Global Eye Genetics Consortium

Newsletter #06

Foreword

Advancing Ophthalmological Genomics Research in Developing Countries

Dr. Raj Ramesar
Professor and head of the Division of Human Genetics
University of Cape Town

The landscape of research into inherited ophthalmological conditions in developing parts of the world, notably in Africa is marked by both immense potential and significant challenges. The vast genetic diversity in African populations has been underutilized in global research, limiting the ability to fully understand and treat conditions like inherited retinal diseases, glaucoma and other eye diseases. However, this is beginning to change as attention shifts toward the unique genetic makeup of African populations and the need for their inclusion in both research and clinical trials.

One of the key obstacles in advancing ophthalmological genomic research in Africa has been the lack of genetic diagnostic infrastructure. Although phenotypic diagnoses of eye diseases have advanced thanks to technological innovations, genetic diagnoses remain scarce. Without these, African patients are often excluded from clinical trials for new gene therapies that are being developed globally. This underscores the urgent need for collaborative efforts that allow African researchers and institutions to access the tools and resources needed for genetic testing. Building this infrastructure will not only improve diagnostic precision but also position Africa to contribute meaningfully to global research.

The Global Eye Genetics Consortium (GEGC) plays a critical role in addressing these gaps. By fostering collaboration between African institutions and the broader international research community, the consortium intends to help to standardize the approaches to diagnosing and treating genetic eye diseases. This structured, systematic approach is particularly important for low-resource settings, where the focus on building capacity and enhancing infrastructure is essential. As African researchers begin to play a larger role in global genetic studies, the GEGC provides a framework that ensures inclusivity and lays the groundwork for the continent to integrate cutting-edge genomic technologies.

Moreover, Africa's genetic diversity presents a unique opportunity to contribute to global datasets that can accelerate the understanding of hereditary eye diseases. In the past, the continent's underrepresentation in genetic research has meant that therapeutic advances have often bypassed African populations. However, recent initiatives are beginning to close this gap by encouraging data sharing and collaboration between African and international researchers. This collaborative effort is key to creating the comprehensive data needed to better understand disease progression in African populations and to develop treatments that are tailored to this unique genetic context.

While progress is being made, the challenges of funding, training, and infrastructure development persist. For Africa to fully engage in the global genomic research landscape, sustained investment in these areas is essential. Capacity building through training programs, the development of laboratory infrastructure, and the creation of networks for data sharing are all vital to ensuring the continent can keep pace with advancements in genetic ophthalmology.

The promise of the GEGC lies in its potential to bridge these gaps and foster an environment where African researchers can lead in the fight against inherited ophthalmological conditions. By leveraging collaboration and supporting the development of genetic diagnostic tools, the consortium is helping to drive a larger research agenda that could revolutionize eye care across the continent. The goal is not just to participate in global initiatives but to shape them, ensuring that the benefits of genomic advances in ophthalmology reach all populations, particularly those in Africa who have been historically overlooked.

As Africa steps into this new era of precision medicine, the role of consortia like the GEGC will be instrumental in transforming research into inherited eye disorders and their treatment. Through continued collaboration, innovation, and investment in infrastructure, Africa is poised to make significant contributions to global efforts in tackling genetic eye diseases.

Raj S Ramesar
Professor and Head:
Division of Human Genetics
Institute for Infectious Diseases and Molecular Medicine
University of Cape Town and Affiliated Hospitals

Ophthalmic Genetics Trivia

Re-arrange the jumbled words to uncover Ophthalmic genetics related terms:

1) NtseitiriPaimtnosge
2) RelbeDrhaieteryCiptoEuponytrha
3) DtaarsgtrEsseaid
4) LraoptNdysoemr
5) PaticoeEnilst
6) LotonuucscuoeaNlsbimai
7) MsaaltoouDamntion
8) GnronimYgolrDmnoresy
9) EedlnfxaRrgeieMdynoesr
10) Cssoiotrnainsyso

Recent Events:

GEGC leadership at the University of Cape Town

The GEGC leadership visited the University of Cape Town on July 24-25. A seminar titled "Current Challenges in Eye Genomics Research in Africa" was held on the 24th, with key participants including Dr. John Prakash from the National Eye Institute (NEI), Dr. Raj Ramesar from the University of Cape Town (UCT), and Dr. Musa Kana from Kaduna State University. Several attendees, such as Dr. Takeshi Iwata (Tokyo Medical Center), Dr. S. Natarajan (Aditya Jyot Eye Hospital), and Dr. Salil Lachke (University of Delaware), joined virtually.

Dr. Raj Ramesar and Dr. Lisa Roberts provided an overview of UCT's programs, stressing their interest in advancing eye genomics through collaboration and the use of genomic technologies for clinical applications. Dr. Sophia Siddiqui (US Health Attaché) discussed NIH programs in South Africa, focusing on partnerships in genetics and ophthalmology to address public health challenges.

Dr. Gyan "John" Prakash shared insights into the NEI's global programs and opportunities for collaboration through the Global Eye Genetics Consortium (GEGC). Dr. Takeshi Iwata highlighted the role of global cooperation in advancing eye genomics, specifically Japan's work in gene-based therapies for eye diseases. Dr. Salil Lachke introduced the Integrated Systems Tool for Eye Gene Discovery (iSyTE), which aids in identifying genes related to eye diseases.

Dr. Musa Kana presented the Kaduna Pediatric Eye Study, which examines childhood eye disorders in Nigeria, emphasizing the NEI's support in advancing research. Dr. S. Natarajan discussed the practical applications of eye genomics research in clinical settings in India, showing how personalized medicine is being integrated into ophthalmic care.
The seminar concluded with a panel discussion on international collaboration and capacity building in eye genomics, exploring how these advancements can reduce global disparities in eye care.

On 25th July a focussed discussion meeting was held between Dr Gyan 'John' Prakash, Dr Raj Ramesar and Dr Musa Kana. The purpose of this meeting was to discuss the establishment of an African group to conduct ophthalmic genetic research in Africa in collaboration with GEGC.

Action Points:

- ***Reach out to other African countries through the African Ophthalmology Council (AOC).***
- ***Focus on existing successful activities and "low-hanging fruits."***
- ***Examine African research priorities, explore funding, and promote the translational potential of genomic research, especially for diagnostics and therapeutics.***
- ***Develop applications for public health surveillance and genetic counselling training.***
- ***Assess the medical genetics capacity and programs in Africa.***

Stakeholder Involvement: Relevant stakeholders should include patient advocates, bioinformatics experts, clinicians, epidemiologists, and public

health professionals. A mapping of these stakeholders is essential. WHO Involvement: Further exploration is needed to define WHO's specific role in the initiative.

Neglected Eye Disease Research in Africa: Genetic research on eye diseases in Africa has been overlooked. This initiative presents an opportunity to build global solidarity, bringing experts to Africa to share knowledge. A grant application for the Global Eye Genetics Consortium (GEGC) congress in Africa is planned, alongside detailed symposium planning focused on "Prevention of Blindness in Africa."

Africa's Role in Future Meetings: Plans will be made to ensure active participation of African countries in future meetings and collaborations.

The Division of Human Genetics at the University of Cape Town (UCT), led by Professor Raj Ramesar, will coordinate the initiative. UCT has the necessary infrastructure and expertise in genetic research, especially in genomics of inherited eye diseases. The National Eye Institute (NEI) has an existing Memorandum of Understanding (MOU) with the Nigerian Eye Clinic (NEC), facilitated by Dr. Musa Kana in Nigeria.

GEGC at the African Ophthalmology Council Congress 2024, July 28th

The Global Eye Genetics Consortium (GEGC) hosted a pivotal plenary session at the African Ophthalmological Council (AOC) annual meeting, with the goal of advancing genetic research on eye diseases across Africa. Experts in ophthalmic genetics, public health, and clinical sciences from both Africa and worldwide gathered to tackle the challenges in understanding and treating inherited eye disorders common in African populations. The session emphasized the need for collaborative research, capacity building, and applying genomic knowledge in clinical settings. Key areas of focus included translational research, early diagnosis, treatment options, and the role of policy in enhancing genetic services. Additionally, the plenary underscored the importance of building regional partnerships and tapping into global expertise to create lasting solutions for Africa's eye health challenges.

GEGC at the Rwanda International Institute of Ophthalmology

The leadership of GEGC made a visit to the Rwanda International Institute of Ophthalmology (RIIO). RIIO was created in response to the gaps in the provision of eye services in Rwanda. It was established in 2011 by two ophthalmologists living in Kigali, Dr. John Nkurikiye and Prof. Wanjiku 'Ciku' Mathenge (President of Africa Ophthalmology Council). The GEGC team visited the Institution's two operational sites in Kigali. The RIIO head office has its secretariat, clinics, theatre, pharmacy, classroom, optometric unit and other supporting units. A second site for community ophthalmology care and training is sited within in a district hospital in another location in Kigali. During the visit the leadership of RIIO indicated that RIIO will be willing to collaborate with GEGC in Rwanda and the sub-region.

GEGC at World Ophthalmology Congress

On August 16th the Global Eye Genetics Consortium hosted a scientific session at the World Ophthalmological Congress, featuring prominent speakers on key topics in genetic eye research. Dr. Ken Nischal opened the session with a talk titled "What Should the Ophthalmologist and Researchers Know About Genetic Implications of Eye Diseases?" He emphasized the importance of understanding genetic factors in the diagnosis and treatment of various eye conditions, advocating for stronger integration of genetics into clinical practice. Dr. Laryssa Huryn presented on "Inherited Eye Diseases: Clinical Perspectives," offering insights into the clinical management of genetic eye disorders. She highlighted advancements in diagnosis and the need for personalized treatment approaches for patients with inherited eye conditions. Dr. Vikas Khetan discussed the "Genetics of Retinoblastoma," detailing the genetic underpinnings of this childhood cancer. He addressed current research on gene mutations linked to the disease and how genetic testing plays a critical role in early detection and family counselling. Dr. Manal Bouhaimed explored "Opportunities for Genetics Eye Research in the Middle East," outlining the unique genetic landscape of the region. She underscored the potential for regional collaborations and research to address hereditary eye conditions prevalent in Middle Eastern populations. Dr. Gyan Prakash concluded with a presentation on "International Collaborations in Genetic Eye Research," stressing the importance of global partnerships to advance genetic research in ophthalmology. He discussed the role of shared resources, expertise, and data in addressing global eye health challenges.

The session ended with a lively Q&A and discussion, where speakers and attendees engaged on the importance of integrating genetic insights into clinical practice and fostering international research collaborations.

Upcoming events

Meet Our Members

Jibran Mohamed-Noriega is a professor in the Department of Ophthalmology at the Faculty of Medicine, University Hospital, Autonomous University of Nuevo León. His career began with a medical degree from the same institution, where he also completed his specialisation in Ophthalmology at the Department of Ophthalmology. To deepen his expertise, Jibran undertook advanced training in Glaucoma at Moorfields Eye Hospital NHS Foundation Trust and Paediatric Glaucoma at Great Ormond Street Hospital for Children NHS Foundation Trust. Further solidifying his expertise, he earned a PhD from the UCL Institute of Ophthalmology.

Jibran has participated in significant studies such as "From Conventional Angle Surgery to 360-Degree Trabeculotomy in Pediatric Glaucoma," which explores advancements in surgical techniques for treating paediatric glaucoma. He also participated in the publication of "Risk Factors for Visual Field Deterioration in the United Kingdom Glaucoma Treatment Study," shedding light on factors impacting visual field decline. Additionally, he has explored the "Relationship of Neutrophil-to-Lymphocyte and Platelet-to-Lymphocyte Ratio With Visual Acuity After Surgical Repair of Open Globe Injury," offering insights into post-surgical recovery metrics. Jibran has also contributed to the field with publications focussed on practical clinical information such as "Neovascular Glaucoma: Prevention and Treatment", which addresses strategies for managing this complex form of Glaucoma, while "Defining and Diagnosing Glaucoma: A Focus on Blindness Prevention" emphasizes the importance of early detection and prevention of patients with an unequivocal diagnosis of glaucoma. Finally, he is part of the ophthalmo-genetics clinic of the University Hospital of the Autonomous University of Nuevo León where he participates in clinical care, teaching and research. Jibran has a strong commitment to advancing the field by promoting research, enhance patient care, and teach future generations of doctors and ophthalmologist, demonstrating his dedication to improving eye health.

Dr (Prof) Zia Chaudhuri, MS, DNB, MNAMS, FRCS (Glasg), FICO, PhD (Genetics), FAMS is a Professor of Ophthalmology at Lady Hardinge Medical College (LHMC) and Associated Hospitals, Faculty of Medical Sciences, University of Delhi, and Dr Ram Manohar Lohia (RML) Hospital and Atal Bihari Vajpayee Institute of Medical Sciences (ABVIMS), New Delhi, India with sub-speciality training and work experience in ophthalmology in strabismus, pediatric ophthalmology, neuro-ophthalmology, cranio-facial abnormalities, nystagm us, and ophthalmic genetics as well as teaching experience of 25 years. She currently heads the Department of Ophthalmology at the Faculty of Medical Sciences, University of Delhi. She has over 150 publications to her credit and has edited 07 books in ophthalmology. Dr Chaudhuri has undergone training in high-resolution surface-coil dynamic functional MRI at the Stein Eye Institute, University of California Los Angeles (UCLA), USA under the mentorship of Prof Joseph L Demer (2011-12) as well as performed her doctoral work in the genetics of strabismus at the Department of Genetics, University of Delhi South Campus, New Delhi, India under the supervision of Prof BK Thelma. High resolution surface coil MRI in different ophthalmic conditions as well as contemporary discovery genomics approaches for identifying novel genes for strabismus comprise Dr Chaudhuri's current research work. She has been the principal investigator of 05 scientific projects funded by the Ministry of Science and Technology, Government of India through the Department of Biotechnology (DBT) and Science and Research Engineering Board (SERB). Dr Chaudhuri had been a MD and PhD mentor with the University of Delhi and has supervised the research of 13 candidates (2 current) so far. She has also

been a mentor with the ARVO Leadership programmes for the past 5 years and has mentored 08 mentees so far (1 current). She has been an ARVO AMPC member for the EY section (2017-20) and is currently a member of the ARVO MIT committee (2022-25) as well as a member of Community for Sustainable Vision Research working group of the ARVO starting 2023. Dr Chaudhuri is the current editor-in-chief of the journal, Strabismus and is on the editorial board of BMC Ophthalmology (Pediatric Ophthalmology and Strabismus) and Ophthalmology and Therapy (Neuro-ophthalmology and pediatric ophthalmology) and reviews extensively for many high impact ophthalmology journals, including the 03 ARVO journals.

Prof. Govindasamy Kumaramanickavel is a distinguished researcher in ocular genomics with a rich and diverse background in medical genetics. Prof. Kumaramanickavel graduated in medicine and MD (physiology) from the University of Madras, India, and furthered his expertise through training at renowned institutions such as the National Eye Institute, USA, and University of Otago, New Zealand. He has been pivotal in pioneering gene mapping, mutational screening, and association studies for complex and Mendelian ophthalmic diseases. With a significant focus on childhood blindness, glaucoma and diabetic retinopathy, his epidemiological and genetic projects have involved over 35,000 subjects. His career spans leadership roles, including Deputy Director of Research at Sankara Nethralaya and Research Director at the Aditya Jyot Eye Hospital, Mumbai, India, where he provided genetic counseling to 15,000 patients and taught medical and ophthalmic genetics. Currently, as Research Director at Narayana Nethralaya, Bangalore, India. He continues to drive basic and applied research supported by funding from prestigious global institutions. Prof. Kumaramanickavel's contributions extend across 134 peer-reviewed publications, including notable work in Nature Genetics, augmenting his impact on advancing ocular genomic research and patient care worldwide.

Dr. Asma Ali Khan is a highly accomplished geneticist with a focus on human molecular genetics, particularly in the context of autosomal recessive genetic disorders such as Hearing Impairment and Intellectual Disability in consanguineous families in Pakistan. With a doctoral degree (PhD) from the University of Lorraine, France, in Life and Health Sciences, specializing in Human Molecular Genetics, Dr. Khan brings a wealth of expertise to her role as Assistant Professor at the Centre of Excellence in Molecular Biology (CEMB) since 2013.

Dr. Khan's research revolves around identifying novel genetic mutations associated with various genetic disorders. Her thesis research focused on the extended mutational spectrum of IL1RAPL1 and MBD5 genes, employing a range of techniques including aCGH microarray, qPCR, FISH, Western Blotting, and Sanger sequencing. Her current work encompasses the molecular and genetic characterization of syndromic and non-syndromic hereditary disorders, with a particular emphasis on Perrault syndrome and rare disorders with hearing impairment. A notable aspect of Dr. Khan's research is her expertise in Whole Exome Sequencing data analysis, utilizing the latest bioinformatics tools to unravel the genetic basis of inherited diseases. Her contributions have resulted in publications in international peer-reviewed journals, demonstrating the significance of her findings to the broader scientific community. Dr. Khan's dedication to advancing genetic research is further evidenced by her involvement in funded projects such as the NRPU project titled "Identification and Characterization of Genes Responsible for Hearing Impairment In Pakistani Population." Additionally, she plays a crucial role in mentorship, having supervised numerous M.Phil

and Ph.D. students under her guidance. Overall, Dr. Asma Ali Khan's work underscores her commitment to improving our understanding of genetic disorders, particularly within the Pakistani population, and her contributions have the potential to inform future diagnostic and therapeutic strategies for these conditions.

Dr. Sunita Mohan, a distinguished Vitreo-retinal surgeon and epidemiologist dedicated to advancing our understanding of retinal diseases, particularly diabetic retinopathy, within Mumbai's urban slums. Dr. Mohan's research focuses on elucidating the prevalence and risk factors associated with diabetic retinopathy in this unique demographic, alongside investigating behavioral patterns concerning these diseases. She also explores the efficacy of various imaging technologies for diagnosing retinal conditions.

In addition to her pioneering research, Dr. Mohan excels in clinical practice, adeptly identifying inherited retinal diseases and managing a spectrum of retinal disorders such as diabetic retinopathy, retinal detachment, and age-related macular degeneration. Her comprehensive approach integrates cutting-edge diagnostics with compassionate patient care. Dr. Mohan's contributions to ophthalmic epidemiology are underscored by numerous publications and accolades, recognizing her impactful work in advancing our knowledge of retinal health. Her dedication and expertise have earned her several prestigious awards, affirming her status as a leader in the field. Beyond her clinical and research endeavors, Dr. Mohan is committed to enhancing community health through her proactive engagement with urban slum populations, advocating for early detection and effective management of retinal diseases. Her efforts reflect a profound commitment to improving eye care accessibility and outcomes for underserved communities. Dr. Sunita Mohan stands as a beacon of excellence in Vitreo-retinal surgery and epidemiology, embodying a steadfast commitment to innovation, patient well-being, and public health advancement.

Dr. Radhika Tandon is a distinguished ophthalmologist with a robust academic background and extensive professional experience. Currently serving as Professor of Ophthalmology at Dr. Rajendra Prasad Centre for Ophthalmic Sciences, AIIMS, she has been involved in various capacities since joining as a Resident in Ophthalmology in 1987. Dr. Tandon holds multiple degrees including MD and DNB in Ophthalmology from AIIMS, and fellowships from the Royal College of Ophthalmologists, London, and the Royal College of Surgeons, Edinburgh.

Her career highlights include significant contributions to eye care, education, and research. She has chaired the National Eye Bank and Low Vision Services at RPC, AIIMS, and served as the Immediate Past President of the Eye Bank Association of India. Dr. Tandon's expertise spans diverse areas such as cataract surgery, femtosecond laser techniques, corneal transplants, genetics of eye disorders, epidemiology and stem cell therapy. She has received numerous awards, including Gold Medals from prestigious institutions, and has been actively involved in national and international ophthalmic organizations. Dr. Tandon's leadership extends beyond clinical practice; she has chaired committees on climate change and eye health, and has been a member of expert panels for eye bank accreditation and corneal blindness eradication. Her commitment to advancing ophthalmology is evident through her roles in curriculum development, advisory committees, and as a reviewer for leading journals in the field. Dr. Radhika Tandon continues to make significant strides in the field of ophthalmology, enhancing both patient care and education globally.

www.gegc.org

Ophthalmic Genetics News

Around the World

Great Promise from Investigational Gene Therapy (ATSN-101) Trial

A groundbreaking gene therapy, ATSN-101, has shown promising results in treating a rare form of congenital blindness. The early-phase clinical trial, funded by Atsena Therapeutics, investigated the gene therapy, ATSN-101, in 15 patients with Leber congenital amaurosis (LCA) 1, which is a type of LCA caused by biallelic mutations in the GUCY2D gene. All 15 enrolled patients with genetically confirmed LCA1 receivedunilateral subretinal injections to determine the safety and preliminary efficacy of ascending doses of ATSN-101. Thirteen of the patients were treated at the Scheie Eye Institute. Two were treated at the Oregon Health & Science University (OHSU) Casey Eye Institute, under the guidance of Paul Yang, MD, PhD, and Andreas Lauer, MD. ATSN-101 is a recombinant AAV5 gene vector that contains human GUCY2D complementary DNA, a key enzyme in photoreceptors. The trial used escalating dosing in three cohorts of adults (N = 9): A low dose, 1 × 1010 vector genomes per eye (vg/eye); a middle dose, 3 × 1010 vg/eye; and a high dose, 1 × 1011 vg/eye. After the high dose showed preliminary evidence of vision improvement, an additional six patients — three adults and three children — were given the high dose. According to Cideciyan, Senior Study Author and codirector of the Center for Hereditary Retinal Degenerations at the Scheie Eye Institute at the University of Pennsylvania in Philadelphia, the main efficacy outcome was not visual acuity but change in ability for the photoreceptors to catch light. Twelve-month results showed the highest dose treatment was linked to an improved ability to see light and fine details; an effect translating into almost two lines on the Early Treatment Diabetic Retinopathy Study (ETDRS) chart which could be the difference between 20/200 vision, the threshold for legal blindness, and 20/125 vision. According to Darius Moshfeghi, MD, chief of the Retina Division at Byers Eye Institute at Stanford University in Stanford, California, and a pediatric retina specialist, this is the first time you show a dramatic change in vision function. These findings offer hope for patients with inherited retinal diseases and suggest that gene therapy could be a viable treatment option for a wider range of visual impairments.

Sources: (1) Medscape Ophthalmology. Gene Therapy Shown to Improve Vision in Congenital Blindness (medscape.com)
(2) Atsena Therapeutics – Atsena Therapeutics Announces 12-Month Safety and Efficacy Data from Phase I/II Clinical Trial of ATSN-101 in LCA1 Published in The Lancet (atsenatx.com)

Case Western Receives Grant for Oral Drug Targeting Inherited Retinal Disease

Case Western Reserve University (CWRU) has been awarded a $1.5 million grant over 3 years to explore a potentially groundbreaking treatment for inherited retinal diseases (IRD). With limited existing options, this research focuses on a potential breakthrough oral medication that could address a wide range of IRD, regardless of the specific genetic cause.

Dr. Shigemi Matsuyama, leading the research, aims to develop a therapeutic for RP patients who are suffering from the fear of blindness. Prior research by his team yielded promising results with a series of novel orally-active cell-death inhibitors (Cytoprotective Small Compounds) which work by blocking the activation of Bax, a protein that contributes to cell death, and in which the lead compound effectively prevented retinal cell death and vision loss in four mouse models.

The grant will support further development of this drug, including safety assessments and formulation testing for both oral and eye drop delivery. The goal is to advance this therapy through clinical trials and potentially bring it to patients suffering from IRD.

Interestingly, the research team sees potential beyond IRD. They believe Bax-related cell death plays a role in other neurodegenerative diseases like glaucoma and Alzheimer's. This opens exciting possibilities for broader applications of this therapeutic approach.

The Foundation Fighting Blindness, who provided the grant, expressed their commitment to supporting such research with the potential to significantly impact the lives of people affected by inherited retinal diseases.

Sources:
(1) Case Western Reserve University receives $1.5M grant from Foundation Fighting Blindness to test possible new treatment for inherited retinal disease. EurekAlert! Accessed July 12, 2024. https://www.eurekalert.org/news-releases/1050251
(2) Case Western Reserve University receives $1.5M grant for oral drug trial to prevent blindness in RP patients (news-medical.net)
(3) Case Western Reserve University receives $1.5 million grant from Foundation Fighting Blindness to test possible new treatment for inherited retinal disease (ophthalmologytimes.com)

Abeona and Beacon Therapeutics Partner on Ophthalmic Gene Therapy

Abeona Therapeutics has teamed up with Beacon Therapeutics to explore the potential of Abeona's AAV204 capsid for developing gene therapies for eye diseases. Beacon will assess AAV204's ability to reach different layers of the retina and its suitability for treating various retinal conditions. Under the agreement, Beacon has the right to evaluate AAV204 for a year and may obtain a non-exclusive license to use it in up to five gene therapy targets. Abeona will receive upfront payments and additional payments based on development milestones. AAV204 is a unique virus capsid developed by Abeona from the University of North Carolina. Studies have shown that it can effectively target the macula, optic nerve, and both inner and outer retina in animal models, such as mice and non-human primates. The AIM capsid library, which includes AAV204, is a collection of new virus serotypes designed to deliver genetic material to specific tissues involved in genetic diseases.

Source:Abeona Therapeutics and Beacon Therapeutics Announce Non-Exclusive Agreement for Beacon to Evaluate Therapeutic Potential of Abeona's Patented AAV204 Capsid for Select Ophthalmology Indications. Press Release; July 11, 2024. Accessed July 11, 2024.
https://www.beacontx.com/news-and-events/abeona-therapeutics-and-beacon-therapeutics-announce-non-exclusive-agreement-for-beacon-to-evaluate-therapeutic-potential-of-abeonas-patented-aav204-capsid-for-select-ophthalmology-indications/

Answers

1) Retinitis Pigmentosa
2) Leber Hereditary Optic Neuropathy
3) Stargardt Disease
4) Alport Syndrome
5) Ectopia Lentis
6) Oculocutaneous Albinism
7) Autosomal Dominant
8) Morning Glory Syndrome
9) Axenfeld-Rieger Syndrome
10) Craniosynostosis

Editorial Members: Dr. Shailja Tibrewal, Ms. Ria Sachdeva, Dr. Purvasha Narang, Dr Onochie Okoye, Ms. Riya Pal
Designed By: Alpana Singh