, which was postponed since 2020 by pandemic. We hope to see you at African Ophthalmology Council in Kigali, Rwanda or at International Society for Eye Research in Buenos Aires, Argentia. In addition to these progress, Dr. Gyan "John" Prakash and myself are also excited to announce the coming publication of Springer Nature, Advances in Vision Research, Volume IV, From Basic to Translational Research - Developing Diagnostics and Therapeutics for Genetic Eye Diseases. Since publication of books take years of preparation, we were desperate to establish more timely publication from GEGC. We hope to bring you this GEGC Newsletter in quarterly interval to introduce individual or labs that can offer new collaborations and projects to whom seeking for help to start eye genetic research. Lastly, I would like to thank our Secretary General, Dr. Shailja Tibrewal and her team at Dr Shroff's Charity Eye Hospital to initiate this GEGC Newsletter.



Dr. Takeshi Iwata, PhD, FARVO

Upcoming Activities

Global Eye Genetics Consortium Meeting at the Annual APAO Meeting at Bali Nusa Dua Convention Center, Indonesia, from February 22 to 25, 2024

Global Eye Genetics Consortium Meeting Join us May 6, 2024, 1:15pm-2:45pm at Seattle Convention Center, Seattle, USA

Recent Event

GEGC was well represented in the 16th Asia-Pacific Vitreo-retina Society (APVRS) Congress held in Hong Kong, from December 8 – 10, 2023. It was attended by clinicians, scientists and policy makers from accross the world. The session was named after Dr Chi Chao Chan who is the former chief of the NEI immunopathology section and the histopathology core. She was honored by a lifetime achievement award in the session. The scientific session reflected the diversity and the inclusivity of GEGC. It began with our patron Dr Gyan 'John' Prakash, (Associate Director for International Programs, National Eye Institute) enlightening the audience about the global collaborations in eye research. Special emphasis on the relationship between the Asia-Pacific region and the US with regard to biomedical research was given in the presentation by Dr Erika Elvander (US Health Attache to China). Therafter Dr Kapil Bharti (Head of the Ocular and Stem Cell Translational Research Section at the National Eye Institute (NEI)) gave insights on his work related to stem cell derived RPE in improving macular sensitivity in age related macular degeneration. Conducting large multicentric clinical studies is always challenging. Dr Emily Chew (Director of the Division of Epidemiology and Clinical Applications (DECA), NEI) highlighted these challenges and provides avenues for solutions in her talk. Dr Michael Chiang (Director, NEI) further provided the standards of Ocular imaging which support global collaboration. Dr Calvin Pang (S.H. Ho Research Professor of Visual Sciences, CUHK) spoke about the global collaborations pertaining specifically to eye genetics, which is at the core of GEGC's mission. Myopia is a growing concern in the Asia Pacific region. This has led to path breaking research from this region guiding the world. One such science showcasing the role of light therapy in myopia was presented by Dr Mingguang HE (Chair Professor of Experimental Ophthalmology at The Hong Kong Polytechnic University). Finally Dr Wei Le (Associate Professor of Computer Science, Iowa State University) described a case of vision research collaboration. The session was interspersed with discussions amongst the panelists and the audiences. Ample opportunities for the attendees to connect with others in their field, fostering collaboration and professional development were provided.

Meet Our Members

Dr. Laura Echandi, MD

Currently living in Argentina, South America. Dr. Laura Echandi, is a Retina specialist (clinical and surgical) in Consultores Oftalmologicos (University of Buenos Aires and Universidad del Salvador, Buenos Aires Argentina). She specializes in Inherited Retinal Dystrophies and Electrophysiology. She is the Founder Professor of PANIRD (Panamerican Society of Inherited Retinal Dystrophies). She has been an active member of the Global Eye Genetics Consortium member (GEGC). She is the honorary Member of the directive Counsel of Fundación Médica para la Salud Visual y Rehabilitacion (FUSAVI), Buenos Aires.

Dr Zi-Bing Jin MD, PhD

Dr Zi-Bing Jin obtained his MD in 2000 from Wenzhou Medical College and received residency training at the Eye Hospital afterward. He has also a PhD in Ophthalmology obtained in 2007 from University of Miyazaki in Japan. After 4-year research working with Dr. Masayo Takahashi at RIKEN, he came back to motherland. Currently, Dr. Jin is a Full Professor of Ophthalmology at the Capital Medical University (CMU) and the Vice President of the Beijing Tongren Hospital, Capital Medical University (CMU) after being appointed as the Director of Beijing Institute of Ophthalmology since 2020. He is also the Chief physician at Beijing Tongren Hospital, CMU. Dr Jin aims to elucidate the disease mechanisms of childhood ocular disorders with special focus on early-onset high myopia, juvenile macular degeneration, translating laboratory technology to improve bedside outcomes. Dr. Jin and his team research and validate new, groundbreaking ways of growing key ocular tissues from fibroblasts through small molecules and culturing retinal organoids in vitro for the disease modeling of retinitis pigmentosa and retinoblastoma. Dr. Jin is an active contributor to the wider scientific community, acting as an editor and reviewer for several academic journals, including Experimental Eye Research, Ophthalmic Research, APJO."

Christina Zeitz

Christina Zeitz is a research director (DR2) at INSERM and is co-leader of the team involved in "Identification of gene defects leading to progressive or non-progressive eye diseases" in the Department of Genetics at the Institut de la Vision (INSERM, UMR_S968, CNRS, UMR_7210, University Pierre and Marie Curie Paris6). She was recruited by an international committee in 2007 as a team leader at the Institut de la Vision with the recruitment at INSERM in 2010 as a CR1 and in 2015 as a DR2 researcher. The work of the team aims to identify genetic defects in large cohorts of patients with different retinal diseases by new technologies such as next-generation sequencing (NGS) using large gene panels involved in these diseases or by sequencing exomes or entire genomes. The team is also interested in studying the function of the proteins encoded bythe identified genes and the pathophysiological mechanisms resulting from these gene defects. Other axes of her work consist of investigations to better understand high myopia in eye diseases and the development of innovative therapies.

Ophthalmic Genetics News around the Globe

Study of more than 11,000 individuals of African descent finds genetic variants linked to glaucoma

One of the leading causes of irreversible blindness around the world, glaucoma affects up to 44 million people. Although people of African ancestry are most frequently and severely affected, little research has been done in the genetic roots of the disease among this population.

Researchers now are unwrapping results that show previously unknown inherited genetic variants that contribute to primary open-angle glaucoma (POAG), the most common form of the disease. The study was based on the analysis of 11,275 individuals of African descent and is being published January 18, 2024, in Cell.1 https://www.cell.com/cell/pdf/S0092-8674(23)01338-7.pdf

Corresponding author Joan O'Brien, MD, the director of the Penn Medicine Center for Genetics of Complex Disease, who was funded by a $17.9 million grant from the National Institutes of Health for this research, noted how the disease impacts the demographic.

"Individuals with African ancestry are five times more likely to be affected by glaucoma and up to 15 times more likely to experience vision loss or blindness from the disease compared to individuals with European ancestry,"

Scientists isolate the cone response in fMRI in Canine models – promising for cone dystrophies therapy models

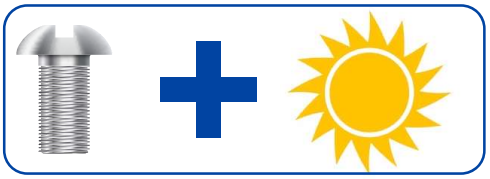
Vision scientists from the Division of Experimental Retinal Therapies at the University of Pennsylvania School of Veterinary Medicine, Gustavo D. Aguirre and William A. Beltran used the functional magnetic resonance imaging (fMRI) to assess the responses in brain stimulated by only cone cells. The experiments were conducted in dogs with three different types of retinal disease and normal controls. Their results reveal that fMRI can detect the anatomical location of the brain responses and the degree of the responses to daylight vision for black and white information as well as color information in dogs. They demonstrated that gene augmentation therapy in a NPHP5 mutation related retinal disorder, restored the response in the cortex to black and white stimulation using this technique (published in Translational Vision Science & Technology). Previously fMRI has been used to detect responses from both rods and cones after gene therapy for Leber congential amaurosis. The new study proves the usefulness of fMRI in diseases affecting specifically the cones as well.

ACDN-01 gets FDA approval and fast track status for application in Stargardt's disease and other ABCA4 retinopathies

Jan 2024 - FDA has cleared the investigational new drug ACDN-01, Ascidian Therapeutics (Boston) and granted Fast Track designation for application in Stargardt disease and other ABCA4 retinopathies. ACDN-01 is the first-ever clinical-stage RNA exon editor and the only clinical-stage therapeutic targeting the genetic cause of Stargardt disease. The company expects to initiate enrollment in the Phase 1/2 STELLAR study of ACDN-01 in Stargardt disease and other ABCA4 retinopathies in the first half of 2024. ACDN-01 has demonstrated efficient, durable in vivo RNA exon editing in non-human primate retina and ex vivo RNA exon editing in human retinal explants.

Sources: Ophthalmology Times, NIH website, Penn News, Pubmed

Genetics Trivia



Hint

World-Renowned cancer geneticist and physician who treated children with retinoblastoma

Editorial Members: Dr. Shailja Tibrewal, Ms. Ria Ratna,
Designed By: Alpana Singh

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