BIOGRAPHICAL SKETCH

CURRICULUM VITAE:	Marian Humphries
OFFICE ADDRESS	Ocular Genetics Unit, Smurfit Institute of Genetics Trinity College Dublin
DEGREES	MSc, Trinity College Dublin 1993. Phd, University of Szeged, 2000
POSTS	Research Assistant, The Queen's University of Belfast, 1979-1983. Research Assistant, Trinity College Dublin, 1983-2000. Senior Research Fellow, Trinity College Dublin, 2000-to date

CURRENT MEMBERSHIPS

Irish Society of Human Genetics, American Society of Human Genetics, Association for Research in Vision and Ophthalmology (ARVO), Trinity College Neurosciences Institute (TCIN), All-Ireland Retinal Researchers Network, Irish Network of Neuronal Stem-cell Investigators, European Vision Institute, Panel Member of Scientific and Medical Advisory Board of Retina International.

RESEARCH INTERESTS

Major interests have included genetics of blistering skin disorders and hereditary retinal diseases.

PUBLICATIONS

1. Humphries P, Barton D, McKay AM, Humphries MM, Carritt B. Isolation of a polymorphic DNA segment unique to human chromosome 7 by molecular cloning of hybrid cell DNA. Mol. Gen. Genet., 190(1):143-9, 1983.
2. Humphries P, MacCabe AP, Spencer RA, Humphries MM, Pearson C. Structural and transcriptional properties associated with a member of a new family of conserved short dispersed repeated elements in human DNA. Gene, 39(2-3):255-61, 1985.
3. Humphries P, Russell SE, McWilliam P, McQuaid S, Pearson C, Humphries MM. Observations on the structure of two human 7SK pseudogenes and on homologous transcripts in vertebrate species. Biochem. J., 245(1):281-4, 1987.
4. 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. Cytogenet.. Cell Genet., 50, 181-187, 1989.
5. 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.
6. 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.
7. 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.
8. Humphries M, Nagayoshi T, Sheils D, Humphries P, Uitto J. Human nidogen gene: identification of multiple RFLP and exclusion as candidate gene in a family with epidermolysis bullosa (EBS2) with evidence for linkage to chromosome 1. J. Invest. Dermatol., 95(5):568-70, 1990.
9. Humphries MM, Sheils D, Lawler M, Farrar GJ, McWilliam P, Kenna P, Bradley DG, Sharp EM, Gaffney EF, Young M, Uitto J and Humphries P. Epidermolysis bullose: evidence for linkage to genetic markers on Chromosome 1 in a family with the autosomal dominant simplex form. Genomics, 7, 377-381, 1990.
10. 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.
11. 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.
12. 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..
13. 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.
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 locala1ization 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. 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.
18. 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.
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20. Humphries MM, Sheils DM, Jordan SA, Farrar GJ, Kumar-Singh R and Humphries P. Alu polymorphism in the human type I Keratin (KRT14) gene. Hum. Mol. Genet., 1 (6), 453, 1992.
21. Humphries MM, Sheils D, Young M, Farrar GJ and Humphries P. Genetic linkage analysis in a pedigree with autosomal dominant simplex epidermolysis Bullosa. J. Invest. Dermatol. 98 (3), 397, 1992.

22. Humphries MM, Sheils DM, Farrar GJ, Kumar-Singh R, Kenna PF, Mansergh FC, Jordan SA, Young M, Humphries P. A mutation (Met-->Arg) in the type I keratin (K14) gene responsible for autosomal dominant epidermolysis bullosa simplex. Hum. Mutat, 2(1):37-42, 1993.
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