

SELECTED RECENT PUBLICATIONS

1) Mecp2²⁷⁰ mutant protein is expressed in astrocytes as well as in neurons and localizes into the nucleus.

Kifayathullah LA*, [Arunachalam JP*](#), Bodda C*, Agbemenyah HY, Laccone FA, Mannan AU. *Cytogenet Genome Res.* 2010;129 (4):290-7. doi: 10.1159/000315906. Epub 2010 Jul 8. *First authors equally contributed.

2) Mild overexpression of Mecp2 in mice causes a higher susceptibility toward seizures.

Bodda C, Tantra M, Mollajew R, [Arunachalam JP](#), Laccone FA, Can K, Rosenberger A, Mironov SL, Ehrenreich H, Mannan AU. *Am J Pathol.* 2013 Jul;183 (1):195-210. doi: 10.1016/j.ajpath.2013.03.019. Epub 2013 May 15.

3) Next-generation sequencing technology detects deletions in the RB1 gene, ranging from a single basepair to whole gene (~170 kb) with a higher sensitivity than FISH.

Ashraf UM, Smita A, [Arunachalam JP](#), Rupali G, Ravi R, Satish S, Vamsi V, Preveen R, Kas S, and Ramesh H. *J Mol Diagn.* 2014 Nov; (Abstract)

4) Prenatal genetic diagnosis of retinoblastoma and report of RB1 gene mutation from India.

Shah PK, Sripriya S, Narendran V, [Pandian AJ](#). *Ophthalmic Genet.* 2016 Feb 25:1-4.

5) Phenotypic characterization of X-linked Retinoschisis: Clinical, Electroretinography and Optical coherence tomography variables.

Srividya N, Sudha D, [Arunachalam JP](#), Rajiv R. Paper accepted in *Indian Journal of Ophthalmology* (IJO_602_15R6)

6) Next generation sequencing based method shows increased mutation detection sensitivity in an Indian retinoblastoma cohort

Jaya S, Avshesh M, [Arunachalam JP](#), Ashwin CM, Sripriya S, Suman K, Smita A, Satish S, Shanmukh K, Vamsi V, Ramesh H, Kalyanasundaram S, Ashraf UM. Paper accepted in *Molecular Vision* (2016MOLVIS0200).

7) Genetic studies in a patient with X-Linked Retinoschisis coexisting with Developmental Delay and Sensorineural Hearing Loss.

Sudha D, Irene R, Aparna G, Smitha A, Shuba K, Srividya N, Sripriya S, Praveen S, Subbulakshmi C and [Arunachalam JP](#). Paper accepted in *Ophthalmic Genetics* (DOI: 10.1080/13816810.2016.1214972) as corresponding author.

MANUSCRIPTS UNDER REVIEW

1) A novel therapeutic approach by TAT-MeCP2 protein delivery prolongs the lifespan and rescues neuronal damage in a mouse model of Rett syndrome.

Prakasha K, [Arunachalam JP](#), Ralf D, Marco P, Wolfgang E and Franco LA. (Reviewed and resubmitted to *Nature Medicine-Manuscript# NMED: BC29956A*. Presently manuscript under review in journal *Brain*, #BRAIN-00554)

2) HIWI2 regulates tight junction proteins in retinal pigment epithelial cells through modulation of AKT/GSK3 signaling pathway.

Suganya S, Karthikka P, Aditya V, [Arunachalam JP](#), Subbulakshmi C. (Manuscript submitted to '*PLoS ONE*', #PONE-D-16-13976)

3) Differing severity among Retinoschisis patients encompassing nonsecreted mutant profile Retinoschisin gene mutants: An insight into the localization of mutant retinoschisin to the plasma membrane of the cell.

Sudha D, Srividya N, Muthukumar S, Al Ameen M, Umashankar V, Lingam G, Vikas K, Rajiv R, Subbulakshmi C and [Arunachalam JP](#) (Manuscript to '*Nature Scientific Reports*' as Corresponding Author)

4) **Proteomic profiling of human intraschisis cavity fluid.**

(Manuscript to 'Journal of Proteome Research' as Corresponding Author)

5) **NDP whole gene deletion in a patient with Norrie Disease.**

(Manuscript to 'European Journal of Medical Genetics' as Corresponding Author)

CHAPTERS IN BOOKS / REVIEW ARTICLE

1) Chapter titled '**Diagnostic procedures for Genetically Transmitted Eye Diseases**'

Sudha D and *Jayamuruga Pandian A*

in the book, 'Diagnostic Procedure in Ophthalmology (III edition)' edited by Dr.HV Nema and published by Jaypee Brothers Medical Publisher. (ISBN number: 978-93-5090-852-5)

2) Chapter titled '**Molecular genetic testing in ophthalmology**'

Soumitra N, Sripriya S and *Jayamuruga Pandian A*

in INSIGHT periodical *Sci J Med & Vis Res Foun October 2013; XXXI:46-49*

3) Chapter entitled '**Current research Perspectives in Understanding Diabetic Retinopathy**'

Jayamuruga Pandian A, Soumitra N, Rajiv R, Sripriya S

in the book, Genetic Eye Research in Asia, as a part of springer series, Essentials in Ophthalmology.

4) Review article entitled, '**Strategies for gene mapping in inherited ophthalmic diseases**'

Srilekha S, Bhavana S, Divya M R, Sudha D, Sathya Priya C, *Jayamuruga Pandian Arunachalam*, Soumitra N, Sripriya S

in the Asia-Pacific Journal of Ophthalmology (APJO) [official publication of the Asia-Pacific Academy of Ophthalmology (APAO)] special Issue on Eye Genetics and Gene Therapy, 5.4, Jul 2016.

ORAL / POSTER PRESENTATIONS

06/2015

Invited talk on, "Genetic Engineering and Functional Characterization of *RS1* Gene and its mutants implicated in X-Linked Retinoschisis (XLRS)" in Continual Medical Education (CME) program and Genomics Workshop entitled 'Genomics: Personalized and Precision Medicine' organized by Frontier Mediville (a unit of Frontier Lifeline hospital), Gummudipoondi, **India**

03/2015

Invited talk on, "Molecular study of *RS1* Gene and its mutants implicated in X-Linked Retinoschisis (XLRS)" in Indian Genetic Congress held at SRM University, Chennai, **India**

2014

Invited talk on, "Functional Characterization of *RS1* Gene and its mutants implicated in X-Linked Retinoschisis (XLRS)" in Indian Eye Research Group (IERG) - Association for Research in Vision and Ophthalmology (ARVO), **USA** [IERG-ARVO] India Chapter 21st Annual Meeting held at L V Prasad Eye Institute, Hyderabad, **India**

2014

Invited talk on, "Engineering and Functional Characterization of genes involved in inherited eye diseases: A significant approach" in international Indo-UK conference & workshop on 'Ophthalmic Genetics and Genetic Counselling for Clinicians and Basic Scientists' conducted at Narayana Nethralaya hospital, Bangalore, **India** in association with the Cardiff University and University of South Wales, **UK**

2014

17th ADNAT international Convention on, 'Genomics in Personalized Medicine and Public Health' at Rajiv Gandhi Center for Biotechnology (RGCB), Trivandrum, **India**

- 2013 Oral presentation entitled, "Ocular Gene therapeutics: An update" in 1st 'SN GENETiCS annual national symposium and Continual Medical Education (CME) program on 'Genetic counseling and Gene testing' at Vision Research Foundation, Sankara Nethralaya, Chennai, **India**
- 2012 Oral presentation entitled, "Molecular Diagnostics: From Gene to Disease" in CME program sponsored by Johnson and Johnson pharmaceuticals conducted at Vision Research Foundation, Chennai, **India**
- 2011 Asia-ARVO (Association for Research in Vision and Ophthalmology) 2011 international conference organized by Singapore Eye Research Institute (SERI), **Singapore**
- 2010 'Diabetic Retinopathy from Bench to Population' international conference organized by Sankara Nethralaya, Chennai, **India**
- 2008 'HD2008: The Milton Wexler celebration of Life' international conference organized by Hereditary Disease Foundation (HDF), Boston, **USA**
- 2008 'Cellular Responses to DNA Damage' international conference organized by Abcam, Boston, **USA**
- 2007 11th Annual Coalition/Scientific HDSA meeting on Huntington's Disease, Wakefield, **USA**
- 2007 CHIP-on-chip scientific meeting by Affymetrix, Boston, **USA**
- 2006 'American Society of Human Genetics (ASHG)' annual meeting and international conference, New Orleans, **USA**
- 2005 DFG (German Research Council) Center for Molecular Physiology of the Brain (CMPB) meeting, Eschwege, **Germany**
- 2004 – 2006 I, II, III International PhD student symposium, 'Horizons in Molecular Biology' by Max Planck Research School, Goettingen, **Germany**
- 2001 XXV All India Cell Biology conference and satellite symposium on cell growth and death held at Indian Institute of Sciences, Bangalore, **India**