

## CURRICULUM VITAE



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**DATE OF BIRTH** 18th January 1950  
**CITIZENSHIP** British

### CURRENT AFFILIATIONS

**01.02.2016 onwards:** Emeritus Professor of Ophthalmology, University College London

**01.04.2016 onwards:** Senior Scientific Advisor, Narayana Nethralaya, Bangalore

### PREVIOUS APPOINTMENTS

**01.05.2016 to 06.05.19:** Distinguished Professor & Principal Investigator, Andalusian Centre for Molecular Biology and Regenerative Medicine (CABIMER)

**01.08.2008 to 30.04.2016:** Director of CABIMER

**01.10.1992 to 31.01.2016:** Sembal Professor of Experimental Ophthalmology, Head Division of Molecular Genetics, Institute of Ophthalmology, University College London, UK

**01.01.2007 to 31.12.11:** Chair de Excellence, Institut de la Vision, Pierre et Marie Curie University, Paris, France

**From: 01.02.87 to 30.09.92.** Top Grade Scientist (with special responsibilities) and Head of Molecular Genetics Unit, Department of Human Genetics, University of Newcastle upon Tyne, UK

**From: 01.06.80 to 31.01.87.** Scientific staff and Senior Research Fellow, MRC Human Genetics Unit, Edinburgh and University of Edinburgh, UK

**From: 01.05.77 to 30.05.80.** Research Associate in the University Department of Clinical Biochemistry, Royal Victoria Infirmary, Newcastle Upon Tyne, UK

## QUALIFICATIONS

**BSc, 1969**, Upper Second Division with Honours in Chemistry, University of Bombay.

**MSc, 1971**, Clinical Biochemistry, University of Newcastle upon Tyne, U.K.

Awarded a tuition fee grant from the British Council.

**PhD, 1977**, Clinical Biochemistry, University of Newcastle upon Tyne, UK.

Awarded a studentship from the Wellcome Foundation for the PhD studies.

## PERSONAL STATEMENT AND CAREER HIGHLIGHTS

My genetic research has been dedicated to the identification of the diseases genes that are involved with visual impairment and blindness. This research has facilitated understanding of the molecular mechanisms of disease and the development of novel approaches for the treatment of patients.

Inherited retinal dystrophies are a major cause of incurable blindness in the Western World. Amongst these a significant proportion is accounted for by a clinically heterogeneous group of diseases collectively known as Retinitis pigmentosa (RP). Based on pedigree analysis, all three modes of Mendelian inheritance (Autosomal dominant, Autosomal recessive and X-linked) have been observed. RP is initially characterised by night blindness and progressive loss of peripheral vision due to the loss of rod photoreceptor cells. As the disease progresses cone cells in the central retina are also involved, often leading to total blindness in later stages of the disease.

Decades of research into the biochemical basis of the disease failed to identify even a single causative factor and therefore no treatment options could be pursued. In the absence of any biochemical defect, a reverse genetic approach should facilitate chromosomal assignment and the isolation of the disease causing genes, thereby raising the prospects for understanding disease mechanisms that may eventually help in the development of gene based therapies. The foundations of Ophthalmic Genetics were laid with the help of recombinant DNA technology for the development of X-chromosome specific DNA markers (**Ref 1**) and their use in the successful mapping of the X-linked form of RP (**Ref 2**). Linkage studies demonstrated the existence of at least two genes for XLRP (RP2 and RP3) which eventually facilitated the isolation of these genes through a positional cloning approach (1997/98). With the finding of genetic heterogeneity in XLRP, involvement of multiple genes in the causation of the dominant or recessive forms of the disease was expected. It soon became clear that the identification of these genes would depend on genetic mapping in single large families. In 1989 the first locus for autosomal dominant retinitis pigmentosa (ADRP) was mapped to chromosome 3q24, in the same interval as the gene for the rod-cell specific photopigment rhodopsin. The first European mutation identified (**Ref 3**) was a small deletion of the rhodopsin gene which may have originated as a result of replication slippage during meiosis due to a 3bp repeating motif (CAT) present at the site of the mutation. At this point it became essential to develop a rapid and simple mutation detection method (**Ref 4**) which would allow large-scale screening of ADRP patients. Our studies indicated that up to 30% of all ADRP patients carry a rhodopsin gene mutation. A positional candidate approach soon implicated peripherin/rds in ADRP however, our studies demonstrated that the same gene is also involved in macular dystrophies (**Ref 5**), making it the first example of two distinct clinical phenotypes resulting from mutations in the same gene. This led to the re-examination of the peripherin/rds cellular localisation data which confirmed its expression in rod as well

as cone cells, providing a possible explanation of the spectrum of retinal diseases associated with this gene.

To further elucidate the genetic basis of retinal degeneration, genealogical studies were undertaken to identify large families with ADRP and cone-rod dystrophy. A novel locus for ADRP was identified in a large 9 generation family on chromosome 7p (**Ref 6**) which was soon followed by the identification of a dominant cone-rod dystrophy (CORD2) locus (**Ref 7**) on chromosome 19q. A positional cloning approach was initiated which subsequently resulted in the identification of the CORD2 gene known as CRX (**Ref 8**). The gene contains a homeobox sequence (typical of most transcription factors), and has been shown to be involved in the regulation of photoreceptor specific genes such as rhodopsin, IRBP and arrestin. Apart from identifying two additional loci for ADRP on chromosome 17q and 19q, linkage studies in a large Cone dystrophy family highlighted the guanylate cyclase activating protein 1 (GCAP1) as a possible candidate gene on chromosome 6p. Soon a Y99C mutation was identified and biochemical studies in-vitro indicated that even in the presence of high levels of Ca (usually a suppressor of GCAP1 activity) GCAP1 remains active (**Ref 9**) leading to a possible constitutive activation of ret GC1 and raised levels of cGMP production. In animal models, high levels of intracellular cGMP have been associated with retinal degeneration and may explain the biological basis of the disease in our Cone-dystrophy family. More recent work allowed the mapping of a further new locus for ADRP on chromosome 14 followed by the identification of a S50T mutation in a second transcription factor gene NRL (**Ref 10**). Transient co-transfection experiments, in-vitro, demonstrated consistent over stimulation of the rhodopsin promoter as a functional impact of the mutant protein in presence of CRX. Transgenic animals over-expressing wild type rhodopsin are known to display retinal degeneration. This may account for the phenotype seen in the chromosome 14-linked ADRP family.

The clinical heterogeneity seen in patients with retinal degeneration have been corroborated by the finding of extensive genetic heterogeneity. This has been borne out by the identification of more than 12 genes for the dominant form of RP with our latest finding of TOPORS mutations causing ADRP (**Ref 11**). As more and more genes are identified, understanding the biochemical basis and functional consequences of the mutant protein may truly revolutionise the 'Post-Genome' era and ultimately lead to the development of more rationale gene or gene based therapies. Proof of principle demonstrating success of such an approach has recently been established by work carried out in our laboratory. Subretinal injection of an adeno-associated viral construct containing a functional copy of the peripherin gene allowed complete ultrastructural and functional rescue of photoreceptor cells in the retinal degeneration slow (rds) mouse (**Ref 12**). As stated earlier, peripherin has been implicated in a variety of human retinopathies and the gene therapy based rescue of a mouse model provides a great deal of hope to patients suffering from degenerative disease of the retina. More recent work supporting further identifications of recessive RP genes (**Ref 13**), preliminary clinical trials in patients (**Ref 14**), generation of retinal progenitor cells for transplantation from ES cells (**Ref 15**) and derivation of retinal pigment epithelial cells for disease modeling using iPSC technology (**Ref 16**) are underway

In summary, for the reasons outlined above it would be critical to undertake careful genotype/phenotype correlation so that the patients who are most likely to benefit are enrolled first for clinical trials, as and when they become available.

### Highlighted publications:

1. Towards a complete linkage map of the human X-chromosome: Regional assignment of 16 cloned single copy DNA sequences employing a panel of somatic cell hybrids. Wiecker, P., Davies, K.E., Cooke, H.J., Pearson, P.L., Williamson, R., **Bhattacharya, S.S.**, Zimmer, J. and Ropers, H. (1984): *Am.J.Hum.Genet.*, **36**: 265-276.
2. Close genetic linkage between X-linked retinitis pigmentosa and a restriction fragment length polymorphism identified by recombinant DNA probe L1.28. **Bhattacharya, S.S.**, Wright, A.F., Clayton, J.F., Price, W.H., Phillips, C.I., McKeown, C.M.E., Jay, M.R., Bird, A.C., Pearson, P.L., Southern, E.M. and Evans, H.J. (1984): *Nature*, **309**: 253-255.
3. A three base pair deletion in the rhodopsin gene in a family with autosomal dominant retinitis pigmentosa. Inglehearn, C.F., Bashir, R., Lester, D.H., Jay, M.R., Bird, A.C. and **Bhattacharya, S.S.** (1991). *Am. J. Hum. Genet.* **48**: 26-30.
4. Rapid detection of single base mismatches as heteroduplexes on hydrolink gels. Keen, J., Lester, D.H., Inglehearn, C.F., Curtis, A. and **Bhattacharya, S.S.** (1991). *Trends in Genetics*, **7**: 5.
5. Mutations in the human retinal degeneration slow (RDS) gene can cause either retinitis pigmentosa or macular dystrophy. Wells, J., Wroblewski, J., Keen, T.K., Inglehearn, C.F., Jubb, A., Eckstein, M., Jay, M.R., Arden, G.B., **Bhattacharya, S.S.**, Fitzke, F.W., Bird, A.C. (1993). *Nature Genetics*, **3**: 213-218.
6. A new locus for autosomal dominant Retinitis Pigmentosa (adRP) on chromosome 7p. Inglehearn, C.F., Carter, S.A., Keen, T.J., Lindsey, J., Stephenson, A.M., Bashir, R., Al-Magthteh, M., Moore, A.T., Jay, M.R., Bird, A.C. and **Bhattacharya, S.S.** (1993). *Nature Genetics*, **4**: 51-53.
7. Genetic linkage of cone-rod dystrophy to chromosome 19q and evidence for segregation distortion. Evans, .E., Fryer, A.F., Inglehearn, C.F., Duvalloung, J., Whittaker, J., Gregory, C.Y., Ebenezer, N., Hunt, D. and **Bhattacharya, S.S.** (1994). *Nature Genetics*, **6**: 210-213.
8. Cone-Rod Dystrophy due to mutations in a novel photoreceptor-specific homeobox gene (CRX) essential for maintenance of the photoreceptor. Freund, C.L., Gregory-Evans, C.Y., Furukawa, T., Papaioannou, M., Looser, J., Ploder, L., Bellingham, J., Ng, D., Herbrick, J.S., Duncan, A., Scherer, S.W., Tsui, L., Loutradis-Anagnostou, A., Jacobson, S.G., Cepko, C.L., **Bhattacharya, S.S.** and McInnes, R.R. (1997). *Cell*, **91**: 543-553.
9. GCAP1(Y99C) Mutant is constitutively active in autosomal dominant cone dystrophy. Sokal, I., Li, N., Surgucheva, I., Warren, M.J., Payne, A.M., **Bhattacharya, S.S.**, Baehr, W. and Palczewski, K. (1998). *Molecular Cell*, **2**: 129-133.
10. Mutation in *NRL* is associated with autosomal dominant retinitis pigmentosa. Bessant, D.A.R., Payne, A.M., Mitton, K.P., Wang, Q-L., Swain, P.K., Plant, C., Bird, A.C., Zack, D.J., Swaroop, A. and **Bhattacharya, S.S.** (1999). *Nature Genetics*, **21**: 355-356.
11. Mutations in *TOPORS* Cause Autosomal Dominant Retinitis Pigmentosa with Perivasculat RPE Atrophy. Chakarova, C.F., Papaioannou, M.G., Khanna, H., Lopez, I., Waseem, N., Shah, A., Theis, T., Friedman, J., Maubaret, C., Bujakowska, K., Veraitch, B., Abd El-Aziz, M.M., Prescott, Q., Parapuram, S., Bickmore, W.A., Munro, P.M/G/, Gal. A/, Hamel, C.P., Marigo, V., Ponting, C.P., Wissinger, B., Zrenner, E., Matter, K., Swaroop, A., Koenekoop, R.K. and **Bhattacharya, S.S.** (2007). *Am J Hum Genet*, **81**: 1098-1103.

12. Restoration of photoreceptor ultrastructure and function in retinal degeneration slow mice by gene therapy. Ali, R.R., Sarra, G.M., Stephens, C., Alwis, M.D., Bainbridge, J.W., Munro, P.M., Fauser, S., Reichel, M.B., Kinnon, C., Hunt, D.M., **Bhattacharya, S.S.** and Thrasher, A.J. (2000). *Nature Genetics*, **25**: 306-310.
13. EYS, encoding an ortholog of *Drosophila* spacemaker, is mutated in autosomal recessive retinitis pigmentosa. Abd El-Aziz MM, Barragan I, O'Driscoll CA, Goodstadt L, Prigmore E, Borrego S, Mena M, Pieras JI, El-Ashry MF, Safieh LA, Shah A, Cheetham ME, Carter NP, Chakarova C, Ponting CP, **Bhattacharya SS**, Antinolo G. (2008) *Nat Genet.***40**: 1285-1287. PMID: 18836446
14. Effect of gene therapy on visual function in Leber's congenital amaurosis. Bainbridge JW, Smith AJ, Barker SS, Robbie S, Henderson R, Balaggan K, Viswanathan A, Holder GE, Stockman A, Tyler N, Petersen-Jones S, **Bhattacharya SS**, Thrasher AJ, Fitzke FW, Carter BJ, Rubin GS, Moore AT, Ali RR. (2008) *N Engl J Med.***358**: 2231-2239. PMID: 18441371
15. Hypoxia increases the yield of photoreceptors differentiating from mouse embryonic stem cells and improves the modeling of retinogenesis in vitro. Garita-Hernández M, Diaz-Corrales F, Lukovic D, González-Guede I, Díez-Lloret A, Valdés-Sánchez ML, Massalini S, Erceg S, **Bhattacharya SS**. *Stem Cells*. 2013 May; 31(5): 966-978. PMID: 23362204
16. Human iPSC derived disease model of MERTK-associated retinitis pigmentosa. Lukovic D, Artero Castro A, Delgado AB, Bernal Mde L, Luna Pelaez N, Díez Lloret A, Perez Espejo R, Kamenarova K, Fernández Sánchez L, Cuenca N, Cortón M, Avila Fernandez A, Sorkio A, Skottman H, Ayuso C, Erceg S, **Bhattacharya SS**. (2015). *Sci Rep.* **5**:12910. doi: 10.1038/srep12910. PMID: 26263531.

## RESEARCH EXPERIENCE AND RESEARCH ACTIVITIES

### Newcastle upon Tyne

**January 1972 to May 1980**

The research work in Newcastle involved investigations into cell membrane permeability and Na-pump activity in heart and skeletal muscle. Net accumulation of cell sodium was taken as an index of membrane permeability and net extrusion from Na-enriched tissues as for Na-pump activity. Total tissue and cellular sodium, potassium chloride and water content were measured. Kinetics of sodium and potassium exchanges in heart and skeletal muscle cells were compared and the mode of action of certain cardioactive drugs (cardiac glycoside, diuretics and beta blockers etc) on membrane permeability and Na-pump activity were investigated.

### Edinburgh

**June 1980 to January 1987**

I have been involved in collaboration with members of Edwin Southern's Mammalian Genome Unit in the construction of a flow sorted human X-chromosome specific genomic library. Extensive screening of this library resulted in the isolation and characterisation of a large number of unique X-chromosome specific DNA probes. A number of restriction fragment length polymorphisms (RFLP) were identified using these probes that can facilitate disease gene mapping as well as construction of a genetic linkage map of the X-chromosome. In collaboration with Dr. Alan Wright of the clinical section of the MRC

Human Genetics Unit, Edinburgh, a locus for the X-linked form of retinitis pigmentosa (XLRP) was mapped to the proximal short arm of the chromosome (Bhattacharya et al Nature, 1984, Vol 309; 253). Two loci for XLRP were identified on the short arm of the X-chromosome and work continued on the identification of the X-linked RP genes.

**Newcastle upon Tyne  
February 1987 to September 1992**

I have been responsible for organising and establishing a Molecular Genetics Laboratory for research and diagnostic work in the Department of Human Genetics, University of Newcastle upon Tyne. The laboratory was mainly equipped from my research grants and is fully operational. The Newcastle Area Health Authority provided me with permanent funding to employ a principal scientist, three senior grade scientists and 4 medical laboratory technicians for the diagnostic service. We provided DNA diagnostic tests for the Duchenne Muscular Dystrophy, Becker Muscular Dystrophy, Cystic Fibrosis and Huntington's Disease registers in the region. With the availability of new resources, molecular diagnostic tests were offered for most inherited genetic diseases in the region for which closely linked DNA markers were identified.

My research group was involved with the isolation and characterisation of retinal genes, studying its developmental and cellular expression patterns and molecular genetic localisation of inherited eye diseases such as Norrie's Disease, Ushers syndrome and autosomal dominant or recessive or X-linked forms of Retinitis Pigmentosa. A spectrum of mutations in candidate genes, such as Rhodopsin and Peripherin in patients with ADRP were identified. We also investigated deletion patterns and phenotypic expression of X-chromosome linked muscular dystrophies, the possibility of genetic heterogeneity in polycystic kidney disease, confirming and extending linkage results in polyposis coli, undertaking novel linkages in incontinentia pigmenti, Retts syndrome and Aarskog syndrome, polymorphisms of mitochondrial DNA and racial variations in the allele frequencies of DNA polymorphisms. The overall aim of this research was to understand the nature and biology of genetic diseases and to provide an efficient diagnostic service for carrier detection and prenatal diagnosis. My laboratory was a participating group in UK and EU Human Genome Mapping Initiative and developed rapid and novel ways of mutation detection and genome analysis. In recognition of significant expansion in my research activity, Wellcome Trust provided substantial refurbishment funds to create state-of-the-art research facilities.

**London  
October 1992 to January 2016**

I was appointed to the Sembal Chair in Experimental Ophthalmology to establish a strong research division dedicated to Ophthalmic Genetics, working in close association with colleagues from Moorfields Eye Hospital. The establishment of the division included complete design and planning of the laboratories and offices and the research facilities. The division is well equipped for biochemical and molecular biological techniques and much of the instrumentation was bought from my research grants. The facilities include automated DNA sequencing (obtained equipment grant funding from the Wellcome Trust for an ABI automated sequencer), high speed and ultracentrifuges, a tissue culture and class 2 containment laboratory, and a radio-isotope laboratory. The techniques of gene mapping,

positional cloning, DNA sequencing, mutation analysis, and structure/function studies of normal and mutant proteins are extensively used in current research projects.

The long-term aim of my research group is to locate, identify and characterise the genes responsible for inherited eye diseases. Development of PCR based microsatellite markers has revolutionised mapping of human genetic diseases. Rapid progress in Human Genome Sequencing projects have resulted in the generation of extensive databases of human genes. Detailed clinical examination and accurate pedigree information recorded over the last twenty five years at Moorfields Eye Hospital has allowed the establishment of an extensive genetic register of inherited eye diseases. Information on over 3000 families are recorded in this register and it is a unique world resource. Many of the eye disease genes identified in my laboratory benefited from the availability of this database. Characterisation and functional analysis of the genes involved in the pathophysiology of these diseases should eventually lead to better clinical management as well as formulating protocols for treatment.

### **AWARDS AND HONOURS**

- Paul Kayser International Award of Merit in Retina Research, Presented at the 7th Bi-Annual Congress of International Society of Eye Research, September 1986, Nagoya, Japan.
- Alcon Research Institute Award for Molecular Genetic Investigations into Inherited Retinal Degenerations, 1991.
- Aida Mann medal, Oxford Eye Infirmary and University of Oxford, awarded 1993.
- Elected Fellow of the Academy of Medical Sciences(UK) 2001-**FMedSci**
- Elected Fellow of the Royal Society of Edinburgh 2006 – **FRSE**
- Awarded Chair of Excellence 2006, France. Full professor appointment held at Pierre et Marie Curie University, Paris from January 2007 to December 2011.
- Scientific recognition from ONCE, Spain, for research on retinal degeneration, 2015
- Board of Directors Special Award for Genetic research, Foundation Fighting Blindness (USA), 2016

### **MEMBERSHIP OF LEARNED SOCIETIES**

- Genetical Society of UK.
- The Galton Institute.
- British Society of Human Genetics.
- American Society of Human Genetics.
- Association for Research in Vision and Ophthalmology (ARVO).

### **INVITED TALKS**

I have given a large number of invited talks at international conferences such as the Am Soc Hum Genet (ASHG), Eu Soc Hum Genet (ESHG) and Association for Vision & Ophthalmology (ARVO) and as external speakers for universities both in the UK and abroad. I have given Keynote lectures at Retinal Degeneration Symposium at the International Congress of Eye Research (ICER) meeting in Nagoya, Japan 1986, New Delhi, India 1994; New Therapeutic Approaches in Inherited Eye Disease (sponsored by EU): Madrid 1996, Athens 1998; Novartis Foundation sponsored meeting in Baltimore 2002; Indian Society of Human Genetics, Kolkata 2006; Andalucian Ministry of Health organized symposium on Genetics and New Treatments, Seville 2007; College de France & INSERM sponsored joint meeting on Vision and Hearing, Paris 2007. Invited speaker at School of Biosciences,

University of Kent, UK, November 2012. Invited speaker at the Chinese University of Hong Kong, University Eye Hospital, Shantou, China and Department of Ophthalmology, University of Chengdu, China, January 2014, October 2015, January 2017. Invited talks at Narayan Nethralaya, Bangalore India (January 2016); University of Newcastle upon Tyne (March 2016) and Vision Conference, Baltimore June 2016 organised by Foundation Fighting Blindness, USA

Regular research presentations (on average two per year since 1985) to branch members of the British Retinitis Pigmentosa Society, to explain the genetic basis of retinal degeneration, research progress and new developments in treatment. I have also given lectures to RP Societies of Italy, Germany, Finland, France, South Africa, Canada, Ireland, Australia and USA.

## TEACHING

Along with Dr. Papiha, I have been responsible for establishing a new course in October 1989 in Medical Genetics at the University of Newcastle-upon-Tyne. The course was approved by the Medical Research Council and an MRC studentship was awarded on a yearly basis. I covered the disease gene mapping and gene identification aspect of the course. Since taking up my current appointment in London I have been giving periodic lectures to undergraduate students in Genetics at UCL.

## Ph.D. THESIS SUPERVISION

Supervised a total of 40 students

Paula Monaghan	1989	Douglas Lester	1990
Ivan Still	1991	Alison Hardcastle	1992
Smaro Kamakari	1994	Peter Clements	1994
Mai Al-Magheteh	1995	Nick Occleston	1996
Dawn Thiselton	1997	Sujeewa Wijesuriya	1997
Francesca Cordeiro	1998	Reshma Patel	1998
Eranga Vithana	1998	Donna MaKay	1998
Marcela Votruba	2000	Sana Kermani	2000
Peter Francis	2000	Mohamed El-Ashry(MD)	2001
Marc Botcherby	2002	Ordan Lehmann	2003
David Bessant(MD)	2003	Leen Abu-Saif	2003
Neil Ebenezer	2003	Aung Tin*	2003
Ashwin Reddy(MD)	2003	Peter Addison(MD)	2006
Mai Abd El-Aziz	2007	Kinga Bujakowska	2008
Brotati Ghosh	2008	Petra Liskova	2008
Amna Shah	2009	Ciara O'Driscoll	2009
Abhay Krishna	2010	Francesca Fiocco	2010
Anna Rose	2012	Margarita Romero	2012
Marcela Garita	2013	Barbara Czub	2015
Przemyslaw Kruczek	2015	Ana Belen	2019

Several of my former Ph.D. students have become senior academics both in UK and abroad including two students who are now full professors of UCL (Alison Hardcastle, 1992 and Francesca Cordeiro, 1998). Several Clinical Fellows who completed their MD/Ph.D. under my supervision have become Consultant/Professor (Alex Ionedes, Kevin



Gregory-Evans, David Bessant, Marcela Votruba, Ashwin Reddy, Aung Tin, Ordan Lehman and Peter Addison). Seven of my former postdoctoral fellows have become full professors (Susan Lindsay, Chris Inglehearn, Alan Shiels, Cheryl Gregory-Evans, Andrew Webster, Robin Ali and Jane Sowden) and Robin Ali was elected to the prestigious Academy of Medical Sciences, UK in 2007 and as a senior investigator of National Institute of Health Research, UK in 2008.

Examined over 40 Ph.D. students from top UK & European Universities including Oxford, Cambridge, London, Edinburgh, Dublin, Ghent, Nijmegen and University of Seville.

### **UNIVERSITY ENABLING ACTIVITIES**

- Academic Appointments Committee member at University of Newcastle
- Founder member of Centre for Human Genetics at UCL
- Executive board member of the UCL Institute of Human Genetics
- Academic Appointments Committee member at Institute of Ophthalmology and Institute of Child Health, University College London
- Senior member of Strategy Committee, Institute of Ophthalmology, UCL

### **PROFESSIONAL ACTIVITIES**

- On the examiners panel of the Royal College of Pathologists for the Degree in Clinical Cytogenetics and Molecular Genetics. Course tutor in Molecular Genetics for the MRC Path. examination.
- Past Committee member of Clinical Molecular Genetic Society.
- Scientific Advisory Panel of the British Retinitis Pigmentosa Society.
- Assessor for British Council sponsored European joint research programmes.
- Member of the Genetics Committee of the Foundation Fighting Blindness (USA).
- Editorial Board member of Annals of Human Genetics and Disease Markers
- Editorial Board member of Molecular Vision
- Grant review panel member of the Irish Health Research Board, Deutsche Forschungsgemeinschaft (DFG) and INSERM (France)
- Member of the visiting sub-committee for the Management and Scientific reviews of the following research centres:
  - Telethon Institute of Genetic Medicine, Naples (September 2003)
  - MRC Human Genetics Unit, Edinburgh (May 2005).
  - GSF(Munich)-Institutes of Bioinformatics, Developmental Genetics, Experimental Genetics, Human Genetics and Department of Comparative Medicine (March 2005).
  - Institut de la Vision, Pierre et Marie Curie University (UPMC), Paris (January 2017)
- International Advisor to National Eye Institute of NIH for EyeGENE Programme (Jan 2006)
- Inauguration of the Medical Genome Centre in Seville (March 2011)
- Grant review panel member of the Irish Health Research Board (May 2011)
- Panel member of European Union review committee for Translational Research March 2012
- Grant review panel member of the Irish Health Research Board and MRCG (September 2013)
- ARVO Foundation Awards Committee member (since 2012)
- Grant review panel member of Fondazione Roma (February 2014)

### **Refereed grant applications for:**

- Medical Research Council
- BBSRC
- The Wellcome Trust
- British Retinitis Pigmentosa Society
- German Medical Research Council
- Irish Retinitis Pigmentosa Society
- Foundation Fighting Blindness of USA
- Irish Department of Health
- March of Dimes Birth Defects Foundation
- Health Research Board of Australia
- New Zealand Medical Research Council
- Guide Dogs for the Blind
- Action Research

### **Refereed manuscripts**

Refereed papers for all top journals in the field including Nature, Science, Cell, Nature Genetics, Nature Medicine, NEJM, EMBO, American Journal of Human Genetics, European Journal of Human Genetics, Human Genetics, Human Molecular Genetics, Journal of Medical Genetics, Human Mutation and Investigative Ophthalmology & Visual Science.

### **INTERNATIONAL CONFERENCES / SESSIONS ORGANISED**

- Retinal Degeneration Symposium at International Congress Eye Research Meeting (ICER) - New Delhi, India 1994.
- Genetics section at the British Society for Eye Research - Cardiff 1994.
- Inherited Retinal Dystrophies Symposium at Association for Research in Vision and Ophthalmology (ARVO) - Fort Lauderdale, Florida 1996.
- New therapeutic approaches in inherited eye disease (sponsored by EU):
  - 1<sup>st</sup> Meeting 1996 - Madrid, Spain
  - 2<sup>nd</sup> Meeting 1997 - Athens, Greece
  - 3<sup>rd</sup> Meeting 1999 - Tübingen, Germany
  - 4<sup>th</sup> Meeting 2001 – Prague, Czech Republic
- Retinal Degeneration Symposium at International Congress Eye Research Meeting (ICER) - Paris, France 1998.
- Mutations Associated with Retinal Disease at Association for Research in Vision and Ophthalmology (ARVO) - Fort Lauderdale, Florida 1999.
- Molecular Characteristics of Cataractogenesis at Association for Research in Vision and Ophthalmology (ARVO) - Fort Lauderdale, Florida 2001.
- Mutations, Modifiers and Complex Genetics at Association for Research in Vision and Ophthalmology (ARVO) - Fort Lauderdale, Florida 2003.
- Mutations, Modifiers and Complex Genetics at Association for Research in Vision and Ophthalmology (ARVO) - Fort Lauderdale, Florida 2008.
- Disease Mechanisms at Association for Research in Vision and Ophthalmology (ARVO) - Fort Lauderdale, Florida 2012

--Proposer and Chair of The Novartis Foundation and Foundation Fighting Blindness (USA) sponsored symposium on “Retinal Dystrophies: Functional Genomics to Gene Therapy” 2002. Symposium volume published (John Wiley & Sons) Jan. 04.

--Course in Eye Genetics. 23-25 September 2010. Bologna (Italy).

--8th International Meeting on Retinal Dystrophies.19-22 May 2010 (Dublín, Reino Unido).

-- 4th Genome Sequencer FLX User Conference, 15-17Jun, 2010, Athens (Greece)

--Course in Rare Diseases, June 2010, organized by the UNIA (International University of Andalusia) Seville (Spain)

--The American Society of Human Genetics, 31Oct-7 Nov, 2010, Washington (USA)

-- Jornadas Salud Investiga 21-23 October 2010. (Cádiz, España).

--ARVO Traslational research: seeing the possibilities. 6-10 May 2010 (Fort-Lauderdale, Florida (USA)

--ARVO Traslational research: seeing possibilities.7-11 May 2012 (Fort- Lauderdale, Florida (USA)

-- International Symposium on cell therapy and gene-based therapies.28-29 June 2012 (Granada-Spain)

-- ARVO AMD poster session moderator May 2014

-- ARVO Retinal dystrophy genetics 2015

-- Invited talks, Hong Kong Eye Hospital and Chinese University of Hong Kong, 17-18<sup>th</sup> October 2015

-- Invited talks, Guandong Ophthalmological Society International Conference, China 29th Oct -1st Nov 2015

-- Invited talk Narayana Nethralaya, Bangalore, 29th January 2016

--Seminar presentation, University of Newcastle upon Tyne, March 2016

International Program Committee member of Asia-ARVO January 2009, 2011, 2013, 2015 and 2016

International Program Committee member of World Ophthalmology Congress (WOC) 2018, Barcelona, Spain

## **BOOK PUBLICATIONS**

*Degenerative Retinopathies: Advances in Clinical and Genetic Research* by Peter Humphries, Shomi Bhattacharya, Alan Bird, CRC Press, Florida, 1991.

*Retinal Dystrophies: Functional Genomics to Gene Therapy*. Novartis Foundation Symposium 255, Chaired by Shomi Bhattacharya, John Wiley & Sons Ltd, UK, 2004.

## **LIST OF PUBLICATIONS**

Published 384 peer reviewed papers and 13 book chapters

1. Effects of ouabain on sodium uptake by frog heart and skeletal muscle. Flear, C.T., Greener, J.S. and Bhattacharya, S.S. (1975). *Recent Adv. Stud. Cardiac Struct. Metab.* **5**: 343-349.
2. Actions of cardiac-glycosides and diuretics on Na-pump activity and Na-permeability

- in frog ventricle fibres. Flear CTG, Bhattacharya SS, Walsh, TD and McCambridge HJ (1977). *Br. Heart Journal*, **39**: 350-350.
3. Actual or standard bicarbonate? Tibi, L., Bhattacharya, S.S. and Flear, C.T.G. (1979). *Lancet*, **2**: 1139.
  4. Solute and water exchanges between cells and extracellular fluids in health, and disturbances after trauma. Flear, C.T.G., Bhattacharya, S.S. and Singh, C.M. (1980). *Journal of parenteral enteral nutrition*, **4**: 98-120.
  5. Variability in pK<sup>1</sup> of human plasma. Tibi, L., Bhattacharya, S.S. and Flear, C.T. (1982). *Clin. Chim. Acta.* **121**: 15-31.
  6. DNA probes in X-linked retinitis pigmentosa. Wright, A.F., Bhattacharya, S. S., Price, W.H., Phillips, C.I., McKeown, C., Crews, S.J., Jay, M.R. and Bird, A.C. (1983): *Transactions of Ophthalmological Society U.K.*, **103**: 467-472.
  7. Recombinant DNA probes in retinitis pigmentosa. Bhattacharya, SS, Wright, AF, Cooke, H, Southern EM and Evans, HJ. (1984). *Genetical Research*, **43**: 213-214.
  8. Towards a complete linkage map of the human X-chromosome: Regional assignment of 16 cloned single copy DNA sequences employing a panel of somatic cell hybrids. Wiecker, P., Davies, K.E., Cooke, H.J., Pearson, P.L., Williamson, R., Bhattacharya, S.S., Zimmer, J. and Ropers, H. (1984): *Am.J.Hum.Genet.*, **36**: 265-276.
  9. Close genetic linkage between X-linked retinitis pigmentosa and a restriction fragment length polymorphism identified by recombinant DNA probe L1.28. Bhattacharya, S.S., Wright, A.F., Clayton, J.F., Price, W.H., Phillips, C.I., McKeown, C.M.E., Jay, M.R., Bird, A.C., Pearson, P.L., Southern, E.M. and Evans, H.J. (1984): *Nature*, **309**: 253-255.
  10. A genetic linkage study of a kindred with X-linked retinitis pigmentosa. Bhattacharya, S.S., Clayton J.F., Harper, P., Hoare, G.W., Jay, M.R., Lyness A.L. and Wright, A.F. (1985): *Br. J. Ophthalmology*, **69**: 340-347.
  11. Genetic linkage between X-linked retinitis pigmentosa and DNA probe DXS7(L1.28): further linkage data, heterogeneity testing and risk estimation. Clayton, J.F., Wright, A.F., Jay, M.R., McKeown, C.M.E., Dempster, M., Jay, B.S., Bird, A.C. and Bhattacharya, S.S. (1986). *Hum. Genet.* **74**: 168-171.
  12. A genetic linkage study of choroideremia. Jay, M., Wright, A.F., Clayton, J.F., Deans, M., Dempster, M., Bhattacharya, S.S. and Jay, B. A. (1986). *Ophthalmic. Paediatr. Genet.* **7**: 201-204.
  13. Human interstitial retinal-binding protein (IRBP): cloning, partial sequence and chromosomal localisation. Liou, G.I., Fong, S.L., Gosden, J.R., van Tuinen, P., Ledbetter, D.H., Christie, S., Rout, D., Bhattacharya, S.S., Cook, R.G., Li, Y., Wang, C. and Bridges, C.D.B. (1987). *Somatic Cell and Molecular Genetics.* **13**: 315-323.

14. Linkage relationships between X-linked retinitis pigmentosa and nine X-chromosome markers: exclusion of the disease locus from Xp2.1 and localisation to between DXS7 and DXS14. Wright, A.F., Bhattacharya, S.S., Clayton, J.F., Dempster, M., Tippett, P., McKeown, C.M.E., Jay, M., Jay, B.S. and Bird, A.C. (1987). *Am. J. Hum. Genet.* **41**: 635-644.
15. Molecular genetic approaches to the analysis of human ophthalmic disease. Cooper, D.N., Jay, M.R., Bhattacharya, S.S. and Jay, B. (1987). *Eye.* **1**: 699-721.
16. The generation of metastatic mosaicism in B-16 murine melanoma. Sherbet, GV, Lakshmi, MS, Lune, J, Bhattacharya, SS and Parker, C (1987). *Br. J. Cancer*, **56**: 174-175.
17. Bgl II RFLP recognised by a human IRBP cDNA localised to chromosome 10. Lious, G.I., Li, Y., Wang, C., Fong, S.L., Bhattacharya, S.S. and Bridges, C.D.B. (1987). *Nucleic Acids Research.* **15**: 3196.
18. A mileage chart of part of the X-chromosome. Bhattacharya, SS, Wright, AF and Clayton, JF. (1987). *Cytogenet. Cell Genet.* **46**: 580-581.
19. A case of disputed maternity. Roberts, D.F., Papiha, S.S. and Bhattacharya, S.S. (1987). *Lancet* **II**: 478-480.
20. DNA polymorphisms, identified by an X-chromosome short-arm probe L1.28 (DXS7), in different racial groups. Papiha, S.S., Bhattacharya, S.S. and Roberts, D.F. (1988). *Hum. Hered.* **38**: 72-75.
21. X-linked retinitis pigmentosa: a molecular genetic approach to isolating the defective genes. Lindsay, S., Jay, M., Bower, D.J., Adam, G., Inglehearn, C.F., Sealey, P.G., Papiha, S.S. and Bhattacharya, S.S. (1989). *Prog. Clin. Biol. Res.* **314**:83-97.
22. Population frequency of three DNA alleles linked to Duchenne muscular dystrophy gene. Papiha, S.S., Roberts, D.F., Clarke, A., Burn, J., Gardner-Medwin, D. and Bhattacharya, S.S. (1989). *J. Med. Genet.* **26**: 390-392.
23. X-chromosome restriction fragment length polymorphisms in five racial groups: Rare variants detected with the RC8 (DXS9) probe in the Marathu population, India. Wadhwa, R., Papiha, S.S., Lester, D.H., Ray, V., Saha, N. and Bhattacharya, S.S. (1989). *Hum. Hered.* **39**: 309-312.
24. Dystrophin in skeletal muscle. II. Immunoreactivity in patients with Xp21 muscular dystrophy. Nicholson, L.V.B., Davison, K., Johnson, M.A., Slater, C.R., Young, C., Bhattacharya, S.S., Gardner-Medwin, D. and Harris, J.B. (1989). *J. Neurol. Sci.* **94**: 137-146.
25. Linkage of internal minisatellite loci on chromosome 1 and exclusion of autosomal dominant retinitis pigmentosa proximal to rhesus. Inglehearn, C.F., Papiha, S.S., Jay, M.R., Wright, A.F., Moore, A.T. and Bhattacharya, S.S. (1990). *J. Med. Genet.* **27**: 14-16.

26. Better fingerprinting with PCR. Bellamy, R.J., Inglehearn, C.F., Lester, D.H., Hardcastle, A. and Bhattacharya, S.S. (1990). *Trends in Genetics*. **6**: 32.
27. Autosomal dominant RP: Evidence for at least two genetic loci. Lester, D.H., Bashir, R., Jay, M.R., Bird, A.C., Wright, A.F., Inglehearn, C.F. and Bhattacharya, S.S. (1990). *J. Med. Genet.* **37**: 647.
28. Linkage studies and deletion screening in choroideremia. Wright, A.F., Nussbaum, R.L., Bhattacharya, S.S., Jay, M.R., Lesko, J.G., Evans, H.J. and Jay, B. (1990). *J. Med. Genet.* **27**: 496-498.
29. No evidence for linkage between late onset autosomal dominant retinitis pigmentosa and chromosome 3 locus D3S47 (C17). Evidence for genetic heterogeneity. Inglehearn, C.F., Jay, M.R., Lester, D.H., Bashir, R., Jay, B., Bird, A.C., Wright, A.F., Evans, H.J., Papiha, S.S. and Bhattacharya, S.S. (1990). *Genomics*. **6**: 168-173.
30. Linkage to D3S47 (C17) in one large family and exclusion in another: confirmation of genetic heterogeneity. Lester, D.H., Inglehearn, C.F., Bashir, R., Ackford, H.E., Esakowitz, L., Jay, M.R., Bird, A.C., Wright, A.F. and Bhattacharya, S.S. (1990). *Am. J. Hum. Genet.* **47**: 536-541.
31. Localising multiple X-chromosome linked retinitis pigmentosa loci using multilocus homogeneity tests. Ott, J., Bhattacharya, S.S., Chen, J.D., et al (1990). *Proc. Natl. Acad. Sci. USA*. **87**: 701-704.
32. Localisation of the microsatellite probe DXS426 between DXS7 and DXS255 on Xp and linkage to X-linked retinitis pigmentosa. Coleman, M., Bhattacharya, S.S., Lindsay, S.J., Wright, A.F., Jay, M.R., Litt, M. and Davies, K.E. (1990). *Am. J. Hum. Genet.* **47**: 935-940.
33. Genomic probes in prenatal diagnosis of Duchenne muscular dystrophy in Indians and Chinese. Papiha, S.S. and Bhattacharya, S.S. (1990). *Nat. Med. J. India*. **3**: 17-19.
34. Autosomal dominant retinitis pigmentosa: Absence of the rhodopsin proline -> histidine substitution (codon 23) in pedigrees from Europe. Farrar, G.J., Kenna, P., Redmond, R., McWilliam, P., Bradley, D.G., Humphries, M.M., Sharp, E.M., Inglehearn, C.F., Bashir, R., Jay, M.R., Watty, A., Ludwig, M., Schinzel, A., Samanns, C., Gal, A., Bhattacharya, S.S. and Humphries, P. (1990). *Am. J. Hum. Genet.* **47**: 941-945.
35. Heterogeneity of dystrophin expression in patients with Duchenne and Becker muscular dystrophy. Nicholson, L.V.B., Johnson, M.A., Gardner-Medwin, D., Bhattacharya, S.S. and Harris, J.B. (1990). *Acta Neuropathol.* **80**: 239-250.
36. Abnormalities of carbohydrate metabolism and of OCT gene function in the Rett syndrome. Clarke, A., Gardner-Medwin, D., Richardson, J., McGann, A., Bonham, J.R., Carpenter, K.H., Bhattacharya, S.S., Haggerty, I.D., Fleetwood, J.A. and Aynsley-Green, A. (1990). *Brain and Development*. **12**: 119-124.

37. Association of less common cystic fibrosis mutations with a mild phenotype. Curtis, A., Nelson, R., Porteous, M., Burn, J. and Bhattacharya, S.S. (1991). *J. Med. Genet.* **28**: 34-37.
38. A new XmnI polymorphism for the DMD probe PERT 87-8. Haggerty, I.D., Keen, J., Curtis, A. and Bhattacharya, S.S. (1991). *Nucleic Acids Res.* **19**:680.
39. Population variation in molecular polymorphisms of the short arm of the human X chromosome. Papiha, S.S., Mastana, S.S., Roberts, D.F., Onyemelukwe, G.C. and Bhattacharya, S.S. (1991). *Am. J. Phys. Anthropol.* **85**:329-334.
40. A three base pair deletion in the rhodopsin gene in a family with autosomal dominant retinitis pigmentosa. Inglehearn, C.F., Bashir, R., Lester, D.H., Jay, M.R., Bird, A.C. and Bhattacharya, S.S. (1991). *Am. J. Hum. Genet.* **48**: 26-30.
41. Rapid detection of single base mismatches as heteroduplexes on hydrolysis gels. Keen, J., Lester, D.H., Inglehearn, C.F., Curtis, A. and Bhattacharya, S.S. (1991). *Trends in Genetics.* **7**: 5.
42. Retinitis pigmentosa and mutations in rhodopsin. Bhattacharya, S., Lester, D., Keen, T.J., Bashir, R., Lauffart, B., Inglehearn, C.F., Jay, M.R., and Bird, A.C. (1991). *Lancet.* **337**: 185.
43. Increased band sharing in DNA fingerprints of an inbred human population. Bellamy, R.J., Inglehearn, C.F., Jalili, I.K., Jeffreys, A.J. and Bhattacharya, S.S. (1991). *Hum. Genet.* **87**: 341-347.
44. Genetic localisation of the RP2 type of X-linked retinitis pigmentosa in a large kindred. Wright, A.F., Bhattacharya, S.S., Aldred, M.A., Jay, M.R., Carothers, A.D., Harper, P.S., Bird, A.C., Jay, B. and Evans, H.E. (1991). *J. Med. Genet.* **28**: 453-457.
45. Identification of a mutation in the promoter region of the dystrophin gene in a patient with atypical Becker muscular dystrophy. Bushby, K., Cleghorn, N.J., Curtis, A., Haggerty, I.D., Nicholson, L., Johnson, A., Harris, J.B. and Bhattacharya, S.S. (1991). *Hum. Genet.* **88**: 195-199.
46. Autosomal dominant retinitis pigmentosa: four new mutations in rhodopsin, one of them in the retinal attachment site. Keen, T.J., Inglehearn, C.F., Lester, D.H., Bashir, R., Jay, M.R., Bird, A.C., Jay, B. and Bhattacharya, S.S. (1991). *Genomics.* **11**: 199-205.
47. The Msh-like homeobox genes define domains in the developing vertebrate eye. Monaghan, A.P., Davidson, D.R., Sime, C., Graham, E., Baldock, R., Bhattacharya, S. S., and Hill, R. E. (1991). *Development.* **112**: 1053-1061.
48. Genetic and physical mapping around the properdin P gene. Coleman, M.P., Murray, J.C., Willard, H.F., Nolan, K.F., Reid, K.B.M., Blake, D.J., Lindsay, S., Bhattacharya, S.S., Wright, A.F. and Davies, K.E. (1991). *Genomics.* **11**: 991-996.

49. Localisation of the gene for Norrie disease to between DXS7 and DXS426 on Xp. Lindsay, S., Thiselton, D.L., Bateman, J.B., Ngo, J.T., Sparkes, R.S., Coleman, M., Davies, K.E. and Bhattacharya, S.S. (1992). *Hum. Genet.* **88**: 349-350.
50. A completed screen for mutations of the rhodopsin gene in a panel of patients with autosomal dominant retinitis pigmentosa. Inglehearn, C.F., Keen, T.J., Bashir, R., Jay, M.R., Fitzke, F.W., Bird, A.C., Crombie, A. and Bhattacharya S.S. (1992). *Hum. Mol. Genet.* **1**: 41-45.
51. Recurrent 3-bp Deletion at Codon 255/256 of the Rhodopsin Gene in a German Pedigree with Autosomal Dominant Retinitis Pigmentosa. Artlich, A., Horn, M., Lorenz, B., Bhattacharya S. S. and Gal, A. (1992). *Am. J. Hum. Genet.* **50**: 876-877.
52. Abnormal dark adaptation kinetics in autosomal dominant sector retinitis pigmentosa. Moore, A. T., Fitzke, F.W., Kemp, C.H., Arden, G. B., Keen, T.J., Inglehearn, C.F., Bhattacharya, S. S. and Bird, A.C. (1992). *Br. J. Ophthal.* **76**: 465-469.
53. Molecular genetics of inherited retinal degenerations. Lindsay S, Inglehearn CF, Curtis A and Bhattacharya SS, (1992). *Current opinions in Genetics and Development.* **2**: 459-466.
54. Exclusion of chromosome 6 and 8 locations in non-rhodopsin autosomal dominant retinitis pigmentosa families: Further locus heterogeneity in adRP. Bashir, R., Inglehearn, C. F. Keen, T.J. Lindsay, J., Atif, U., Carter, S.A., Stephenson, A. M., Jackson, A., Jay, M.R., Bird, A. C., Papiha, S and Bhattacharya S. S. (1992). *Genomics.* **14**: 191-193.
55. The Gene for Aarskog syndrome is located between DXS255 and DXS256. Porteous MEM, Curtis A, Lindsay S, Williams O, Goudie D, Kamakari S, Bhattacharya S.S. (1992). *Genomics.* **14**: 298-301.
56. Two dinucleotide repeat polymorphisms at the DXS571 locus. Curtis, A.R.J., Roustan, P., Kamakari, S., Thiselton, D.L., Lindsay, S. and Bhattacharya S.S. (1992). *Hum. Mol. Genet.* **1**: 776.
57. Dinucleotide repeat polymorphism at the DXS559 locus. Roustan, P., Curtis, A.R.J., Kamakari, S., Thiselton, D.L., Lindsay, S. and Bhattacharya, S.S. (1992). *Hum. Mol. Genet.* **1**: 778.
58. Dinucleotide repeat polymorphism at the DXS537 locus. Roustan, P., Curtis, A.R., Kamakari, S., Thiselton, D.L., Lindsay, S. and Bhattacharya, S.S. (1993). *Hum. Mol. Genet.* **2**: 92.
59. A large deletion at the 3' end of the rhodopsin gene in an Italian family with a diffuse form of autosomal dominant retinitis pigmentosa. Restagno, G., Maghtheh, M., Bhattacharya, S.S., Ferrone, M., Garnerone, S., Samuelly, R. and Carbonara, A. (1993). *Hum. Mol. Genet.* **2**: 207-208.
60. Genetic mapping of the X chromosome in familial Rett Syndrome. Curtis ARJ,



- Headland S, Lindsay S, Thomas NST, Boye E, Kamakari S, Roustan P, Anvret M, Wahlstrom J, McCarthy G, Clarke AJ, Bhattacharya, SS. (1993). *Hum. Genet.* **90**: 551-555.
61. Absence of cystic fibrosis mutations in a large Asian population sample and occurrence of a homogyous S549N mutation in an inbred Pakistani family. Curtis, A., Richardson, R.J., Boohene, J., Jackson, A., Nelson, R. and Bhattacharya, S.S. (1993). *J. Med. Genet.* **30**: 164-166.
  62. The clinical, genetic and dystrophin characteristics of Becker muscular dystrophy. Bushby, K.M.D., Gardner-Medwin, D., Nicholson, L.V.B., Johnson, M.A., Haggerty, I.K., Cleghorn, N.J., Harris J.B. and Bhattacharya S.S. (1993). *J. Neurol.* **240**: 105-112.
  63. Variation in DNA polymorphisms of the short arm of the human X chromosome genetic affinity of Parsi from Western India. Al-Maghteh, M., Ray, V., Mastana, S.S., Garalda, M.D., Bhattacharya, S.S. and Papiha, S.S. (1993). *Hum Hered.* **43**: 239-243.
  64. Isolation and characterisation of 3 microsatellite markers in the proximal long arm of the human X chromosome. Lindsay, S., Curtis, A.R.J., Roustan, P., Kamakari, S., Thiselton, D.L., and Bhattacharya, S.S. (1993). *Genomics.* **17**: 208-210.
  65. Evidence against a second autosomal dominant Retinitis pigmentosa locus close to Rhodopsin on chromosome 3q. Inglehearn, C., Farrar, J., Denton, M.J., Gal, A., Humphries, P. and Bhattacharya, S.S. (1993). *Am. J. Hum. Genet.* **33**: 536-537.
  66. Autosomal dominant retinitis pigmentosa with apparent incomplete penetrance: a clinical, electrophysiological, psychophysical and molecular genetic study. Moore, A.T., Fitzke, F.W., Jay, M.R., Arden, G.B., Inglehearn, C.F., Keen, T.J., Bhattacharya, S.S. and Bird, A.C. (1993). *Br. J. Ophthal.* **77**: 473-479.
  67. Exclusion of the involvement of all known retinitis pigmentosa loci in the disease present in a family of Irish origin provides evidence for a sixth autosomal dominant locus (RP8). Kumar-Singh, R., Farrar, F.J., Mansergh, F., Kenna, P., Bhattacharya, S.S., Gal, A. and Humphries, P. (1993). *Hum. Mol. Genet.* **2**: 875-878.
  68. Rhodopsin mutations in autosomal dominant retinitis pigmentosa. Al-Maghteh, M., Gregory, C.Y., Inglehearn, C.F., and Bhattacharya, S.S. (1993). *Human Mutation.* **2**: 249-255.
  69. Mutations in the human retinal degeneration slow (RDS) gene can cause either retinitis pigmentosa or macular dystrophy. Wells, J., Wroblewski, J., Keen, T.K., Inglehearn, C.F., Jubb, A., Eckstein, M., Jay, M.R., Arden, G.B., Bhattacharya, S.S., Fitzke, F.W., and Bird, A.C. (1993). *Nature Genetics.* **3**: 213-218.
  70. A new locus for autosomal dominant Retinitis Pigmentosa (adRP) on chromosome 7p. Inglehearn, C.F, Carter, S.A., Keen, T.J., Lindsey, J., Stephenson, A.M., Bashir, R., Al-Maghteh, M., Moore, A.T., Jay, M.R., Bird, A.C., and Bhattacharya, S.S. (1993). *Nature Genetics.* **4**: 51-53.

71. Confirmation of the rod cGMP phosphodiesterase b subunit (PDEb) nonsense mutation in affected red-1 Irish setters in the UK and development of a diagnostic test. Clements, P.J.M., Gregory, C.Y., Peterson-Jones, S.M., Sargan, D.R., and Bhattacharya S.S. (1993). *Current Eye Research*. **12**: 861-866.
72. Dominant retinitis pigmentosa associated with two rhodopsin gene mutations: Leu-40-Arg and an insertion disrupting the 5'-splice junction of exon 5. Kim, R.Y., Al-Magthteh, M., Fitzke, F.W., Arden, G.B., Jay, M., Bhattacharya, S.S. and Bird, A.C. (1993). *Arch. Ophthal.* **111**: 1518-1524.
73. A 150 bp insertion in the rhodopsin gene of an autosomal dominant retinitis pigmentosa family. Al-Magthteh, M., Kim, R., Hardcastle, A., Inglehearn, C.F. and Bhattacharya, S.S. (1994). *Hum. Mol. Genet.* **3**: 205-206.
74. Identification of a sixth locus for autosomal dominant retinitis pigmentosa on chromosome 19. Al-Magthteh, M.G., Inglehearn, C.F., Keen, T.J., Evans, K.E., Moore, A.T., Jay, M., Bird, A.C. and Bhattacharya, S.S. (1994). *Hum. Mol. Genet.* **3**: 351-354.
75. Genetic linkage of cone-rod dystrophy to chromosome 19q and evidence for segregation distortion. Evans, .E., Fryer, A.F., Inglehearn, C.F., Duvalloung, J., Whittaker, J., Gregory, C.Y., Ebenezer, N., Hunt, D. and Bhattacharya, S.S. (1994). *Nature Genetics*. **6**: 210-213.
76. Further refinement of the location for autosomal dominant retinitis pigmentosa on chromosome 7p (RP9). Inglehearn, C.F., Keen, T.J., Al-Magthteh, M., Gregory, C.Y., Jay, M.R., Moore, A.T., Bird, A.C. and Bhattacharya, S.S. (1994) *Am. J. Hum. Genet.* **54**: 675-680.
77. Retinal pattern dystrophy caused by a 4 bp insertion at codon 140 in the Rds-peripherin gene. Keen, T.J., Inglehearn, C.F., Kim, R.C., Bird, A.C. and Bhattacharya, S.S. (1994). *Hum. Mol. Genet.* **3**: 376-368.
78. Two new rhodopsin transversion mutations at codons 40 and 216 in families with autosomal dominant retinitis pigmentosa. Al-Magthteh, M., Inglehearn, C.F., Lunt, P., Jay, M., Bird, A.C. and Bhattacharya, S.S. (1994). *Human Mutation*. **3**: 409-410.
79. A study of X chromosome activity in two Incontinentia Pigmenti families with probable linkage with Xq28. Curtis, A.R.J., Lindsay, S., Boye, E., Clarke, A.J., Landy, S.J., and Bhattacharya, S. (1994). *Eu. J. Hum. Genet.* **2**: 51-58.
80. Ocular manifestation in autosomal dominant retinitis pigmentosa with a lys-296-Glu rhodopsin mutation at the retinal binding site. Owens, S., Fitzke, F.W., Inglehearn, C.F., Jay, M.R., Keen, T., Arden, G.B., Bhattacharya, S.S., Bird, A.C. (1994). *Br. J. Ophthal.* **78**: 353-358.
81. Macular dystrophy associated with mutations at codon 172 in the human retinal degeneration slow (RDS) gene. Wroblewski, J.J., Wells, J.A., Eckstein, A., Fitzke, F.W., Jubb, C., Keen, T.J., Inglehearn, C., Bhattacharya, S., Arden, G.B., Jay, M., and Bird, A.C. (1994). *Ophthalmology*. **101**: 12-22.

82. Loci for autosomal dominant retinitis pigmentosa and dominant cystoid macular dystrophy on chromosome 7p are not allelic. Inglehearn, C.F., Keen, T.J., Al-Maghteh, M. and Bhattacharya, S. (1994). *Am. J. Hum. Genet.* **55**: 581-582.
83. Refinement of the cone-rod retinal dystrophy locus on chromosome 19q. Gregory C.Y., Evans K, Whittaker J, Fryer A, Weissenbach J and Bhattacharya S.S. (1994). *Am. J. Hum. Genet.* **55**: 1061-1063.
84. The role of molecular genetics in the prenatal diagnosis of retinal dystrophies. Evans K, Gregory C.Y., Fryer A, Whittaker J, Duvall-Young J, Bird A.C. and Bhattacharya S.S. (1994). *Eye.* **9**: 24-28.
85. Ocular findings associated with a three-base-pair deletion in the peripherin-RDS gene in autosomal dominant Retinitis Pigmentosa. Wroblewski, J.J., Wells, J.A., Eckstein, A., Fitzke, F., Jubb, C., Keen, T.J., Inglehearn, C.F., Bhattacharya, S.S., Arden, G.B., Jay, M. and Bird, A.C. (1994). *Br. J. Ophthalmol.* **78**: 381-386.
86. Three novel rhodopsin mutations (C110F, L131P, A164V) in patients with autosomal dominant retinitis pigmentosa. Fuchs, S., Kranich, H, Denton, M.J., Zrenner, E., Bhattacharya, S.S., Humphries, P. and Gal, A. (1994). *Hum. Mol. Genet.* **3**: 1203.
87. Retinitis pigmentosa families showing apparent X linked inheritance but unlinked to the RP2 or RP3 loci. Aldred, M.A., Teague, P.W., Jay, M., Bunday, S., Redmond, R.M., Jay, B., Bird, A.C., Bhattacharya, S.S. and Wright, A.F. (1994). *J. Med. Genet.* **31**: 848-852.
88. Genetic heterogeneity in hereditary haemorrhagic telangiectasia. Porteous, M.E., Curtis, A., Williams, O., Marchuk, D., Bhattacharya, S.S. and Burn, J. (1994). *J. Med. Genet.* **31**: 925-926.
89. Dinucleotide repeat polymorphism at the DXS977 locus. Yan, D., Wong, D., Zheng, K., Thiselton, D., Fujita, R., Sieving, P.A., Bhattacharya, S.S., Yang-Feng, T.L., Richards, J.E. and Swaroop, A. (1994). *Hum. Mol. Genet.* **3**: 1030.
90. Autosomal dominant macular dystrophy simulating North Carolina macular dystrophy. Holz F.G., Evans K, Gregory C. Y., Bhattacharya S.S. and Bird A.C. (1995). *Arch. Ophthalmol.* **113**: 176-193.
91. Chromosome 19q cone-rod retinal dystrophy: ocular phenotype. Evans K Duvall-Young J, Fitzke F, Arden G.B., Bhattacharya S.S. and Bird A.C. (1995). *Arch. Ophthalmol.* **113**: 195-201.
92. Genetic Refinement of the chromosome 5q lattice corneal dystrophy type I to within two centimorgan interval. Gregory C.Y., Evans K, and Bhattacharya S.S. (1995). *J. Med. Genet.* **32**: 224-226.
93. Autosomal dominant pattern dystrophy of the retina associated with a 4-base pair

- insertion at codon 140 in the peripherin/RDS gene. Kim, R.Y., Dollfus, H., Keen, T.J., Fitzke, G.B., Arden, G.B., Bhattacharya, S.S. and Bird, A.C. (1995) *Arch. Ophthalmol.* **113**: 451-455.
94. Autosomal dominant retinitis pigmentosa mapping to chromosome 7p exhibits variable expression. Kim, R.Y., Fitzke, F.W., Moore, A.T., Inglehearn, C., Arden, G.B., Bhattacharya, S.S. and Bird, A.C. (1995). *Br. J. Ophthal.* **79**: 23-27.
  95. Genetic and Physical Mapping of Five Novel Microsatellite Markers on Human Xp21.1-p11.22. Thiselton, D.L., Lindsay, S., Kamakari, S., Hardcastle, A.J., Roustan, P. and Bhattacharya, S.S. (1995). *Genomics.* **25**: 279-281.
  96. Linkage refinement localises Sorsby fundus dystrophy between markers D22S275 and D22S278. Gregory, C.Y., Wijesuriya, S., Evans, K., Jay, M.R., Bird, A.C. and Bhattacharya, S.S. (1995). *J. Med. Genet.* **32**: 240-241.
  97. Localisation of the aquaporin 1 (AQP1) gene within a YAC contig containing the polymorphic markers D7S632 and D7S526. Keen, T.J., Inglehearn, C.F., Patel, R.J., Green, E.D., Peluso, D.C., and Bhattacharya, S.S. (1995). *Genomics.* **25**: 599-600.
  98. A YAC contig spanning the dominant retinitis pigmentosa locus (RP9) on chromosome 7p. Keen, T.J., Inglehearn, C.F., Green, E.D., Cunningham, A.F., Patel, R.J., Peacock, R.E., Gerken, S., White, R., Weissenbach, J. and Bhattacharya, S.S. (1995). *Genomics.* **28**: 383-388.
  99. An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. Bardien, S., Ebenezer, N., Greenberg, J., Inglehearn, C.F., Bartmann, L., Goliath, R., Beighton, P., Ramesar, R. and Bhattacharya, S.S. (1995). *Hum.Mol.Genet.* **4**: 1459-1462.
  100. Linkage of congenital hereditary endothelial dystrophy to chromosome 20. Toma, N.M.G., Ebenezer, N.D., Inglehearn, C.F., Plant, C., Ficker, L.A. and Bhattacharya, S.S. (1995). *Hum. Mol. Genet.* **4**: 2395-2398.
  101. Regional assignment of thirty expressed sequence tags (ESTs) on human chromosome 7 using a somatic cell hybrid panel. Patel, R.J., Keen, T.J., Grzeschik, K-H, Nierman, W.C., Hayes, P., Bhattacharya, S.S. and Inglehearn, C.F. (1995). *Genomics.* **30**: 112-114.
  102. cDNA sequence and gene locus of human retinal phosphoinositide-specific phospholipase-Cb4 (PLCb4). Alvarez, R.A., Ghalayini, A.J., Xu, P., Hardcastle, A., Bhattacharya, S.S., Rao, P.N., Pettenati, M.J., Anderson, R.E. and Baehr, W. (1995). *Genomics.* **29**: 53-61.
  103. Gene transfer into the mouse retina mediated by an adeno-associated viral vector. Ali, R.R., Reichel, M.B., Thrasher, A.J., Levinsky, R.J., Kinnon, C., Kanuga, N., Hunt, D.M. and Bhattacharya, S.S. (1996). *Hum. Mol. Genet.* **6**: 591-594.

104. Simple tests for rhodopsin involvement in Retinitis pigmentosa. Tarttelin, E.E., Al-Magthteh, M., Keen, T.J., Bhattacharya, S.S. and Inglehearn, C.F. (1996). *J. Med. Genet.* **33**: 262-263.
105. A new family linked to the RP13 locus for autosomal dominant retinitis pigmentosa on distal 17p. Tarttelin, E.E., Plant, C., Weissenbach, J., Bird, A.C., Bhattacharya, S.S. and Inglehearn, C.F. (1996). *J. Med. Genet.* **33**: 518-520.
106. Evidence for a major retinitis pigmentosa locus on 19q 13.4 (RP11) and association with a unique bimodal expressivity phenotype. Al-Magthteh, M., Vithana, E., Tarttelin, E.E., Jay, M., Evans, K., Moore, T., Bhattacharya, S.S. and Inglehearn, C.F. (1996). *Am. J. Hum. Genet.* **59**: 864-871.
107. A locus for autosomal dominant anterior polar cataract on chromosome 17p. Berry, V., Ionides, A.C.W., Moore, A.T., Plant, C., Bhattacharya, S.S. and Shiels, A. (1996). *Hum. Mol. Genet.* **5**: 415-419.
108. Sorsby's fundus dystrophy in the British Isles: demonstration of a striking founder effect by microsatellite generated haplotypes. Wijesuriya, S.D., Evans, K., Jay, M.R., Davison, C., Weber, B.H.G., Bird, A.C., Bhattacharya, S.S. and Gregory, C.Y. (1996). *Genome Res.* **6**: 92-101.
109. The gene responsible for autosomal dominant Doyme's honeycomb retinal dystrophy (DHRD) maps to chromosome 2p16. Gregory, C.Y., Evans, K., Wijesuriya, S.D., Kermani, S., Jay, M.R., Plant, C., Cox, N., Bird, A.C. and Bhattacharya, S.S. (1996). *Hum. Mol. Genet.* **5**: 1055-1059.
110. Mapping the RP2 locus for X-linked retinitis pigmentosa on proximal Xp; A genetically defined 5cM critical region and exclusion of candidate genes by physical mapping. Thiselton, D.L., Hardcastle, A.J., Hampson, R.M., Nayudu, M., Van Maldergen, L., Wolf, M.L., Saha, B.K. and Bhattacharya, S.S. (1996). *Genome Res.* **6**: 1093-1102.
111. A locus for autosomal dominant posterior polar cataract on chromosome 1p. Ionides, A.C.W., Berry, V., Mackay, D.S., Moore, A.T., Bhattacharya, S.S. and Shiels, A. (1997). *Hum. Mol. Genet.* **6**: 47-51.
112. Genetic refinement of dominant optic atrophy (OPA1) locus to within a 2cM interval of chromosome 3q. Votruba, M., Moore, A.T. and Bhattacharya, S.S. (1997). *J. Med. Genet.* **34**: 117-121.
113. Genomic organisation of the human TIMP-1 gene: Investigation of a causative role in the pathogenesis of X-linked retinitis pigmentosa-2. Hardcastle, A.J., Thiselton, D.L., Nayudu, M., Hampson, R.M. and Bhattacharya, S.S. (1997). *Invest. Ophthalmol. Vis. Sci.* **38**: 1893-1896.
114. Isk and KvLQT1: mutation in either of the two subunits of the slow component of the delayed rectifier potassium channel can cause Jervell and Lange-Nielsen syndrome. Tyson, J., Tranebjaerg, L., Bellman, S., Wren, C., Taylor, JF, Bathen, J,

- Aslaksen, B, Sorland, SJ, Lund, O, Malcolm, S, Pembury, M, Bhattacharya, S, and Bitner-Glindzicz, M, (1997). *Hum. Mol. Genet.* **6**: 2179-2185.
115. A new locus for dominant 'Zonular Pulverulent' cataract on chromosome 13. Mackay, D., Ionides, A., Berry, V., Moore, A., Bhattacharya, S. and Shiels, A. (1997). *Am. J. Hum. Genet.* **60**: 1474-1478.
116. Gene therapy for inherited eye disease. Ali, R.R., Reichel, M.B., Hunt, D.M. and Bhattacharya, S.S. (1997). *Brit. J. Ophthalmol.* **81**: 795-802.
117. Localisation of CSNBX (CSNB4) between the retinitis pigmentosa loci RP2 and RP3 on proximal Xp. Hardcastle, A.J., David-Gray, Z.K., Jay, M., Bird, A.C. and Bhattacharya, S.S. (1997). *Invest. Ophthalmol. Vis. Sci.* **38**: 2750-2755.
118. Retinitis pigmentosa locus on 17q (RP17): fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes. Bardien, S., Ramesar, R., Bhattacharya, S. and Greenberg, J. (1997). *Hum. Genet.* **101**: 13-17.
119. Familial Glaucoma Iridogoniodysplasia Maps to a 6p25 Region Implicated in Primary Congenital Glaucoma and Iridogoniodysgenesis Anomaly. Jordan, T., Ebenezer, N., Manners, R., McGill, J. and Bhattacharya, S.S. (1997). *Am. J. Hum. Genet.* **61**: 882-888.
120. Cone-Rod Dystrophy due to mutations in a novel photoreceptor-specific homeobox gene (CRX) essential for maintenance of the photoreceptor. Freund, C.L., Gregory-Evans, C.Y., Furukawa, T., Papaioannou, M., Looser, J., Ploder, L., Bellingham, J., Ng, D., Herbrick, J.S., Duncan, A., Scherer, S.W., Tsui, L., Loutradis-Anagnostou, A., Jacobson, S.G., Cepko, C.L., Bhattacharya, S.S. and McInnes, R.R. (1997). *Cell.* **91**: 543-553.
121. Effect of varying the mitomycin-C treatment area in glaucoma filtration surgery in the rabbit. Cordeiro, M.F., Constable, P.H., Alexander, R.A., Bhattacharya, S.S. and Khaw, P.T. (1997). *Invest Ophthalmol Vis Sci.* **38**: 1639-1646.
122. Single exposures to antiproliferatives: long-term effects on ocular fibroblast wound-healing behavior. Occleston, N.L., Daniels, J.T., Tarnuzzer, R.W., Sethi, K.K., Alexander, R.A., Bhattacharya, S.S., Schultz, G.S. and Khaw, P.T. (1997). *Invest Ophthalmol Vis Sci.* **38**: 1998-2007.
123. Gene therapy for retinal degeneration. Reichel, M.B., Ali, R.R., Hunt, D.M. and Bhattacharya, S.S. (1997). *Ophthalmic Res.* **29**: 261-268.
124. High frequency of hyperplastic primary vitreous in p53-deficient mice. Reichel, M.B., Ali, R.R., D'Esposito, F., Clarke, A.R., Luthert, P.J., Bhattacharya, S.S. and Hunt, D.M. (1998). *Cell Death and Differentiation.* **5**: 156-162.
125. Adeno-associated virus gene transfer to mouse retina. Ali, R.R., Reichel, M.B., de Alwis, M., Kanuga, N., A.J., Levinsky, Hunt, D.M., Bhattacharya, S.S. and Thrasher, A.J. (1998). *Human Gene Therapy.* **9**: 81-86.

126. Demonstration of a founder effect and fine mapping of dominant optic atrophy locus on 3q28-qter by linkage disequilibrium method. Votruba, M., Moore, A.T. and Bhattacharya, S.S. (1998). *Hum. Genet.* **102**: 79-86.
127. A mutation in guanylate cyclase activator 1A (GUCA1A) in an autosomal dominant cone dystrophy pedigree mapping to a new locus on chromosome 6p21.1. Payne, A.M., Downes, S.M., Bessant, D.A., Taylor, R., Holder, G.E., Warren, M.J., Bird, A.C. and Bhattacharya, S.S. (1998). *Hum. Mol. Genet.* **7**: 273-277.
128. GCAP1(Y99C) Mutant is constitutively active in autosomal dominant cone dystrophy. Sokal, I., Li, N., Surgucheva, I., Warren, M.J., Payne, A.M., Bhattacharya, S.S., Baehr, W. and Palczewski, K. (1998). *Molecular Cell.* **2**: 129-133.
129. A missense mutation in the human connexin50 gene (GJA8) underlies autosomal dominant "zonular pulverulent" cataract, on chromosome 1q. Shiels, A., Mackay, D., Ionides, A., Berry, V., Moore, A. and Bhattacharya, S.S. (1998). *Am. J. Hum. Genet.* **62**: 526-532.
130. A locus for autosomal recessive congenital microphthalmia maps to chromosome 14q32. Bessant, D. R., Khaliq, S., Hameed, A., Anwar, K., Mehdi, S.Q., Payne, A. M. and Bhattacharya, S.S. (1998). *Am. J. Hum. Genet.* **62**: 1113-1116.
131. Segregation of a PRKCG mutation in two RP11 families. Al Maghtheh, M., Vithana, E. N., Iglehearn, C.F., Moore, T.A., Bird, A.C. and Bhattacharya, S.S. (1998). *Am. J. Hum. Genet.* **62**: 1248-1252.
132. Genetic blindness: current concepts in the pathogenesis of human outer retinal dystrophies. Gregory-Evans, K. and Bhattacharya, S.S. (1998). *Trends in Genetics.* **14**: 103-108.
133. Founder effect seen in the British population, of the peripherin/RDS mutation and further refinement of genetic positioning of the peripherin/RDS gene. Payne, A.M., Downes, S.M, Bessant, D.A.R., Bird, A.C. and Bhattacharya, S.S. (1998). *Am. J. Hum. Genet.* **62**: 192-195.
134. Clinical features in affected individuals from 21 pedigrees with dominant optic atrophy. Votruba, M., Fitzke, F.W., Holder, G.E., Carter, A., Bhattacharya, S.S. and Moore, A.T. (1998). *Arch. Ophthalmol.* **116**: 351-358.
135. Anterior polar cataract: clinical spectrum and genetic linkage in a single family. Ionides, A., Berry, V., Mackay, D., Shiels, A., Bhattacharya, S., and Moore, A. (1998). *Eye*, **12**: 224-226.
136. A linkage survey of 20 dominant retinitis pigmentosa families: frequencies of the nine known loci and evidence for further heterogeneity. Iglehearn, C.F., Tartelin, E.E., Peacock, R.E., Al-Maghtheh, M., Vithana, E., Bird, A.C. and Bhattacharya, S.S. (1998). *J. Med. Genet.* **35**: 1-5.

137. RP11 is the second most common locus for dominant retinitis pigmentosa. Vithana, E., Al-Magthteh, M., Bhattacharya, S.S. and Inglehearn, C.F. (1998). *J. Med. Genet.* **35**: 174-175.
138. A new family of Greek origin maps to the CRD locus for autosomal dominant cone-rod dystrophy on 19q. Papaioannou, M., Bessant, D., Payne, A., Bellingham, J., Rougas, C., Loutradis-Anagnostou, A., Gregory-Evans, C., Balassopoulou, A. and Bhattacharya S. (1998). *J. Med. Genet.* **35**: 429-431.
139. Immune responses limit adenovirally-mediated gene expression in the adult mouse eye. Reichel, M.B., Ali, R.R., Thrasher, A.J., Hunt, D.M., Bhattacharya, S.S. and Baker, D. (1998). *Gene Therapy.* **5**: 1038-1046.
140. Co-injection of adenovirus expressing CTLA4-Ig prolongs adenovirally-mediated LacZ reporter gene expression in the mouse retina. Ali, R.R, Reichel, M.B., Baker, D., Byrnes, A.P., Kanuga, N., Hunt, D.M., and Bhattacharya, S.S. (1998). *Gene Therapy.* **5**: 1561-1565.
141. Absence of p53 delays apoptotic photoreceptor cell death in the rds mouse. Ali, R. R., Reichel, M. B., Kanuga, N., Munro, P. M., Alexander, R. A., Clarke, A. R., Luthert, P. J., Bhattacharya, S. S., and Hunt, D. M. (1998). *Curr. Eye Res.* **17**: 917-923.
142. Dominant optic atrophy: exclusion and fine genetic mapping of the candidate gene, HRY. Votruba, M., Payne, A., Moore, A.T., and Bhattacharya, S.S. (1998). *Mam. Genome* **9**: 784-787.
143. Further refinement of the Usher 2A locus at 1q41. Bessant, D.A., Payne, A.M., Plant, C., Bird, A.C. and Bhattacharya, S.S. (1998). *J. Med. Genet.* **35**: 773-774.
144. A new dominant retinitis pigmentosa family mapping to the RP18 locus on chromosome 1q11-21. Inglehearn, C.F., Tarttelin, E.E., Keen, T.J., Bhattacharya, S.S., Moore, A.T., Taylor, R., and Bird, A.C. (1998). *J. Med. Genet.* **35**: 788-789.
145. Clinical features, molecular genetics, and pathophysiology of dominant optic atrophy. Votruba, M., Moore A.T., and Bhattacharya, S.S. (1998). *J. Med. Genet.* **35**: 793-800.
146. Mutations of the Forkhead/Winged-Helix gene, FKHL7, in patients with Axenfeld-Rieger anomaly. Mears, A.J., Jordan, T., Mirzayans, F., Dubois, S., Kume, T., Parlee, M., Ritch, R., Koop, B., Kuo, W-L., Collins, C., Marshall, J., Gould, D.B., Pearce, W., Carlsson, P., Enerback, S., Morissette, J., Bhattacharya, S.S., Hogan, B., Raymond, V. and Walter, M.A. (1998). *Am. J. Hum. Genet.* **63**: 1316-1328.
147. New model of conjunctival scarring in the mouse eye. Reichel, M.B., Cordeiro, M.F., Alexander, R.A., Cree, I.A., Bhattacharya, S.S. and Khaw, P.T. (1998). *Br. J. Ophthalmol.* **82**: 1072-1077.
148. Refined genetic and physical positioning of the gene for Doyme honeycomb



- retinal dystrophy. Kermani, S., Gregory-Evans, K., Tarttelin, E.E., Bellingham, J., Plant, C., Bird, A.C., Fox, M., Bhattacharya, S.S. and Gregory-Evans, C.Y. (1999). *Hum Genet.* **104**: 77-82.
149. Mutations in the RP2 Gene cause disease in 10% of Familial XLRP assessed in this study. Hardcastle, A.J., Thiselton, D.L., Maldergem, L.V., Saha, B.K., Jay, M., Plant, C., Taylor, R., Bird, A.C. and Bhattacharya, S.S. (1999). *Am. J. Hum. Genet.* **64**: 1210-1215.
150. Molecular Genetic Study of Autosomal Dominant Retinitis pigmentosa in Lithuanian Patients. Kucinskas, V., Payne, A.M., Ambrasiene, D., Jurgelevicius, V., Steponaviciaumacrite, D., Arciuliene, J.V., Daktaraviciene, E. and Bhattacharya, S. (1999). *Hum. Hered.* **49**: 71-74.
151. Mutation in *NRL* is associated with autosomal dominant retinitis pigmentosa. Bessant, D.A.R., Payne, A.M., Mitton, K.P., Wang, Q-L., Swain, P.K., Plant, C., Bird, A.C., Zack, D.J., Swaroop, A. and Bhattacharya, S.S. (1999). *Nat. Genet.* **21**: 355-356.
152. Lens biology, development and human cataractogenesis. Francis, P., Berry, V., Moore, A. and Bhattacharya, S.S. (1999). *Trends in Genetics*, **15**: 191-196.
153. Connexin-46 mutations in autosomal dominant congenital cataract. Mackay, D., Ionides, A., Kibar, Z., Rouleau, G., Berry, V., Moore, A., Shiels, A. and Bhattacharya, S. (1999). *Am. J. Hum. Genet.* **64**: 1357-1364.
154. Severe autosomal dominant retinitis pigmentosa caused by a novel rhodopsin mutation (Ter349Glu). Mutations in brief no.208. Online. Bessant, D.A., Khaliq, S., Hameed, A., Anwar, K., Payne, A.M., Mehdi, S.Q. and Bhattacharya, S.S. (1999). *Hum. Mutat.* **13**: 83.
155. Refinement of the locus for autosomal recessive retinitis pigmentosa (RP25) linked to chromosome 6q in a family of Pakistani origin. Khaliq, S., Hameed, A., Ismail, M., Mehdi, S.Q., Bessant, D.A., Payne, A.M. and Bhattacharya, S.S. (1999). *Am. J. Hum. Genet.* **65**: 571-574.
156. Phenotype of autosomal recessive congenital microphthalmia mapping to chromosome 14q32. Bessant, D.A., Anwar, K., Khaliq, S., Hameed, A., Ismail, M., Payne, A.M., Mehdi, S.Q. and Bhattacharya, S.S. (1999). *Br. J. Ophthalmol.* **83**: 919-922.
157. Connexin 50 mutation in a family with congenital “zonular nuclear” pulverulent cataract of Pakistani origin. Berry, V., Mackay, D., Khaliq, S., Francis, P.J., Hameed, A., Anwar, K., Mehdi, S.Q., Newbold, R.J., Ionides, A., Shiels, A., Moore, A. and Bhattacharya, S.S. (1999). *Hum. Genet.* **105**: 168-170.
158. Mutations in the RP1 gene causing autosomal dominant retinitis pigmentosa. Bowne, S.J., Daiger, S.P., Hims, M.W., Sohocki, M.M., Malone, K.A., McKie, A.B., Heckenlively, J.R., Birch, D.R., Inglehearn, C.F., Bhattacharya, S.S., Bird, A., and Sullivan, L.S. (1999). *Hum. Mol.Genet.* **11**: 2121-2128.

159. Clinical and genetic heterogeneity in autosomal dominant cataract. Ionides, A., Francis, P., Berry, V., Mackay, D., Bhattacharya, S., Shiels, A., and Moore, A. (1999). *Br. J. Ophthalmol.* **83**: 802-808.
160. Identification of novel RPGR (retinitis pigmentosa GTPase regulator) mutations in a subset of X-linked retinitis pigmentosa families segregating with the RP3 locus. Zito, I., Thiselton, D.L., Gorin, M.B., Stout, J.T., Plant, C., Bird, A.C., Bhattacharya, S.S. and Hardcastle, A.J. (1999). *Hum Genet.* **105**: 57-62.
161. Genetic analysis of the guanylate cyclase activator 1B (GUCA1B) gene in patients with autosomal dominant retinal dystrophies. Payne, A.M., Downes, S.M., Bessant, D.A., Plant, C., Moore, T., Bird, A.C. and Bhattacharya, S.S. (1999). *J. Med. Genet.* **36**: 691-693.
162. Mutations in a human homolog of *Drosophila* crumbs cause retinitis pigmentosa with preserved para-arteriolar retinal pigment epithelium (RP12). den Hollander, A.I., ten Brink, J.B., de Kok, Y.J.M., van Soest, S., van den Born, L. I., van Driel, M.A., van de Pol, D.J.R., Payne, A.M., Bhattacharya, S.S., Kellner, U., Hoyng, C.B., Westerveld, A., Brunner, H.G., Bleeker-Wagemakers, E.M., Deutman, A.F., Heckenlively, J.R., Cremers F.P.M., and Bergen, A.A.B. (1999). *Nat. Genet.* **23**: 217-221.
163. Characterization of the human diacylglycerol kinase epsilon gene and its assessment as a candidate for inherited retinitis pigmentosa. Tang, W., Bardien, S., Bhattacharya, S.S. and Prescott, S.M. (1999). *Gene*, **239**: 185-92.
164. Molecular therapy in ocular wound healing. Cordeiro, M.F., Schultz, G.S., Ali, R.R., Bhattacharya, S.S. and Khaw, P.T. (1999). *Br J Ophthalmol.* **83**: 1219-1224.
165. Clinical features of codon 172 RDS macular dystrophy: similar phenotype in 12 families. Downes, S.M., Fitzke, F.W., Holder, G.E., Payne, A.M., Bessant, D.A., Bhattacharya, S.S. and Bird, A.C. (1999). *Arch. Ophthalmol.* **117**: 1373-1383
166. Electrophysiological findings in dominant optic atrophy (DOA) linking to the OPA1 locus on chromosome 3q 28-qter. Holder, G.E., Votruba, M., Carter, A.C., Bhattacharya, S.S., Fitzke, F.W. and Moore, A.T. (1998-99). *Doc. Ophthalmol.* **95**: 217-228.
167. An analysis of ABCR mutations in British patients with recessive retinal dystrophies. Papaioannou, M., Ocaka, L., Bessant, D., Lois, N., Bird, A., Payne, A. and Bhattacharya, S. (2000). *Invest. Ophthalmol. Vis.Sci.* **41**: 16-19.
168. A novel locus for Leber Congenital Amaurosis with Anterior Keratoconus mapping to chromosome 17p13. Hameed, A., Khaliq, S., Ismail, M., Anwar, K., Ebenezer, N. D., Jordan, T., Mehdi, S.Q., Payne, A.M. and Bhattacharya S.S. (2000). *Invest. Ophthalmol. Vis. Sci.* **41**: 629-633.
169. Mutations in a novel photoreceptor-pineal gene on 17p cause Leber congenital amaurosis (LCA4). Sohocki, M.M., Bowne, S.J., Sullivan, L.S., Blackshaw, S., Cepko,

- C.L., Payne, A.M., Bhattacharya, S.S., Khaliq, S., Qasim Mehdi, S., Birch, D.G., Harrison, W.R., Elder, F.F., Heckenlively, J.R. and Daiger, S.P. (2000). *Nat. Genet.* **24**: 79-83.
170. Novel mutations of the RPGR gene in RP3 families. Zito, I., Gorin, M.B., Plant, C., Bird, A.C., Bhattacharya, S.S. and Hardcastle, A.J. (2000). *Hum. Mutat.* (Online), **15**: 386.
171. TGF-beta1,-beta2, and -beta3 in vitro: biphasic effects on Tenon's fibroblast contraction, proliferation, and migration. Cordeiro, M.F., Bhattacharya S.S., Schultz, G.S. and Khaw, P.T. (2000). *Invest. Ophthalmol. Vis. Sci.* **41**: 756-763.
172. Importance of the autosomal recessive retinitis pigmentosa locus on 1q31-q32.1 (RP12) and mutation analysis of the candidate gene *RGS16* (*RGS-r*). Bessant, D.A.R., Payne, A.M., Snow, B., Antinolo, G., Mehdi, S.Q., Bird, A.C., Siderovski, D.P. and Bhattacharya, S.S. (2000). *J. Med. Genet.* **37**: 384-387.
173. Novel frameshift mutations in the RP2 gene and polymorphic variants. Thiselton, D.L., Zito, I., Plant, C., Jay, M., Hodgson, S.V., Bird, A.C., Bhattacharya, S.S. and Hardcastle, A.J. (2000). *Hum. Mutat.* (Online), **15**: 580.
174. Missense mutations in *MIP* underlie autosomal dominant 'polymorphic' and lamellar cataracts linked to 12q. Berry, V., Francis, P., Kaushal, S., Moore, A. and Bhattacharya, S. (2000). *Nat. Genet.* **25**: 15-17.
175. Restoration of photoreceptor ultrastructure and function in retinal degeneration slow mice by gene therapy. Ali, R.R., Sarra, G.M., Stephens, C., Alwis, M.D., Bainbridge, J.W., Munro, P.M., Fauser, S., Reichel, M.B., Kinnon, C., Hunt, D.M., Bhattacharya, S.S. and Thrasher, A.J. (2000). *Nat. Genet.* **25**: 306-310.
176. Mutations in a novel photoreceptor-pineal gene on 17p cause Leber congenital amaurosis (LCA4). Sohocki, M.M., Bowne, S.J., Sullivan, L.S., Blackshaw, S., Cepko, C.L., Payne, A.M., Bhattacharya, S.S., Khaliq, S., Qasim Mehdi, S., Birch, D.G., Harrison, W.R., Elder, F.F., Heckenlively, J.R. and Daiger, S.P. (2000). *Am. J. Ophthalmol.* **129**: 834-5.
177. MRI of the intraorbital optic nerve in patients with autosomal dominant optic atrophy. Votruba, M., Leary, S., Losseff, N., Bhattacharya, S.S., Moore, A.T., Miller, D.H. and Moseley, I.F. (2000). *Neuroradiology*, **42**: 180-183.
178. The genetics of childhood cataract. Francis, P.J., Berry, V., Bhattacharya, S.S. and Moore, A.T. (2000). *J. Med. Genet.* **37**: 481-488.
179. Abnormalities of the transforming growth factor-beta pathway in ocular melanoma. Myatt, N., Aristodemou, P., Neale, M.H., Foss, A.J., Hungerford, J.L., Bhattacharya, S. and Cree I. A. (2000). *J Pathol.* **192**: 511-518.
180. Prevalence of AIPL1 mutations in inherited retinal degenerative disease. Sohocki, M.M., Perrault, I., Leroy, B.P., Payne, A.M., Dharamraj, S., Bhattacharya, S.S., Kaplan,

- J., Maumenee, I.H., Koenekoop, R., Meire, F.M., Birch, D.G., Heckenlively, J.R. and Daiger, S.P. (2000). *Mol. Genet. Metab.* **70**: 142-150.
181. Characterization of the human TBX20 gene, a new member of the T-Box gene family closely related to the drosophila H15 gene. Meins, M., Henderson, D.J., Bhattacharya, S.S. and Sowden, J.C. (2000). *Genomics* **67**: 317-332.
182. Evidence for a new locus for X-linked retinitis pigmentosa (RP23). Hardcastle, A.J., Thiselton, D.L., Zito, I., Ebenezer, N., Mah, T.S., Gorin, M.B. and Bhattacharya, S.S. (2000). *Invest. Ophthalmol. Vis. Sci.* **41**: 2080-2086.
183. Sequence variation within the RPGR gene: Evidence for a founder complex allele. Zito, I., Morris, A., Tyson, P., Winship, I., Sharp, D., Gilbert, D., Thiselton, D.L., Bhattacharya, SS, and Hardcastle, A.J. (2000). *Hum. Mutat.* **16**: 273-274.
184. Functional impairment of lens aquaporin in two families with dominantly inherited cataracts. Francis, P., Chung, J-J., Yasui, M., Berry, V., Moore, A., Wyatt, M.K., Wistow, G., Bhattacharya, S.S. and Agre, P. (2000). *Hum. Mol. Genet.* **9**: 2329-2334.
185. *OPA1*, encoding a dynamin-related GTPase, is mutated in autosomal dominant optic atrophy linked to chromosome 3q28. Alexander, C., Votruba, M., Pesch, U.E.A., Thiselton, D.L., Mayer, S., Moore, A., Rodriguez, M., Kellner, U., Leo-Kottler, B., Auburger, G., Bhattacharya, S.S. and Wissinger, B. (2000). *Nat. Genet.* **26**: 211-215.
186. Chromosomal duplication involving the Forkhead Transcription Factor Gene FOXC1 causes Iris Hypoplasia and Glaucoma. Lehmann, O.J., Ebenezer, N.D., Jordan, T., Fox, M., Ocaka, L., Payne, A., Leroy, B.P., Clark, B.J., Hitchings, R.A., Povey, S., Khaw, P.T. and Bhattacharya, S.S. (2000). *Am. J. Hum. Genet.* **67**: 1129-1135.
187. Novel Locus for Autosomal Recessive Cone-Rod Dystrophy CORD8 Mapping to Chromosome 1q12-q24. Khaliq, S., Hameed, A., Ismail, M., Anwar, K., Leroy, B., Mehdi, S.Q., Payne, A.M. and Bhattacharya, S.S. (2000). *Invest. Ophthalmol. Vis.Sci.* **41**: 3709-3712.
188. Congenital progressive polymorphic cataract caused by a mutation in the major intrinsic protein of the lens, MIP. Francis, P., Berry, V., Bhattacharya, S. and Moore, A. (2000). *Br. J. Ophthalmol.* **84**: 1376-1379.
189. NRL S50T mutation and the importance of 'founder effects' in inherited retinal dystrophies. Bessant, D.A., Payne, A.M., Plant, C., Bird, A.C., Swaroop, A. and Bhattacharya, S.S. (2000). *Eur. J. Hum. Genet.* **8**: 783-787.
190. Functional characterization of missense mutations at codon 838 in retinal guanylate cyclase correlates with disease severity in patients with autosomal dominant cone-rod dystrophy. Wilkie, S.E., Newbold, R.J., Deery, E., Walker, C.E., Stinton, I., Ramamurthy, V., Hurley, J.B., Bhattacharya, S.S., Warren, M.J. and Hunt, D.M. (2000). *Hum. Mol. Genet.* **9**: 3065-3073.

191. RP1 protein truncating mutations predominate at the RP1 adRP locus. Payne, A., Vithana, E., Khaliq, S., Hameed, A., Deller, J., Abu-Safieh, L., Kermani, S., Leroy, B.P., Mehdi, S.Q., Moore, A.T., Bird, A.C. and Bhattacharya, S.S. (2000). *Invest. Ophthalmol. Vis. Sci.* **41**: 4069-4073.
192. Abnormalities of the transforming growth factor-beta pathway in ocular melanoma. Myatt, N., Aristodemou, P., Neale, M.H., Foss, A.J., Hungerford, J.L., Bhattacharya, S. and Cree, I.A. (2000). *J. Pathol.* **192**: 511-518.
193. The destabilisation of human GCAP1 by a proline to leucine mutation might cause cone-rod dystrophy. Newbold, R.J., Deery, E.C., Walker, C.E., Wilkie, S.E., Srinivasan, N., Hunt, D.M., Bhattacharya, S.S. and Warren, M.J. (2001). *Hum. Mol. Genet.* **10**: 47-54.
194. Autosomal Dominant Cone and Cone-Rod Dystrophy With Mutations in the Guanylate Cyclase Activator 1A Gene-Encoding Guanylate Cyclase Activating Protein-1. Downes, S.M., Holder, G.E., Fitzke, F.W., Payne, A.M., Warren, M.J., Bhattacharya, S.S. and Bird, A.C. (2001). *Arch Ophthalmol.* **119**: 96-105.
195. An immune response after intraocular administration of an adenoviral vector containing a beta galactosidase reporter gene slows retinal degeneration in the rd mouse. Reichel, M.B., Bainbridge, J., Baker, D., Thrasher, A.J., Bhattacharya, S.S. and Ali, R.R. (2001). *Br. J. Ophthalmol.* **85**: 341-344.
196. A new locus for autosomal recessive rp (rp29) mapping to chromosome 4q32-q34 in a Pakistani family. Hameed, A., Khaliq, S., Ismail, M., Anwar, K., Mehdi, S.Q., Bessant, D., Payne, A.M. and Bhattacharya, S.S. (2001). *Invest Ophthalmol Vis Sci.* **42**:1436-1438.
197. Molecular genetics and prospects for therapy of the inherited retinal dystrophies. Bessant, D.A., Ali, R.R. and Bhattacharya, S.S. (2001). *Curr Opin Genet Dev.* **11**: 307-316.
198. Spectrum of mutations in ush2a in British patients with usher syndrome type ii. Leroy, B.P., Aragon-Martin, J.A., Weston, M.D., Bessant, D.A., Willis, C., Webster, A.R., Bird, A.C., Kimberling, W.J., Payne, A.M., and Bhattacharya, S.S. (2001). *Exp Eye Res.* **72**: 503-509.
199. Assignment (1) of BCL2L11 to human chromosome band 2p13 with somatic cell and radiation hybrids. Murray, S., Halford, S., Ebenezer, N.D., Gregory-Evans, C.Y. and Bhattacharya, S.S. (2001). *Cytogenet. Cell Genet.* **92**: 353.
200. Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). McKie, A.B., McHale, J.C., Keen, T.J., Tarttelin, E.E., Goliath, R., van Lith-Verhoeven, J.J., Greenberg, J., Ramesar, R.S., Hoyng, C.B., Cremers, F.P., Mackey, D.A., Bhattacharya, S.S., Bird, A.C., Markham, A.F. and Inglehearn, C.F. (2001). *Hum. Mol. Genet.* **10**: 1555-1562.
201. Identification and functional consequences of a new mutation (e155g) in the gene

- for *gcap1* that causes autosomal dominant cone dystrophy. Wilkie, S.E., Li, Y., Deery, E.C., Newbold, R.J., Garibaldi, D., Bateman, J.B., Zhang, H., Lin, W., Zack, D.J., Bhattacharya, S.S., Warren, M.J., Hunt, D.M. and Zhang, K. (2001). *Am. J. Hum. Genet.* **69**: 471-480.
202. A human homolog of yeast pre-mrna splicing gene, *prp31*, underlies autosomal dominant retinitis pigmentosa on chromosome 19q13.4 (rp11). Vithana, E.N., Abu-Safieh, L., Allen, M.J., Carey, A., Papaioannou, M., Chakarova, C., Al-Magthteh, m., Ebenezer, N.D., Willis, C., Moore, A.T., Bird, A.C., Hunt, D.M. and Bhattacharya, S.S. (2001). *Mol. Cell*, **8**: 375-381.
203. Locus for autosomal recessive nonsyndromic persistent hyperplastic primary vitreous. Khaliq S, Hameed, A., Ismail, M., Anwar, K., Leroy, B., Payne, A.M., Bhattacharya, S.S. and Mehdi, S.Q. (2001). *Invest. Ophthalmol. Vis. Sci.*, **42**: 2225-2228.
204. Clustering and frequency of mutations in the retinal guanylate cyclase (*GUCY2D*) gene in patients with dominant cone-rod dystrophies. Payne, A.M., Morris, A.G., Downes, S.M., Johnson, S., Bird, A.C., Moore, A.T., Bhattacharya, S.S. and Hunt, D.M. (2001). *J. Med. Genet.* **38**: 611-614.
205. Alpha-b crystallin gene (*cryab*) mutation causes dominant congenital posterior polar cataract in humans. Berry, V., Francis, P., Reddy, M.A., Collyer, D., Vithana, E., MacKay, I., Dawson, G., Carey, A.H., Moore, A., Bhattacharya, S.S. and Quinlan, R.A. (2001). *Am. J. Hum. Genet.* **69**: 1141-1145.
206. A frameshift mutation in exon 28 of the *OPA1* gene explains the high prevalence of dominant optic atrophy in the Danish population: evidence for a founder effect. Thiselton, D.L., Alexander, C., Morris, A., Brooks, S., Rosenberg, T., Eiberg, H., Kjer, B., Kjer, P., Bhattacharya, S.S. and Votruba, M. (2001). *Hum Genet.* **109**: 498-502.
207. A novel keratocan mutation causing autosomal recessive cornea plana. Lehmann, O.J., El-ashry, M.F., Ebenezer, N.D., Ocaka, L., Francis, P.J., Wilkie, S.E., Patel, R.J., Ficker, L., Jordan, T., Khaw, P.T. and Bhattacharya, S.S. (2001). *Invest Ophthalmol Vis Sci.* **42**: 3118-22.
208. Expression of *Drosophila* omb-related T-box genes in the developing human and mouse neural retina. Sowden, J.C., Holt, J.K., Meins, M., Smith, H.K. and Bhattacharya, S.S. (2001). *Invest Ophthalmol Vis Sci.* **42**: 3095-3102.
209. Mutations in *HPRP3*, a third member of pre-mRNA splicing factor genes, implicated in autosomal dominant retinitis pigmentosa. Chakarova, C.F., Hims, M.M., Bolz, H., Abu-Safieh, L., Patel, R.J., Papaioannou, M.G., Inglehearn, C.F., Keen, T.J., Willis, C., Moore, A.T., Rosenberg, T., Webster, A.R., Bird, A.C., Gal, A., Hunt, D., Vithana, E.N. and Bhattacharya, S.S. (2002). *Hum Mol Genet.* **11**: 87-92.
210. Identification of novel mutations in the carbohydrate sulfotransferase gene (*CHST6*) causing macular corneal dystrophy. El-Ashry, M.F., El-Aziz, M.M., Wilkins, S., Cheetham, M.E., Wilkie, S.E., Hardcastle, A.J., Halford, S., Bayoumi, A.Y., Ficker,

- L.A., Tuft, S., Bhattacharya, S.S. and Ebenezer, N.D. (2002). *Invest Ophthalmol Vis Sci.* **43**: 377-82.
211. A major marker for normal tension glaucoma: association with polymorphisms in the OPA1 gene. Aung, T., Ocaka, L., Ebenezer, N.D., Morris, A.G., Krawczak, M., Thiselton, D.L., Alexander, C., Votruba, M., Brice, G., Child, A.H., Francis, P.J., Hitchings, R.A., Lehmann, O.J. and Bhattacharya, S.S. (2002). *Hum Genet.* **110**: 52-56.
212. An integrated, functionally annotated gene map of the DXS8026-ELK1 interval on human Xp11.3-Xp11.23: potential hotspot for neurogenetic disorders. Thiselton, D.L., McDowall, J., Brandau, O., Ramser, J., d'Esposito, F., Bhattacharya, S.S., Ross, M.T., Hardcastle, A.J., and Meindl, A. (2002). *Genomics* **79**: 560-572.
213. A locus for isolated cataract on human Xp. Francis, P.J., Berry, V., Hardcastle, A.J., Maher, E.R., Moore, A.T. and Bhattacharya, S.S. (2002). *J Med Genet.* **39**:105-109.
214. Mutations in a protein target of the Pim-1 kinase associated with the RP9 form of autosomal dominant retinitis pigmentosa. Keen, T.J., Hims, M.M., McKie, A.B., Moore, A.T., Doran, R.M., Mackey, D.A., Mansfield, D.C., Mueller, R.F., Bhattacharya, S.S., Bird, A.C., Markham, A.F. and Inglehearn, C.F. (2002). *Eur J Hum Genet.* **10**: 245-249.
215. The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. Mauerer, A., Flothmann, K., Hemmrich, N., Ingvast, S., Jorge, P., Paloma, E., Patel, R., Rozet, J.M., Tammur, J., Testa, F., Balcells, S., Bird, A.C., Brunner, H.G., Hoyng, C.B., Metspalu, A., Simonelli, F., Allikmets, R., Bhattacharya, S.S., D'Urso, M., Gonzalez-Duarte, R., Kaplan, J., te Meerman, G.J., Santos, R., Schwartz, M., Van Camp, G., Wadelius, C., Weber, B.H. and Cremers, F.P. (2002). *Eur J Hum Genet.* **10**: 197-203.
216. Ocular developmental abnormalities and glaucoma associated with interstitial 6p25 duplications and deletions. Lehmann, O.J., Ebenezer, N.D., Ekong, R., Ocaka, L., Mungall, A.J., Fraser, S., McGill, J.I., Hitchings, R.A., Khaw, P.T., Sowden, J.C., Povey, S., Walter, M.A., Bhattacharya, S.S. and Jordan, T. (2002). *Invest Ophthalmol Vis Sci.* **43**: 1843-1849.
217. A comprehensive survey of mutations in the OPA1 gene in patients with autosomal dominant optic atrophy. Thiselton, D.L., Alexander, C., Taanman, J.W., Brooks, S., Rosenberg, T., Eiberg, H., Andreasson, S., Van Regemorter, N., Munier, F.L., Moore, A.T., Bhattacharya, S.S. and Votruba, M. (2002). *Invest Ophthalmol Vis Sci.* **43**: 1715-1724.
218. Investigating the association between OPA1 polymorphisms and glaucoma: comparison between normal tension and high tension primary open angle glaucoma. Aung, T., Ocaka, L., Ebenezer, N.D., Morris, A.G., Brice, G., Child, A.H., Hitchings, R.A., Lehmann, O.J. and Bhattacharya, S.S. (2002). *Hum Genet.* **110**: 513-514.
219. Characterisation of two genes for guanylate cyclase activator protein (GCAP1 and

- GCAP2) in the Japanese pufferfish, *Fugu rubripes*. Wilkie, S.E., Stinton, I., Cottrill, P., Deery, E., Newbold, R., Warren, M.J., Bhattacharya, S.S. and Hunt, D.M. (2002). *Biochim Biophys Acta*. **1577**: 73-80.
220. Cloning and characterization of WDR17, a novel WD repeat-containing gene on chromosome 4q34. Stohr, H., Mohr, N., Frohlich, S., Mehdi, S.Q., Bhattacharya, S.S., and Weber, B.H. (2002). *Biochim Biophys Acta*. **1579**: 18-25
221. Disease mechanism for retinitis pigmentosa (RP11) caused by mutations in the splicing factor gene PRPF31. Deery, E.C., Vithana, E.N., Newbold, R.J., Gallon, V.A., Bhattacharya, S.S., Warren, M.J., Hunt, D.M., and Wilkie, S.E. (2002). *Hum Mol Genet*. **11**: 3209-3219.
222. Optic disc morphology of patients with OPA1 autosomal dominant optic atrophy. Votruba, M., Thiselton, D., and Bhattacharya, S.S. (2003). *Br J Ophthalmol*. **87**: 48-53.
223. RPGR mutation associated with retinitis pigmentosa, impaired hearing, and sinorespiratory infections. Zito, I., Downes, S.M., Patel, R.J., Cheetham, M.E., Ebenezer, N.D., Jenkins, S.A., Bhattacharya, S.S., Webster, A.R., Holder, G.E., Bird, A.C., Bamiou, D.E., and Hardcastle, A.J. (2003) *J Med Genet*. **40**: 609-615.
224. Single-nucleotide polymorphisms and glaucoma severity. Bunce, C., Hitchings, R.A., Bhattacharya, S.S., and Lehmann, O.J. (2003). *Am J Hum Genet*. **72**:1593-1594; author reply 1594-5.
225. A clinical, histopathological, and genetic study of Avellino corneal dystrophy in British families. El-Ashry, M.F., El-Aziz, M.M., Larkin, D.F., Clarke, B., Cree, I.A., Hardcastle, A.J., Bhattacharya, S.S., and Ebenezer, N.D. (2003). *Br J Ophthalmol*. **87**: 839-842.
226. Fox's in development and disease. Lehmann, O.J., Sowden, J.C., Carlsson, P., Jordan, T., and Bhattacharya, S.S. (2003). *Trends Genet*. **19**: 339-344.
227. Phenotype of retinitis pigmentosa associated with the Ser50Thr mutation in the NRL gene. Bessant, D.A., Holder, G.E., Fitzke, F.W., Payne, A.M., Bhattacharya, S.S., and Bird, A.C. (2003). *Arch Ophthalmol*. **121**: 793-802.
228. Novel anterior segment phenotypes resulting from forkhead gene alterations: evidence for cross-species conservation of function. Lehmann, O.J., Tuft, S., Brice, G., Smith, R., Blixt, A., Bell, R., Johansson, B., Jordan, T., Hitchings, R.A., Khaw, P.T., John, S.W., Carlsson, P., and Bhattacharya, S.S. (2003). *Invest Ophthalmol Vis Sci*. **44**: 2627-2633.
229. The contribution of USH1C mutations to syndromic and non-syndromic deafness in the UK. Blaydon, D.C., Mueller, R.F., Hutchin, T.P., Leroy, B.P., Bhattacharya, S.S., Bird, A.C., Malcolm, S., and Bitner-Glindzicz, M. (2003). *Clin Genet*. **63**:303-307.
230. A clinical and molecular genetic study of a rare dominantly inherited syndrome (MRCS) comprising of microcornea, rod-cone dystrophy, cataract, and posterior



- staphyloma. Reddy, M.A., Francis, P.J., Berry, V., Bradshaw, K., Patel, R.J., Maher, E.R., Kumar, R., Bhattacharya, S.S., and Moore, A.T. (2003). *Br J Ophthalmol.* **87**:197-202.
231. The phenotype of normal tension glaucoma patients with and without OPA1 polymorphisms. Aung, T., Okada, K., Poinoosawmy, D., Membrey, L., Brice, G., Child, A.H., Bhattacharya, S.S., Lehmann, O.J., Garway-Heath, D.F., and Hitchings, R.A. (2003). *Br J Ophthalmol.* **87**: 149-152.
232. Prevalence of optineurin sequence variants in adult primary open angle glaucoma: implications for diagnostic testing. Aung, T., Ebenezer, N.D., Brice, G., Child, A.H., Prescott, Q., Lehmann, O.J., Hitchings, R.A., and Bhattacharya, S.S. (2003). *J Med Genet.* **40**: e101.
233. Expression of PRPF31 mRNA in patients with autosomal dominant retinitis pigmentosa: a molecular clue for incomplete penetrance? Vithana, E.N., Abu-Safieh, L., Pelosini, L., Winchester, E., Hornan, D., Bird, A.C., Hunt, D.M., Bustin, S.A., and Bhattacharya, S.S. (2003). *Invest Ophthalmol Vis Sci.* **44**: 4204-4209.
234. Characterisation of the G91del CRYBA1/3-crystallin protein: a cause of human inherited cataract. Reddy, M.A., Bateman, O.A., Chakarova, C., Ferris, J., Berry, V., Lomas, E., Sarra, R., Smith, M.A., Moore, A.T., Bhattacharya, S.S., and Slingsby, C. (2004). *Hum Mol Genet.* **13**: 945-953.
235. Dominant cone and cone-rod dystrophies: functional analysis of mutations in retGC1 and GCAP1. Hunt, D.M., Wilkie, S.E., Newbold, R., Deery, E., Warren, M.J., Bhattacharya, S.S., and Zhang, K. (2004). *Novartis Found Symp.* **255**: 37-49; discussion 49-50, 177-178.
236. BIGH3 mutation in a Bangladeshi family with a variable phenotype of LCDI. El-Ashry, M.F., Abd El-Aziz, M.M., Ficker, L.A., Hardcastle, A.J., Bhattacharya, S.S., and Ebenezer, N.D. (2004). *Eye* **18**: 723-728.
237. Developmental expression profile of the optic atrophy gene product: OPA1 is not localised exclusively in the ganglion cell layer of the mammalian retina. Aijaz, S., Erskine, L., Jeffery, G., Bhattacharya, S.S., and Votruba, M. (2004). *Invest. Ophthalmol. Vis. Sci.* **45**: 1667-1673.
238. Molecular genetic basis of inherited cataract and associated phenotypes. Reddy, M.A., Francis, P.J., Berry, V., Bhattacharya, S.S. and Moore, A.T. (2004). *Surv Ophthalmol.* **49**: 300-315.
239. Purification, characterisation and intracellular localisation of aryl hydrocarbon interacting protein-like 1 (AIPL1) and effects of mutations associated with inherited retinal dystrophies. Gallon, V.A., Wilkie, S.E., Deery, E.C., Newbold, R.J., Sohocki, M.M., Bhattacharya, S.S., Hunt, D.M. and Warren, M.J. (2004). *Biochim Biophys Acta.* **1690**: 141-149.
240. Mutations of VMD2 Splicing Regulators Cause Nanophthalmos and Autosomal

- Dominant Vitreoretinopathies (ADVIRC). Yardley, J., Leroy, B.P., Hart-Holden, N., Lafaut, B.A., Loeys, B., Messiaen, L.M., Perveen, R., Reddy, M.A., Bhattacharya, S.S., Traboulsi, E., Baralle, D., De Laey, J.J., Puech, B., Kestelyn, P., Moore, A.T., Manson, F.D. and Black, G.C. (2004). *Invest Ophthalmol Vis Sci.* **45**: 3683-3689.
241. Recurrent 17 bp duplication in PITX3 is primarily associated with posterior polar cataract (CPP4). Berry, V., Yang, Z., Addison, P.K., Francis, P.J., Ionides, A., Karan, G., Jiang, L., Lin, W., Hu, J., Yang, R., Moore, A., Zhang, K. and Bhattacharya, S.S. (2004). *J Med Genet.* **41**: e109.
242. The phenotype of Leber congenital amaurosis in patients with AIPL1 mutations. Dharmaraj, S., Leroy, B.P., Sohocki, M.M., Koeneke, R.K., Perrault, I., Anwar, K., Khaliq, S., Devi, R.S., Birch, D.G., De Pool, E., Izquierdo, N., Van Maldergem, L., Ismail, M., Payne, A.M., Holder, G.E., Bhattacharya, S.S., Bird, A.C., Kaplan, J. and Maumenee, I.H. (2004). *Arch Ophthalmol.* **122**: 1029-1037.
243. Mutant carbonic anhydrase 4 impairs pH regulation and causes retinal photoreceptor degeneration. Yang, Z., Alvarez, B.V., Chakarova, C., Jiang, L., Karan, G., Frederick, J.M., Zhao, Y., Sauve, Y., Li, X., Zrenner, E., Wissinger, B., Den Hollander, A.I., Katz, B., Baehr, W., Cremers, F.P., Casey, J.R., Bhattacharya, S.S., Zhang, K. (2005). *Hum Mol Genet.* **14**: 255-265
244. Posterior polar cataract is the predominant consequence of a recurrent mutation in the PITX3 gene. Addison, P.K., Berry, V., Ionides, A.C., Francis, P.J., Bhattacharya, S.S. and Moore, A.T. (2005). *Br J Ophthalmol.* **89**: 138-141.
245. Novel CHST6 nonsense and missense mutations responsible for macular corneal dystrophy. El-Ashry, M.F., Abd El-Aziz, M.M., Shalaby, O., Wilkins, S., Poopalasundaram, S., Cheetham, M., Tuft, S.J., Hardcastle, A.J., Bhattacharya, S.S. and Ebenezer, N.D. (2005). *Am J Ophthalmol.* **139**: 192-193.
246. Clinical Features and Course of Patients with Glaucoma with the E50K Mutation in the Optineurin Gene. Aung, T., Rezaie, T., Okada, K., Viswanathan, A.C., Child, A.H., Brice, G., Bhattacharya, S.S., Lehmann, O.J., Sarfarazi, M. and Hitchings, R.A. (2005). *Invest Ophthalmol Vis Sci.* **46**: 2816-2822.
247. A clinical and molecular genetic study of autosomal-dominant stromal corneal dystrophy in british population. El-Ashry, M.F., El-Aziz, M.M., Hardcastle, A.J., Bhattacharya, S.S., and Ebenezer, N.D. (2005). *Ophthalmic Res.* **37**: 310-317.
248. Exclusion of Four Candidate Genes, KHDRBS2, PTP4A1, KIAA1411 and OGFRL1, as Causative of Autosomal Recessive Retinitis Pigmentosa. Abd El-Aziz, M.M., Patel, R.J., El-Ashry, M.F., Barragan, I., Marcos, I., Borrego, S., Antinolo, G. and Bhattacharya, S.S. (2005). *Ophthalmic Res.* **38**: 19-23.
249. A new locus (RP31) for autosomal dominant retinitis pigmentosa maps to chromosome 9p. Papaioannou, M., Chakarova, C.F., Prescott, D.Q., Waseem, N., Theis, T., Lopez, I., Gill, B., Koeneke, R.K. and Bhattacharya, S.S. (2005). *Hum*

Genet. **118**: 501-503.

250. Posterior Polymorphous Corneal Dystrophy in Czech Families Maps to Chromosome 20 and Excludes the VSX1 Gene. Gwilliam, R., Liskova, P., Filipec, M., Kmoch, S., Jirsova, K., Huckle, E.J., Stables, C.L., Bhattacharya, S.S., Hardcastle, A.J., Deloukas, P., Ebenezer, N.D. (2005). *Invest Ophthalmol Vis Sci.* **46**: 4480-4484.
251. Molecular genetic analysis of two functional candidate genes in the autosomal recessive retinitis pigmentosa, RP25, locus. Abd El-Aziz, M.M., El-Ashry, M.F., Barragan, I., Marcos, I., Borrego, S., Antinolo, G. and Bhattacharya, S.S. (2005). *Curr Eye Res.* **30**: 1081-1087.
252. A novel GJA8 mutation is associated with autosomal dominant lamellar pulverulent cataract: further evidence for gap junction dysfunction in human cataract. Arora, A., Minogue, P.J., Liu, X., Reddy, M.A., Ainsworth, J.R., Bhattacharya, S.S., Webster, A.R., Hunt, D.M., Ebihara, L., Moore, A.T., Beyer, E.C., Berthoud, V.M. (2006). *J Med Genet.* **43**: e2.
253. A study of the nuclear trafficking of the splicing factor protein PRPF31 linked to autosomal dominant retinitis pigmentosa (ADRP). Wilkie, S.E., Morris, K.J., Bhattacharya, S.S., Warren, M.J. and Hunt, D.M. (2006). *Biochim Biophys Acta.* **1762**: 304-311
254. A large deletion in the adRP gene *PRPF31*: evidence that haplo-insufficiency is the cause of disease. Abu-Safieh, L., Vithana, E.N., Mantel, I., Holder, G.E., Pelosini, L., Bird, A.C. and Bhattacharya, S.S. (2006) *Mol Vis.* **12**: 384-388.
255. Molecular genetics of retinitis pigmentosa in two Romani (Gypsy) families. Chakarova CF, Cherninkova S, Tournev I, Waseem N, Kaneva R, Jordanova A, Veraitch BK, Gill B, Colclough T, Nakova A, Oscar A, Mihaylova V, Nikolova-Hill A, Wright AF, Black GC, Ramsden S, Kremensky I, Bhattacharya SS. (2006) *Mol Vis.* **12**: 909-914.
256. A novel mutation in the connexin 46 gene (GJA3) causes autosomal dominant zonular pulverulent cataract in a Hispanic family. Addison PK, Berry V, Holden KR, Espinal D, Rivera B, Su H, Srivastava AK, Bhattacharya SS. (2006). *Mol Vis.* **12**: 791-795.
257. Maculopathy due to the R345W substitution in fibulin-3: distinct clinical features, disease variability, and extent of retinal dysfunction. Michaelides M, Jenkins SA, Brantley MA Jr, Andrews RM, Waseem N, Luong V, Gregory-Evans K, Bhattacharya SS, Fitzke FW, Webster AR. (2006). *Invest Ophthalmol Vis Sci.* **47**: 3085-3097.
258. Clinical characterisation of a family with retinal dystrophy caused by mutation in the Mertk gene. Tschernutter M, Jenkins SA, Waseem NH, Saihan Z, Holder GE, Bird AC, Bhattacharya SS, Ali RR, Webster AR. (2006). *Br J Ophthalmol.* **90**: 718-23.
259. Retinitis pigmentosa associated with rhodopsin mutations: Correlation between phenotypic variability and molecular effects. Iannaccone A, Man D, Waseem N,

- Jennings BJ, Ganapathiraju M, Gallaher K, Reese E, Bhattacharya SS, Klein-Seetharaman J. (2006). *Vision Res.* **46**: 4556-4567.
260. Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration. Friedman JS, Chang B, Kannabiran C, Chakarova C, Singh HP, Jalali S, Hawes NL, Branham K, Othman M, Filippova E, Thompson DA, Webster AR, Andreasson S, Jacobson SG, Bhattacharya SS, Heckenlively JR, Swaroop A. (2006). *Am J Hum Genet.* **79**: 1059-1070.
261. A Novel Genetic Study of Chinese Families with Autosomal Recessive Retinitis Pigmentosa. Abd El-Aziz MM, El-Ashry MF, Chan WM, Chong KL, Barragan I, Antinolo G, Pang CP, Bhattacharya SS. (2007). *Ann Hum Genet.* **71**: 281-294.
262. A Clinical and Molecular Genetic Study of Egyptian and Saudi Arabian Patients With Primary Congenital Glaucoma (PCG). El-Ashry, M.F., Abd El-Aziz, M.M. and Bhattacharya, S.S. (2007). *J Glaucoma.* **16**: 104-111.
263. Mutations in the Gene Coding for the Pre-mRNA Splicing Factor, PRPF31, in Patients with Autosomal Dominant Retinitis Pigmentosa. Waseem, N.H., Vaclavik, V., Webster, A., Jenkins, S.A., Bird, A.C. and Bhattacharya, S.S. (2007). *Invest Ophthalmol Vis Sci.* **48**: 1330-1334.
264. Novel mutations in the ZEB1 gene identified in Czech and British patients with posterior polymorphous corneal dystrophy. Liskova, P., Tuft, S.J., Gwilliam, R., Ebenezer, N.D., Jirsova, K., Prescott, Q., Martinova, R., Pretorius, M., Sinclair, N., Boase, D.L., Jeffrey, M.J., Deloukas, P., Hardcastle, A.J., Filipec, M. and Bhattacharya, S.S. (2007). *Hum Mutat.* **28**: 638.
265. Novel SLC4A11 mutations in patients with recessive congenital hereditary endothelial dystrophy (CHED2). Ramprasad, V.L., Ebenezer, N.D., Aung, T., Rajagopal, R., Yong, V.H., Tuft, S.J., Viswanathan, D., El-Ashry, M.F., Liskova, P., Tan, D.T., Bhattacharya, S.S., Kumaramanickavel, G. and Vithana, E.N. (2007). *Hum Mutat.* **28**: 522-523.
266. Mutations in splicing factor PRPF3, causing retinal degeneration, form detrimental aggregates in photoreceptor cells. Comitato, A., Spanpanato, C., Chakarova, C., Sanges, D., Bhattacharya, S.S. and Marigo, V. (2007). *Hum Mol Genet.* **16**: 1699-1707.
267. Lisko Study of p.N247S KERA mutation in a British family with cornea plana. va, P., Hysi, P.G., Williams, D., Ainsworth, J.R., Shah, S., de la Chapelle, A., Tuft, S.J. and Bhattacharya, S.S. (2007). *Mol Vis.* **13**: 1339-1347.
268. Mutations in *TOPORS* Cause Autosomal Dominant Retinitis Pigmentosa with Perivascular RPE Atrophy. Chakarova, C.F., Papaioannou, M.G., Khanna, H., Lopez, I., Waseem, N., Shah, A., Theis, T., Friedman, J., Maubaret, C., Bujakowska, K., Veraitch, B., Abd El-Aziz, M.M., Prescott, Q., Parapuram, S., Bickmore, W.A., Munro, P.M/G/, Gal. A/, Hamel, C.P., Marigo, V., Ponting, C.P., Wissinger, B., Zrenner, E., Matter, K., Swaroop, A., Koenekoop, R.K. and Bhattacharya, S.S. (2007). *Am J Hum*

Genet, **81**: 1098-1103.

269. The Roles of PAX6 and SOX2 in Myopia: Lessons from the 1958 British Birth Cohort. Simpson, C.L., Hysi, P., Bhattacharya, S.S., Hammond, C.J., Webster, A., Peckham, C.S., Sham, P.C., Rahi, J.S. (2007) *Invest Ophthalmol Vis Sci.* **48**: 4421-4425.
270. An assessment of the apex microarray technology in genotyping patients with Leber congenital amaurosis and early-onset severe retinal dystrophy. Henderson, R.H., Waseem, N., Searle, R., van der Spuy, J., Russell-Eggitt, I., Bhattacharya, S.S., Thompson, D.A., Holder, G.E., Cheetham, M.E., Webster, A.R. and Moore, A.T. (2007). *Invest Ophthalmol Vis Sci.* **48**: 5684-5689. PMID: 18055820
271. British family with early-onset Fuchs' endothelial corneal dystrophy associated with p.L450W mutation in the COL8A2 gene. Liskova, P., Prescott, Q., Bhattacharya, S.S. and Tuft, S.J. (2007). *Br J Ophthalmol.* **91**: 1717-1718. PMID: 18024822
272. Genetic Analysis of FAM46A in Spanish Families with Autosomal Recessive Retinitis Pigmentosa: Characterisation of Novel VNTRs. Barragan, I., Borrego, S., Abd El-Aziz, M.M., El-Ashry, M.F., Abu-Safieh, L., Bhattacharya, S.S. and Antinolo, G. (2008). *Ann. Hum. Genet.* **72**: 26-34. PMID 17803723
273. Bilateral giant macular schisis in a patient with enhanced S-cone syndrome from a family showing pseudo-dominant inheritance. Vaclavik, V., Chakarova, C., Bhattacharya, S.S., Robson, A.G., Holder, G.E., Bird, A.C. and Webster, A.R. (2008). *Br J Ophthalmol.* **92**: 299-300. PMID: 18227217
274. Phenotype associated with the H626P mutation and other changes in the TGFBI gene in Czech families. Liskova, P., Klintworth, G.K., Bowling, B.L., Filipec, M., Jirsova, K., Tuft, S.J., Bhattacharya, S.S., Hardcastle, A.J. and Ebenezer, N.D. (2008). *Ophthalmic Res.* **40**:105-108. PMID: 18259096
275. Sequencing of the CHST6 gene in Czech macular corneal dystrophy patients supports the evidence of a founder mutation. Liskova, P., Veraitch, B., Jirsova, K., Filipec, M., Neuwirth, A., Ebenezer, N.D., Hysi, P.G., Hardcastle, A.J., Tuft, S.J. and Bhattacharya, S.S. (2008). *Br J Ophthalmol.***92**: 265-267. PMID: 17962390
276. Disease mechanism for retinitis pigmentosa (RP11) caused by missense mutations in the splicing factor gene PRPF31. Wilkie SE, Vaclavik V, Wu H, Bujakowska K, Chakarova CF, Bhattacharya SS, Warren MJ, Hunt DM. (2008). *Mol Vis.***14**: 683-690. PMID: 18431455
277. Linkage Validation of RP25 Using the 10K GeneChip Array and Further Refinement of the Locus by New Linked Families. Barragán I, Abd El-Aziz MM, Borrego S, El-Ashry MF, O'Driscoll C, Bhattacharya SS, Antiñolo G. (2008). *Ann Hum Genet.* **72**: 454-462. PMID: 18510647
278. Large-scale Molecular Analysis of a 34 Mb Interval on Chromosome 6q: Major Refinement of the RP25 Interval. Abd El-Aziz MM, Barragan I, O'Driscoll C, Borrego

- S, Abu-Safieh L, Pieras JI, El-Ashry MF, Prigmore E, Carter N, Antinolo G, Bhattacharya SS. (2008). *Ann Hum Genet.* 72: 463-477 PMID: 18510646
279. Effect of gene therapy on visual function in Leber's congenital amaurosis. Bainbridge JW, Smith AJ, Barker SS, Robbie S, Henderson R, Balaggan K, Viswanathan A, Holder GE, Stockman A, Tyler N, Petersen-Jones S, Bhattacharya SS, Thrasher AJ, Fitzke FW, Carter BJ, Rubin GS, Moore AT, Ali RR. (2008) *N Engl J Med.* **358**: 2231-2239. PMID: 18441371
280. EYS, encoding an ortholog of *Drosophila* spacemaker, is mutated in autosomal recessive retinitis pigmentosa. Abd El-Aziz MM, Barragan I, O'Driscoll CA, Goodstadt L, Prigmore E, Borrego S, Mena M, Pieras JI, El-Ashry MF, Safieh LA, Shah A, Cheetham ME, Carter NP, Chakarova C, Ponting CP, Bhattacharya SS, Antinolo G. (2008) *Nat Genet.* **40**: 1285-1287. PMID: 18836446
281. Dominant cataract formation in association with a vimentin assembly-disrupting mutation. Müller M, Bhattacharya SS, Moore T, Prescott Q, Wedig T, Herrmann H, Magin TM. (2009) *Hum Mol Genet.* **18**: 1052-1057. PMID: 19126778
282. Mutations of the EPHA2 receptor tyrosine kinase gene cause autosomal dominant congenital cataract. Zhang T, Hua R, Xiao W, Burdon KP, Bhattacharya SS, Craig JE, Shang D, Zhao X, Mackey DA, Moore AT, Luo Y, Zhang J, Zhang X. (2009). *Hum Mutat.* **30**: E603-611. PMID: 19306328
283. Mutations in TOPORS: a rare cause of autosomal dominant retinitis pigmentosa in continental Europe? Schob C, Orth U, Gal A, Kindler S, Chakarova CF, Bhattacharya SS, Rüther K. (2009). *Ophthalmic Genet.* **30**: 96-98. PMID: 19373681
284. Mutations in a BTB-Kelch protein, KLHL7, cause autosomal-dominant retinitis pigmentosa. Friedman JS, Ray JW, Waseem N, Johnson K, Brooks MJ, Hugosson T, Breuer D, Branham KE, Krauth DS, Bowne SJ, Sullivan LS, Ponjavic V, Gränse L, Khanna R, Trager EH, Gieser LM, Hughbanks-Wheaton D, Cojocarui RI, Ghiasvand NM, Chakarova CF, Abrahamson M, Göring HH, Webster AR, Birch DG, Abecasis GR, Fann Y, Bhattacharya SS, Daiger SP, Heckenlively JR, Andréasson S, Swaroop A. (2009). *Am J Hum Genet.* **84**: 792-800. PMID: 19520207
285. A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Khanna H, Davis EE, Murga-Zamalloa CA, Estrada-Cuzcano A, Lopez I, den Hollander AI, Zonneveld MN, Othman MI, Waseem N, Chakarova CF, Maubaret C, Diaz-Font A, Macdonald I, Muzny DM, Wheeler DA, Morgan M, Lewis LR, Logan CV, Tan PL, Beer MA, Inglehearn CF, Lewis RA, Jacobson SG, Bergmann C, Beales PL, Attié-Bitach T, Johnson CA, Otto EA, Bhattacharya SS, Hildebrandt F, Gibbs RA, Koenekoop RK, Swaroop A, Katsanis N. (2009). *Nat Genet.* **41**: 739-745. PMID: 19430481
286. Study of gene targeted mouse models of splicing factor gene Prpf31 implicated in human autosomal dominant retinitis pigmentosa (RP). Bujakowska KM, Maubaret C, Chakarova CF, Tanimoto N, Beck SC, Fahl E, Humphries MM, Kenna P, Makarov E, Makarova O, Paquet-Durand F, Ekström P, van Veen T, Leveillard T, Humphries P,

- Seeliger M, Bhattacharya SS. (2009). *Invest Ophthalmol Vis Sci.* Jul 2. PMID: 19578015
287. TRPM1 is mutated in patients with autosomal-recessive complete congenital stationary night blindness. Audo I, Khol S, Leroy BP, Munier FL, Guillon X, Mohand-Saïd S, Bujakowska K, Nandrot EF, Lorenz B, Preising M, Kellner U, Renner AB, Bernd A, Antonio A, Moskova-Doumanova V, Lancelot ME, Poloschek CM, Drumare I, Defoort-Dhellemmes S, Wissinger B, Léveillard T, Hamel CP, Schorderet DF, De Baere E, Berger W, Jacobson SG, Zrenner E, Sahel JA, Bhattacharya SS, Zeit C. (2009). *Am J Hum Genet.* **85**:720-729. PMID: 19896113
288. Molecular genetic study of Egyptian patients with macular corneal dystrophy. El-Ashry MF, Abd El-Aziz MM, Shalaby O, Bhattacharya SS. (2010). *Br J Ophthalmol.* **94**: 250-255. PMID:19734134
289. Biallelic mutation of photocadherin-21 (PCDH21) causes retinal degeneration in humans. Henderson RH, Li Z, Abd El Aziz MM, Mackay DS, Eljinini MA, Zeidan M, Moore AT, Bhattacharya SS, Webster AR. (2010). *Mol Vis.***16**: 46-52. PMID: 20087419
290. Spectrum of rhodopsin mutations in French autosomal dominant rod-cone dystrophy patients. Audo I, Manes G, Mohand-Saïd S, Friedrich A, Lancelot ME, Antonio A, Moskova-Doumanova V, Poch O, Zanlonghi X, Hamel CP, Sahel JA, Bhattacharya SS, Zeit C. (2010). *Invest Ophthalmol Vis Sci.* **51**: 3687-3700. PMID: 20164459
291. Alterations of the 5' untranslated region of SLC16A12 lead to age-related cataract. Zuercher J, Neidhardt J, Magyar I, Moore AT, Tanner F, Waseem NH, Schorderet D, Muniel FL, Bhattacharya SS, Berger W, Kloeckener-Gruissem B. (2010). *Invest Ophthalmol Vis Sci.* **51**: 3354-3361. PMID: 20181839
292. Photoreceptor degeneration: genetic and mechanistic dissection of a complex trait. Wright AF, Chakarova CF, Abd El-Aziz MM, Bhattacharya SS. (2010). *Nat Rev Genet.***11**: 273-284. PMID: 20212494
293. Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes. Tomwms KV, Kipioti A, Long V, McKibbin M, Maubaret C, Vaclvik V, Ehsani P, Springell K, Kamal M, Ramesar RS, Mackey DA, Moore AT, Mukhopadhyay R, Webster AR, Black GC, O'Sullivan J, Bhattacharya SS, Pierce EA, Beggs JD, Inglehearn CF. (2010). *Hum Mutat.* **31**: E1361-1376. PMID: 20232351
294. Identification of Novel Mutations in the ortholog of Drosophila eyes shut Gene (EYS) Causing Autosomal Recessive Retinitis Pigmentosa. Abd El-Aziz MM, O'Driscoll CA, Kaye RS, Barragan I, El-Ashry MF, Borrego S, Antiñolo G, Pang CP, Webster A, Bhattacharya SS. (2010). *Invest Ophthalmol Vis Sci.* **51**: 4266-4277. PMID: 20237254
295. Novel mutations in MERTK associated with childhood onset rod-cone dystrophy.

- Mackay DS, Henderson RH, Sergouniotis PI, Li Z, moradi P, Holder GE, Waseem N, Bhattacharya SS, Aldahmesh MA, Alkuraya FS, meyer B, Webster AR, Moore AT. (2010). *Mol Vis*. **16**: 369-377. PMID:20300561
296. EYS in a major gene for rod-cone dystrophies in France. Audo I, Sahel JA, Mohand-Saïd S, Lancelot ME, Antonio A, Moskova-Doumanova V, Nandrot EF, Doumanov J, Barragan I, Antiñolo G, Bhattacharya SS, Zeitz C. (2010). *Hum Mutat*. **31**: E1406-35. PMID: 20333770.
297. Efficient differentiation of human embryonic stem cells into functional cerebellar-like cells. Erceg S, Ronaghi M, Ivan Z, Lainez S, Garcia Rosello M, Moreno-Manzano V, Xiong C, Rodríguez-Jiménez FJ, Planeéis R, Alvarez Dolado M, Bhattacharya SS, Stojkovic M. (2010). *Stem Cells Dev*. **19**: 1745-1756. PMID: 20521974
298. Transplanted oligodendrocytes and motoneuron progenitors generated from human embryonic stem cells promote locomotor recovery after spinal cord transection. Erceg S, Ronaghi M, Oria M, Roselló MG, Aragón MA, Lopez MG, Radojevic I, Moreno-Manzano V, Rodríguez-Jiménez FJ, Bhattacharya SS, Cordoba J, Stojkovic M. (2010). *Stem Cells*. **28**:1541-1549. PMID: 20665739
299. Loss of lysophosphatidylcholine acyltransferase 1 leads to photoreceptor degeneration in rd11 mice. Friedman JS, Chang B, Krauth DS, Lopez I, Waseem NH, Hurd RE, Feathers KL, Branham KE, Shaw M, Thomas GE, Brooks MJ, Liu C, Bakeri HA, Campos MM, Maubaret C, Webster AR, Rodriguez IR, Thompson DA, Bhattacharya SS, Koenekoop RK, Heckenlively JR, Swaroop A. (2010). *Proc Natl Acad Sci U S A*. **107**:15523-15528. PMID: 20713727
300. Three Gene Targeted Mouse Models of RNA Splicing Factor RP Show Late Onset RPE and Retinal Degeneration. Graziotto JJ, Farkas MH, Bujakowska KM, Deramaudt BM, Zhang Q, Nandrot EF, Inglehearn CF, Bhattacharya SS, Pierce EA. (2011). *Invest Ophthalmol Vis Sci*. **52**: 190-198. PMID: 20811066
301. Evidence for keratoconus susceptibility locus on chromosome 14: a genome-wide linkage screen using single-nucleotide polymorphism markers. Liskova P, Hysi PG, Waseem N, Ebenezer ND, Bhattacharya SS, Tuft SJ. (2010). *Arch Ophthalmol*. **128**: 1191-1195. PMID: 20837804
302. Prevalence and novelty of PRPF31 mutations in French autosomal dominant rod-cone dystrophy patients and a review of published reports. Audo I, Bujakowska K, Mohand-Saïd S, Lancelot ME, Moskova-Doumanova V, Waseem NH, Antonio A, Sahel JA, Bhattacharya SS, Zeitz C. (2010). *BMC Med Genet*. **11**:145. PMID: 20939871
303. Mutation spectrum of EYS in Spanish patients with autosomal recessive retinitis pigmentosa. Barragán I, Borrego S, Pieras JI, González-del Pozo M, Santoyo J, Ayuso C, Baiget M, Millan JM, Mena M, El-Aziz MM, Audo I, Zeitz C, Littink KW, Dopazo J, Bhattacharya SS, Antiñolo G. (2010). *Hum Mutat*. **31**(11): E1772-800. PMID: 21069908



304. TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. Chakarova CF, Khanna H, Shah AZ, Patil SB, Sedmak T, Murga-Zamalloa CA, Papaioannou MG, Nagel-Wolfrum K, Lopez I, Munro P, Cheetham M, Koenekoop RK, Rios RM, Matter K, Wolfrum U, Swaroop A, Bhattacharya SS. (2011). *Hum Mol Genet.* **20**: 975-987. PMID: 21159800
305. Novel C2orf71 mutations account for ~1% of cases in a large French arRP cohort. Audo I, Lancelot ME, Mohand-Saïd S, Antonio A, Germain A, Sahel JA, Bhattacharya SS, Zeitz C. (2011). *Hum Mutat.* **32**(4): E2091-2103. PMID:21412943
306. Concise review: Stem Cells for the Treatment of Cerebellar Related Disorders. Erceg S, Moreno-Manzano V, Garita-Hernandez M, Stojkovic M, Bhattacharya SS. (2011). *Stem Cells.* **29** (4): 564-569. PMID:21319272
307. Copy-Number Variations in EYS. A significant event in the appearance of arRP. Pieras JI, Barragán I, Borrego S, Audo I, González-Del Pozo M, Bernal S, Baiget M, Zeitz C, Bhattacharya SS, Antiñolo G. (2011). *Invest Ophthalmol Vis Sci.* **52**(8): 5625-5631. PMID: 21519034
308. A novel 1-bp deletion in PITX3 causing congenital posterior polar cataract. Berry V, Francis PJ, Prescott Q, Waseem NH, Moore AT, Bhattacharya SS. (2011). *Mol Vis.* **17**: 1249-1253. PMID: 21633712
309. 112kb deletion in chromosome 19q13.42 leads to retinitis pigmentosa. Rose AM, Mukhopadhyay R, Webster AR, Bhattacharya SS, Waseem NH. (2011). *Invest Ophthalmol Vis Sci.* **52**(9): 6597-6603. PMID: 21715351.
310. A novel locus for autosomal dominant congenital cerulean cataract maps to chromosome 12q. Berry V, Ionides AC, Moore AT, Bhattacharya SS. (2011). *Eur J Hum Genet.* **19** (12): 1289-1291. PMID: 21731060.
311. Autosomal dominant retinitis pigmentosa with intrafamilial variability and incomplete penetrance in two families carrying mutations in PRPF8. Maubaret CG, Vaclavik V, Mukhopadhyay R, Waseem NH, Churchill A, Holder GE, Moore AT, Bhattacharya SS, Webster AR. (2011). *Invest Ophthalmol Vis Sci.* **52**(13): 9304-9309. PMID: 22039234.
312. Autosomal dominant Best disease with an unusual electrooculographic light rise and risk of angle-closure glaucoma: a clinical and molecular genetic study. Low S, Davidson AE, Holder GE, Hogg CR, Bhattacharya SS, Black GC, Foster PJ, Webster AR. (2011). *Mol Vis.* **17**: 2272-2282. PMID: 21921978
313. RP1 and autosomal dominant rod-cone dystrophy: novel mutations, a review of published variants, and genotype-phenotype correlation. Audo I, Mohand-Saïd S, Dhaenens CM, Germain A, Orhan E, Antonio A, Hamel C, Sahel JA, Bhattacharya SS, Zeitz C. (2012). *Hum Mutat.* **33**(1): 73-80. PMID: 2052604.
314. CRB1 mutations in inherited retinal dystrophies. Bujakowska K, Audo I, Mohand-Saïd S, Lancelot ME, Antonio A, Germain A, Léveillard T, Letexier M,

- Saraiva JP, Lonjou C, Carpentier W, Sahel JA, Bhattacharya SS, Zeitz C. (2012). *Hum Mutat.* **33**(2): 306-315. PMID: 22065545.
315. Hypoxia enhances the generation of retinal progenitor cells from human induced pluripotent and embryonic stem cells. Bae D, Mondragon-Teran P, Hernandez D, Ruban L, Mason C, Bhattacharya SS, Veraitch FS. (2012). *Stem Cells Dev.* **21**(8): 1344-1355. PMID: 21875341.
316. Age-related macular degeneration: the importance of family history as a risk factor. Shahid H, Khan JC, Cipriani V, Sepp T, Matharu BK, Bunce C, Harding SP, Clayton DG, Moore AT, Yates JR; Genetic Factors in AMD Study Group (2012). *Br J Ophthalmol.* **96**(3): 427-431. PMID: 21865200.
317. RDH12 retinopathy: novel mutations and phenotypic description. Mackay DS, Dev Borman A, Moradi P, Henderson RH, Li Z, Wright GA, Waseem N, Gandra M, Thompson DA, Bhattacharya SS, Holder GE, Webster AR, Moore AT. (2011). *Mol Vis.* **17**: 2706-2716. PMID: 22065924.
318. Common polymorphisms in the SERPINI2 gene are associated with refractive error in the 1958 British Birth Cohort. Hysi PG, Simpson CL, Fok YK, Gerrelli D, Webster AR, Bhattacharya SS, Hammond CJ, Sham PC, Rahi JS. (2012). *Invest Ophthalmol Vis Sci.* **53**(1): 440-447. PMID: 22110064.
319. Complement factor H genetic variant and age-related macular degeneration: effect size, modifiers and relationship to disease subtype. Sofat R, Casas JP, Webster AR, Bird AC, Mann SS, Yates JR, Moore AT, Sepp T, Cipriani V, Bunce C, Khan JC, Shahid H, Swaroop A, Abecasis G, Branham KE, Zarepari S, Bergen AA, Klaver CC, Baas DC, Zhang K, Chen Y, Gibbs D, Weber BH, Keilhauer CN, Fritsche LG, Lotery A, Cree AJ, Griffiths HL, Bhattacharya SS, Chen LL, Jenkins SA, Peto T, Lathrop M, Leveillard T, Gorin MB, Weeks DE, Ortube MC, Ferrell RE, Jakobsdottir J, Conley YP, Rahu M, Seland JH, Soubrane G, Topouzis F, Vioque J, Tomazzoli L, Young I, Whittaker J, Chakravarthy U, de Jong PT, Smeeth L, Fletcher A, Hingorani AD. (2012). *Int J Epidemiol.* **41**(1): 250-262. PMID: 22253316.
320. Functional characterization of a novel c.614-622del rhodopsin mutation in a French pedigree with retinitis pigmentosa. Maubaret C, Kosmaoglou M, Low S, Chakarova CF, Bidot S, Thauvin-Robinet C, Robson AG, Waseem N, Cheetham ME, Bhattacharya SS. (2012). *Mol Vis.* **18**: 581-587. PMID: 22419850.
321. Control of neuromal differentiation by sumoylation of BRAF35, a subunit of the LSD1-CoREST histone demethylase complex. Ceballos-Chávez M, Rivero S, García-Gutiérrez P, Rodríguez-Paredes M, García-Domínguez M, Bhattacharya SS, Reyes JC. (2012). *Proc Natl Acad Sci (USA)* **109**(21): 8085-90. PMID: 22570500
322. Whole-exome sequencing identifies mutations in GPR179 leading to autosomal-recessive complete congenital stationary night blindness. Audo I, Bujakowska K, Orhan E, Poloschek CM, Defoort-Dhellemmes S, Drumare I, Kohl S, Luu TD, Lecompte O, Zrenner E, Lancelot ME, Antonio A, Germain A, Michiels C, Audier C, Letexier M, Saraiva JP, Leroy BP, Munier FL, Mohand-Saïd S, Lorenz B, Friedburg C, Preising M,

- Kellner U, Renner AB, Moskova-Doumanova V, Berger W, Wissinger B, Hamel CP, Schorderet DF, De Baere E, Sharon D, Banin E, Jacobson SG, Bonneau D, Zanlonghi X, Le Meur G, Casteels I, Koenekoop R, Long VW, Meire F, Prescott K, de Ravel T, Simmons I, Nguyen H, Dollfus H, Poch O, Léveillard T, Nguyen-Ba-Charvet K, Sahel JA, Bhattacharya SS, Zeitz C. (2012). *Am J Hum Genet.* **90**(2): 321-330. Erratum in: *Am J Hum Genet.* 2012. 91(1): 209. PMID: 22325361.
323. Expression of PRPF31 and TFPT: regulation in health and retinal disease. Rose AM, Shah AZ, Waseem NH, Chakarova CF, Alfano G, Coussa RG, Ajlan R, Koenekoop RK, Bhattacharya SS. (2012). *Hum Mol Genet.* **21**(18): 4126-4137. PMID: 22723017.
324. A map of human microRNA variation uncovers unexpectedly high levels of variability. Carbonell J, Alloza E, Arce P, Borrego S, Santoyo J, Ruiz-Ferrer M, Medina I, Jimenez-Almazan J, Mendez-Vidal C, Gonzalez-Del Pozo M, Vela A, Bhattacharya SS, Antinolo G, Dopazo J. (2012). *Genome Med.* **4**(8): 62. PMID: 22906193.
325. Development and application of a next-generation-sequencing (NGS) approach to detect known and novel gene defects underlying retinal diseases. Audo I, Bujakowska KM, Léveillard T, Mohand-Saïd S, Lancelot ME, Germain A, Antonio A, Michiels C, Saraiva JP, Letexier M, Sahel JA, Bhattacharya SS, Zeitz C. (2012). *Orphanet J Rare Dis.* **7**: 8. PMID: 22277662.
326. Derivation of cerebellar neurons from human pluripotent stem cells. Erceg S, Lukovic D, Moreno-Manzano V, Stojkovic M, Bhattacharya SS. (2012). *Curr Protoc Stem Cell Biol.* Mar; Chapter 1:Unit 1H.5. PMID: 22415839.
327. Concise review: human pluripotent stem cells in the treatment of spinal cord injury. Lukovic D, Moreno Manzano V, Stojkovic M, Bhattacharya SS, Erceg S. (2012). *Stem Cells.* **30**(9): 1787-1792. PMID: 22736576.
328. NMNAT1 mutations cause Leber congenital amaurosis. Falk MJ, Zhang Q, Nakamaru-Ogiso E, Kannabiran C, Fonseca-Kelly Z, Chakarova C, Audo I, Mackay DS, Zeitz C, Borman AD, Staniszevska M, Shukla R, Palavalli L, Mohand-Said S, Waseem NH, Jalali S, Perin JC, Place E, Ostrovsky J, Xiao R, Bhattacharya SS, Consugar M, Webster AR, Sahel JA, Moore AT, Berson EL, Liu Q, Gai X, Pierce EA. (2012). *Nat Genet.* **44**(9): 1040-1045. PMID: 22842227.
329. Genome-wide association analyses identify three new susceptibility loci for primary angle closure glaucoma. Vithana EN, Khor CC, Qiao C, Nongpiur ME, George R, Chen LJ, Do T, Abu-Amero K, Huang CK, Low S, Tajudin LS, Perera SA, Cheng CY, Xu L, Jia H, Ho CL, Sim KS, Wu RY, Tham CC, Chew PT, Su DH, Oen FT, Sarangapani S, Soumitra N, Osman EA, Wong HT, Tang G, Fan S, Meng H, Huong DT, Wang H, Feng B, Baskaran M, Shantha B, Ramprasad VL, Kumaramanickavel G, Iyengar SK, How AC, Lee KY, Sivakumaran TA, Yong VH, Ting SM, Li Y, Wang YX, Tay WT, Sim X, Lavanya R, Cornes BK, Zheng YF, Wong TT, Loon SC, Yong VK, Waseem N, Yaakub A, Chia KS, Allingham RR, Hauser MA, Lam DS, Hibberd ML, Bhattacharya SS, Zhang M, Teo YY, Tan DT, Jonas JB, Tai ES, Saw SM, Hon do

- N, Al-Obeidan SA, Liu J, Chau TN, Simmons CP, Bei JX, Zeng YX, Foster PJ, Vijaya L, Wong TY, Pang CP, Wang N, Aung T. (2012). *Nat Genet.* **44**(10): 1142-1146. PMID: 22922875.
330. Cross species analysis of Prominin reveals a conserved cellular role in invertebrate and vertebrate photoreceptor cells. Nie J, Mahato S, Mustill W, Tipping C, Bhattacharya SS, Zelhof AC. (2012). *Dev Biol.* **371**(2): 312-320. PMID: 22960282.
331. High prevalence of posterior polymorphous corneal dystrophy in the czech republic; linkage disequilibrium mapping and dating an ancestral mutation. Liskova P, Gwilliam R, Filipic M, Jirsova K, Reinstein Merjava S, Deloukas P, Webb TR, Bhattacharya SS, Ebenezer ND, Morris AG, Hardcastle AJ. (2012). *PLoS One*, **7**(9): e45495. PMID: 23049806.
332. A novel locus for autosomal dominant cone-rod dystrophy maps to chromosome 10q. Kamenarova K, Cherninkova S, Romero Durán M, Prescott D, Valdés Sánchez ML, Mitev V, Kremensky I, Kaneva R, Bhattacharya SS, Tournev I, Chakarova C. (2012). *Eur J Hum Genet.* **21**(3): 338-342. PMID: 22929024.
333. Stem cell based therapy for spinal cord injury. Volarevic V, Erceg S, Bhattacharya SS, Stojkovic P, Horner P, Stojkovic M. (2012). *Cell Transplant.* **22**(8): 1309-1323. PMID: 23043847.
334. CNOT3 Is a Modifier of PRPF31 Mutations in Retinitis Pigmentosa with Incomplete Penetrance. Venturini G, Rose AM, Shah AZ, Bhattacharya SS, Rivolta C. (2012). *PLoS Genet.* **8**(11): e1003040 . PMID: 23144630.
335. Whole-exome sequencing identifies LRIT3 mutations as a cause of autosomal-recessive complete congenital stationary night blindness. Zeitz C, Jacobson SG, Hamel CP, Bujakowska K, Neullé M, Orhan E, Zanlonghi X, Lancelot ME, Michiels C, Schwartz SB, Bocquet B; Congenital Stationary Night Blindness Consortium, Antonio A, Audier C, Letexier M, Saraiva JP, Luu TD, Sennlaub F, Nguyen H, Poch O, Dollfus H, Lecompte O, Kohl S, Sahel JA, Bhattacharya SS, Audo I. (2013). *Am J Hum Genet.* **92**(1): 67-75. PMID: 23246293.
336. ATR localizes to the photoreceptor connecting cilium and deficiency leads to severe photoreceptor degeneration in mice. Valdés-Sánchez L, De la Cerda B, Diaz-Corrales FJ, Massalini S, Chakarova CF, Wright AF, Bhattacharya SS. (2013). *Hum Mol Genet.* **22**(8): 1507-1515. PMID: 23297361.
337. Hypoxia Increases the Yield of Photoreceptors Differentiating from Mouse Embryonic Stem Cells and Improves the Modeling of Retinogenesis in Vitro. Garita-Hernández M, Diaz-Corrales F, Lukovic D, Guede IG, Lloret AD, Sánchez ML, Massalini S, Erceg S, Bhattacharya SS. (2013). *Stem Cells.* **31**(5): 966-978. PMID: 23362204.
338. Wolfram gene (WFS1) mutation causes autosomal dominant congenital nuclear cataract in humans. Berry V, Gregory-Evans C, Emmett W, Waseem N, Raby J, Prescott D, Moore AT, Bhattacharya SS. (2013). *Eur J Hum Genet.* **21**(12): 1356-1360.

PMID: 23531866.

339. Disease-causing mutation in BEST1 gene area associated with altered sorting of bestrophin-1 protein. Doumanov JA, Zeitz C, Dominguez Gimenez P, Audo I, Krishna A, Alfano G, Diaz ML, Moskova-Doumanova V, Lancelot ME, Sahel JA, Nandrot EF, Bhattacharya SS. (2013). *Int J Mol Sci.* **14**(7): 15121-40. PMID: 23880862.
340. Perspectives and future directions of human pluripotent stem cell-based therapies. Lessons from Geron's clinical trial for spinal cord injury. Lukovic D, Stojkovic M, Moreno-Manzano V, Bhattacharya SS, Erceg S. (2013). *Stem Cells Dev.* **23**(1): 1-4. PMID: 23980630.
341. Novel GUCA1A Mutations Suggesting Possible Mechanism of Pathogenesis in Cone, ConeRod and Macular Dystrophy Patients. Kamenarova K, Corton M, García-Sandoval B, Fernández-San Jose P, Panchev V, Avila-Fernández A, López-Molina MI, Chakarova C, Ayuso C, Bhattacharya SS. (2013). *Biomed Res. Int.* 2013:517570. doi: 10.1155/2013/517570. PMID: 24024198.
342. The familial dementia gene revisited: a missense mutation revealed by whole exome sequencing identifies ITM2B as a candidate gene underlying a novel autosomal dominant retinal dystrophy in a large family. Audo I, Bujakowska K, Orhan E, El Shamieh S, Sennlaub F, Guillonneau X, Antonio A, Michiels C, Lancelot ME, Letexier M, Saraiva JP, Nguyen H, Luu TD, Léveillard T, Poch O, Dollfus H, Paques M, Goureau O, Mohand-Säïd S, Bhattacharya SS, Sahel JA, Zeitz C. (2013). *Hum. Mol. Genet.* **23**(2): 491-501. PMID: 24026677.
343. Further insights into GPR179: expression, localization, and associated pathogenic mechanisms leading to complete congenital stationary night blindness. Orhan E, Prézeau L, El Shamieh S, Bujakowska KM, Michiels C, Zagar Y, Vol C, Bhattacharya SS, Sahel JA, Sennlaub F, Audo I, Zeitz C. (2013). *Invest Ophthalmol Vis Sci.* **54**(13): 8041-8050. PMID: 24222301.
344. Dominant PRPF31 mutations are hypostatic to a recessive CNOT3 polymorphism in retinitis pigmentosa: a novel phenomenon of "linked trans-acting epistasis". Rose AM, Shah AZ, Venturini G, Rivolta C, Rose GE, Bhattacharya SS. (2014). *Ann Hum Genet.* **78**(1): 62-71. PMID: 24116917.
345. Brief Report: Astrogliosis Promotes Functional Recovery of Completely Transected Spinal Cord Following Transplantation of hESC-Derived Oligodendrocyte and Motoneuron Progenitors. Lukovic D, Valdés-Sanchez L, Sanchez-Vera I, Moreno-Manzano V, Stojkovic M, Bhattacharya SS, Erceg S. (2014). *Stem Cells.* **32**(2): 594-599. PMID: 24115357.
346. ABCC5, a Gene That Influences the Anterior Chamber Depth, Is Associated with Primary Angle Closure Glaucoma. Nongpiur ME, Khor CC, Jia H, Cornes BK, Chen LJ, Qiao C, Nair KS, Cheng CY, Xu L, George R, Tan D, Abu-Amero K, Perera SA, Ozaki M, Mizoguchi T, Kurimoto Y, Low S, Tajudin LS, Ho CL, Tham CC, Soto I, Chew PT, Wong HT, Shantha B, Kuroda M, Osman EA, Tang G, Fan S, Meng H, Wang H, Feng B, Yong VH, Ting SM, Li Y, Wang YX, Li Z, Lavanya R, Wu RY,

- Zheng YF, Su DH, Loon SC, Allingham RR, Hauser MA, Soumitra N, Ramprasad VL, Waseem N, Yaakub A, Chia KS, Kumaramanickavel G, Wong TT, How AC, Chau TN, Simmons CP, Bei JX, Zeng YX, Bhattacharya SS, Zhang M, Tan DT, Teo YY, Al-Obeidan SA, Hon do N, Tai ES, Saw SM, Foster PJ, Vijaya L, Jonas JB, Wong TY, John SW, Pang CP, Vithana EN, Wang N, Aung T. (2014). *PLoS Genet.* **10**(3): e1004089. doi: 10.1371/journal.pgen.1004089. PMID: 24603532.
347. Biallelic variants in *TLL5*, encoding a tubulin glutamylase, cause retinal dystrophy. Sergouniotis PI, Chakarova C, Murphy C, Becker M, Lenassi E, Arno G, Lek M, MacArthur DG; UCL-Exomes Consortium, Bhattacharya SS, Moore AT, Holder GE, Robson AG, Wolfrum U, Webster AR, Plagnol V. (2014). *Am J Hum Genet.* **94**(5): 760-769. doi: 10.1016/j.ajhg.2014.04.003. PMID: 24791901.
348. Non-coding RNAs in pluripotency and neural differentiation of human pluripotent stem cells. Lukovic D, Moreno-Manzano V, Klabusay M, Stojkovic M, Bhattacharya SS, Erceg S. (2014). *Front Genet.* **5**:132. doi: 10.3389/fgene.2014.00132. eCollection 2014. Review. PMID: 24860598.
349. Rare and common variants in extracellular matrix gene *Fibrillin 2* (*FBN2*) are associated with macular degeneration. Ratnapriya R, Zhan X, Fariss RN, Branham KE, Zipprer D, Chakarova CF, Sergeev YV, Campos MM, Othman M, Friedman JS, Maminishkis A, Waseem NH, Brooks M, Rajasimha HK, Edwards AO, Lotery A, Klein BE, Truitt BJ, Li B, Schaumberg DA, Morgan DJ, Morrison MA, Souied E, Tsironi EE, Grassmann F, Fishman GA, Silvestri G, Scholl HP, Kim IK, Ramke J, Tuo J, Merriam JE, Merriam JC, Park KH, Olson LM, Farrer LA, Johnson MP, Peachey NS, Lathrop M, Baron RV, Igo RP Jr, Klein R, Hagstrom SA, Kamatani Y, Martin TM, Jiang Y, Conley Y, Sahel JA, Zack DJ, Chan CC, Pericak-Vance MA, Jacobson SG, Gorin MB, Klein ML, Allikmets R, Iyengar SK, Weber BH, Haines JL, Léveillard T, Deangelis MM, Stambolian D, Weeks DE, Bhattacharya SS, Chew EY, Heckenlively JR, Abecasis GR, Swaroop A. (2014). *Hum Mol Genet.* **23**(21): 5827-5837. doi: 10.1093/hmg/ddu276. Epub 2014 Jun 4. PMID: 24899048.
350. Mutations in pre-mRNA processing factors 3, 8, and 31 cause dysfunction of the retinal pigment epithelium. Farkas MH, Lew DS, Sousa ME, Bujakowska K, Chatagnon J, Bhattacharya SS, Pierce EA, Nandrot EF. (2014). *Am J Pathol.* **184**(10): 2641-2652. doi: 10.1016/j.ajpath.2014.06.026. Epub 2014 Aug 8. PMID: 25111227.
351. Cleavage of Mer tyrosine kinase (MerTK) from the cell surface contributes to the regulation of retinal phagocytosis. Law AL, Parinot C, Chatagnon J, Gravez B, Sahel JA, Bhattacharya SS, Nandrot EF. (2015). *J Biol Chem.* **290**(8): 4941-4952. doi: 10.1074/jbc.M114.628297. Epub 2014 Dec 23. PMID: 25538233.
352. *HMG20A* is required for *SNAI1*-mediated epithelial to mesenchymal transition. Rivero S, Ceballos-Chávez M, Bhattacharya SS, Reyes JC. (2015). *Oncogene.* **34**(41): 5264-5276. doi: 10.1038/onc.2014.446. [Epub ahead of print] PMID: 25639869.
353. Concise review: reactive astrocytes and stem cells in spinal cord injury: good guys or bad guys? Lukovic D, Stojkovic M, Moreno-Manzano V, Jendelova P, Sykova E, Bhattacharya SS, Erceg S. (2015). *Stem Cells.* **33**(4): 1036-1041. doi:

10.1002/stem.1959. PMID: 25728093.

354. Diverse clinical phenotypes associated with a nonsense mutation in FAM161A. Rose AM, Sergouniotis P, Alfano G, Muspratt-Tucker N, Barton S, Moore AT, Black G, Bhattacharya SS, Webster AR. (2015). *Eye (Lond)*. **29**(9): 1226-1232. doi: 10.1038/eye.2015.93. [Epub ahead of print] PMID: 26113502.
355. Human iPSC derived disease model of MERTK-associated retinitis pigmentosa. Lukovic D, Artero Castro A, Delgado AB, Bernal Mde L, Luna Pelaez N, Díez Lloret A, Perez Espejo R, Kamenarova K, Fernández Sánchez L, Cuenca N, Cortón M, Avila Fernandez A, Sorkio A, Skottman H, Ayuso C, Erceg S, Bhattacharya SS. (2015). *Sci Rep*. **5**:12910. doi: 10.1038/srep12910. PMID: 26263531.
356. Genotype and Phenotype Studies in Autosomal Dominant Retinitis Pigmentosa (adRP) of the French Canadian Founder Population. Coussa RG, Chakarova C, Ajlan R, Taha M, Kavalec C, Gomolin J, Khan A, Lopez I, Ren H, Waseem N, Kamenarova K, **Bhattacharya SS**, Koenekoop RK. (2015). *Invest Ophthalmol Vis Sci*. **56**(13): 8297-8305. doi: 10.1167/iovs.15-17104. PMID: 26720483
357. 267 Spanish Exomes Reveal Population-Specific Differences in Disease-Related Genetic Variation. Dopazo J, Amadoz A, Bleda M, Garcia-Alonso L, Alemán A, García-García F, Rodriguez JA, Daub JT, Muntané G, Rueda A, Vela-Boza A, López-Domingo FJ, Florido JP, Arce P, Ruiz-Ferrer M, Méndez-Vidal C, Arnold TE, Spleiss O, Alvarez-Tejado M, Navarro A, **Bhattacharya SS**, Borrego S, Santoyo-López J, Antiñolo G. (2016). *Mol Biol Evol*. **33**(5): 1205-1218. doi: 10.1093/molbev/msw005. Epub 2016 Jan 13. PMID: 26764160
358. Transcriptional regulation of PRPF31 gene expression by MSR1 repeat elements causes incomplete penetrance in retinitis pigmentosa. Rose AM, Shah AZ, Venturini G, Krishna A, Chakravarti A, Rivolta C, **Bhattacharya SS**. (2016). *Sci Rep*. **6**:19450. doi: 10.1038/srep19450. PMID: 26781568
359. Variant haploinsufficiency and phenotypic non-penetrance in PRPF31-associated retinitis pigmentosa. Rose AM, **Bhattacharya SS**. (2016). *Clin Genet*. **90**(2): 118-126. doi: 10.1111/cge.12758. Epub 2016 Mar 4. Review. PMID: 26853529
360. TOPORS, a Dual E3 Ubiquitin and Sumo1 Ligase, Interacts with 26 S Protease Regulatory Subunit 4, Encoded by the PSMC1 Gene. Czub B, Shah AZ, Alfano G, Kruczek PM, Chakarova CF, **Bhattacharya SS**. (2016). *PLoS One*. **11**(2): e0148678. doi: 10.1371/journal.pone.0148678. eCollection 2016. PMID: 26872363
361. Genetic association and stress mediated down-regulation in trabecular meshwork implicates MPP7 as a novel candidate gene in primary open angle glaucoma. Vishal M, Sharma A, Kaurani L, Alfano G, Mookherjee S, Narta K, Agrawal J, Bhattacharya I, Roychoudhury S, Ray J, Waseem NH, **Bhattacharya SS**, Basu A, Sen A, Ray K, Mukhopadhyay A. (2016). *BMC Med Genomics*. **9**:15. doi: 10.1186/s12920-016-0177-6. PMID: 27001270
362. Genome-wide association study identifies five new susceptibility loci for primary

angle closure glaucoma. Khor CC, Do T, Jia H, Nakano M, George R, Abu-Amero K, Duvsh R, Chen LJ, Li Z, Nongpiur ME, Perera SA, Qiao C, Wong HT, Sakai H, Barbosa de Melo M, Lee MC, Chan AS, Azhany Y, Dao TL, Ikeda Y, Perez-Grossmann RA, Zarnowski T, Day AC, Jonas JB, Tam PO, Tran TA, Ayub H, Akhtar F, Micheal S, Chew PT, Aljasim LA, Dada T, Luu TT, Awadalla MS, Kitnarong N, Wanichwecharunguang B, Aung YY, Mohamed-Noor J, Vijayan S, Sarangapani S, Husain R, Jap A, Baskaran M, Goh D, Su DH, Wang H, Yong VK, Yip LW, Trinh TB, Makornwattana M, Nguyen TT, Leuenberger EU, Park KH, Wiyogo WA, Kumar RS, Tello C, Kurimoto Y, Thapa SS, Pathanapitoon K, Salmon JF, Sohn YH, Fea A, Ozaki M, Lai JS, Tantisevi V, Khaing CC, Mizoguchi T, Nakano S, Kim CY, Tang G, Fan S, Wu R, Meng H, Nguyen TT, Tran TD, Ueno M, Martinez JM, Ramli N, Aung YM, Reyes RD, Vernon SA, Fang SK, Xie Z, Chen XY, Foo JN, Sim KS, Wong TT, Quek DT, Venkatesh R, Kavitha S, Krishnadas SR, Soumittra N, Shantha B, Lim BA, Ogle J, de Vasconcellos JP, Costa VP, Abe RY, de Souza BB, Sng CC, Aquino MC, Kosior-Jarecka E, Fong GB, Tamanaja VC, Fujita R, Jiang Y, Waseem N, Low S, Pham HN, Al-Shahwan S, Craven ER, Khan MI, Dada R, Mohanty K, Faiq MA, Hewitt AW, Burdon KP, Gan EH, Pruthipongsit A, Patthanathamrongkasem T, Catacutan MA, Felarca IR, Liao CS, Rusmayani E, Istiantoro VW, Consolandi G, Pignata G, Lavia C, Rojanapongpun P, Mangkornkanokpong L, Chansangpetch S, Chan JC, Choy BN, Shum JW, Than HM, Oo KT, Han AT, Yong VH, Ng XY, Goh SR, Chong YF, Hibberd ML, Seielstad M, Png E, Dunstan SJ, Chau NV, Bei J, Zeng YX, Karkey A, Basnyat B, Pasutto F, Paoli D, Frezzotti P, Wang JJ, Mitchell P, Fingert JH, Allingham RR, Hauser MA, Lim ST, Chew SH, Ebstein RP, Sakuntabhai A, Park KH, Ahn J, Boland G, Snippe H, Stead R, Quino R, Zaw SN, Lukasik U, Shetty R, Zahari M, Bae HW, Oo NL, Kubota T, Manassakorn A, Ho WL, Dallorto L, Hwang YH, Kiire CA, Kuroda M, Djamal ZE, Peregrino JI, Ghosh A, Jeoung JW, Hoan TS, Srisamran N, Sandragasu T, Set SH, Doan VH, **Bhattacharya SS**, Ho CL, Tan DT, Sihota R, Loon SC, Mori K, Kinoshita S, Hollander AI, Qamar R, Wang YX, Teo YY, Tai ES, Hartleben-Matkin C, Lozano-Giral D, Saw SM, Cheng CY, Zenteno JC, Pang CP, Bui HT, Hee O, Craig JE, Edward DP, Yonahara M, Neto JM, Guevara-Fujita ML, Xu L, Ritch R, Liza-Sharmini AT, Wong TY, Al-Obeidan S, Do NH, Sundaresan P, Tham CC, Foster PJ, Vijaya L, Tashiro K, Vithana EN, Wang N, Aung T. (2016). *Nat Genet.* **48**(5): 556-562. doi: 10.1038/ng.3540. Epub 2016 Apr 4. PMID: 27064256

363. A missense mutation in ASRGL1 is involved in causing autosomal recessive retinal degeneration. Biswas P, Chavali VR, Agnello G, Stone E, Chakarova C, Duncan JL, Kannabiran C, Homsher M, **Bhattacharya SS**, Naeem MA, Kimchi A, Sharon D, Iwata T, Riazuddin S, Reddy GB, Hejtmancik JF, Georgiou G, Riazuddin SA, Ayyagari, R. (2016). *Hum Mol Genet.* **25**(12): 2483-2497. PMID: 27106100
364. First insights into the expression of VAX2 in humans and its localization in the adult primate retina. Alfano G, Shah AZ, Jeffery G, **Bhattacharya SS**. (2016). *Exp Eye Res.* **148**: 24-29. doi: 10.1016/j.exer.2016.05.008. Epub 2016 May 11. PMID: 27179412
365. Span poly-L-arginine nanoparticles are efficient non-viral vectors for PRPF31 gene delivery: An approach of gene therapy to treat retinitis pigmentosa. Pensado A, Diaz-Corrales FJ, De la Cerda B, Valdés-Sánchez Del Boz AA, Rodriguez-Martinez D, García-Delgado AB, Seijo B, **Bhattacharya SS**, Sanchez A. (2016). *Nanomedicine.* **12**:



2251-2260. PMID: 27381066

366. EYS Is a Protein Associated with the Ciliary Axoneme in Rods and Cones. Alfano G, Kruczek PM, Shah AZ, Kramarz B, Jeffery G, Zelhof AC, **Bhattacharya SS**. (2016). *PLoS One*; **11**(11): e0166397. doi: 10.1371/journal.pone.0166397. eCollection 2016. PMID: 27846257
367. Highly Efficient Neural Conversion of Human Pluripotent Stem Cells in Adherent and Animal-Free Conditions. Lukovic D, Diez Lloret A, Stojkovic P, Rodríguez-Martínez D, Perez Arago MA, Rodríguez-Jimenez FJ, González-Rodríguez P, López-Barneo J, Sykova E, Jendelova P, Kostic J, Moreno-Manzano V, Stojkovic M, **Bhattacharya SS**, Erceg S. (2017). *Stem Cells Transl Med.*; **6**(4): 1217-1226. doi: 10.1002/sctm.16-0371. Epub 2017 Feb 18. PMID: 28213969
368. Gene of the month: *PRPF31*. Rose AM, Luo R, Radia UK, **Bhattacharya SS**. (2017). *J Clin Pathol.*; **70**(9): 729-732 pii: jclinpath-2016-203971. doi: 10.1136/jclinpath-2016-203971. [Epub ahead of print] Review. PMID:28663330
369. Effects of Ca<sup>2+</sup> ions on bestrophin-1 surface films. Mladenova K, Petrova SD, Andreeva TD, Moskova-Doumanova V, Topouzova-Hristova T, Kalvachev Y, Balashev K, **Bhattacharya SS**, Chakarova C, Lalchev Z, Doumanov JA. (2017). *Colloids Surf B Biointerfaces*; **149**: 226-232. doi: 10.1016/j.colsurfb.2016.10.023. Epub 2016 Oct 13. PMID: 27768912
370. Prevalence of Age-Related Macular Degeneration in Europe: The Past and the Future. Colijn JM, Buitendijk GHS, Prokofyeva E, Alves D, Cachulo ML, Khawaja AP, Cougnard-Gregoire A, Merle BMJ, Korb C, Erke MG, Bron A, Anastasopoulos E, Meester-Smoor MA, Segato T, Piermarocchi S, de Jong PTVM, Vingerling JR, Topouzis F, Creuzot-Garcher C, Bertelsen G, Pfeiffer N, Fletcher AE, Foster PJ, Silva R, Korobelnik JF, Delcourt C, Klaver CCW; **EYE-RISK consortium**; European Eye Epidemiology (E3) consortium. (2017). *Ophthalmology*; **124**(12): 1753-1763. doi: 10.1016/j.optha.2017.05.035. Epub 2017 Jul 14. PMID: 28712657
371. Rasagiline delays retinal degeneration in a mouse model of retinitis pigmentosa via modulation of Bax/Bcl-2 expression. Garcia-Delgado AB, Valdés-Sánchez L, Calado SM, Diaz-Corrales FJ, **Bhattacharya SS**. (2018). *CNS Neurosci Ther.*; **24**(5): 448-455. doi: 10.1111/cns.12805. Epub 2018 Jan 25. PMID: 29372592
372. MSR1 repeats modulate gene expression and affect risk of breast and prostate cancer. Rose AM, Krishan A, Chakarova CF, Moya L, Chambers SK, Hollands M, Illingworth JC, Williams SMG, McCabe HE, Shah AZ, Palmer CNA, Chakravarti A, Berg JN, Batra J, **Bhattacharya SS**. (2018). *Ann Oncol.* **29**(5): 1292-1303. doi: 10.1093/annonc/mdy082. PMID: 29509840
373. Identification and characterization of the VAX2 p.Leu139Arg variant: possible involvement of VAX2 in cone dystrophy. Alfano G, Waseem NH, Webster AR, **Bhattacharya SS**. (2018). *Ophthalmic Genet.* **39**(4): 539-543. doi: 10.1080/13816810.2018.1484927. PMID: 29947570

374. Generation of a human iPS cell line from a patient with retinitis pigmentosa due to EYS mutation. Calado SM, Garcia-Delgado AB, De la Cerda B, Ponte-Zuñiga B, **Bhattacharya SS**, Díaz-Corrales FJ. (2018). *Stem Cell Res.*; **33**: 251-254. doi: 10.1016/j.scr.2018.11.002. Epub 2018 Nov 16. PMID: 30471616
375. Formation of 53BP1 foci and ATM activation under oxidative stress is facilitated by RNA:DNA hybrids and loss of ATM-53BP1 expression promotes photoreceptor cell survival in mice. Bhatia V, Valdés-Sánchez L, Rodríguez-Martínez D, **Bhattacharya SS**. (2018). *F1000Res.*; **7**:1233. doi: 10.12688/f1000research.15579.1. eCollection 2018. PMID: 30345028
376. Increased High-Density Lipoprotein Levels Associated with Age-Related Macular Degeneration: Evidence from the EYE-RISK and European Eye Epidemiology Consortia. Colijn JM, den Hollander AI, Demirkan A, Cougnard-Grégoire A, Verzijden T, Kersten E, Meester-Smoor MA, Merle BMJ, Papageorgiou G, Ahmad S, Mulder MT, Costa MA, Benlian P, Bertelsen G, Bron AM, Claes B, Creuzot-Garcher C, Erke MG, Fauser S, Foster PJ, Hammond CJ, Hense HW, Hoyng CB, Khawaja AP, Korobelnik JF, Piermarocchi S, Segato T, Silva R, Souied EH, Williams KM, van Duijn CM, Delcourt C, Klaver CCW; European Eye Epidemiology Consortium; **EYE-RISK Consortium**. (2019). *Ophthalmology*; **126**(3): 393-406. doi: 10.1016/j.ophtha.2018.09.045. Epub 2018 Oct 10. PMID: 30315903
377. Mediterranean Diet and Incidence of Advanced Age-Related Macular Degeneration: The EYE-RISK Consortium. Merle BMJ, Colijn JM, Cougnard-Grégoire A, de Koning-Backus APM, Delyfer MN, Kieft-de Jong JC, Meester-Smoor M, Féart C, Verzijden T, Samieri C, Franco OH, Korobelnik JF, Klaver CCW, Delcourt C; **EYE-RISK Consortium**. (2019). *Ophthalmology*. **126**(3): 381-390. doi: 10.1016/j.ophtha.2018.08.006. Epub 2018 Aug 13. PMID: 30114418
378. Generation and characterization of the human iPSC line CABi001-A from a patient with retinitis pigmentosa caused by a novel mutation in PRPF31 gene. de la Cerda B, Díez-Lloret A, Ponte B, Vallés-Saiz L, Calado SM, Rodríguez-Bocanegra E, Garcia-Delgado AB, Moya-Molina M, **Bhattacharya SS**, Díaz-Corrales FJ. (2019). *Stem Cell Res.*; **36**: 101426. doi: 10.1016/j.scr.2019.101426. [Epub ahead of print] PMID: 30921587
379. Subretinal Transplant of Induced Pluripotent Stem Cell-Derived Retinal Pigment Epithelium on Nanostructured Fibrin-Agarose. García Delgado AB, de la Cerda B, Alba Amador J, Valdés Sánchez ML, Fernández-Muñoz B, Relimpio López I, Rodríguez de la Rúa E, Díez Lloret A, Calado SM, Sánchez Pernaute R, **Bhattacharya SS**, Díaz Corrales FJ. (2019). *Tissue Eng Part A*. **25**:799-808. doi: 10.1089/ten.TEA.2019.0007. PMID: 30963803
380. Generation of a human iPS cell line (CABi003-A) from a patient with age-related macular degeneration carrying the CFH Y402H polymorphism. Garcia-Delgado AB, Calado SM, Valdes-Sanchez LM, Montero-Sanchez A, Ponte-Zuñiga B, de la Cerda B, **Bhattacharya SS**, Diaz-Corrales FJ. (2019). *Stem Cell Res*. **38**:101473. doi: 10.1016/j.scr.2019.101473. Epub 2019 May 29. PMID: 31176916

381. Dysfunctional LAT2 Amino Acid Transporter Is Associated With Cataract in Mouse and Humans. Knöpfel EB, Vilches C, Camargo SMR, Errasti-Murugarren E, Stäubli A, Mayayo C, Munier FL, Miroshnikova N, Poncet N, Junza A, **Bhattacharya SS**, Prat E, Berry V, Berger W, Heon E, Moore AT, Yanes Ó, Nunes V, Palacín M, Verrey F, Kloeckener-Gruissem B. *Front Physiol.* (2019). **10**: 688. doi: 10.3389/fphys.2019.00688. eCollection 2019. PMID: 31231240
382. The Resveratrol Prodrug JC19 Delays Retinal Degeneration in rd10 Mice. Valdés-Sánchez L, García-Delgado AB, Montero-Sánchez A, de la Cerda B, Lucas R, Peñalver P, Morales JC, **Bhattacharya SS**, Díaz-Corrales FJ. (2019). *Adv Exp Med Biol.* **1185**: 457-462. doi: 10.1007/978-3-030-27378-1\_75. PMID: 31884654
383. Retinal pigment epithelium degeneration caused by aggregation of PRPF31 and the role of HSP70 family of proteins. Valdés-Sánchez L, Calado SM, de la Cerda B, Aramburu A, García-Delgado AB, Massalini S, Montero-Sánchez A, Bhatia V, Rodríguez-Bocanegra E, Díez-Lloret A, Rodríguez-Martínez D, Chakarova C, **Bhattacharya SS**, Díaz-Corrales FJ. (2019). *Mol Med.* Dec 31; **26**(1):1. doi: 10.1186/s10020-019-0124-z. PMID: 31892304
384. Mutations in SPATA13/ASEF2 cause primary angle closure glaucoma. Waseem NH, Low S, Shah AZ, Avisetti D, Ostergaard P, Simpson M, Niemiec KA, Martin-Martin B, Aldehlawi H, Usman S, Lee PS, Khawaja AP, Ruddle JB, Shah A, Sackey E, Day A, Jiang Y, Swinfield G, Viswanathan A, Alfano G, Chakarova C, Cordell HJ, Garway-Heath DF, Khaw PT, **Bhattacharya SS**, Waseem A, Foster PJ. (2020). *PLoS Genet.* Apr 27; **16**(4): e1008721. doi: 10.1371/journal.pgen.1008721. [Epub ahead of print] PMID: 32339198

#### BOOK CHAPTERS:

1. Cellular exchanges and happenings after injury and in the critically ill. Flear, C.T.G., Bhattacharya, S.S. and Nandra, G.S. (1977): in *Nutritional aspects of care in the critically ill*, edited by Richards, J.R. and Kinney, J.M. Published by Churchill Livingstone (Edinburgh, London and N.Y.) p195-224.
2. Transmembrane sodium exchanges in health and heart disease: actions of beta-blockers. Flear, C.T.G., Bhattacharya, S.S. and Jackson, A. (1981): on *Current themes in cardiology*, edited by G.F.B. Birdwood and J.G. Russel, published by Geigy Pharmaceuticals, Horsham, England, p29-32.
3. DNA probes in the diagnosis of X-linked retinitis pigmentosa. Wright, A.F., Dempster, D., Clayton, J.F., Jay, M.R., Bird, A.C. and Bhattacharya, S.S. (1985). In *Biotechnology in Diagnostics*, Koprowski, H., Ferrone, S., Alvertini, A. (eds), Elsevier Science Publishers, p261-268.
4. The detection of X-linked retinitis pigmentosa by DNA hybridization. Wright, A.F., Dempster, M., Jay, M.R., Clayton, J.F., Bhattacharya, S.S. and Bird, A.C. (1985): In *"Retinal Degeneration: Experimental and Clinical Studies*, Hollyfield, J.G., La Vail, M.M. (eds) Alan R. Liss Inc., New York.

5. Recombinant DNA mapping of retinitis pigmentosa genes. Wright, A.F., Bhattacharya, S.S., Craig, I.W., Jay, M.R., Dempster, M., Fraser, N., Meitinger, T., Jay, B., Bird, A.C. and Evans, H.J. (1988): *Molecular Biology of the Eye: Genes, Vision and Ocular Diseases* (Eds Paitagorsky, Shinohara and Zelinka), Alan R. Liss (Pub), p293-303.
6. X-linked retinitis pigmentosa: A molecular genetic approach to isolating the defective genes. Lindsay, S., Jay, M.R., Bower, D.J., Adam, G., Inglehearn, C.F., Sealey, P.G., Papiha, S.S. and Bhattacharya, S.S. (1989): In "Inherited and environmentally induced retinal degenerations. Published by Alan R. Liss Inc., New York, p83-97.
7. Molecular genetic studies of inherited eye diseases. Inglehearn, C.F., Bashir, R., Curtis, A., Lindsay, S.J. and Bhattacharya, S.S. (1990): In: *Application of molecular genetics to the diagnosis of inherited disease*, K. Davies (ed), Royal College of Physicians, London (Pub.), p17-27.
8. Molecular genetic studies in autosomal dominant retinitis pigmentosa. Bashir, R., Inglehearn, C.F., Lester, D., Lauffart, B., Keen, T.J., Papiha, S.S., Jay, M.R., Bird, A.C. and Bhattacharya, S.S. (1991). In "Degenerative Retinopathies: Advances in Clinical and Genetic Research", Humphries, Bhattacharya and Bird (eds) CRC Press/ Boston p13-19.
9. Linkage studies and rhodopsin mutation detection in autosomal dominant retinitis pigmentosa: an update. Bhattacharya, S.S., Bashir, R., Keen, T.J., Lester, D.H., Lauffart, B., Jay, M.R., Bird, A.C. and Inglehearn, C.F. (1991). *Retinal Degenerations*, Anderson, Hollyfield and LaVail (eds), CRC Press publication, p375-382.
10. Extensive genetic heterogeneity in autosomal dominant retinitis pigmentosa. Farrar, G.J., Siobhan, A., Jordan, Kumar-Singh, R., Inglehearn, C.F., Gal, A., Gregory, C., Al-Magthteh, A., Kenna, P.F., Kenna, Humphries, M.M., Sharp, E.M., Sheils, D.M., Bunge, S., Hargrave, P.A., Denton, M.J., Schwinger, E., Bhattacharya, S.S. and Humphries P. (1993) In *Retinal Degeneration: Clinical and Laboratory Applications*, edited by Hollyfield, J.G., Anderson, R.E., LaVail, M.M. Plenum Press, p63-77.
11. Molecular Genetic Studies of retinal dystrophies principally affecting the macula. Evans, K., Gregory, C.Y., Wijesuriya, S., Jay, M., Chopdar, A. and Bhattacharya, S.S. (1995). In "Retinal Degenerations" eds Anderson, R.E., LaVail, M.M. and Hollyfield, J.C. Plenum Press, London, p323-330.
12. Rhodopsin mutations in photoreceptor degeneration: implications for protein dysfunction. Bhattacharya, S.S., Al-Magthteh, M. and Inglehearn, C. (1997) in *Protein Dysfunction in Human Genetic Disease*, Swallow, D.M. and Edwards, Y.H. (eds.), Bios Scientific Publ. U.K., pp 187-202.
13. Francis PJ, Berry V, Kaushal S, Moore AT, Bhattacharya SS. Len aquaporin mutations. *Molecular Biology and Physiology of Water and Solute Transport*, Kluwer Academic/Plenum, New York 2000, Eds. Hohmann and Nielsen, p 205-207.

## **RESEARCH GRANT SUPPORT OBTAINED (1986 to present):**

I have obtained grant income in excess of £18 million based on 70 successful grant applications over the last 31 years. I have also sponsored 11 Fellowship applicants.

Genetic Mapping in X-Linked RP, National RP Foundation of USA, July 1986-June 1989, SS Bhattacharya and AF Wright, £31,400

Molecular Genetic Analysis of Retinal Specific Genes, British RP Society, October 1986-September 1989, SS Bhattacharya, £26,250

Gene Mapping in Autosomal Dominant RP, National RP Foundation of USA, October 1987-September 1993, SS Bhattacharya, £334,000

Gene Mapping in Usher's Syndrome, British RP Society, October 1987 - September 1989, SS Bhattacharya and SS Papiha, £14,500

Isolation and Identification of Candidate Genes for XLRP and other X-Linked Eye Diseases, The Wellcome Trust, November 1987-October 1991, SS Bhattacharya £123,300

Isolation of Retinal Genes, Fight for Sight, October 1988 - September 1991, SS Bhattacharya, £24,000

Deletions in DNA and Phenotypic Expression of Muscular Dystrophy, Medical Research Council, October 1988-September 1989, J Harris and SS Bhattacharya, £19,088

Isolation of Retinal Genes Using Antibody Probes to Retinal Proteins, British RP Society, October 1988 - September 1991, SS Bhattacharya, £12,000

Molecular Genetic Studies of X-Linked Hypohidrotic Ectodermal Dysplasia. The Wellcome Trust, January 1989 - December 1991, SS Bhattacharya and P. Harper, £82,247

Incontinentia Pigmenti and Retts Syndrome: Two X-linked Dominant, Male-Lethal Causes of Mental Retardation in Females, The Sir Jules Thorn Trust, January 1989 - December 1990, SS Bhattacharya, £83,533

The Molecular Biology of a Feline Model for Retinitis Pigmentosa, The Wellcome Trust, February 1989 to January 1992, SS Bhattacharya and G. Arden, £89,918

Identification of the Genes in X-Linked Retinitis Pigmentosa and Norrie's Disease, The British RP Society, May 1989 - April 1992, SS Bhattacharya, £54,244

Molecular Genetic Studies of inherited eye disease, University Research Committee - Newcastle upon Tyne, December 1989 - November 1992, SS Bhattacharya, £39,750

Development and application of Molecular techniques for the study of inherited eye diseases, Special Trustees of Royal Victoria Infirmary, Newcastle upon Tyne, September 1990 to August 1992, S S Bhattacharya and A L Crombie, £36,000

Autosomal dominant hereditary haemorrhagic telangiectasia (HHT): A genomic search for the locus, Medical Research Council, June 1991 - May 1993, M Porteous, J Burn and SS Bhattacharya, £46,520

Towards identifying the causative genetic defects in autosomal dominant retinitis pigmentosa, The British RP Society, September 1991 to August 1994, SS Bhattacharya, £151,156

Cornelia de Lange Syndrome: A molecular genetic Study, Medical Research Council, October 1991 to September 1994, M Ireland and SS Bhattacharya, £88,659

Molecular Genetic studies of inherited retinal Degeneration, The Wellcome Trust, October 1991 to September 1996, S Lindsay and SS Bhattacharya, £211,193

Isolation and characterisation of the genes encoding canine phospho-diesterases and their role in progressive atrophy, The Wellcome Trust, October 1991 to September 1994, SS Bhattacharya, £43,036

Molecular and biochemical studies on the etiology of retinal degeneration in the Royal College of Surgeons rat, The Wellcome Trust, December 1991 to November 1994, C Gregory and SS Bhattacharya, £92,263

Characterisation of functional loss in the outer retinal dystrophies with known genetic mutations, Medical Research Council, October 1993 to September 1998, A.C. Bird, SS Bhattacharya and F Fitzke, £680,000

Development for a delivery system for gene therapy of inherited retinal degenerations, Medical Research Council, September 1994 to August 1997, SS Bhattacharya, DM Hunt and AC Bird, £287,393

Positional Cloning of the X-linked Retinitis Pigmentosa gene (RP2) in proximal Xp, The Wellcome Trust, October 1994 to September 1997, SS Bhattacharya, £134,889

Mapping and identification of genes for autosomal dominant cataracts in humans, The Wellcome Trust, January 1995 to December 1997, A Shiels and SS Bhattacharya, £260,120

The isolation of genes expressed in the retina from defined regions of the X chromosome, Guide Dogs for the Blind, April 1996 to March 1999, SS Bhattacharya, £103,437

Autosomal dominant retinitis pigmentosa, cloning and characterisation of a novel gene on chromosome 7p, Foundation Fighting Blindness, September 1996 to August 1999, SS Bhattacharya, £105,000

Preservation of vision by the preservation of apoptosis in the retina, Guide Dogs for the Blind, December 1996 to November 1999, SS Bhattacharya and CY Gregory, £122,923

Expression and characterisation of proteins involved in eye development and disease, The WellcomeTrust, July 1997 to June 1998, MJ Warren, SS Bhattacharya and DM Hunt, £86,758

Positional cloning of the X-linked retinitis pigmentosa-2 (RP2) gene, The WellcomeTrust, October 1997 to February 2001, SS Bhattacharya, £160,054

Mapped cataract loci and candidate genes, The WellcomeTrust, March 1998 to May 2001, SS Bhattacharya and AT Moore, £299,055

Identification, cloning and characterisation of a dominant RP gene on chromosome 19q, The WellcomeTrust, April 1998 to March 1999, SS Bhattacharya, £48,808

Expression and characterisation of proteins involved in retinal disease, The WellcomeTrust, Jan 1999 to Dec 2003, M Warren, DM Hunt and SS Bhattacharya, £985,000

Cloning and characterisation of the chromosome 3q28 autosomal dominant optic atrophy gene, The WellcomeTrust, April 1999 to March 2002, M Votruba, SS Bhattacharya and AT Moore, £158,255

Autosomal dominant retinitis pigmentosa: Identification of new loci by genetic linkage studies and positional cloning of RP genes from chromosomes 19q(RP11), 17p(RP13) and 1q(RP18), Foundation Fighting Blindness, September 1999 to August 2002, SS Bhattacharya, £130,980

Functional genomics and retinal degenerations, The European Union, September 2000 to August 2004, SS Bhattacharya, 200,000 EURO

**Co-ordinator** for a successful MRC Co-operative group application entitled: “Retinal degenerative diseases: An integrated approach from functional genomics to therapies”. Grant ref: G9900417, tenure 60 months from September 2000.

Autosomal dominant retinitis pigmentosa; identification and functional analysis of the RP11 disease gene, Medical Research Council, October 2000 to June 2004 SS Bhattacharya and DM Hunt, £231,504

Human DNA sample collection and phenotypic characterisation of AMD: A key national resource to determine predisposing genetic factors, Medical Research Council, February 2001 to January 2004, AC Bird, SS Bhattacharya, P Luthert, A Webster and B Clarke, £394,336

Development of animal models for retinal disease, The Wellcome Trust, March 2001 to February 2004, DM Hunt, D Wells and S S Bhattacharya, £340,450

Identification of genes responsible for inherited cataract in man, The Wellcome Trust, June 2001 to Feb 2005, SS Bhattacharya and AT Moore £362,997

X-linked Progressive Cone-Dystrophies: Identification of the genes causing COD1 and COD2, The Wellcome Trust, April 2002 to March 2005, A Hardcastle, SS Bhattacharya, AT Moore, DM Hunt and M Cheetham, £172,685

Development of an inducible photoreceptor specific system for use in gene therapy for retinal degenerations, Foundation Fighting Blindness, July 2002 to June 2005 RR Ali and S S Bhattacharya, \$195,102

Autosomal dominant retinitis pigmentosa: Identification of new loci by genetic linkage studies and positional cloning of RP genes from chromosomes 19q(RP11), 17p(RP13) and 1q(RP18), Foundation Fighting Blindness, July 2003 to June 2005, SS Bhattacharya, \$136,500

An investigation into the genetic basis of keratoconus, Special Trustees of Moorfields Eye Hospital, January 2004 to December 2005, S Tuft and SS Bhattacharya, £46,000

Cloning of retinal dystrophy genes from chr 1q, 6cen, 4q, British Retinitis Pigmentosa Society, July 2003 to June 2006 SS Bhattacharya, £136,257

Genotyping in ophthalmic diseases, Special Trustees of Moorfields Eye Hospital, June 2002 to December 2006, SS Bhattacharya and AC Bird , £840,000

**Scientific co-ordinator** of a Euro 3.68 million grant for a retinal research training network (RETNET) involving 10 European laboratories (January 2004-December 2007). Grant ref: MRTN-CT-2003-504003

Identification of novel retinal disease genes, The European Union, January 2004 to December 2007, SS Bhattacharya, Euro 375,474

Genetic basis of partial penetrance in adRP, Fight for Sight, September 2004 to August 2007, SS Bhattacharya, £65,800

An investigation of the genetic basis of myopia in the 1958 British birth cohort, Medical Research Council, June 2005 to July 2008, Dr J. Rahi, SS Bhattacharya, A Webster and C Peckham, £272,350

**Component leader** of the Mechanisms of Disease module of EU funded EURO 10 Million integrated project (IP) on Functional Genomics of the Retina in Health and Disease (GENORET) Grant ref: LSHG-CT-2005-512036

Genetic mapping and gene identification of a novel monogenic retinal dystrophy, The European Union, April 2005 to March 2009, SS Bhattacharya, Euro 440,000

A genomewide association study for refractive error in the 1958 British Birth Cohort, The Wellcome Trust, May 2008 to April 2009, Dr J. Rahi, C Hammond, A Webster, SS Bhattacharya and P Sham, £591,024



Gene therapy for childhood diseases, Department of Health, August 2004 to July 2009, R Ali, SS Bhattacharya, L Da Cruz, AT Moore and AJ Thrasher, £915,552

Genetic Studies, Functional Genomics and Animal Models of Retinal Disease, The Foundation Fighting Blindness, July 2005 to June 2010, SS Bhattacharya and DM Hunt, £345,000

**Scientific co-ordinator** of a Euro 1.8 million grant for an Early Stage Training (EST) in Neurodegeneration research (NEUROTRAIN) involving 10 EU partners (January 2006-December 2009). Grant ref: MEST-CT-2005-020235

Genetics and Functional Genomics of Retinal Degeneration, The European Union, January 2006 to December 2009, SS Bhattacharya, Euro 105,000

Genotyping in ophthalmic diseases, National Institute of Health Research, Biomedical Research Centre at Moorfields Eye Hospital, April 2007 to March 2012, SS Bhattacharya, £285,000

Identification of a major gene (RP25) on chromosome 6q for autosomal recessive RP, British Retinitis Pigmentosa Society, April 2007 to March 2010 SS Bhattacharya and A Webster, £159,257

Monolithically integrated interferometric biochips for label-free early detection of human diseases (PYTHIA), European Union funded consortium of 8 laboratories, May 2008 to December 2011, SS Bhattacharya, UCL share Euro 216,000

Molecular mechanisms of disease associated with a major gene (PRPF31) for ADRP, British RP Society, December 2008 to February 2012, SS Bhattacharya, £176,000

To investigate the molecular basis of partial penetrance associated with PRPF31 gene in RP, Fight For Sight, October 2008 to March 2012, SS Bhattacharya £149,928

Identification of interacting partners for TOPORS. Fight For Sight, October 2010 to September 2013, SS Bhattacharya, £92,500

Molecular mechanisms associated with PRPF31 for autosomal dominant retinitis pigmentosa. Shom Shanker Bhattacharya.P09-CTS-04967, January 2011 -December 2013 Consejería de Economía, Innovación y Ciencia de la Junta de Andalucía (Excellence Project).255.774 €

Functional studies of retina expressed genes. Rosetrees Trust with Special Trustees of MEH, July 2012 to June 2013, SS Bhattacharya, £ 45,000

Y2H technology for identification of interacting partners for EYS. Fight For Sight, January 2012 to December 2014, SS Bhattacharya, £96,500

Functional analysis and characterization of a novel gene for autosomal recessive retinitis pigmentosa, British Retinitis Pigmentosa Society, April 2011 to March 2015, SS Bhattacharya, £156,545

Identification and functional characterization of retinal genes. Rosetrees and Butterfield Trust, August 2012 to November 2015, SS Bhattacharya, £206,350

Gene identification, Functional Characterisation and Animal Models of Retinal Degeneration, The Foundation Fighting Blindness, (USA), July 2010 to July 2016, SS Bhattacharya, £467,460

Genotyping in ophthalmic diseases, National Institute of Health Research, Biomedical Research Centre at Moorfields Eye Hospital, April 2012 to June 2017, SS Bhattacharya, £420,000

Functional studies and characterization of retinal genes. Rosetrees and Butterfield Trust, July 2013 to June 2017, SS Bhattacharya, £219,250

Convenio de colaboración para el desarrollo del Programa de Captación del Conocimiento C2A (#) SS Bhattacharya. Expediente: P09-CTS-04967 €259,169 per year

EYERISK-Genetic and Functional Characterisation of Age related macular degeneration, European Union H2020, June 2015-May 2019, SS Bhattacharya, €196,750

## **FELLOWSHIPS SPONSORED**

Molecular genetic analysis of retinitis pigmentosa, The Wellcome Trust, October 1992 to September 1997, CF Inglehearn and SS Bhattacharya, £468,530

Molecular genetic approaches to macular disease, The Wellcome Trust, May 1995 to April 1999, CY Gregory and SS Bhattacharya, £312,903

Autosomal dominant optic atrophy: positional cloning of OPA 1 gene on chromosome 3q, The Wellcome Trust, December 1996 to November 1998  
M Votruba and SS Bhattacharya, £111,675

The effects of TGF- $\beta$  on conjunctival fibroblasts, extracellular matrix interactions and wound healing, The Wellcome Trust, September 1996 to August 1998, F Cordeiro and SS Bhattacharya, £97,217

Molecular genetics of autosomal dominant retinitis pigmentosa, The Wellcome Trust, October 1997 to September 2000, CF Inglehearn and SS Bhattacharya, £482,749

Identification of the human homologue of the *Drosophila* optomotor-blind gene and investigation of its role in development of the visual system, Medical Research Council, November 1996 to October 2000, JC Sowden and SS Bhattacharya, £213,722

A search for quantitative trait that influence the development of myopia in humans, The Wellcome Trust, October 1999 to September 2002, AR Webster and SS Bhattacharya, £221,563

Identification and characterisation of novel retina-enriched cDNAs as candidate genes for retinal degenerations, The European Union, Dec 2000 to November 2002, M Papaioannou and SS Bhattacharya, €114,572

Gene editing and iPSC technology for the generation of neural progenitor cells, Miguel-Servet Fellowship, September 2012 to August 2017, Slaven Erceg, €320,000

Therapeutic approaches for inherited retinal dystrophies, Miguel-Servet Fellowship, September 2016 to August 2021, Francisco Diaz Correlas, €350,000

DNA damage and repair in retinal photoreceptor cells, Juan de la Cierva Fellowship, January 2016 to December 2017, Vaibhav Bhatia, €75,000