



Department of Biotechnology
Ministry of Science and Technology
Government of India

DBT



National Institute of
Advanced Industrial Science
and Technology

AIST

**DBT - AIST International Laboratory
for Advanced Biomedicine**

DAIILAB

Classroom for Advanced & Frontier Education

CAFE

DAIILAB - CAFE

Series - 017

Date & Time - Sept 28, 2016 - 3 p.m.

Venue - Central 5-41; 2F (Conference Room No. 1)

Speaker - Dr. Akshay ANAND

Affiliation - Neuroscience Research Lab, Department of Neurology, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India

E-mail: akshay2anand@gmail.com



Title: AMD Genetics in India - the missing links

Age related macular degeneration (AMD) is a degenerative eye disease. Photoreceptors present in macular region begin to degenerate due to accumulation of drusen between retinal pigment epithelium (RPE) cells and Bruch's membrane (BM). Further pathological deterioration can stimulate choroidal blood vessels to leak. We described the knockout study of CCL2 gene mice which develop the AMD like features prompting us to examine associated SNPs in Indian population. This showed positive results. Both environmental and genetic factors are found to equally contribute to AMD pathobiology. Recently, GWAS done by Dr Swaroop (Nat Gen, 2016), about 52 genetic loci have been identified as a independently causative agent of AMD in Caucasian populations. ARMS2 and CFH have been explored in various populations and also found to be associated with AMD pathogenesis. Other genes are also being investigated in AMD pathogenesis in various populations, some with conflicting and unverified reports, others which are validated, however, equally large comprehensive genetic studies from India, particularly from North-West India remain limited thus impacting India's reduced role in pioneering AMD diagnostics and therapies. We have investigated various genetic SNPs like *CFH*, *MCP-1*, *VEGFR2*, *CCR-3* in Indian population which were significantly associated with AMD pathology. We also plan to investigate the impact of environmental factors and associated comorbidities with genetic data and/or expression levels of various protein in these Indian AMD patients. Our aim is to incorporate the integrative approach in AMD genetics by including gene-protein, gene-gene, gene-socio-demographic data, follow up studies, statistical modeling and bioinformatics approach to deal such complex nature of AMD is ongoing.

DBT - AIST International Laboratory for Advanced Biomedicine

DAILAB

Classroom for Advanced & Frontier Education CAFE

SERIES - 17

Speaker: Dr. Akshay Anand

Affiliation: PGI, Chandigarh

Topic: AMD Genetics in India: The Missing Links

Date: 28th September 2016 (15:00-16:00 hours JST)

Host: AIST, Japan



National Institute of Advanced Industrial Science & Technology, Japan



PMUC, Beijing, China

IIT-Delhi, India



Brawijaya University, Indonesia



Hanyang University, South Korea

USJP, Sri Lanka



Manipal University, India



Thanks for participation!

Abstract-

Age related macular degeneration (AMD) is a degenerative eye disease. Photoreceptors present in macular region begin to degenerate due to accumulation of drusen between retinal pigment epithelium (RPE) cells and Bruch's membrane (BM). Further pathological deterioration can stimulate choroidal blood vessels to leak. We described the knockout study of CCL2 gene mice which develop the AMD like features prompting us to examine associated SNPs in Indian population. This showed positive results. Both environmental and genetic factors are found to equally contribute to AMD pathobiology. Recently, GWAS done by Dr Swaroop (Nat Gen, 2016), about 52 genetic loci have been identified as a independently causative agent of AMD in Caucasian populations. ARMS2 and CFH have been explored in various populations and also found to be associated with AMD pathogenesis. Other genes are also being investigated in AMD pathogenesis in various populations, some with conflicting and unverified reports, others which are validated, however, equally large comprehensive genetic studies from India, particularly from North-West India remain limited thus impacting India's reduced role in pioneering AMD diagnostics and therapies. We have investigated various genetic SNPs like *CFH*, *MCP-1*, *VEGFR2*, *CCR-3* in Indian population which were significantly associated with AMD pathology. We also plan to investigate the impact of environmental factors and associated comorbidities with genetic data and/or expression levels of various protein in these Indian AMD patients. Our aim is to incorporate the integrative approach in AMD genetics by including gene-protein, gene-gene, gene-socio-demographic data, follow up studies, statistical modeling and bioinformatics approach to deal with complex nature of AMD is ongoing.