CURRICULUM VITAE

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DATE OF BIRTH	18th January 1950
CITIZENSHIP	British

CURRENT APPOINTMENT

01.10.92 – Present Sembal Professor of Experimental Ophthalmology and Head of Molecular Genetics, Institute of Ophthalmology, University College London.

QUALIFICATIONS

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

MSc, 1971, Clinical Biochemistry, University of Newcastle upon Tyne, U.K.
Awarded a tuition fee grant from the British Council for the MSc Course.

PhD, 1977, Department of Clinical Biochemistry (Faculty of Medicine), University of Newcastle upon Tyne, UK.
Awarded a fellowship from the Wellcome Trust for the PhD studies.

DISSERTATION AND THESIS SUBMITTED

Dissertation submitted in partial fulfilment for the degree of Master of Science to the University of Newcastle upon Tyne, 1971.
“Investigations of electrolyte permeability of frog sartorius muscle membranes”.

Thesis entitled 'Na-exchanges in skeletal and heart muscle membrane of the frog' submitted for the degree of Doctor of Philosophy to the University of Newcastle upon Tyne, 1977.

PREVIOUS APPOINTMENTS

From : 01.05.77
To : 30.05.80

Research Associate in the University Department of Clinical Biochemistry, Royal Victoria Infirmary, Newcastle Upon Tyne

From : 01.06.80
To : 31.01.87

Scientific staff and Senior Research Fellow, MRC Human Genetics Unit, Edinburgh and University of Edinburgh.

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.

RESEARCH EXPERIENCE AND RESEARCH ACTIVITIES

Newcastle upon Tyne

From : 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

From : June 1980
To : September 1980

Department of Clinical Surgery

Assessment of oestrogen receptor activity. This involved the comparison of the sensitivities of three methods (sucrose density gradient analysis; intranuclear translocation of radioactive oestrogen; and standard saturation analysis using dextran coated charcoal) for assaying oestrogen receptor activity on the same tissue sample and to assess if they measured the same parameter for receptor activity.

From : October 1980
To : September 1981

Department of Medicine (Western General Hospital, Edinburgh)

The role of intracellular electrolytes and platelets in the vascular changes associated with hypertension. The hypertension work involved the setting up of a continuous flow in situ animal model system whereby changes in perfusate pressure on the electrolyte and H2O composition of the arterial endothelium was measured and platelet adhesion and aggregation was studied using Scanning Electron Microscopy.

From : October 1981
To : March 1983

MRC Mammalian Genome Unit (Kings Buildings, University of Edinburgh)

I spent 18 months training in recombinant DNA techniques in Dr. Edwin Southern's world-renowned MRC laboratory. The training included DNA extraction from cells and various tissues, bacterial culture techniques, cell transformation, construction of cDNA and genomic libraries, recombinant selection, amplification and purification of plasmid and bacteriophage DNA and nucleic acid hybridization and sequencing techniques.

From : April 1983
To : January 1987

MRC Clinical & Population Cytogenetics Unit (Western General Hospital, Edinburgh)

I have been involved in collaboration with members of the 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) have been identified using these

probes which 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). Further studies have been undertaken with 40 XLRP families and 14 DNA markers spanning the short arm and the proximal long arm of the X-chromosome to address the question of genetic heterogeneity. Two loci for XLRP have now been identified on the short arm of the X-chromosome. Also, work is in progress in identifying expressed sequences that map to appropriate regions of the X-chromosome as suitable candidates for the putative RP genes.

Newcastle upon Tyne

From : 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. This laboratory has been mainly equipped from my research grants and is fully operational. The Newcastle Area Health Authority has provided me with permanent funding to employ a principal scientist, three senior grade scientists and 4 medical laboratory techicians for the diagnostic service. At present, DNA diagnostic tests are provided for the Duchenne Muscular Dystrophy, Becker Muscular Dystrophy, Cystic Fibrosis and Huntington's Disease registers in the region. On the availability of new resources, molecular diagnostic tests can be provided for most inherited genetic diseases in the region for which closely linked DNA markers have been identified. I do not anticipate any technical or operational problems with the expansion of such services. Along with the DNA based tests, my laboratory also provides a wide spectrum of diagnostic tests using biochemical and serological markers.

My research group is 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. Work is in progress to define the mutation spectrum in candidate genes such as Rhodopsin and Peripherin in patients with ADRP. We are also investigating 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 mitrochondrial DNA and racial variations in the allele frequencies of DNA polymorphisms. The overall aim of this research is to understand the nature and biology of genetic diseases and to provide an efficient diagnostic service for carrier detection and prenatal diagnosis. My laboratory is a participating group in UK and EEC Human Genome Mapping Initiative and is developing 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

From : October 1992
To : Present

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 ofices and the research facilities. The division is well equiped for biochemical and molecular biological techniques. 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 of the various 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. 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.

MEMBERSHIP OF 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).

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.

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 February 2007

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 I have been giving occasional lectures to genetics students at University College London.

Ph.D. THESIS SUPERVISION

Paula Monaghan	1989	Douglas Lester	1990
Ivan Still	1991	Alison Hardcastle	1992
Smaro Kamakari	1994	Peter Clements	1994
Mai Al-Maghetheh	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

Currently supervising 6 Ph.D students and 1 MD student: Mai Abd El-Aziz, Brotati Ghosh, Kinga Bujakowska, Amna Shah, Ciara O'Driscoll, Francesca Fiocco and Petra Liskova

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
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 (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).
International Advisor to National Eye Institute of NIH for EyeGENE Programme (Jan 2006)

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 papers for all top journals in the field including Nature, Science, Cell, Nature Genetics, American Journal of Human Genetics, Human Molecular Genetics and Investigative Ophthalmology & Visual Science.

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):
1st Meeting 1996 - Madrid, Spain
2nd Meeting 1997 - Athens, Greece
3rd Meeting 1999 - Tubingen, Germany
4th 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.

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.

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

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. Actual or standard bicarbonate? Tibi, L., Bhattacharya, S.S. and Flear, C.T.G. (1979). Lancet, **2**: 1139.

3. 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.

4. Variability in pK'1 of human plasma. Tibi, L., Bhattacharya, S.S. and Flear, C.T. (1982). Clin. Chim. Acta. **121**: 15-31.

5. 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.

6. 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.

7. 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.

8. 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.

9. 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.

10. 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.

11. Human interstitial retinal-binding protein (IRBP): cloning, partial sequence and chromosomal localisation. Lious, 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.

12. 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.

13. 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.

14. 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.

15. A case of disputed maternity. Roberts, D.F., Papiha, S.S. and Bhattacharya, S.S. (1987). Lancet **II**: 478-480.

16. 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.

17. 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.

18. 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.

19. 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.

20. 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.

21. 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.

22. 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.

23. 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.

24. 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.

25. 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.

26. 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.

27. 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.

28. 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.

29. 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.

30. 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.

31. 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.

32. 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.

33. 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.

34. A new Xmnl 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.

35. 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. Anthropoll. **85**:329-334.

36. 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.

37. 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.

38. 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.

39. Increased band sharing in DNA fingerprints of an inbred human population. Bellamy, R.J., Inglehearn, C.F., Jaliki, I.K., Jeffreys, A.J. and Bhattacharya, S.S. (1991). Hum. Genet. **87**: 341-347.

40. 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.

41. 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.

42. 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.

43. 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.

44. 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.

45. 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.

46. 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.

47. 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.

48. 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.

49. 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.

50. 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.

51. 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.

52. 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. Gent. **1**: 776.

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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

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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

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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-diesteras 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) and1q(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) and1q(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 Hospita, 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

Active Grants

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, 375,474 EURO

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 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 May 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, 440,000 EURO

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, 105,000 EURO

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

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 dominat 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-β 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 theDrosophila 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 candidategenes for retinal degenerations, The European Union, Dec 2000 to November 2002, M Papaioannou and SS Bhattacharya, 114,572 EURO