CURRICULUM VITAE

VERONICA VAN HEYNINGEN 12 November 1946

Current details:
MRC Human Genetics Unit, Western General Hospital, Edinburgh EH4 2XU
Tel: +44 131 467 8405; FAX: +44 131 467 8456; email: v.vanheyningen@hgu.mrc.ac.uk

EDUCATION
1968 BA (Cantab) Natural Sciences, (Genetics) 2.1;
1970 MS (Northwestern, Illinois);
1973 DPhil (Oxon) with Walter Bodmer on "Mitochondrial enzymes in somatic cell hybrids"

POSTS HELD

1973-76	*Beit Memorial Fellow:* Oxford, Genetics Laboratory,1973-4; Edinburgh, MRC Mammalian Genome Unit, 1974-6
MRC Human Genetics Unit, Edinburgh:	
1977-1981	MRC Postdoctoral Scientist
1981	MRC Appointment of unlimited tenure
1986	MRC Appointment to Senior Scientist grade
1991	MRC Special Appointment grade - band 1
1992-	Head of Cell and Molecular Genetics Section (now renamed Medical Genetics)

HONOURS
1993-7 Howard Hughes International Scholar - awarded for five years for work on "Molecular, genetic and functional studies on PAX6 and other aniridia-associated genes in the WAGR region"
1994-0 Trustee of National Museums of Scotland
1995 Honorary Professor, Faculty of Medicine, University of Edinburgh
1997 Fellow of the Royal Society of Edinburgh
1999 Fellow Academy of Medical Sciences
1999 Member Scottish Executive Science Strategy Review Group
2000-5 Member UK Government's Human Genetics Commission, chair of Horizon-scanning subgroup
2002 Member of EMBO
2003 Fellowship Japan Society for the Promotion of Science – to visit Japan for 3 week lecture tour
2003-4 President, European Society of Human Genetics
2006 ESHG Award – presented at ESHG 2006 meeting in Amsterdam
2007 Elected Fellow of the Royal Society
2008 Henry Dryerre Prize Lectureship, Royal Society of Edinburgh

COMMITTEES
1992- Senior GDB Editor For Chromosome 11, Organiser, 1994 Chromosome 11 Workshop
1994-7 Treasurer of Genetical Society and running Membership Office with a full time Membership Secretary
1990- Member MRC advisory committee to the European Collection Of Animal Cell Cultures
1990-4 Member Research and Medical Advisory Committee to Cystic Fibrosis Trust (UK)
1994-7 Member MRC Molecular Cell and Medicine Board Grant Committee B
1994 Member of US DOE Visiting Committee to assess work and funding for the Genome Database, Baltimore
1994 Member Human Genome Mapping Committee of HUGO
1995/6 Co-Chair/Chair, Human Genome Mapping Committee of HUGO
1995-7 Member Scientific Organising Committee, HUGO Human Genome Mapping Conferences
1996-9 Member, Conseil Scientifique de l'IFREM (Institut Fédératif de Recherche des Enfants Malades), Paris
1996-8 Member, Medical Research Committee, Muscular Dystrophy Group
1997 Member Sir Henry Wellcome Commemorative Awards Panel, Wellcome Trust
1997-0 Member International Interest Group, Wellcome Trust
1997- Member Scientific Programme Committee, European Society for Human Genetics
1998 Member Peer Review Committee at ICRF
1999 Member of Scientific Programme Committee, British Society for Human Genetics

1999 Member Steering Committee at HUGO EU FP5 advisory meeting on genome research
1999-03 Member MRC Physiology Medicine and Infections Board
1999- Member Caledonian Research Foundation Board of Governors
1999 Member site visit committee to TIGEM laboratory of human genetics in Milan
2000-1 Chair local organising committee for HGM2001
2000- MRC Advisory Committee on Scientific Advances Genetics
2000- Member of Telethon Scientific Advisory Board
2001 Berlin Site visit for assessment of large joint DFG application "Molecular Basis of Clinical Variability in Mendelian Disorders"
2003 Member of review board for Institute for Clinical Genetics of the University of Zurich
2003 Scientific Organising Committee for International Congress of Human Genetics, Brisbane 2006
2004 Berlin Site visit for re-assessment of joint DFG MPI project "Molecular Basis of Clinical Variability in Mendelian Disorders"
2004 Genome Canada International Science Review Board
2004 Member of expert group to review genetics services in Scotland
2004 Scientific Advisory Board, EU BioSapiens Network of Excellence, European Virtual Institute for Genome Annotation
2005 Member of grant panel, Academy of Finland, Helsinki
2005 External assessor pre-RAE exercise Institute of Genetics, University of Newcastle
2006 Member HUGO Council
2006 Member Council of Academy of Medical Sciences
2008 ERC Grant panel

Selected publications

1. HILL R E, FAVOR J, HOGAN B L M, TON C C T, SAUNDERS G F, HANSON I M, PROSSER J, JORDAN T, HASTIE N D and VAN HEYNINGEN V, Mouse *Small eye* results from mutations in a paired-like homeobox-containing gene. *Nature* **354,** 522-525, 1991.
2. TON C C T, HIRVONEN H, MIWA H, WEIL M M, MONAGHAN P, JORDAN T, VAN HEYNINGEN V, HASTIE N D, MEIJERS-HEIJBOER H, DRECHSLER M, ROYER-POKORA B, COLLINS F, SWAROOP A, STRONG L C and SAUNDERS G F, Positional cloning and characterization of a paired box- and homeobox- containing gene from the aniridia region. *Cell* **67,** 1059-1074, 1991
3. JORDAN T, HANSON I, ZALETAYEV D, HODGSON S, PROSSER J, SEAWRIGHT A, HASTIE N and VAN HEYNINGEN V, The human PAX6 gene is mutated in two patients with aniridia. *Nature Genet* **1,** 328-332, 1992
4. HANSON IM, SEAWRIGHT A, HARDMAN K, HODGSON S, ZALETAYEV D, FEKETE G and VAN HEYNINGEN V. PAX6 mutations in aniridia. *Human Molecular Genetics* **2,** 915-920, 1993
5. HANSON IM, FLETCHER JM, JORDAN T, BROWN A, TAYLOR D, ADAMS RJ, PUNNETT HH, and VAN HEYNINGEN V. Mutations at the PAX6 locus found in heterogeneous anterior segment malformations including Peters' anomaly. *Nature Genetics,* **6,** 168-173, 1994
6. FANTES J, REDEKER B, BREEN M, BOYLE S, BROWN J, FLETCHER J, JONES S, BICKMORE W, FUKUSHIMA Y, MANNENS M, DANES S, VAN HEYNINGEN V and HANSON IM Aniridia-associated cytogenetic rearrangements suggest that a position effect may cause the mutant phenotype. *Human Molecular Genetics* **4:** 415-422 1995
7. HANSON IM and VAN HEYNINGEN V PAX6: more than meets the eye. *Trends in Genetics* 11: 268-272 1995
8. SCHEDL A, ROSS A, LEE M, ENGELKAMP D, RASHBASS P, VAN HEYNINGEN V and HASTIE ND. Influence of PAX6 gene dosage on development: Overexpression causes severe eye abnormalities. *Cell* **86:** 71-82, 1996
9. ERICSON J, RASHBASS P, SCHEDL A, BRENNER-MORTON S, KAWAKAMI A, VAN HEYNINGEN V, JESSELL TM and BRISCOE J. Pax6 controls progenitor cell identity and neuronal fate in response to graded Shh signalling. *Cell* **90:** 169-180, 1997
10. KLEINJAN DA and VAN HEYNINGEN V. Position effect in human genetic disease. *Hum Mol Genet* **7:** 1611-1617, 1998
11. PROSSER J AND VAN HEYNINGEN V. PAX6 mutations reviewed. *Human Mutation* **11:** 93-108, 1998
12. MILES C, ELGAR G, COLES E, KLEINJAN DJ, VAN HEYNINGEN V and HASTIE ND. Complete sequencing of the Fugu WAGR region from WT1 to PAX6 - dramatic compaction and conservation of synteny with human chromosome 11p13. *Proc Natl Acad Sci US* **95**: 13068-13072, 1998
13. HANSON IM, CHURCHILL A, LOVE J, AXTON R, MOORE A, CLARKE M, MEIRE F and VAN HEYNINGEN V. Missense mutations in the most ancient residues of the PAX6 paired domain underlie a spectrum of human congenital eye malformations. *Hum Mol Genet* **8:** 165-172, 1999
14. ENGELKAMP D, RASHBASS P, SEAWRIGHT A and VAN HEYNINGEN V. Role of *Pax6* in development of the cerebellar system. *Development* **126:** 3585-3596, 1999
15. SISODIYA SM, FREE SL, WILLIAMSON KA, MITCHELL TH, WILLIS C, STEVENS JM, KENDALL BE, SHORVON SD, HANSON I, MOORE AT and VAN HEYNINGEN V. *PAX6* haploinsufficiency causes cerebral malformation and olfactory dysfunction in humans. *Nature Genetics* **28:** 214-216, 2001
16. KLEINJAN DA, SEAWRIGHT A, SCHEDL A, QUINLAN RA, DANES S and VAN HEYNINGEN V. Aniridia-associated translocations, DNase hypersensitivity, sequence comparison, and transgenic analysis redefine the functional domain of PAX6. *Hum Mol Genet* **10:** 2049-2059, 2001
17. MORRISON D, FITZPATRICK D, HANSON I, WILLIAMSON K, VAN HEYNINGEN V, FLECK B, JONES I, CHALMERS J and CAMPBELL H. National study of microphthalmia, anophthalmia and coloboma (MAC) in Scotland: investigation of genetic aetiology *J Med Genet* **39:** 16-22, 2002
18. JAMIESON RV, PERVEEN R, KERR B, CARETTE M, YARDLEY J, HEON E, WIRTH MG, VAN HEYNINGEN V, DONNAI D, MUNIER F & BLACK GCM. Domain disruption and mutation of the bZIP transcription factor, *MAF*, associated with cataract, ocular anterior segment dysgenesis and coloboma. *Hum Mol Genet* **11:** 33-42, 2002

19. ESTIVILL-TORRUS G, PEARSON H, VAN HEYNINGEN V, PRICE D and RASHBASS P. Pax6 is required to regulate the cell cycle and the rate of progression from symmetrical to asymmetrical division in mammalian cortical progrenitors. *Development* **129:** 455-466, 2002
20. GRIFFIN C, KLEINJAN DA, DOE B and VAN HEYNINGEN V. New 3' elements control Pax6 expression in the developing pretectum, neural retina, and olfactory region. *Mechanisms of Development* **112:** 89-100, 2002
21. KLEINJAN DA, SEAWRIGHT A, ELGAR G and VAN HEYNINGEN V. Characterisation of a novel gene adjacent to PAX6, revealing synteny conservation with functional significance. *Mammalian Genome* **13:** 102-107, 2002
22. VAN HEYNINGEN V & WILLIAMSON KA. *PAX6* in sensory development. *Hum Mol Genet* **11:** 1161-1167 2002
23. CROLLA JA and VAN HEYNINGEN V. Frequent chromosome aberrations revealed by molecular cytogenetic studies in referred aniridia cases. *Am J Hum Genet* **71:** 1138-1149, 2002
24. FANTES J, RAGGE NK, LYNCH SA, MCGILL N, COLLIN JRO, HOWARD-PEEBLES PN, HAYWARD C, VIVIAN AJ, WILLIAMSON K, VAN HEYNINGEN V and FITZPATRICK DR Mutations in SOX2 cause human anophthalmia *Nat Genet* **33:** 461-463, 2003
25. MITCHELL TN, FREE SL, WILLIAMSON KA, STEVENS JM, CHURCHILL AJ, HANSON IM, SHORVON SD, MOORE AT, VAN HEYNINGEN V, SISODIYA SM. Polymicrogyria and absence of pineal gland due to *PAX6* mutation. *Annals of Neurology* **53**: 658-663, 2003
26. KLEINJAN DA, SEAWRIGHT A, CHILDS A and VAN HEYNINGEN V. Conserved elements in *Pax6* intron 7 involved in (auto)regulation and alternative transcription. *Dev Biol* **265:** 462-477, 2004
27. BAMIOU DE, MUSIEK FE, SISODIYA SM, FREE SL, DAVIES RA, MOORE AT, VAN HEYNINGEN V and LUXON LM. Deficient auditory interhemispheric transfer in patients with *PAX6* mutations. *Annals of Neurology,* in press
28. VAN HEYNINGEN V and YEYATI PL. Mechanisms of non-Mendelian inheritance in genetic disease. *Hum Mol Genet* **13:** R225-R333, 2004
29. KLEINJAN DA and VAN HEYNINGEN V. Long-range control of gene expression: emerging mechanisms and disruption in disease. *Am J Hum Genet* **76:** 8-32, 2005
30. RAGGE NK, LORENZ B, SCHNEIDER A, BUSHBY K, DE SANCTIS L, DE SANCTIS U, SALT A, COLLIN JRO, VIVIAN AJ, FREE SL, THOMPSON P, WILLIAMSON KA, SISODIYA SM, VAN HEYNINGEN V and FITZPATRICK DR. SOX2 anophthalmia syndrome. *Am.J.Med.Genet. A*, **135:** 1-7, 2005
31. RAGGE NK, BROWN AG, POLOSCHEK CM, LORENZ B, HENDERSON RA, CLARKE MP, RUSSELL-EGGITT I, FIELDER A, GERRELLI D, MARTINEZ-BARBERA JP, RUDDLE P, HURST J, COLLIN JRO, SALT A, COOPER ST, THOMPSON PJ, SISODIYA SM, WILLIAMSON KA, FITZPATRICK DR, VAN HEYNINGEN V, HANSON IM. Heterozygous mutations of OTX2 cause severe but variable ocular malformations. *Am J Hum Genet* **76:** 1008-1022, 2005
32. FAIVRE L, WILLIAMSON KA, FABER V, LAURENT N, GRIMALDI M, THAUVIN-ROBINET C, DURAND C, MUGNERET F, GOUYON JB, BRON A, HUET F, HAYWARD C, VAN HEYNINGEN V, FITZPATRICK DR. Recurrence of SOX2 anophthalmia syndrome with gonosomal mosaicism in a phenotypically normal mother. *Am J Med Genet A*. **140A:** 636-639, 2006
33. SISODIYA SM, RAGGE NK, CAVALLERI GL, HEVER A, LORENZ B, SCHNEIDER A, WILLIAMSON KA, STEVENS JM, FREE SL, THOMPSON PJ, VAN HEYNINGEN V, FITZPATRICK DR. Role of SOX2 mutations in human hippocampal malformations and epilepsy. *Epilepsia* **47:** 534-542, 2006
34. WILLIAMSON KA, HEVER AM, RAINGER J, ROGERS RC, MAGEE A, FIEDLER Z, KENG WT, SHARKEY FH, MCGILL N, HILL CJ, SCHNEIDER A, MESSINA M, TURNPENNY PD, FANTES JA, VAN HEYNINGEN V, FITZPATRICK DR. Mutations in SOX2 Cause Anophthalmia-Esophageal-Genital (AEG) Syndrome. *Hum Mol Genet.* **15:** 1413-1422, 2006
35. HEVER AM, WILLIAMSON KA and VAN HEYNINGEN V. Developmental malformations of th eye: the role of *PAX6*, *SOX2* and *OTX2*. *Clin Genet.* **69:** 459-470, 2006
36. KLEINJAN DA, SEAWRIGHT A, MELLA S, CARR CB, TYAS DA, SIMPSON TI, MASON JO, PRICE DJ, VAN HEYNINGEN V. Long-range downstream enhancers are essential for Pax6 expression. *Dev Biol.* **299:** 563-581, 2006
37. MANUEL M, GEORGALA PA. CARR CB, CHANAS S, KLEINJAN DA, MARTYNOGA B, MASON JO, MOLINEK M, PINSON J, PRATT T, QUINN JC, T. SIMPSON I, TYAS DA, VAN HEYNINGEN V, WEST JD, PRICE DJ. Controlled overexpression of Pax6 in vivo negatively auto-regulates the *Pax6* locus, causing cell-autonomous defects of late cortical progenitor proliferation with little effect on cortical arealization. *Development* **134:** 545-555, 2007
38. YEYATI PL, BANCEWICZ RM, MAULE J and VAN HEYNINGEN V. HSP90 selectively modulates phenotype in vertebrate development. *PLoS Genetics,* **3:** e43, 2007
39. HENDERSON A, WILLIAMSON K, CUMMING S, CLARKE M, LYNCH SA, HANSON IM, FITZPATRICK D, SISODIYA S, VAN HEYNINGEN V Inherited *PAX6*, *NF1* and *OTX2* mutations in a child with microphthalmia and aniridia *Eur J Hum Genet* **15:** 898-901, 2007
40. BAMIOU DE, CAMPBELL NG, MUSIEK FE, TAYLOR R, CHONG WK, MOORE A, VAN HEYNINGEN V, FREE S, SISODIYA S, LUXON LM Auditory and verbal working memory deficits in a child with congenital aniridia due to a PAX6 mutation. *Int J of Audiol* **46:** 196-202, 2007
41. BAMIOU DE, FREE SL, SISODIYA SM, CHONG WK, MUSIEK F, WILLIAMSON KA, VAN HEYNINGEN V, MOORE AT, GADIAN D, LUXON LM. Auditory interhemispheric transfer deficits, hearing difficulties and brain MRI abnormalities in children with congenital aniridia due to a PAX6 mutation. *Arch Pediatr Adolesc Med.* **161:** 463-469, 2007
42. ROBINSON DO, HOWARTH RJ, WILLIAMSON KA, VAN HEYNINGEN V, BEAL SJ and CROLLA JA. Genetic analysis of Chromosome 11p13 and the PAX6 gene in a series of 125 cases referred with aniridia. *Am J Med Genet* in press, 2008
43. KLEINJAN DA, BANCEWICZ RM, GAUTIER P, DAHM R, SCHONTHALER HB, DAMANTE G, SEAWRIGHT A, HEVER AM, YEYATI PL, VAN HEYNINGEN V and COUTINHO P. Subfunctionalisation of duplicated zebrafish *pax6* genes by cis-regulatory divergence. *PLoS Genetics*,**4:** e29, 2008
44. FANTES JA, BOLAND E, RAMSAY J, DONNAI D, SPLITT M, GOODSHIP JA, STEWART H, WHITEFORD M, GAUTIER P, HAREWOOD L, HOLLOWAY S, SHARKEY F, MAHER E, VAN HEYNINGEN V, CLAYTON-SMITH J, FITZPATRICK DR, BLACK GC. FISH Mapping of de novo Apparently Balanced Chromosome Rearrangements Identifies Characteristics Associated with Phenotypic Abnormality. *Am J Hum Genet.* **82:** 916-926, 2008