

## BIOGRAPHICAL SKETCH

CURRICULUM VITAE: PETER HUMPHRIES

DATE OF BIRTH 20<sup>th</sup> April 1948

PLACE OF BIRTH Newcastle-upon-Tyne, UK

OFFICE ADDRESS Ocular Genetics Unit,  
Smurfit Institute of Genetics  
Trinity College Dublin,  
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DEGREES/DISTINCTIONS Moderator, Natural Science, Trinity  
College Dublin, 1967-1971.  
Molecular Genetics, PhD Trinity College  
Dublin, 1971-74.  
Fellow Trinity College Dublin 1987.  
Alcon Prize for Vision Research 1993.  
Member of Royal Irish Academy 1986.  
Dsc [HC] University of Szeged 1997.  
Member European Molecular Biology  
Organization (EMBO) 2000.

POSTS Fellow of EMBO, University Louis  
Pasteur, Strasbourg 1974-1977.  
Research Fellow, Beatson Institute  
for Cancer Research 1977-1979.  
Lecturer in Medical Genetics, The  
Queen's University of Belfast 1979-1983  
Lecturer in Medical Genetics, Trinity  
College Dublin 1983-1991.  
Chair of Medical Molecular Genetics  
Trinity College Dublin 1991.

CURRENT/RECENT MEMBERSHIPS  
Retina International (IRPA)-Scientific and Medical Advisory Board; Foundation  
Fighting Blindness (USA)-Focus Group on Genetics and Genetic Technology;  
Dystrophic Epidermolysis Bullosa Research Association (DEBRA)-International  
Medical and Scientific Advisory Board; Member, Alcon Research Institute  
(USA); The Wellcome Trust-Neurosciences Panel, Vision Research Working  
Party, Genetics Advisory Group; Communicating Editor, Human Mutation;  
Editorial Board, Human Molecular Genetics; The Human Genome Organisation  
(HUGO)- Founder Irish Member; Irish Society of Human Genetics (President),  
American Society of Human Genetics, Association for Research in Vision and  
Ophthalmology, Fighting Blindness Ireland Medical and Scientific Advisory  
Board, Fellow of European Molecular Biology Organization; AMD (Age-Related  
Macular Dystrophy) Alliance International Scientific Advisory Panel. Medical  
Research Council (UK) Advisory Panel. Member, College of Reviewers for the  
Canada Research Chairs Programme, European Vision Institute, Honorary  
Member Association of Physicians of Great Britain and Ireland.

## RESEARCH INTERESTS

Human Molecular/Medical Genetics with special interest in visual neuroscience

## SELECTED PUBLICATIONS (IN THE AREA OF VISION RESEARCH)

1. Farrar GJ, Geraghty MG, Moloney JMB, McConnell DJ and Humphries P. Linkage analysis of X-linked retinitis pigmentosa in the Irish population. *J. Med. Genet.*, 25, 222-226, 1988.
2. Daiger SP, Humphries MM, Giesenschlag N, Sharp EM, McWilliam P, Farrar GJ, Bradley DG, McConnell DJ, Kenna P, Sparkes RS, Spence MA, Heckenlively JR and Humphries P. Linkage analysis of human chromosome 4: exclusion of autosomal dominant retinitis pigmentosa and detection of new linkage groups. *Cytogen. Cell Genet.*, 50, 181-187, 1989.
3. Bradley D, Farrar GJ, Sharp EM, Kenna P, Humphries MM, McConnell DJ, Daiger SP, McWilliam P and Humphries P. Autosomal dominant retinitis pigmentosa: exclusion of the gene from the short arm of chromosome 1, including the region around the Rhesus locus. *Am. J. Hum. Genet.*, 44, 570-576, 1989.
4. Farrar GJ, McWilliam P, Sharp EM, Kenna P, Bradley DG, Humphries MM, McConnell DJ and Humphries P. Autosomal dominant retinitis pigmentosa: exclusion of a gene from extensive regions of chromosomes, 6, 13, 20 and 21. *Genomics*, 5, 619-622, 1989.
5. McWilliam P, Farrar GJ, Kenna P, Bradley DG, Humphries MM, Sharp EM, McConnell DJ, Lawler M, Sheils D, Stephens K, Daiger SP and Humphries P. Autosomal dominant retinitis pigmentosa: localization of an adRP gene to the long arm of chromosome 3. *Genomics*, 5, 612-619, 1989.
6. Ott J, Bhattacharya S, Chen JD, Denton MJ, Donald J, Dubar C, Farrar GJ, Fishman GA, Frey D, Gal A, Humphries P, Jay B, Jay M, Litt M, Mochler M, Musarella M, Neugebauer M, Nussbaum RL, Terwillinger JD, Weleber RG, Wirth B, Wong R, Worton RG and Wright AF. Localizing multiple X-chromosome linked retinitis pigmentosa loci using multilocus homogeneity tests. *Proc. Natl. Acad. Sci. USA*, 87, 701-704, 1990.
7. Humphries P. Retinitis pigmentosa: genetic mapping in X-linked and autosomal forms of the disease. *Clin. Genet.*, 38, 1-13, 1990.
8. Farrar GJ, McWilliam P, Bradley DG, Kenna P, Sharp EM, Humphries MM, Lawler M, Eiberg H, Conneally MP, Trofatter JA and Humphries P. Autosomal dominant retinitis pigmentosa: linkage to rhodopsin and evidence for genetic heterogeneity. *Genomics*, 8, 35-40, 1990.
9. Blanton SH, Cottingham AW, Giesenschlag N, Heckenlively JR, Humphries P and Daiger SP. Further evidence for exclusion of linkage between type II autosomal dominant retinitis pigmentosa adRP and D3S47. *Genomics*, 8, 179-181, 1990.
10. Kumar-Singh R, Bradley DG, Kenna P, Farrar GJ, Lawler M, Humphries MM, Sharp E, Jordan SA, McWilliam P and Humphries P. Autosomal dominant retinitis pigmentosa (RP4): A new multi-allelic marker genetically linked to the disease locus. *Hum. Genet.*, 86, 502-504, 1990.
11. Farrar GJ, Kenna P, Jordan S, Kumar-Singh R, Humphries MM, Sharp EM, Sheils D and Humphries P. A 3 base-pair deletion in the peripherin gene in one form of retinitis pigmentosa. *Nature*, 354, 478-480, 1991.
12. Farrar GJ, Kenna P, Redmond R, McWilliam P, Bradley DG, Humphries MM, Sharp EM, Inglehearn C, Bashir R, Jay M, Watty A, Ludwig M, Schinzel A, Sammans C, Gal A, Bhattacharya S and Humphries P. Autosomal dominant retinitis pigmentosa (RP4): absence of the rhodopsin codon 23 proline->histidine transversion in pedigrees of European origin. *Am. J. Hum. Genet.*, 47, 941-945,

- 1991.
13. Jordan S, McWilliam P, O'Brian DS and Humphries P. Dinucleotide repeat polymorphism at the D14S42 locus. *Nucl. Acids Res.*, 19, 1171, 1991.
  14. Farrar GJ, Kenna P, Redmond R, McWilliam P, Bradley DG, Humphries MM, Sharp EM, Fishman G, Marchese C, Fusi L, Dufier JL, Abitbol M and Humphries P. Autosomal dominant retinitis pigmentosa (RP4): analysis of mutations within the rhodopsin gene. *Retinitis Pigmentosa: Advances in Clinical and Genetic Research*. (Pbl. CRC Press. Florida: Eds. Humphries P. Bhattacharya S and Bird A), 1991.
  15. McWilliam P, Jordan S, Kenna P, Humphries MM, Kumar-Singh R, Sharp EM and Humphries P. Progress in the localization of a late-onset adRP gene. *Retinal Degenerations*. (Pbl. CRC Press. Florida: Eds. Hollyfield. Anderson and LaVail) 369-373, 1991.
  16. Farrar GJ, Kenna P, Redmond R, Sheils D, McWilliam P, Humphries MM, Sharp EM, Jordan SA, Kumar-Singh R and Humphries P. Autosomal dominant retinitis pigmentosa: a mutation in codon 178 of the rhodopsin gene in an adRP family of Celtic origin. *Genomics*, 11, 1170-1171, 1991.
  17. Doak S, Jordan SA, Kumar-Singh R and Humphries P. A sequence polymorphism in the human peripherin/RDS gene. *Nucl. Acids Res.*, 19, 6982, 1991.
  18. Farrar GJ, Jordan SA, Kenna P, Humphries MM, Kumar-Singh R, McWilliam P, Allamand Y Sharp EM and Humphries P. Autosomal Dominant retinitis pigmentosa: localization of a disease gene (RP6) to the short arm of chromosome 6. *Genomics*. 11.870-874, 1991.
  19. Kumar-Singh R, Jordan S, Farrar GJ and Humphries P. Poly (TIA) polymorphism at the human retinal degeneration slow (RDS) locus. *Nucl. Acids Res.*, 19.5800, 1991.
  20. Doak S, Jordan SA, McWilliam P and Humphries P. Tetranucleotide repeat polymorphism at the ACPP locus. *Nucl. Acids Res.*, 19, 4793. 1991.
  21. Farrar GJ, Kenna P, Jordan SA, Kumar-Singh R and Humphries P. A sequence polymorphism in the human peripherin/RDS gene. *Nucl. Acids Res.*, 19, 6982, 1991.
  22. Kumar-Singh R, Bradley DG, Farrar GJ, Lawler M, Jordan SA and Humphries P. Autosomal dominant retinitis pigmentosa: a new multiallelic marker (D35621) genetically linked to the disease locus (RP4). *Human Genetics*, 86, 502-504, 1991.
  23. Jordan SA, Farrar GJ, Kenna P, Kumar-Singh R, Humphries MM, Allamand V, Sharp EM and Humphries P. Autosomal dominant retinitis pigmentosa (adRP; RP6): Co-segregation of RP6 and the peripherin-RDS locus in a late onset family of Irish origin. *Am. J. Hum. Genet.*, 50, 634-639, 1992.
  24. Farrar GJ, Kenna P, Jordan SA, Kumar-Singh R, Humphries MM, Sharp EM, Sheils D and Humphries P. Autosomal dominant retinitis pigmentosa: A novel mutation at the peripherin/RDS locus in the original 6p-linked pedigree. *Genomics*, 14, 805-807, 1992.
  25. Jordan SA, Farrar GJ, Kenna P and Humphries P. Polymorphic variation within 'conserved' sequences at the 3' end of the human RDS gene which results in amino acid substitutions. *Human Mutation*, 1(3), 240-247, 1992.
  26. Humphries P, Kenna P and Farrar GJ. On the molecular genetics of retinitis pigmentosa. *Science*, 256, 804-808, 1992.
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  28. Inglehern CF, Lester DH, Bashir R, Atif U, Keen J, Sertedaki A, Linksey J, Jay M, Bird AC, Farrar GJ, Humphries P and Bhattacharya SS. Recombination between rhodopsin and locus D3S47 (C17) in rhodopsin linked retinitis pigmentosa families. *Am. J. Hum. Genet.*, 50, 590, 1992.
  29. Jordan SA, del Rio T, Soriano N, Garcia-Sandoval B, Kenna P, Ayuso C, Benitez I and Humphries P. Autosomal dominant retinitis pigmentosa (adRP): exclusion

- of a gene from three mapped loci provides evidence for the existence of a fourth locus. *Hum. Mol. Genet.*, 1(6), 411-415, 1992.
30. Bleeker-Wagemakers L, Bergen O, Gal A, Kumar-Singh R, Kenna P, Humphries P and Farrar GJ. Retinitis pigmentosa: exclusion of a locus from the long arm of chromosome 3 in the region of the rhodopsin gene provides evidence of genetic heterogeneity in the autosomal recessive form of the disease. *Genomics*, 14, 811-812, 1992.
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  33. Li Y Muller B, Fuhrmann C, van Nouhuys E, Horst L, Humphries P, Schwinger E and Gal A. Autosomal dominant familial exudative vitreoretinopathy locus maps on 11q and is closely linked to D11S533. *Am. J. Hum. Genet.*, 51, 749-754, 1992.
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  35. Kumar-Singh R, Wang H, Humphries P and Farrar GJ. Autosomal dominant retinitis pigmentosa: no evidence for non-allelic genetic heterogeneity on chromosome 3q. *Am. J. Hum. Genet.*, 52, 319-326, 1993.
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  39. Jordan SA, Farrar GJ, Kenna P, Humphries MM, Sheils D, Kumar-Singh R, Sharp EM, Soriano N, Ayuso C, Benitez J and Humphries P. Localisation of an autosomal dominant retinitis pigmentosa gene to chromosome 7q. *Nature Genetics*, 4, 54-57, 1993.
  40. Farrar GJ, Jordan SA, Kumar-Singh R, Inglehearn CF, Gal A, Gregory C, Al-Magthteh M, Kenna PF, Humphries MM, Sharp EM, Sheils D, Gunge S, Hargrave PA, Denton MJ, Schwinger E, Bhattacharya SS and Humphries P. Extensive Genetic Heterogeneity in Autosomal Dominant Retinitis Pigmentosa. *Retinal Degeneration*, Plenum Press, New York, 1993 (Eds. Hollyfield et al), 63-77, 1993.
  41. Kumar-Singh R, Farrar GJ, Mansergh F, Kenna P, Bhattacharya S, Gal A and Humphries P. 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). *Hum. Mol. Genet.*, 2, 875-878, 1993.
  42. Humphries P, Kenna P and Farrar GJ. Autosomal Dominant Retinitis Pigmentosa. *The Molecular Genetics of Inherited Eye Disorders: Modern Genetics 2* (AF Wright & B Jay. Eds), 153-172, 1994.
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  45. Mansergh FC, Jordan S, Farrar GJ, Kumar-Singh R and Humphries P. Three sequence polymorphisms in the phosphodiesterase (PDE) gene on chromosome 1q25-32.

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  52. Humphries MM, Rancourt D, Farrar GJ, Kenna P, Hazel M, Bush RA, Sieving PA, Sheils DM, McNally N, Creighton P, Erven A, Boros A, Gulya K, Capocchi MR and Humphries P. Retinopathy induced in mice by targeted disruption of the rhodopsin gene. *Nature Genetics*, 15, 216-219, 1997. News and views article by Gabriel Travis in same edition of *Nature Genetics*, pgs. 116-117.
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  57. al-Jandal N, Farrar GJ, Kiang AS, Humphries MM, Bannon N, Findlay JBC, Humphries P and Kenna PF. A Novel Mutation within the Rhodopsin Gene (Thr-94-Ile) Causing Autosomal Dominant Congenital Stationary Night Blindness. *Human Mutation*, 13,75-81, 1999.
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