

## Welcome address by President of EAIRDs

Prof Kaoru Fujinami

Dear colleagues, dear friends, dear supporters,

I would like to extend a warm welcome on behalf of East Asia Inherited Retinal Disease Society (EAIRDs: <https://www.eairds.org/>) for the 1st EAIRDs symposium associated with 2nd International Ophthalmic Genetics Congress. I am honoured to have the opportunity to hold this symposium in Seoul, Republic of Korea, thanks to the huge support of Prof Se Joon Woo, Seoul National University Bundang Hospital, South Korea and colleagues in Republic of Korea.



Inherited retinal disease (IRD) is a leading cause of blindness both in children and adults of working age. Clinical treatment trials are ongoing in North America and Europe. However, at day-to-day ophthalmology clinics in East Asian countries, there remains little or no access to effective treatment, for patients since there have been no large-scale studies of well-characterised patient cohorts in the East Asian population. The objective of the East Asia IRD studies is to establish a large cohort of East Asian Patients with IRD in the international multicenter studies.

A number of internationally highly renowned clinicians/scientists from East Asian countries (Republic of Korea, Japan, China, Singapore, Australia) have followed our invitation to give an overview of their most recent cutting-edge research in national/international cohorts with IRD. Major contributions from brilliant young clinicians/scientists also help us to promote academic activities of IRD both inside and outside of East Asia.

I am grateful to all contributors whose efforts helped in establishing the program starting with the scientific committee (Prof. Se Joon Wee, Prof. Ruifang Sui, Prof. Shiyong Li, Prof. Bo Lei, Prof. Chan Choi Mun, and Prof. Fred Chen) and local organisation team. In creating the website, online membership registration, online congress registration, and online abstract submission, my very special thanks go to Dr Yu Yokokawa-Fujinami, National Institute of Sensory Organs, Tokyo, Japan.

Finally, without the generous support of sponsors, we would not have been able to establish this symposium setting. Therefore, I would like the attendees to communicate closely with people from the sponsors.

I sincerely wish all of you a pleasant stay, exciting discussions, beneficial encounters for your scientific success, and stimulating exchanges with old and new friends.



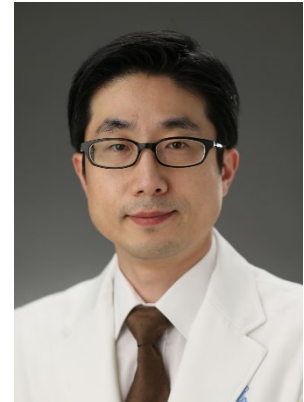
Kaoru Fujinami  
President of EAIRDs  
President of International Ophthalmic Genetics Congress  
Kaoru Fujinami, MD, PhD

Laboratory Head/Consultant Ophthalmic Surgeon  
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## Welcome address by Chair of EAIRDs 1<sup>st</sup> Symposium

Dear Friends and colleagues,

It is my great pleasure and honor to host the 1st symposium of East Asia Inherited Retinal Disease Society (EAIRDs). I would like to thank my friend, Pf. Fujinami, the president of EAIRDs, for his great contribution and enthusiastic effort to lead the EAIRDs to include such an extensive number of researchers and experts of IRD in East Asia. I would also appreciate all of you, the participants and members of EAIRDs, for your interest to our society and for the active participation in collaborative studies. Without your support, we could not have reached this point. I really thank you all with my heart and I also ask your help in our future work.



Compared to the western countries, research on IRDs in East Asia was quite delayed. Most of the clinical trials were conducted on western patients and the targets of gene therapy products are mostly aimed for western populations. At this time, it is imperative that researchers of IRD in East Asia should cooperate together to have knowledge and to gain the treatments of IRDs in East Asian patients. I firmly believe that this symposium is an important footstep for the future progress of the collaboration and achievement in the field of East Asian IRDs.

Recently, there are many young Korean ophthalmologists and investigators who work hard for the IRDs. More number of publications are coming from their studies to reveal the genetics and clinical features of Korean IRDs. I hope that by collecting the Korean researchers and by their collaborations through our EAIRD society, we can contribute to the knowledge and care of IRDs in East Asia. We feel that the venue of this 1st symposium has a great meaning and I, on behalf of Korean members, feel huge responsibility and expectations for the EAIRDs.

I welcome all of you to our hospital, Seoul National University Bundang Hospital in Korea, the venue of the 1st symposium of EAIRDs. Seoul is a beautiful city with exciting dynamics and delicious cuisines. I hope you enjoy Seoul and Korea during your stay.

I really look forward to welcoming you in the symposium.

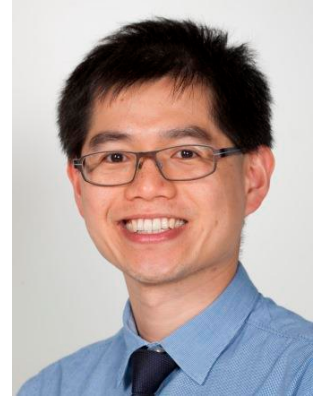


Se Joon Woo, MD, PhD  
Chairman of the 1st symposium of EAIRDs,  
Korean Leader of EAIRDs  
Associate Professor, Department of Ophthalmology,  
Seoul National University Bundang Hospital, Seoul, South Korea.

## Messages from national leader of Australia

Message from the Australian Lead for EAIRDs

Dr Fred Chen



Dear colleagues,

As the Australian lead for the East Asia Inherited Retinal Disease Society, I congratulate Professors Kaoru Fujinami and Se Joon Woo for organising the first EAIRDs symposium.

The proportion of Australians from East Asia ancestry is 6.5% and increasing. This percentage is one of the highest in the Western countries compared to only 1% in the United Kingdom and 2% in the United States of America. However, IRD patients with East Asian ancestry in the Australian Inherited Retinal Diseases Registry (AIRDR) is under represented (3.4%) and only 1.4% of those with a confirmed genetic diagnosis in our national registry had an East Asian background.

In this 1<sup>st</sup> EAIRDs symposium, Ms Enid Chelva from Australia, will provide an overview of the historical background and the current status of the AIRDR. We look forward to future collaboration with clinician-scientists within the EAIRDs so we can provide East Asian Australians with IRDs the same opportunities to be genetically characterized and enrolled into international observational and treatment trials as any Australian.

A handwritten signature in cursive script that reads "Fred Chen".

Fred Chen, MBBS PhD (UCL) FRANZCO  
Australian Lead of EAIRDs

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## **Principal investigator of EAIRDs**

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National University of Singapore, Singapore, Singapore.

**Fred Chen, MD, PhD**

Lions Eye Institute, Nedlands, Western Australia, Australia

## Program

8:50-09:00	Opening remark	Se Joon Woo (Seoul National University Bundang Hospital) Kaoru Fujinami (National Institute of Sensory Organs, Tokyo Medical Centre)
9:00-11:00	Session 1: Overview	Chairs: Kaoru Fujinami, Se Joon Woo
11:00-11:10	Coffee Break	
11:10-12:00	Session 2: Retinal cases and others	Chairs: Kaoru Fujinami, Se Joon Woo
12:00-12:05	Group photo	
12:05-13:30	Lunch/Board meeting (12:30-)	
13:30-14:45	Session 3: Occult Macular Dystrophy	Chairs: Kaoru Fujinami, Ruifang Sui
14:45-14:55	Coffee Break	
14:55-15:40	Session 4: Stargardt Disease	Chairs: Kaoru Fujinami, Shiyong Li
15:40-16:20	Session 5: Retinitis Pigmentosa and other IRDs	Chairs: Se Joon Woo, Bo Lei
16:20-16:25	Award ceremony	Kaoru Fujinami (National Institute of Sensory Organs, Tokyo Medical Centre)
16:25-16:30	Closing Remark	Se Joon Woo (Seoul National University Bundang Hospital)

## Program Overview

- 8:15- Registration
- 8:50-09:00 Opening remark  
Se Joon Woo (Seoul National University Bundang Hospital)  
Kaoru Fujinami (National Institute of Sensory Organs, Tokyo Medical Centre)

### **Session 1: Overview 9:00-11:00 Chairs; Kaoru Fujinami, Se Joon Woo**

- 09:00-09:10 Overview of EAIRDs. Kaoru Fujinami, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 09:10-09:20 East Asia inherited retinal disease studies: clinical and genetic characteristics of inherited retinal disease in Japanese population. Kaoru Fujinami, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 09:20-09:35 East Asia inherited retinal disease studies: clinical and genetic characteristics of inherited retinal disease in Korean population. Se Joon Woo, Department of Ophthalmology, Seoul National University Bundang Hospital, Seoul National University College of Medicine, Seongnam, South Korea.
- 09:35-09:50 East Asia Inherited Retinal Disease Studies: clinical and genetic characteristics of inherited retinal disease in a large Chinese cohort in Peking Union Medical College Hospital. Ruifang Sui, Peking Union Medical College Hospital, Beijing, China
- 09:50-10:05 East Asia inherited retinal disease studies: clinical and genetic characteristics of inherited retinal disease in a South West China population. Shiyong Li.
- 10:05-10:20 East Asia inherited retinal disease studies: clinical and genetic characteristics of inherited retinal disease in Henan Eye Institute/Henan Provincial People's Hospital. Bo Lei, Henan Provincial People's Hospital / Henan Eye Institute, Henan Eye Hospital, Henan, China.
- 10:20-10:35 The Australian Inherited Retinal Disease Registry and DNA Bank. Enid Chelva, Sir Charles Gairdner Hospital, Perth, Australia.
- 10:35-11:00 Discussion

### **Session 2: Retinal cases and others 11:10-12:00 Chairs: Kaoru Fujinami, Se Joon Woo**

- 11:10-11:40 Rapid-fire case presentations by onsite registers
- 11:40-12:00 Free Discussion

**Session 3: Occult Macular Dystrophy 13:30-14:45 Chair; Kaoru Fujinami, Ruifang Sui**

- 13:30-13:40 EAOMD Study Report 1. Kaoru Fujinami, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 13:40-13:50 Functional Characteristics of East Asian Patients with Occult Macular Dystrophy (Miyake's disease); EAOMD Report No.2 Lizhu Yang
- 13:50-14:00 EAOMD Study Report 3. Kwangic Joo, Department of Ophthalmology, Seoul National University Bundang Hospital, Seoul National University College of Medicine, Seongnam, South Korea.
- 14:00-14:10 Genotype-Phenotype Association in East Asian Patients with Occult Macular Dystrophy (Miyake's disease); EAOMD Report No.4. Yu Fujinami-Yokokawa, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 14:10-14:20 Spectrum of Occult Macular Dystrophy: Investigation of RP1L1 Positive and RP1L1 Negative OMD; EAOMD Report 5 Lizhu Yang, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 14:20-14:30 Investigation of the origin of p.R45W causative for Occult Macular Dystrophy (Miyake's disease) in the East Asian population; EAOMD Report No.6. Yu Fujinami-Yokokawa, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 14:30-14:45 Discussion

**Session 4: Stargardt Disease 14:55-15:40 Chairs: Kaoru Fujinami, Shiyong Li**

- 14:55-15:05 EAStar Study Report 1. Kaoru Fujinami, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 15:05-15:15 EAStar Study Report 2; the imaging study on EAStar data. Seong Joon Ahn, Hanyang University Hospital, Seoul, South Korea.
- 15:15-15:25 Clinical and Genetic Investigation of Stargardt disease in a Chinese Cohort: EAStar China Report 1. Xiao Liu, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 15:25-15:40 Discussion

**Session 5: Retinitis Pigmentosa 15:40-16:20 Chairs: Se Joon Woo, Bo Lei**

- 15:40-15:50 EARP Studies. Kaoru Fujinami, Laboratory of Visual Physiology, Division of Vision Research, National Institute of Sensory Organs, National Hospital Organization, Tokyo Medical Center, Tokyo, Japan.
- 15:50-16:00 Genetic and clinical characteristics of patients with retinitis pigmentosa associated with Pde6b mutations. Kim You Na, Department of Ophthalmology, University of Ulsan College of Medicine, Asan Medical Center, Seoul, Republic of Korea
- 16:00-16:20 Discussion





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