Professor Chris Inglehearn; Short Curriculum Vitae, prepared 12/1/10

NAME: Christopher Francis Inglehearn

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DATE OF BIRTH: 25th March 1962

EDUCATION
BSc Biological Sciences (Genetics), Edinburgh University.
Second class (division one) Honours. July 1984.
PhD Molecular Genetics, Edinburgh University. June 1988.

CURRENT POST
Professor of Molecular Ophthalmology, School of Medicine, Leeds University. From February 2001; HEFCE funded permanent post, end date 31.3.2027
Section Head, Section of Ophthalmology and Neuroscience, Leeds Institute of Molecular Medicine

PREVIOUS POSTS
Senior Wellcome Fellow/Senior Lecturer, School of Medicine, Leeds University. October 1997 to January 2001.
Senior Wellcome Fellow, Institute of Ophthalmology, University College London. October 1992 to September 1997.

PROFESSIONAL MEMBERSHIPS
Chair, Candlelighters Children's Cancer Charity Scientific Advisory Board (current)
Yorkshire Eye Research Management Board (current)
Wellcome Trust Neuroscience Panel member October 2004 –July 2008
Guide Dogs for the Blind Ophthalmic Research Advisory Grp 2001–08
Association for Research into Vision and Ophthalmology (ARVO)
American Society of Human Genetics (ASHG)
British Society of Human Genetics (BSHG)

POSTGRADUATE SUPERVISION
Previously supervised 1 BMedSci student, 2 MSc Students and 11 PhD students to successful conclusion of studies. Currently cosupervise 10 PhD and 1 MD students.

EXAMINATIONS
Four first year transfer vivas, three MPhil vivas as internal, three PhD vivas as internal and nine PhD vivas as external

GRANTS REVIEWED
Grants reviewed for Wellcome Trust, Guide Dogs for the Blind, Candlelighters, MRC and many other grant bodies as external peer-reviewer

PAPERS REVIEWED
Papers reviewed in last two years for American Journal of Human Genetics, Human Molecular Genetics, Investigative Ophthalmology and Visual Sciences and others.

PUBLICATIONS:

1. **Inglehearn CF,** Papiha SS, Jay M, Wright AF, Moore AT and Bhattacharya SS (1990). Linkage of internal minisatellite loci on chromosome 1 and exclusion of autosomal dominant retinitis pigmentosa proximal to rhesus**. J Med Genet** 27:14-16

2. **Inglehearn CF**, Jay M, Lester DH, Bashir R, Jay B, Bird AC, Wright AF, Evans HJ, Papiha SS and Bhattacharya SS (1990). No evidence for linkage between late onset autosomal dominant retinitis pigmentosa and chromosome 3 locus D3S47 (C17): Evidence for genetic heterogeneity. **Genomic**s 6:168-173

3. **Inglehearn CF** and Cooke HJ (1990). A VNTR immediately adjacent to the human pseudoautosomal telomere. **Nucl Acids Res** 18:471-476

4. Bellamy RJ, **Inglehearn CF,** Lester DH, Hardcastle A and Bhattacharya SS (1990). Better fingerprinting with PCR. **Trends Genet** 6:32

5. Lester DH, **Inglehearn CF**, Bashir R, Ackford H, Esakowitz L, Jay M, Bird AC, Wright AF, Papiha SS and Bhattacharya SS (1990). Linkage to D3S47 (C17) in one large autosomal dominant retinitis pigmentosa family and exclusion in another: Confirmation of genetic heterogeneity. **Am J Hum Genet** 47:536-541

6. Farrar GJ, Kenna P, Redmond R, McWilliam P, Bradley PG, Humphries MM, Sharp EM, **Inglehearn CF**, Bashir R, Jay M, Watty A, Ludwig M, Schinzel A, Samanns C, Gal A, Bhattacharya S and Humphries P (1990). Autosomal dominant retinitis pigmentosa: Absence of the rhodopsin codon 23 Proline → Histidine substitution in pedigrees from Europe. **Am J Hum Genet** 47:941-945

7. Jay M and **Inglehearn CF** (1990). New strategies in molecular genetic studies of X-linked retinitis pigmentosa. **Eye** 4:737-742

8. **Inglehearn CF**, Bashir R, Lester DH, Jay M, Bird AC and Bhattacharya SS (1991). A three base pair deletion in the rhodopsin gene in a family with autosomal dominant retinitis pigmentosa. **Am J Hum Genet** 48:26-30.

9. Keen J, Lester DH, **Inglehearn CF**, Curtis A and Bhattacharya SS (1991). Rapid detection of single base mismatches as heteroduplexes on hydrolink gels. **Trends Genet** 7:5

10. Bhattacharya SS, Lester DH, Keen TJ, Bashir R, Lauffart B, **Inglehearn CF**, Jay M and Bird AC (1991). Retinitis pigmentosa and mutations in rhodopsin**. Lancet** 337:185

11. Bellamy RJ, **Inglehearn CF,** Jalili IK, Jeffreys AJ and Bhattacharya SS (1991). Increased band sharing in DNA fingerprints of an inbred human population. **Hum Genet** 87:341-347

12. Keen TJ, **Inglehearn CF**, Lester DH, Bashir R, Jay M, Bird AC, Jay B and Bhattacharya SS (1991) Autosomal dominant retinitis pigmentosa: four new mutations in rhodopsin, one of them in the retinal attachment site. **Genomics** 11:199-205

13. **Inglehearn CF,** Lester DH, Bashir R, Atif U, Keen TJ, Sertedaki A, Lindsey J, Jay M, Bird A, Farrar GJ, Humphries P and Bhattacharya SS (1991). Recombination between rhodopsin and locus D3S47 (C17) in Rhodopsin retinitis pigmentosa families. **Am J Hum Genet** 50:590-597

14. **Inglehearn CF**, Keen TJ, Bashir R, Jay M, Fitzke F, Bird AC, Crombie A and Bhattacharya SS (1991). A completed screen for mutations in the rhodopsin gene in a panel of patients with autosomal dominant retinitis pigmentosa. **Hum Mol Genet** 1:41-45

15. Moore AT, Fitzke FW, Kemp CH, Arden GB, Keen TJ, **Inglehearn CF**, Bhattacharya SS and Bird, AC (1992). Abnormal dark adaptation kinetics in autosomal dominant sector retinitis pigmentosa. **Brit J Ophthalmol** 76:465-469

16. Bashir R, **Inglehearn CF,** Keen TJ, Lindsay J, Atif U, Carter SA, Stephenson AM, Jackson A, Jay M, Bird AC, Papiha S and Bhattacharya SS (1992). Exclusion of chromosome 6 and 8 locations in non-rhodopsin autosomal dominant retinitis pigmentosa families: Further locus heterogeneity in adRP. **Genomics** 14:191-193

17. Wells J, Wroblewski J, Keen TJ, **Inglehearn CF**, Jubb C, Eckstein A, Jay M, Arden G, Bhattacharya SS, Fitzke F and Bird AC (1993). Mutations in the human retinal degeneration slow (rds) gene can cause either retinitis pigmentosa or macular dystrophy. **Nat Genet** 3:213-218

18. **Inglehearn CF**, Carter SA, Keen TJ, Lindsey J, Stephenson AM, Bashir R, Al-Maghtheh M, Moore AT, Jay M, Bird AC and Bhattacharya SS (1993). A new locus for autosomal dominant retinitis pigmentosa (adRP) on chromosome 7p. **Nat Genet** 4:51-53

19. **Inglehearn CF,** Farrar GJ, Denton M, Gal A, Humphries P and Bhattacharya, S.S. (1993). Evidence against a second autosomal dominant retinitis pigmentosa locus close to rhodopsin on chromosome 3q. **Am J Hum Genet** 53:536-537

20. Moore AT, Fitzke F, Jay M, Arden GB, **Inglehearn CF**, Keen TJ, Bhattacharya SS and Bird AC (1993). Autosomal dominant retinitis pigmentosa with apparent incomplete penetrance: a clinical, electrophysiological, psychophysical and molecular genetic study**. Brit J Ophthalmol** 77:473-479

21. Al-Maghtheh M, Gregory C, **Inglehearn CF** and Bhattacharya SS (1993). Rhodopsin mutations in autosomal dominant retinitis pigmentosa. **Human Mutation** 2:249-255

22. Wroblewski JJ, Wells J, Eckstein A, Fitzke F, Jubb C, Keen TJ, **Inglehearn CF,** Bhattacharya S, Arden GB, Jay M and Bird AC (1993). Macular dystrophy associated with mutations at codon 172 in the human retinal degeneration slow (rds) gene. **Ophthalmology** 101:12-22

23. Owens SL, Fitzke FW, Jay M, Keen TJ, **Inglehearn CF,** Bhattacharya SS and Bird AC (1994). Lys-296-Glu rhodopsin mutation at the retinal binding site: Ocular manifestations in autosomal dominant retinitis pigmentosa**. Br J Ophthalmol** 78:353-358

24. Wroblewski JJ, Wells JA, Eckstein A, Fitzke F, Jubb C, Keen TJ, **Inglehearn CF**, Bhattacharya SS, Arden GB, Jay M and Bird AC (1994). Ocular findings associated with a three-base-pair deletion in the peripherin-RDS gene in autosomal dominant Retinitis Pigmentosa**. Br J Ophthalmol** 78:381-386

25. Al-Maghtheh M, **Inglehearn CF**, Lunt P, Jay M, Bird AC and Bhattacharya SS (1994). Two new mutations at codons 40 and 216 in families with autosomal dominant Retinitis Pigmentosa. **Human Mutation** 3:409-410

26. Al-Maghtheh M, **Inglehearn CF,** Keen TJ, Evans KE, Moore AT, Jay M, Bird A and Bhattacharya SS (1994). Identification of a sixth locus for autosomal dominant Retinitis Pigmentosa on chromosome 19. **Hum Mol Genet** 3:351-354

27. **Inglehearn CF**, Keen TJ, Al-Maghtheh M, Gregory C, Jay M, Moore AT, Bird AC and Bhattacharya SS (1994). Further refinement of the location for autosomal dominant Retinitis Pigmentosa on chromosome 7p (RP9). **Am J Hum Genet** 54:675-680

28. Evans K, Fryer AF, **Inglehearn CF,** Duvall-Young J, Whittaker J, Gregory CY, Ebenezer N, Hunt D and Bhattacharya SS (1994). Genetic linkage of cone-rod dystrophy to chromosome 19q and evidence for segregation distortion. **Nat Genet** 6:210-213

29. Keen TJ, **Inglehearn CF**, Kim RC, Bird AC and Bhattacharya SS (1994). Retinal Pattern Dystrophy caused by a 4 bp insertion at codon 140 in the Rds-peripherin gene. **Hum Mol Genet** 3:367-368

30. Al-Maghtheh M, Kim R, Hardcastle A, **Inglehearn CF** and Bhattacharya SS (1994). A 150 bp insertion in the rhodopsin gene of an autosomal dominant Retinitis Pigmentosa family**. Hum Mol Genet** 3:205-206

31. **Inglehearn CF**, Keen TJ, Al-Maghtheh M and Bhattacharya SS (1994). Loci for autosomal dominant Retinitis Pigmentosa and Dominant Cystoid Macular Dystrophy on chromosome 7p are not allelic. **Am J Hum Genet** 55:581-582

32. Kim RY, Fitzke FW, Moore AT, Jay M, **Inglehearn CF**, Arden GB, Bhattacharya SS and Bird AC (1994). Autosomal dominant Retinitis Pigmentosa mapping to chromosome 7p exhibits variable expression. **Brit J Ophthalmol** 79:23-27

33. Keen TJ, **Inglehearn CF**, Patel RJ, Green ED, Peluso DC and Bhattacharya SS (1995). Localisation of the aquaporin 1 (AQP1) gene within a YAC contig containing the polymorphic markers D7S632 and D7S526. **Genomics** 25:599-600

34. Bardien S, Ebenezer N, Greenberg J, **Inglehearn CF**, Bartmann L, Goliath R, Beighton P, Ramesar R and Bhattacharya SS (1995). An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q. **Hum Mol Genet** 4:1459-1462

35. Keen TJ, **Inglehearn CF**, Green ED, Cunningham AF, Patel RJ, Peacock RE, Gerken S, White R, Weissenbach J and Bhattacharya SS (1995). A YAC contig spanning the dominant retinitis pigmentosa locus (RP9) on chromosome 7p**. Genomics** 28:383-388

36. Toma NM, Ebenezer ND, **Inglehearn CF**, Plant C, Ficker LA and Bhattacharya SS (1995). Linkage of congenital hereditary endothelial dystrophy to chromosome 20. **Hum Mol Genet** 4:2395-2398

37. Evans K, Al-Maghtheh M, Fitzke FW, Moore AT, Jay M, **Inglehearn CF,** Arden GB and Bird AC (1995). Bimodal expressivity in dominant retinitis pigmentosa genetically linked to chromosome 19q. **Br J Ophthalmology** 79:841-846

38. Patel RJ, Keen TJ, Grzeschik K-H, Nierman WC, Hayes P, Bhattacharya SS and **Inglehearn CF** (1995). Regional assignment of thirty expressed sequence tags (ESTs) on human chromosome 7 using a somatic cell hybrid panel**. Genomics** 30:112-114

39. **Inglehearn CF** and Hardcastle AJ (1996). Nomenclature for inherited diseases of the retina. **Am J Hum Genet** 58:433-435

40. Tarttelin EE, Al-Maghtheh M, Keen TJ, Bhattacharya SS and **Inglehearn CF** (1996). Simple tests for rhodopsin involvement in Retinitis pigmentosa. **J Med Genet** 33:262-263

41. Tarttelin EE, Plant C, Weissenbach J, Bird AC, Bhattacharya SS and **Inglehearn CF** (1996). A new family linked to the RP13 locus for autosomal dominant retinitis pigmentosa on distal 17p. **J Med Genet** 33:518-520

42. Keen TJ and **Inglehearn CF** (1996). Mutations and polymorphisms in the human peripherin/Rds gene and their involvement in inherited retinal degeneration. **Human Mutation** 8:297-303

43. Reynolds PA, Powlesland RM, Keen TJ, **Inglehearn CF,** Henthorn K, Green ED and Brown KW (1996). Localisation of a novel t(1;7) translocation associated with Wilm's tumor predisposition and skeletal abnormalities. **Genes Chromosomes and Cancer** 17:151-155

44. Al-Maghtheh M, Vithana E, Tarttelin EE, Jay M, Evans K, Moore T, Bhattacharya SS and **Inglehearn CF** (1996). Evidence for a major retinitis pigmentosa locus on 19q 13.4 (RP11) and association with a unique bimodal expressivity phenotype. **Am J Hum Genet** 59:864-871

45. Keen TJ, Morris AG and **Inglehearn CF** (1997). Exclusion of CAG repeat expansion as the cause of disease in autosomal dominant Retinitis pigmentosa families. **J Med Genet** 34:130-132

46. **Inglehearn CF** and Gregory CY (1997). Meiotic drive at the myotonic dystrophy and cone-rod dystrophy loci on chromosome 19q13.3. **Am J Hum Genet** 60:1562-1563

47. **Inglehearn CF** (1997). Intelligent linkage analysis using gene density estimates. **Nat Genet** 16:15

48. **Inglehearn CF** (1997). Nightmare antibiotics. **Nature** 388:416

48. Peacock RE Keen TJ and **Inglehearn CF** (1997). Analysis of a Human Gene homologous to Rat Ventral Prostate 1 protein**. Genomics** 46:443-449

49. **Inglehearn CF**, Tarttelin EE, Plant C, Peacock RE, Al-Maghtheh M, Keen TJ, Vithana E, Bird AC and Bhattacharya SS (1998). A linkage survey of twenty dominant Retinitis pigmentosa families: Frequencies of the nine known loci and evidence for further heterogeneity. **J Med Genet** 35:1-5

50. Vithana EN, Al-Maghtheh M, Bhattacharya SS and **Inglehearn CF** (1998). RP11 is the second

most common locus for dominant Retinitis Pigmentosa. **J Med Genet** 35:174-175

51. Al-Maghtheh M, Vithana EN, **Inglehearn CF,** Bird AC and Bhattacharya SS (1998). Segregation of a PRKCG mutation in two RP11 families. **Am J Hum Genet** 62:1248-1252

52. **Inglehearn CF,** Tarttelin EE, Keen TJ, Bhattacharya SS, Moore AT, Taylor R and Bird AC (1998). A new dominant retinitis pigmentosa family mapping to the RP18 locus on chromosome 1q11-21. **J Med Genet** 35:788-789

53. **Inglehearn CF** (1998). Lod scores, location scores and X-linked cone dystrophy. **Am J Hum Genet** 63:900-901

54. **Inglehearn CF** (1998). Molecular Genetics of Human Retinal Dystrophies. **Eye** 12:571-579

55. **Inglehearn CF**, McHale JC, Keen TJ, Skirton HS and Lunt P (1999). A new family linked to the RP1 dominant Retinitis pigmentosa locus on 8q. **J Med Genet** 36:646-648

56. Sullivan LS, Heckenlively JR, Bowne SJ, Zuo J, Hide WA, Gal A, Denton M, **Inglehearn CF**, Blanton, SH and Daiger SP (1999). Mutations in a novel retina-specific gene cause autosomal dominant retinitis pigmentosa. **Nat Genet** 22:255-259

57. Bowne SJ, Daiger SP, Hims MW, Sohocki MM, Malone KA, McKie AB, Heckenlively JR, Birch DR, **Inglehearn CF**, Bhattacharya SS, Bird AC and Sullivan LS (1999). Mutations in the RP1 gene causing autosomal dominant retinitis pigmentosa. **Hum Mol Genet** 8:2121-2128

58**.** Bamashmus MA, Downey LM, **Inglehearn CF,** Gupta SR and Mansfield DC (2000). Genetic heterogeneity in familial exudative vitreoretinopathy; clinical investigation and exclusion of the EVR1 locus on chromosome 11q in a large autosomal dominant pedigree. **Brit J Ophthalmol** 84:358-363 (Featured on front cover of Journal).

59. Jackson A and **Inglehearn CF** (1999). This should not be the end for terminator technology in GM crops. **Nature** 402;457

60. McHale JC, McKie AB, Tarttelin EE and **Inglehearn CF** (2000). Expression map of the chromosome 17p13.3 region spanning the RP13 dominant Retinitis pigmentosa locus, the Miller-Dieker Lissencephaly Syndrome (MDLS) region and a putative tumour suppresser locus. **Cytogenet Cell Genet** 88;225-229

61. Murton NJ, Rehman I, Black GCM, **Inglehearn CF** and Churchill AJ (2000). A novel deletion (IVS11+3del4) identified in the human PAX-6 gene in a patient with aniridia. **Human Mutation** 15;582

62 Murton NJ, French L, Toomes C, Joseph SS, Rehman I, Hopkins BL, **Inglehearn CF** and Churchill AJ (2000). A high-density transcript map of the human dominant optic atrophy locus OPA1 and re-evaluation of evidence for a founder haplotype. **Cytogenet Cell Genet** 92:97-102

63. Mohamed MD, McKibbin MA, Jafri H, Rashid Y, Woods CG and **Inglehearn CF** (2000). A new pedigree with recessive CHED mapping to the CHED2 locus on 20p13. **Brit J Ophthalmol** 85:758-59

64. Downey LM, Keen TJ, Roberts E, Mansfield DC, Bamashmus M and **Inglehearn CF** (2001). A new locus for autosomal dominant Familial Exudative Vitreoretinopathy (EVR3) maps to chromosome 11p12-13. **Am J Hum Genet** 68:778 -781

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66. **Inglehearn CF** (2001). Genetic testing: a research perspective**. British Society for Human Genetics Newsletter** 18:3

67. McKie AB, McHale JC, Keen TJ, Tarttelin EE, Goliath R, van Lith-Verhoeven JJC, Greenberg J, Ramesar RS, Hoyng CB, Cremers FPM, Mackey DA, Bhattacharya SS, Bird AC, Markham AF and **Inglehearn CF** (2001). Mutations in the pre-mRNA splicing factor gene PRPC8 cause autosomal dominant Retinitis pigmentosa (RP13). **Hum Mol Genet** 10:1555-1562

68. Toomes C, Murton NJ, Mackey DA, Craig J, Newbury-Ecob R, Bennett CP, Vize CJ, Desai SP, Black GCM, Patel N, Teimory M, Markham AF, **Inglehearn CF** and Churchill AJ (2001). Spectrum, frequency and penetrance of OPA1 mutations in dominant optic atrophy. **Hum Mol Genet** 10:1369-1378

69. Downey LM, Keen TJ, Jalili IK, McHale JC, Mighell A, Fayle S, Aldred M Wissinger B and **Inglehearn CF** (2002). Identification of a recessive locus on chromosome 2q11 at which Amelogenesis Imperfecta and Cone-rod dystrophy co-segregate. **Eur J Hum Genet** 10:865-869

70. Chakarova CF, Hims MM (joint 1st), Bolz H, Abu-Safien L, Patel RJ, Papaioannou M**, Inglehearn CF,** Keen TJ, Willis C, Moore AT, Rosenberg T, Webster AR, Bird AC, Gal A, Hunt D, Vithana EN and Bhattacharya SS (2002). Mutations in HPRP3, a third member of the pre-mRNA splicing factor genes, implicated in autosomal dominant retinitis pigmentosa. **Hum Mol Genet** 11:87-92

71. Patel N, Churchill A, Toomes C, Marchbank N, **Inglehearn CF**, Foulds N, Moosavi A, Teimory M. Importance of molecular testing in dominant optic atrophy (2002). **Br J Ophthalmol** 86:1314-5

72. Marchbank NJ, Craig JE, Leek JP, Toohey M, Churchill AJ**,** Markham AF, Mackey DA, Toomes C and **Inglehearn CF** (2002). Deletion of the *OPA1* gene in a dominant optic atrophy family: evidence that haploinsufficiency is the cause of disease. **J Med Genet** 39:e47

73. Keen TJ, Hims MM, McKie AB, Moore AT, Doran RM, Mackey DA, Mansfield DC, Mueller RF, Bhattacharya SS, Bird AC, Markham AF and **Inglehearn CF** (2002). Mutations in a protein target of the Pim-1 kinase associated with the RP9 form of autosomal dominant Retinitis pigmentosa. **Eur J Hum Genet** 10:245-249

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75. Mohamed MD, Topping NC, Jafri H, Rashid Y, McKibbin MA and **Inglehearn CF**. (2003). Progession of phenotype in Leber's congenital amaurosis with a mutation at the LCA5 locus. **Br J Ophthalmol** 87:473-475

76. Keen TJ, Mohamed M, McKibbin M, Rashid Y, Jafri H**,** Maumenee IH and **Inglehearn CF** (2003). Identification of a new locus (LCA9) for autosomal recessive Leber's congenital amaurosis. **Eur J Hum Genet** 11;420-423

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79. Powell BL, Toomes C, Yeung A, Scott S, Marchbank NJ, Spry P, Lumb R, **Inglehearn CF** and Churchill AJ (2003). Polymorphisms in *OPA1* are associated with normal tension glaucoma. **Mol Vis** 9:460-464

80. Ponchel F, Toomes C, Douglas SH, Combaret V, Puisieux A, Bransfield K, Bell SM, Robinson PA, **Inglehearn CF,** Markham AF and Isaacs JD (2003). Real-time PCR based on SYBR-green fluorescence: An alternative to the TaqMan assay for the quantification of gene rearrangements, gene amplifications and micro gene deletions. **BMC Biotechnol** 3:18

81. Toomes C, Downey LM, Bottomley HM, Scott S, Woodruff G, Trembath RC and **Inglehearn CF** (2003). Identification of a fourth locus (EVR4) for familial exudative vitreoretinopathy (FEVR). **Mol Vis** 10:37-42.

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Black GCM, Markham AFM, Downey LM and **Inglehearn CF** (2003). Mutations in *LRP5* or *FZD4* underlie the common FEVR locus on chromosome 11q13. **Am J Hum Genet** 74:721-730.

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84. Pal B, Mohamed MD, Keen TJ, Williams G, Jacob A, Bradbury J, Sheridan E and **Inglehearn CF** (2003). Recessively inherited Foveal Hypoplasia and Anterior Segment Dysgenesis; a new locus on chromosome 16q23-24**. J Med Genet.** 41:772-7

85. Toomes C, Downey LM, Bottonley HM, Mintz-Hittner H and **Inglehearn CF** (2003). Further evidence of genetic heterogeneity in familial exudative vitreoretinopathy; exclusion of EVR1, EVR3 and EVR4 in a large autosomal dominant pedigree**. Brit J Ophthalmol.** 89:194-197.

86. Maita H, Kitaura H, Keen TJ, **Inglehearn CF,** Ariga H and Iguchi-Ariga SM (2004). PAP-1, the mutated gene underlying the RP9 form of dominant Retinitis pigmentosa, is a splicing factor. Exp Cell Res 300:283-96

87. Bottomley HM, Downey LM, **Inglehearn CF**, Toomes C (2006). Comment on 'cosegregation of two unlinked mutant alleles in some cases of autosomal dominant familial exudative vitreoretinopathy' Eur J Hum Genet. 14:6-7

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89. Downey LM, Bottomley HM, Sheridan E, Ahmed M, Gilmour DF, **Inglehearn CF**, Reddy A, Agrawal A, Bradbury J, Toomes C (2006). Reduced bone mineral density and hyaloid vasculature remnants in a consanguineous recessive FEVR family with a mutation in LRP5. **Br J Ophthalmol**. 90:1163-1167

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93. Majava M, Bishop PN, Hägg P, Scott PG, Rice A, **Inglehearn C**, Hammond CJ, Spector TD, Ala-Kokko L, Männikkö M. Novel mutations in the small leucine-rich repeat protein/proteoglycan (SLRP) genes in high myopia. **Hum Mutat.** 2007 Apr;28(4):336-44

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