

# *1st International Conference of Global Eye Genetics Consortium (GEGC), Mumbai, India*

Registration site: [www.gegc.org](http://www.gegc.org)

**Nov 19 – 20, 2022**

## **Scientific Program**

### **Summary**

The **1st International Conference** will focus on global partnerships to advance research into the genetics of eye diseases and research programs being carried out by the GEGC members and collaborators across thirty plus countries. The conference is being organized in Mumbai by the Maharashtra Ophthalmological Society and the India Chapter of GEGC. The USA's National Eye Institute (NEI) -National Institutes of Health (NIH), and the Japan's National Institute of Sensory Organs- National Hospital Organization at Tokyo Medical Center entered into an agreement to support vision research collaboration in 2014 to encourage collaborative research programs in genetic eye research and bring researchers from around the world on one common platform. The initiative and combined efforts have led to the establishment of the Global Eye Genetics Consortium (GEGC) which has over two hundred members in more than thirty countries. The 1<sup>st</sup> International Congress will highlight the achievements and will address research gaps such as the need to collect and openly share data from underrepresented populations. It will also address the lessons learned from GEGC activities and eye GENE®, primarily a USA based network led by NEI, that has built a patient registry, biorepository, and genotype-phenotype database. The event is expected to bring together experts from the U.S., Japan, India, China, Australia, the Netherlands, Brazil, South Africa, Turkey, and other countries.

- **1<sup>st</sup> GEGC International Program Organizing Chairman**

**Prof. Dr. S. Natarajan,**  
*[Awarded Padmashree by the President of India]*  
*Secretary General GEGC and Organizing Chairman,*  
*1<sup>st</sup> International GEGC Conference, Mumbai, India.*

- **Co-ordinator, 1<sup>st</sup> International GEGC Conference – Hybrid**  
**Dr. Karobi Lahiri Coutinho**

- **1<sup>st</sup> International GEGC Conference Coordination Team from**
- **MOS Office Bearers :**  
**Dr. Santhosh Bhide** **Dr. Anagha Heroor**  
**Dr. Vivek Motewar** **Dr. Vardhaman Kankaria**
- **Kamala Sundaram Foundation:**  
**Mr. Joseph Thomas**

**Aditya Jyot Eye Hospital:**

**Mrs. Shubha Urankar**

**Ms. Samrudhi Dani**

**Mr. Samir Bhekare,**

**Ms. Meena Visaria,**

**Ms. Charishma Jonnadula,**

**Ms. Shruti Dudwadkar.**

**Mr. Benjamin Prasad,**

**Mr. Prashant Jadhav.**

**Nov 19, 2022**

**Session: 01**

<b>Time</b>	<b>Program</b>
<b>09:00 - 10:30</b>	<i>Opening Session</i>  <i>Chairman: Dr. Gyan "John" Prakash</i> <i>Co-chairman: Dr. Umang Mathur</i> <i>Co-chairman: Dr. Phiroze Patel</i> <i>Moderator: Dr. Ajay Dudhani</i> <i>Convener: Dr. Jaydeep Walinjar</i>
<b>09:00 - 09:15</b>	<b>Ophthalmic Genetics in India-Padmashree Prof. Dr. S. Natarajan</b>
<b>09:15 - 09:30</b>	<b>Dr. Michael Chiang, Director, National Eye Institute, USA – Welcome Note</b>
<b>09:45 - 10:00</b>	<b>Dr. Gyan "John" Prakash USA Founding President, and current Patron of GEGC</b> <b>Title: International Research Collaborations are needed if You Love Your Eyes</b>
<b>09:30 - 09:45</b>	<b>Dr. Takeshi Iwata, Head of JEGC &amp; President of AEGC/GEGC, National Hospital Organization Tokyo Medical Center, Japan [JEGC, AEGC, and GEGC: fruitful scientific outputs from three research consortiums]</b>
<b>10:00 - 10:15</b>	<b>Dr. Umang Mathur GEGC program in India</b>
<b>10:15 - 10:30</b>	<b>Prof. I. C. Verma [Molecular Landscape of OCA (oculocutaneous albinism) in INDIA]</b>

**Session: 02**

<b>Time</b>	<b>Program</b>
<b>10:30 – 12:00</b>	<i>Chairman: Dr. P. Sundaresan</i>

	<p><i>Chairman: Dr. Keiki Mehta</i>  <i>Co-Chairman: Dr. Kumaramanickavel</i>  <i>Co-Chairman: Dr. Mamta Muranjan</i>  <i>Moderator: Dr. Vardhaman Kankaria,</i>  <i>Convener: Dr. Susha Sugathan</i></p>
10:30 – 10:45	<p><b>Chairman Dr.P.Sundaresan</b>  <b>Title: Translational Genomics of Eye Genetics</b></p>
10:45 – 11:00	<p><b>Co-Chairman Dr. Kumaramanickavel</b>  <b>Title: Precision Medicine in Ophthalmic Clinical Practice</b></p>
11:00 – 11:15	<p><b>Dr. Sarath Babu –</b>    <b>“Identification and Characterization of Novel Genotype Associated with Clinical Grades of Stargardt Patients”.</b></p>
11:15 – 11:30	<p><b>Dr. Rajiv Raman</b>  <b>[Genetics of Diabetic Retinopathy in India]</b></p>
11:30 – 11:45	<p><b>Padmashree Prof. Dr. S. Natarajan</b>    <b>Vitreous Surgery for Gene Therapy</b></p>
11:45 - 12:00	<p><i>Coffee break to prepare for Inauguration</i></p>

<b>Time</b>	<b>Program</b>
12:00 - 13:00	<b>Inauguration</b>

<b>Time</b>	<b>Program</b>
13:00 - 14:00	<b>Lunch</b>

### Session:03

<b>Time</b>	<b>Program</b>
14:00 - 16:00	<p><i>Ophthalmic Genetics for Practicing Ophthalmologists-Case based Discussion &amp; FAQs (MOS-BOA) Session</i>  <b>chairman:</b>  <i>Dr.RumiJehangir</i>  <b>Chairman: Dr. Preetam Samant</b>  <b>Co-Chairman: Dr. Santosh Bhide</b>  <b>Co-Chairman: Dr. Anand Subramaniyam</b>  <b>Moderator: Dr. Siddharth Kesarwani</b>  <b>Convener: Dr. Sonal Lakdawala Jadhav</b></p>
14.00 – 14.10	<p><b>Dr. Parveen Sen</b>  <b>[Phenotype and Genotype co-relation in RP]</b></p>
14.10 – 14.18	<p><b>Dr. Priti Kamdar</b>  <b>[Glaucoma Genetics and the Clinician]</b></p>
	<p><b>Dr. Mamta Muranjan</b></p>

14.18 – 14.28	<b>[Phenotypes to Diagnosis: Identification of Pediatric Syndromes]</b>
14.28 – 14.36	<b>Dr. Shruti Bajaj</b> <b>[Beyond the karyotype: New Age Genetic Tests]</b>
14.36 – 14.44	<b>Dr. Karobi Lahiri</b> <b>[Is it roper? -Case Based]</b>
14.44 – 14.52	<b>Dr. Paritosh Kamdar</b> <b>[Gene Testing in a private vitreoretinal practice]</b>
14.52 – 15.00	<b>Dr. Nikhil Nasta</b> <b>[Ocular Albinism] – Giving them a chance to see better</b>
15.00 – 15.10	<b>Dr. krishna kumar/Jyotrimay Biswas</b> <b>[Primary VR Lymphomas and MYD 88L265 Mutation]</b>
15.10 – 15.18	<b>Dr. Himika Gupta</b> <b>[Retinoblastoma Genetics – what clinician should know – case based]</b>
15.18 – 15.26	<b>Dr. Mayur Morekar</b> <b>[Lhon- The Genetic Basis]</b> <b>Lebers hereditary optic neuropathy – normal therapeutic molecules &amp; gene therapy – A near future approach</b>
15.26-15.34	<b>Dr. Ajay Dudani</b> <b>A Bouquet of Weil Marchesanis Syndrome (Cases )</b>

15.34-16.00	<p><b><u>Panel discussion</u></b>  <b><u>Panelists</u></b>  <b><u>-all Speakers plus</u></b>  <b><i>Padmashree Prof. Dr. S. Natarajan</i></b>  <b><i>Dr. Saumil Sheth</i></b>  <b><i>Dr. Nipa Thakkar</i></b>  <b><i>Dr. Anagha Heroor</i></b>  <b><i>Dr. Mamta Muranjan</i></b>  <b><i>Dr. Nishikant Borse</i></b></p>
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**Session:04**

<b>Time</b>	<b>Program</b>
16:00 - 18:00	<p><b><i>Women in Ophthalmology Genetics &amp; Research</i></b></p> <p><b><i>Chairperson: Prof. Bhavna Chawla</i></b>  <b><i>Co-Chairperson: Dr. Usha Johari</i></b>  <b><i>Co-Chairperson:Dr. Karobi Lahiri</i></b>  <b><i>Moderator: Dr. Mamta Muranjan</i></b>  <b><i>Convener: Dr. Shruti Bajaj</i></b>  <b><i>Co convenor: Dr. Nipa Thakkar</i></b></p>
16.00-16.15	<p><b>Prof. Martine Jager, (<u>keynote speaker</u>)</b>  <b>The netherlands</b>  <b>Title: Eye Color and Eye Melanoma.</b></p>
16.15-16.24	<p><b>Dr Jesse Berry</b>  <b>Liquid Biopsy Application for Retinoblastoma - From</b>  <b>Diagnosis to Prognosis of Ocular Salvage</b></p>
16.24-16.39	<p><b>Dr. Shilpa Bisht, Johns Hopkins, USA</b>  <b>[The role of next generation sequencing in retinoblastoma]</b></p>

16.39-16.	<p><b>Dr. Shruti Bajaj</b>  <b>Title: There is an ‘I’ in Genetics]</b>  <b>When and why should an ophthalmologist think of a geneticist</b></p>
17.00-17.15	<p><b>Ms. Kerry Goetz, USA</b>  <b>Title: [‘Casting a wide net to engage participants for rare disease registries’]</b></p>
17.15- 17.25	<p><b>Dr.Prachi Aghashe</b>  <b>[Case of Cutis Marmorata Telangiectasia Congenita]</b></p>
17.25-17.40	<p><b>Dr. Ratna Puri</b>  <b>[Eye Genetics in Inborn Errors of Metabolism]</b></p>
17.40-17.55	<p><b>Dr.Sunita Birjania</b>  <b>Congenital Cataracts - Genetic and Non Genetic Causes)</b></p>
17.55- 18.00	<p><b>Closing Remarks</b></p>

**NOV 20,2022**

**Session: 01**

<b>Time</b>	<b>Program</b>
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<b>09:00 – 11:00</b>	<i>Genetic Research Worldwide</i> <i>Chairman: Dr. Takeshi Iwata</i> <i>Co-Chairman: Dr. Paul Baird</i> <i>Co-Chairman: Dr. Calvin Pang</i> <i>Moderator: Dr. Hitesh sharma</i> <i>Convener: Dr.Ashish Ahuja</i>
<b>09:00 – 09:15</b>	<b>Dr. Janey Wiggs</b>  [International Consortia Providing the Next Milestone of Research Data]
<b>09:15 – 09:30</b>	<b>Dr. Paul Baird</b>  [Technical considerations in GEGC–Now & Future]
<b>09:30 – 09:45</b>	<b>Dr. Calvin pang</b> [Associations of Gene Polymorphisms and lifestyle with Anisometropia in Children]
<b>09:45 – 10:00</b>	<b>Dr. Mohita Sharma</b> [Rare Eye Disease – Project RED]
<b>10:00 – 10:15</b>	<b>Dr. Robert Hufnagel</b>  [Clingen: Clinical Genome Resource]
<b>10:15 – 10:30</b>	<b>Dr. Surabhi Kandaswamy</b>  [NGS Application in diagnosing Inherited Retinal Dystrophies]
<b>10:30 – 10:45</b>	<b>Dr Parag Tamhankar - Clinical Geneticist DM</b> Medgenome labs [Interesting Cases in Ophthalmic Genetics]
<b>10:45 - 11:00</b>	<b>Dr Parag Tamhankar - Clinical Geneticist DM</b> Medgenome labs [Interesting Cases in Ophthalmic Genetics]  <i>(Refreshments will be served alongside)</i>

## Session:02

<b>Time</b>	<b>Program</b>
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<p><b>11:00 - 13:00</b></p>	<p><i>Young Ophthalmologists session (YOs)</i></p> <p><i>Chairman: Dr.Shailja Tibrewal</i>  <i>Co-Chairman: Dr. Fulya Tuncay</i>  <i>Co-Chairman: Dr. Nishikant Borse</i>  <i>Moderator: Dr.Nayana Poddar</i>  <i>Convener: Dr.akshay nair</i></p>
<p><b>11.00-11.15</b></p>	<p><b>KEYNOTE LECTURE</b></p> <p><b>Dr Anne Slavotinek, Professor, Cincinnati Childrens hospital and medical center</b></p> <p><b>Syndromic associations of Microphthalmos, anophthalmos and coloboma</b></p>
<p><b>11:15 – 11:25</b></p>	<p><b>Dr. Shailja Tibrewal</b></p> <p><b>[What did I miss? Genetic Implications of common and uncommon Pediatric Eye Disorders]</b></p>
<p><b>11:25 – 11:35</b></p>	<p><b>Dr. Fulya Tuncay</b></p> <p><b>[Approach to Genetic Eye Disease in Turkey]</b></p>
<p><b>11:35-11:45</b></p>	<p><b>Dr. Meenakshi Lallar</b></p> <p><b>[Targeted Genetic Testing for Ocular Disorders]</b></p>
<p><b>11:45 – 11:55</b></p>	<p><b>Dr. Nazmul Haque Kallol</b></p> <p><b>[A Genomic and Molecular Biological Approach to find complex Mutations in Retinal Disease]</b></p>
<p><b>11:55-12:05</b></p>	<p><b>Dr. Ria Ratna</b></p> <p><b>[The Concept of Genetic Counselling]</b></p>
<p><b>12:05-12:15</b></p>	<p><b>Dr. Srijana Adhikari</b></p> <p><b>[Genetic Testing in Pediatric Ophthalmology: The Challenges in Developing World Setup]</b></p>
<p><b>12:15-12:25</b></p>	<p><b>Dr. Deepika C Parameswarappa</b>  <b>[ Evolution of CERKL Gene Related Autosomal Recessive Retinitis Pigmentosa in a Single Family</b></p>
<p><b>12:25-12:35</b></p>	<p><b>Dr. Muna Bhende</b>  <b>STICKLERS SYNDROME</b></p>



12.35-12.45	Lea Kovac  Reversible vision loss mimicking LHON in a patient with cystic fibrosis
12.45-1.00	Q&A Discussion

<b>Time</b>	<b>Program</b>
13:00 - 14:00	Lunch

### Session:03

Time	Program
14:00 - 15:45	Genetic Analysis for Eye Disease  <i>Chairman: Dr. Takeshi Iwata &amp; Padmashree Prof. Dr. S. Natarajan</i> <i>Co-Chairman: Dr. Paul Baird</i> <i>Co-Chairman: Dr. Calvin Pang</i> <i>Moderator: Dr. Kirit Mody</i>  <i>Convener: Dr. Minu Ramakrishnan</i>
14:00 - 14:20	Dr. Umang Mathur / Dr. Ria Ratna & Padmashree Prof. Dr. S. Natarajan  [Unique population in India for Vision Research]
14:20 - 14:35	Dr. Calvin Pang  [Unique population in China for Vision Research]
14:35 - 14:50	Dr. Namrata Sharma – “Genetics of Keratoconus”
14:50 - 15:05	Padmashree Prof. Dr. S. Natarajan [Advantages of 27ga Vitrectomy]

15:05 - 15:25	<b>Dr. Tania Barragan</b>  [Genetic profile & Mutational Spectrum of Leber Congenital Amaurosis in a Mexican Cohort]
15:25 _ 15:45	<b>Padmashree Prof. Dr. S. Natarajan</b> [Vitreous Surgery in Gene Therapy] – <b>Ozurdex Allergan Session</b>
15:45 - 16:00	<b>Dr. Divyansh Mishra</b>  [ Melanoma Genetics] <i>(Refreshments will be served alongside)</i>

#### Session:04

<b>Time</b>	<b>Program</b>
16:00 – 18:00	<i>Vision Research in India</i>  <i>Chairman: Dr. kirit mody</i> <i>Co-Chairman: Padmashree Prof. Dr. S. Natarajan</i> <i>Co-chairman: Dr.anuprita ghosh</i> <i>Moderator: Dr.Hitesh Sharma</i> <i>Convener: Dr. Chhaya shinde</i> <i>Co-convener: Dr. Mayur Morekar</i>
16:00 – 16:15	<b>Dr. Anuprita Ghosh –</b>  [ Genetic Landscape and Genetic Counselling in Inherited Retinal Diseases Patients]
16:15 – 16:30	<b>Dr. Chitra Gopinath –</b> <b>AAV Medicated Gene Therapy for Retinal Disease</b>
16:30 – 16:45	<b>Dr. Shridhar Sivasibhu</b> <b>Sr principal scientist</b> <b>CSIR Institute of Genomics and Integrated Biology,</b> <b>Delhi</b>  [Title: Personal Genomes to Precision Medicine: Experiences from India]
16:45 – 17:00	<b>Dr. Choi Mun Chan</b>  [Genomic testing in Macula Dystrophy : The Singapore National Eye Centre Experience]

<b>17:00 – 17:10</b>	<b>Padmasree Prof. Dr. S. Natarajan [Luxturna treatment for RPE65 Associated Retinal Dystrophy]</b>
<b>17:10 – 17:25</b>	<b>AJEH Birac Team – Multicentric Registry on Clinical Inherited Retinal disorders</b>
<b>17:25 – 17:40</b>	<b>Dr. Sunita Birjania  Mitochondrial Disorders Involving the Eye- Lhon and Melas</b>
<b>17:40 – 17:50</b>	<b>Dr. Koumudi Godbole – Gender issues in Oculo Genetics</b>
<b>17:50 – 18:00</b>	<b>Ms. Afroz Patel – Low Vision aids for Inherited Retinal Diseases</b>