Curriculum vitae

Leslie, Jacqueline (Jacquie) Greenberg

1.1 Personal

Full name	Leslie, Jacqueline, Harriett, Leviticus, Greenberg
Date of birth	22 January, 1950
Citizenship	South African
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1.2 Academic and Professional Qualifications/Experience

B.Sc.	University of Stellenbosch (Physiology & Chemistry)	1970
Ph.D.	University of Cape Town (Human Genetics)	1990
Registered as a Medical Scientist	(HPCSA)	1994
Registered as a Genetic Counsellor	(HPCSA)	1996
C1 Rated Scientist	(NRF)	2003

1971-1972: Lecturer: Histology & Physiology, Dept. of Physiology, UCT Medical School
1973-1974: Cytogenetics Technician (1973) Biochemistry Technician (1974)
1974-1981: Interrupted service to start a family.
1981-1982: Cytogenetics Technician, Dept. of Human Genetics, UCT Medical School
1983-1986: Molecular Genetics Research Assistant
1987-1991: Molecular Genetics Research Technical Officer
1992-1995: Senior Scientific Officer, Registered Medical Scientist
1995-1996: Research Officer, Registered Genetic Counsellor, Head: Mol. Gen. Diagnostic Lab
1996-1998: Honorary Senior Lecturer, Genetic Counsellor, Head: Mol. Gen. Diagnostic Lab.
1998-2007: Associate Professor, Genetic Counsellor & Head Mol. Gen. Diagnostic Lab
Programme Co-ordinator for the MSc in Genetic Counselling course started in 2004
2009-present: Professor, Genetic Counsellor, MSc in Genetic Counselling Programme Co-ordinator, Honorary member of the NHLS in the Molecular Genetics Diagnostic Laboratory

1.3 Publications Total : 81

1.3.1 Articles in peer-reviewed journals

1.3.1.1 International

59. Allele-specific silencing of mutant Ataxin-7 in SCA7 patient-derived fibroblasts. Scholefield J, Watson L, Smith D, **Greenberg J**, Wood MJ. Eur J Hum Gen 2014 Dec;22(12):1369-75

58. Screening of a large cohort of Leber congenital amaurosis and retinitis pigmentosa patients identifies novel LCA5 mutations and new genotype-phenotype correlations. Mackay DS, Dev Borman A, Sui R, van den Born LI, Berson EL, Ocaka LA, Davidson AE, Heckenlively JR, Branham K, Ren H, Lopez I, Maria M, Azam M, Henkes A, Blokland E; [LCA5 Study Group (see acknowledgements for Universities): Sten Andreasson, Elfride de Baere, Jean Bennett,; Gerald J. Chader, Wolfgang Berger, Irina Golovleva, **Greenberg J,** Anneke I. den Hollander, Caroline C.W. Klaver, B. Jeroen Klevering, Birgit Lorenz, Markus N. Preising, Raj Ramsear, Lisa Roberts, Ronald Roepman, Klaus Rohrschneider, Bernd Wissinger], Qamar R, Webster AR, Cremers FP, Moore AT, Koenekoop RK. Hum Mutat. 2013 Nov;34(11):1537-1546

57. Caution regarding the interpretation of homoallelism in polyglutamine multiplex assays. A recommendation for confirmatory testing of homozygous alleles. Smith DC, Esterhuizen A, **Greenberg J** J Mol Diagn. 2013 Sep;15(5):706-9.

56. Huntington disease in the South African population occurs on diverse and ethnically distinct genetic haplotypes. Baine FK, Kay C, Ketelaar ME, Collins JA, Semaka A, Doty CN, Krause A, **Jacquie Greenberg L,** Hayden MR. Eur J Hum Genet. 2013 Oct;21(10):1120-7

55. RNA therapy for polyglutamine neurodegenerative diseases. Smith, Danielle C; Watson, Lauren M; **Greenberg, Jacquie LJ;** Wood, Matthew JA; and Scholefield, Janine (April 2013) eLS Review: eLS. John Wiley & Sons, Ltd: Chichester. DOI: 10.1002/9780470015902.a0024909

54. Understanding of Genetic Inheritance among Xhosa-Speaking Caretakers of Children with Hemophilia Gabriele Solomon & **Jacquie Greenberg** & Merle Futter, Lauraine Vivian & Claire Penn. J Genet Couns. 2012 Mar 10. Epub ahead of print

53 . Stargardt macular dystrophy: common ABCA4 mutations in South Africa-establishment of a rapid genetic test and relating risk to patients. Roberts LJ, Nossek CA, **Greenberg LJ**, Ramesar RS. Mol Vis. 2012;18:280-9. Epub 2012 Feb 1.

52. Design of RNAi Hairpins for Mutation-Specific Silencing of Ataxin-7 and Correction of a SCA7 Phenotype. Scholefield J, **Greenberg LJ**, Weinberg MS, Arbuthnot PB, Abdelgany A, Wood MJA 2009 PLoS ONE 4(9): e7232. doi:10.1371/journal.pone.0007232
http://www.plosone.org/article/info:doi%2F10.1371%2Fjournal.pone.0007232

51. Clinical utility of the ABCR400 microarray: basing a genetic service on a commercial gene chip. Roberts LJ, Ramesar RS, **Greenberg J.** Arch Ophthalmol. 2009 Apr;127(4):549-54.

50. Predictive testing for Huntington disease in a developing country. Futter MJ, Heckmann JM, **Greenberg LJ.** Clin Genet. 2009 Jan;75(1):92-7.

49. Management of a South African family with retinitis pigmentosa—should potential therapy influence translational research protocols? Lisa Roberts, George Rebello, Rajkumar Ramesar, **Jacquie Greenberg** J Ocul Biol Dis & Inf (JOBDI). 2008 Vol 1(1) 55-58. (New on-line journal)

48. A South African mixed ancestry family with Huntington Disease-Like 2: clinical and genetic features. Bardien, S Abrahams F, Soodyall H, Van Der Merwe L**, Greenberg J**, Brink T, Carr J. Mov Disord. 2007 Vol. 22, No. 14, 2083–2089

47. A common SNP haplotype provides molecular proof of a founder effect of Huntington disease linking two South African populations. Janine Scholefield, **Jacquie Greenberg.** Eur J of Hum Genet. 2007 : 15 : 590-595

46. Qualitative Research Methodology in the exploration of Patients' Perceptions of Participating in a Genetic Research Programme. Frieda Basson, Merle Futter, **Jacquie Greenberg.** Ophthalmic Genetics, 28:143–149, 2007

45. Origin of the *SCA7* gene mutation in South Africa and implications for molecular diagnostics in the South African indigenous Black population**. J Greenberg,** GAE Solomon, AA Vorster, J Heckmann, A Bryer. Clin Genet. Letter to the Editor. Clin Genet. 2006 Nov;70(5):415-7

44. Novel variants in the hotspot region of the RP1 gene in a South African study of patients with autosomal dominant retinitis pigmentosa. L. Roberts, L. Bartmann, R. Ramesar, **J.Greenberg.** Mol Vis 2006; 12:177-183 http://www.molvis.org/molvis/v12/a19/

43 . The Guanine-thymine dinucleotide repeat polymorphism within the tenascin-C gene is associated with achilles tendon injuries. Mokone GG, Gajjar M, September AV, Schwellnus MP, **Greenberg J**, Noakes TD, Collins M. Am J Sports Med. 2005 Jul;33(7):1016-21.

42. Mutation Spectrum and Founder Chromosomes for the ABCA4 gene in South African Stargard Disease patients. September AV, Vorster AV, Ramesar RS and **Greenberg LJ.** Invest Ophthalmol Vis Sci 2004; 45:1705-11

41. Apoptosis-inducing signal sequence mutation in carbonic anhydrase IV identified in patients with the RP17 form of retinitis pigmentosa. George Rebello, Rajkumar Ramesar, Alvera Vorster, Lisa Roberts, Liezle Ehrenreich, Ekow Oppon, Dumisani Gama, Soraya Bardien, **Jacquie Greenberg**, Giuseppe Bonapace, Abdul Waheed, Gul N. Shah, and William S. Sly PNAS 2004 101: 6617-6622.

40. Arg120stop nonsense mutation in the RP2 gene: mutational hotspot and germ line mosaicism? A.A. Vorster, M.T. Rebello, N. Coutts, L. Ehrenreich, A.D. Gama, L.J. Roberts, R. Goliath, R. Ramesar**, L. J. Greenberg** . Clinical Genetics; Jan 2004, Vol. 65 Issue 1, p7-11

39. Retinal degenerative disorders in Southern Africa: a molecular genetic approach. Ramesar RS, Roberts L, Rebello G, Goliath R, Vorster A, September A, Ehrenreich L, Gama D, **Greenberg J.** Adv Exp Med Biol. 2003;533:35-40.

38. A rare homozygous rhodopsin splice site mutation : The issue of when and whether to offer presymptomatic testing. **J. Greenberg,** L. Roberts, R. Ramesar. Ophthalmic Genet. 2003 ; 24(4): 225-32.

37. The Hereditary Adult Onset Ataxias in South Africa. Alan Bryer, Amanda Krause, Pierre Bill, Virginia Davids, Daphne Bryant, James Butler, Jeannine Heckmann, Rajkumar Ramesar, **Jacqueline Greenberg.** 2003 : J Neurol Sci. 2003 Dec 15;216(1):47-54.

36. Analysis of RPGR in a South African Family with X-linked Retinitis Pigmentosa; research and diagnostic implications. George Rebello, Alvera Vorster, **Jacquie Greenberg,** Natalie Coutts, Lisa Roberts, Liezle Ehrenreich, Dumisani Gama, Rajkumar Ramesar. 2003 : Clin Genet 2003 Aug, 64(2):137-41

35. Clinical diagnoses that overlap with choroideremia. Lee TK, McTaggart KE, Sieving PA, Heckenlively JR, Levin AV, **Greenberg J,** Weleber RG, Tong PY, Anhalt EF, Powell BR, MacDonald IM. Can J Ophthalmol. 2003 Aug;38(5):364-372

34. CDH23 Mutation and Phenotype Heterogeneity: A Profile of 107 Diverse Families with Usher Syndrome and Nonsyndromic Deafness. Astuto LM, Bork JM, Weston MD, Askew JW, Fields RR, Orten DJ, Ohliger SJ, Riazuddin S, Morell RJ, Khan S, Riazuddin S, Kremer H, Van Hauwe P, Moller CG, Cremers CW, Ayuso C, Heckenlively JR, Rohrschneider K, Spandau U, **Greenberg J,** Ramesar R et al. 2002 Am J Hum Genet Aug;71(2):262-75

33. A computer based register for inherited retinal dystrophies in Southern Africa. M.T. Rebello**, L.J. Greenberg** , R.S. Ramesar 2002 Ophthalmic Genetics Vol 22, No3. 61-5

32. Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa (RP13). Arthur B. McKie, John C. McHale, T. Jeffrey Keen, Emma E. Tarttelin, Rene Goliath, Janneke J.C. van Lith-Verhoeven, **Jacquie Greenberg,** Rajkumar S. Ramesar et al. 2001 Hum Mol Genet 10: 1555-1562.

31. Genetic heterogeneity of Usher syndrome: analysis of 151 families with Usher type I. Astuto LM, Weston MD, Carney CA, Hoover DM, Cremers CW, Wagenaar M, Moller C, Smith RJ, Pieke-Dahl S, **Greenberg J,** Ramesar R, et al. 2000 Dec Am J Hum Genet.67 (6):1569-74.

30. Low frequency of rhodopsin mutations in South African patients with autosomal dominant retinitis pigmentosa. Letter to the Editor Roberts L, R Ramesar, **J Greenberg.** 2000 Clin Genet 58: 77-78.

29. Genomic Structure & Identification of Novel Mutations in Usherin, the gene responsible for Usher Syndrome Type IIa. Weston MD, Eudy JD, Fujita S, Yao S, Usami S, Cremers C, **Greenburg J,(sic)** Ramesar R, et al. 2000 Am J Hum Genet. Apr; 66 (4) : 1199-1210.

28. Alstrom syndrome: further evidence for linkage to human chromosome 2p13. Collin GB, Marshall J, Boerkoel CF, Levin A, Weksberg R, **Greenberg J,** Michaud J, Naggert J, Nishina PM. **1999** Hum Genet 105(5):474-9

27. Refinement of the RP17 locus for autosomal dominant retinitis pigmentosa, construction of a YAC contig and investigation of the candidate gene retinal fascin. S.Bardien-Kruger, **J Greenberg,** B Tubb, J Bryan, L Queimado, M Lovette, R Ramesar. 1999 European Journal of Human Genetics 7: 332-338

26. Correlation between Waardenburg Syndrome phenotype and genotype in a population of individuals with identified PAX3 mutations. AL DeStefano, LA. Cupples, KS Arnos, JH Asher Jnr, CT Baldwin, S Blanton, ML Curey, ES da Silva, TB Friedlman, **J Greenberg,** AK Lalwani, A Milunsky, W Nance, A Pandya, RR Ramesar, AP Read, M Tassabejhi, ER Wilcox, LA Farrer. 1998 Hun Genet 102 : 499-506

25. Rhodopsin mutation Gly109Arg in a family with autosomal dominant retinitis pigmentosa. Rene Goliath, Soraya Bardien, Alison September, Rebecca Martin, Rajkumar Ramesar & **Jacquie Greenberg.** 1998 Human Mutation : Supplement 1: S40-41

24. Sorsby Fundus Dystrophy: Reevaluation of Variable Expressivity in Patients Carrying a TIMP3 Founder Mutation. Felbor U, Benkwitz C, Klein M, **Greenberg J,** Gregory C, Weber BHF. 1997 Arch Ophthalmol. 115:1569-1571.

23. Ethnic variation and in vivo effects of the -93t_g promoter variant in the lipoprotein lipase gene. Ehrenborg E, Clee SM, Pimstone SN, Reymer Pwa, Benlian P, Hoogendijk CF, Davis HI, Bissada N, Miao L, Gagne SE, **Greenberg LJ,** et al. 1997 Arterioscler Thromb Vasc Biol. 17 (11) :2672-2678.

22. Familial Streptomycin ototoxicity in a South African family: a mitochondrial disorder. Gardner J, **Greenberg J,** Goliath R, Viljoen D, Sellars S, Cortopassi GA, Hutchin T, Beighton P. 1997 J Med Genet : 34 : 904-906.

21. Retinitis Pigmentosa locus on 17q (RP17) : Fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes. Soraya Bardien, Rajkumar Ramesar, Shomi Bhattacharya, **Jacquie Greenberg.** 1997 Human Genetics : 101 : 13-17.

20. The gene for PEDF, a retinal growth factor, is a prime candidate for retinitis pigmentosa and is tightly linked to the RP13 locus on chromosome 17p13.3. Rene Goliath, Joyce Tombran-Tink, Ignacio R Rodriquez, Gerald Chader, Rajkumar Ramesar and **Jacquie Greenberg.** 1996 http://www.cc.emory.edu/MOLECULAR_VISION/goliath_96004/goliath.html

19. The gene for Cleidocranial Dysplasia in the historical Cape Town (Arnold) kindred is on chromosome 6. Rajkumar Ramesar, **Jacquie Greenberg,** Rebecca Martin, Rene Goliath, Soraya Bardien, Stefan Mundlos, Peter Beighton. 1996 J Med Genet 33 : 511-514

18. Fine localization of the locus for autosomal dominant retinitis pigmentosa on chromosome 17p. R Goliath, Y Shugardt, P Janssens, R Ramesar, J Weissenbach, P Beighton, **J Greenberg.** 1995 Am J Hum Genet 57 : 962-965

17. Sorsby's Fundus Dystrophy: A South African family with a point mutation on the tissue inhibitor of Metalloproteinases-3 gene on chromosome 22. A Peters and **J Greenberg.** 1995 Retina 15 : 480-485

16. An eighth locus for autosomal dominant retinitis pigmentosa is genetically linked to chromosome 17q Soraya Bardien, Neil Ebenezar, **Jacquie Greenberg,** Chris F Inglehearn, Lecia Bartmann, Rene Goliath, Peter Beighton, Rajkumar Ramesar, Shomi Bhattacharya. 1995 Hum Mol Genet 4:8 1459-1460.

15. Gene mapping of Usher syndrome type II: Localization of the gene to a 2.1 centimorgan segment on chromosome 1q41. W Kimberling, MD Weston, C Moller, J Sumegi, CWRJ Cremers, PS Ing, A Van Aarem, C Connolly, A Martini, ML Tamayo, **J Greenberg** and C Ayuso. 1995 Am J Hum Genet 56: 216-223.

14. DNA haplotype analysis of Huntington disease reveals clues to the origins and mechanisms of CAG expansion and reasons for geographic variations of prevalence Squitieri F, Andrew SE, Goldberg YP, Kremer B, Spence N, Zeisler J, Nichol K, Theilmann J, **Greenberg J**, Goto J, Kanazawa I, Vesa J, Peltonen L, Almqvist E, Anvret M, Telenius H, Lin B, Napolitano G, Morgan K, Hayden MR Hum Mol Genet 3 (12): 2103-2114 Dec 1994

13. Locus Heterogeneity for Waardenburg Syndrome is predictive of clinical subtypes. LA Farrer, JH Asher, CT Baldwin, TB Friedman, **J Greenberg,** KM Grundfast, C Hoth, AK Lalwani, A Milunsky, R Morell, V Newton, R Ramesar, VS Rao, TB San Agustin, ER Wilcox, I Winship, AP Read. 1994 Am J Hum Genet: 55: 728-737.

12. A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17. **J Greenberg,** R Goliath, P Beighton and R Ramesar. 1994 Hum Mol Genet. 3: 913-918

11. A splice junction mutation in PAX3 causes Waardenburg syndrome in a South African Family. Butt J, **J Greenberg,** I Winship, S Sellars, P Beighton, R Ramesar. 1994 Hum Mol Genet: 3: 197-198

10. Huntington Disease: Prenatal Screening for late onset disease. **J Greenberg** : 1993 J Med Ethics. Letter to the Editor 19: 121-122.

9. Retinitis Pigmentosa in Southern Africa. **LJ Greenberg,** L Bartmann, R Ramesar, P Beighton. 1993 Clin Genet: 44: 232-235.

8. An Alw44 I RFLP of the human S-antigen gene. **LJ Greenberg,** R Ramesar, P Beighton. 1993 Hum Mol Genet: DNA Markers and Polymorphism section.

7. Molecular Analysis of Juvenile Huntington Disease: the major influence (CAG)n on repeat length is the sex of the affected parent. H Telenius, HP Kremer, J Theilmann, SE Andrew, E Almqvist, M Anvert, C Greenberg, **J Greenberg,** G Lucotte, FS Squitieri, E Starr, YP Goldberg and MR Hayden. 1993 Hum Mol Genet: 2: 1535-1540.

6. Retinitis Pigmentosa, AD Type I: Exclusion of Linkage to D3S47 (C17) in a large South African Family of British Origin. **LJ Greenberg,** M Babaya, R Ramesar, P Beighton. 1992 Clin Genet : 322-325.

5. Waardenburg Syndrome (WS) Type 1. is caused by defects at multiple loci, one of which is between ALPP and FNI on chromosome 2 - First report of the WS Consortium. L Farrer, KM Grundfast, J Amos, KS Arnos, JH Asher, Jr, P Beighton, SR Diehl, J Fex, C Foy, TB Friedman, **J Greenberg,** C Hoth, M Marazita, A Milunsky, R Morell, W Nance, V Newton, R Ramesar, TB San Agustin, J Skare, CA Stevens, RG Wagner Jr, ER Wilcox I Winship and AP Read. 1992 Am J Hum Genet 50: 902-913

4. Linkage of Usher Syndrome, Type I gene (USH1b) to the long arm of chromosome 11. Kimberling WJ, Moller C, Davenport S, Priluck I, Beighton P, **Greenberg J**, Reardon W, Kenyon JB, Grunkmeyer JA, Pieke Dahl S, Overbeck LD, Blackwood DJ, Brower AM, Hoover DM, Rowland P and Smith RJH. 1992 Genomics 14: 988-994.

3. Mutations of the KIT (Mast/Stem Cell Growth Factor Receptor)Proto-Oncogene account for a range of Phenotypes in Human Piebaldism Spritz R, Holmes S, Ramesar R, **Greenberg J,** Curtis D, Beighton P. 1992 Am J Hum Genet 51: 1058-1065.

2. Genetic linkage between Huntington Disease and the D4S10 locus in South African families: Further evidence against non-allelic heterogeneity. **LJ Greenberg**, RW Martell, J Theilman, MR Hayden, J Joubert 1991 Hum Genet 87: 701-708.

1. Emery-Dreifus syndrome and X-linked muscular dystrophy with contractures: evidence for homogeneity. J Goldblatt, **LJ Schram,** G Wallis, A Oswald, P Beighton. 1988 Clinical Genetics 1989:35:1-4

4.1.2 Local

22. Duchenne muscular dystrophy: High-resolution melting curve analysis as an affordable diagnostic mutation scanning tool in a South African cohort. AI Esterhuizen, JM Wilmshurst, RG Goliath, **LJ Greenberg**. S Afr Med J 2014; Vol. 104 (11): 779-784

21. The value of genetic testing for inherited retinal disease caused by mutations in the ABCA4 gene in South Africans. LJ Roberts, S Hardie, T Goolam Hoosen, R S Ramesar, **LJ Greenberg.** Correspondence S Afr Med J. 2013; 103 (10):702-3

20. Direct-to-consumer genetic testing: To test or not to test, that is the question. Dandara C, **Greenberg J**, Lambie L, Lombard Z, Naicker T, Ramesar R, Ramsay M, Roberts L, Theron M, Venter P, Bardien-Kruger S. S Afr Med J. 2013; 103(8):510-2.

19. Stem cells on South African shores: Proposed guidelines for comprehensive informed consent. **Greenberg J,** Smith D, Pope A. Correspondence S Afr Med J 2013;103(1):6

18. A New Class of Stem Cells in South Africa: Introducing Induced Pluripotent Stem cells (iPS cells). Ballo R, **Greenberg LJ**, Kidson SH. S Afr Med J. 2012 Dec 6;103(1):16-7

17. Inherited polyglutamine spinocerebellar ataxias in South Africa. DC Smith, A Bryer, LM Watson, **LJ Greenberg.** S Afr Med J. 2012 Jun 14;102(8):683-6.

16. UCT's contribution to medical genetics in Africa - from the past into the future
P Beighton, K Fieggen, A Wonkam, R Ramesar, **J Greenberg.** S Afr Med J June 2012 (102) 6 : 446-448

15. Polyglutamine disease: from pathogenesis to therapy. Lauren M Watson, Janine Scholefield, L **Jacquie Greenberg**, Matthew J A Wood. S Afr Med J June 2012 (102) 6 : 481-484

14. The parent trap: Unexpected revelations that influenced the genetic management of a family with Stargardt disease. **Jacquie Greenberg,** Lisa Roberts. SA Ophthalmology Journal, Summer Edition 2012. Vol 17 No1 P 18-21 (Approved South African Journal ISSN 2218-8304)

13. Delivery of an ophthalmic genetic service in South Africa. **J Greenberg,** L Roberts, Z Bruwer, M Schoeman, K Loggenberg, F Loubser. SA Ophthalmology Journal, Autumn Edition 2010. Vol 5 No2 P 14-19 (Approved South African Journal ISSN 2218-8304)

12. Genetic testing For Huntington's disease In South Africa. Amanda Krause. **Jacquie Greenberg** Editorial S Afr Med J March 2008 (98) 3 : 193-194

11 Huntington's disease-like 2 in South Africa. **J Greenberg,** S Bardien, J Carr Letter to the Editor S Afr Med J March 2008 (98) 3 : 166

10. The á2CDel322-325 adrenergic receptor polymorphism is not associated with heart failure due to idiopathic dilated cardiomyopathy in black Africans. J Du Preez, LO Matolweni**, J Greenberg,** P Mntla, AA Adeyemo, BM Mayosi. Cardiovasc J Afr. 2008 Jan-Feb;19(1):15-6 (Approved South African Journal ISSN 1015-9657)

9. Genetic testing and related ethical issues. **J Greenberg.** 2005 CME January 23 (1) 18-21

8. When is it not Huntington Disease? JM Heckmann, A Bryer **J. Greenberg,** 2001 Scientific Letter : February SAMJ Vol 91 : 132-133

7. Ophthalmic Genetics: A review of the Molecular Genetics of Familial Retinal Dystrophies in Southern Africa. **Jacquie Greenberg,** George Rebello and Rajkumar Ramesar. 1999 (February) Specialist Medicine, 108-112

6. A photoreceptor gene mutation in an indigenous black African family with retinitis pigmentosa identified using a rapid screening approach for common rhodopsin mutations. **J. Greenberg,** T. Franz, R. Goliath and R. Ramesar. 1997 August SAMJ Vol 89 : 8: 877-878

5. A predictive testing service for Huntington Disease (HD) and late onset Spinocerebellar Ataxia (SCA1) in Cape Town. **Jacquie Greenberg,** Sue Beatty, Hans Soltau, Alan Bryer. 1996 CME Review : 14: 1364-1367.

4. Genetic Blindness: Macular Dystrophies and Retinitis Pigmentosa. **Jacquie Greenberg** and Anna Peters. *1995 SAMJ Editorial 85: 6 492-493.*

3. Genetic Mapping of Retinitis Pigmentosa - Implications for South African Patients. **J Greenberg,** R Ramesar and P Beighton. 1994 SAMJ 84: 410-412.

2. A molecular service for Huntington Disease in Southern Africa. **LJ Greenberg** 1989 SAMJ Editorial 76: 135-136

1. Biochemical Screening for inherited metabolic disorders in the mentally retarded. HE Hendersen, R Goodman, **J Schram,** E Diamond, A Daneel. 1981 SAMJ 7: 731-733

1.3.2 Articles in non-peer reviewed journals.

1.3.2.1 International

3. Global Awakening in Genetic Counseling. Edwards, Janice, **Greenberg, Jacquie,** and Sahhar, Margaret. Available from Nature Proceedings 2008 http://hdl.handle.net/10101/npre.2008.1574.1

2. Use of the Schleicher and Schuell DNA sample isolation device for effective sample collection and rapid DNA preparation from whole blood for PCR templates. **Jacquie Greenberg,** Sue Schultz, Najmeeyah Brown & Raj Ramesar. 1997 Schleicher and Schuell : SEQUENCES newsletter.

1. Genetics of hearing impairment. Hearing impairment and pigmentary disturbance.
P Beighton, R Ramesar, **J Greenberg**, K Young, D Curtis, S Sellars. Ann NY Acad Sci 152-166

1.3.2. 2 Local

1. Chromosomal breakages in a family with X-linked MR and in routine cytogenetic cases, induced using culture medium 199 without serum.
RD Smart, I Doidge, J Hamilton, N Kovensky, **J Schram,** P Smart
1982 SAMJ Lab Techn 28 (3) 25-27

1.3.3 Books/proceedings - authored or edited

11. Chapter 45: *Genetic Counseling in South Africa* **Jacquie Greenberg**, Jennifer Kromberg, Kelly Loggenberg, and Tina-Marié Wessels (P 531-546) in *Genomics and Health in the Developing World* Edited by Dhavendra Kumar: Series : Oxford Monographs on Medical Genetics, Oxford University Press: 14 June 2012

10. Chapter in SA Ethics Book : *Medical Ethics, Law and Human Rights: A South African Perspective.* Co-authored chapter entitled : *Genetic and ethical complexity*. **Jacquie Greenberg**. Editor: Keymanthri Moodley ISBN Number: 978 0 627 02809 0 : Associate Professor & Head, Bioethics Unit - Tygerberg Division, Faculty of Health Sciences & Centre for Applied Ethics, University of Stellenbosch. Van Schaik Publishers 2011: This book is recommended by all medical schools for undergraduate teaching. Published January 2011 P291-316

9. A book chapter was published in 2010 entitled : *"Great Expectations: RPE65 mutations in South Africa "* . Roberts L, G. Rebello**, J. Greenberg,** R. Ramesar. In: Retinitis Pigmentosa: Causes, Diagnosis and Treatment Editors: Michaël Baert and Cédric Peeters ISBN: 978-1-60876-884-4 2009 Nova Science Publishers, Inc. P 89-110

8. *Carbonic anhydrase inhibitors as a possible therapy for RP17, an Autosomal Dominant Retinitis Pigmentosa associated with the R14W mutation, apoptosis, and the unfolded protein response.* George Rebello, **Jacquie Greenberg** and Raj Ramesar (Chapter 24: p 455- 459)
In " Opportunity for Retinal Degenerations: Genetics, Progression and Therapeutics. "" Eds J. Tombran-Tink and C.J. Barnstable, Humana Press, Totowa, NJ. 2007

7. *Genetics of ocular vascular disease*. **Jacquie Greenberg,** Ari Ziskind and Stephen P. Daiger (Chapter1.10: p173-188) in " Ocular Angiogenesis: Diseases, Mechanisms and Therapeutics" Eds J. Tombran-Tink and C.J. Barnstable, Humana Press, Totowa, NJ. 2006

6. *Retinal degenerative disorders in Southern Africa: a molecular genetic approach.* Ramesar RS, Roberts L, Rebello G, Goliath R, Vorster A, September A, Ehrenreich L, Gama D, **Greenberg J.** Adv Exp Med Biol. 2003;533:35-40.

5. *Ophthalmic Genetics.* Ziskind A, **Greenberg LJ.** Pub Jaypee Brothers, Medical Publishers (P) Ltd. New Delhi. in "The Textbook of Ophthalmology". Ed : Agarwal S, Apple D, Buratto A, Agarwal A, Alio JL, Pandey SK, Agarwal A Vol 1. Basic Science section. Chapter 12, P99-116, 2002

4. *"An Ophthalmic Genetics Handbook for Ophthalmologists in Southern Africa."* **J. Greenberg** and A. Ziskind. > 100 copies were printed and distributed by Retina SA in 2001 and 2002.

3. *Unusual frequencies of Rhodospin mutations and polymorphisms in Southern African patients with Retinitis Pigmentosa.* **Jacquie Greenberg,** Lisa Roberts and Raj Ramesar. Proceedings published in "New Insights into Retinal Degenerative Diseases", edited by Anderson et al., Kluwer Academic/Plenum Publishers, pp 335-338. 2001 after the IX International Symposium on Retinal Degeneration : held in DURANGO, USA in 2000.

2. *Migratory history of populations and its use in determining research direction for retinal degenerative disorders.* Ramesar, R., September, A., Rebello, G., **Greenberg, J.**, Goliath, R. Proceedings published in "New Insights into Retinal Degenerative Diseases", edited by Anderson et al., Kluwer Academic/Plenum Publishers, pp 338-339. 2001 after the IX International Symposium on Retinal Degeneration : held in DURANGO, USA in 2000.

1. *Growth factors in the retina: Pigment epithelium-derived factor (PEDF) now fine mapped to RP17p13.3 and tightly linked to the RP13 locus.* **J Greenberg,** R Goliath, J Tombran-Tink, G Chader, R Ramesar. Proceedings of the VII International Symposium on Retinal Degeneration : SENDAI, JAPAN 1997 Page 291-294 Degenerative Retinal Diseases, edited by La Vail et al. Plenum Press, New York

1.3.4. Other contributions/reports (e.g. policy research documents)

3. Was invited and has been involved with the National Department of Health, Sub-directorate: Human Genetics with the drafting of regulations and guidelines for section 57 of the National Health Act 2004, Act no 61 of 2003. We have been and are still are consulted and tasked to address the prohibition of reproductive cloning of human beings as well as stem cell research in SA (from August 2003 till 2007).

2. A special sub-committee of the Experimental Biology Group (EBG) of the Western Cape (1994-1995) submitted an extensive document with recommendations for the Science and Technology Initiative to Minister S. Bengu, whilst vice-chair of the EBG.

1. Recommendations for Biology courses and training. L. Bohm, H.Hoffman, D.Bellstedt, M.Cherry, V.Corfield**, J.Greenberg,** M.Quobela, D.Hendricks, S. Kidson, G.Littlejohn, M.Madikizela and P.Swart Published as a letter in : S.Afr.J.Sci 91: 67 , 1995.

2 Personal scientific/scholarly presentations at congresses including by colleagues & students

2.1. International

28. Attended the XVIth International Symposium on Retinal Degeneration in California, USA : July 13-18, 2014. Presented a poster with PhD student D Smith entitled *: "A stem cell-derived model of retinal degeneration associated with Spinocerebellar ataxia type 7"* (DC Smith, LM Watson, R Ballo, J Scholefield, SA Cowley, MJA Wood, SH Kidson, **J Greenberg**).

27. International Conference on Tissue Engineering and Regenerative Medicine, Tshwane, SA (August 2014, oral presentation*): Modeling neural and retinal degeneration in Spinocerebellar Ataxia type 7, using induced pluripotent stem cells.* LM Watson, DC Smith, R Ballo, J Scholefield, L Tyers, SA Cowley, MJA Wood, SH Kidson, **LJ Greenberg**

26. Keystone Symposium on Stem Cells and Reprogramming, Olympic Valley, CA, USA (April 2014, poster presentation): *Transcriptional alterations and enhanced susceptibility to proteasomal stress in an iPSC-derived neural model of Spinocerebellar ataxia type 7.* LM Watson, J Scholefield, R, SA Cowley, DC Smith, SH Kidson, **LJ Greenberg**, MJA Wood

* 25. Attended the 3rd (every 2 years) Transnational Alliance of Genetic Counsellors (TAGC) & the 12th (every 5 years) International Congress of Human Genetics (ICHG) in October 2011 in Montreal Canada. Tina Wessels (from Wits) and I presented a poster entitled on *"Genetic Counselling Training in South Africa"* T Wessels, J Kromberg, **J Greenberg.** I then went to the USA where I was invited to visit the Eastside Preparatory School, Kirkland, Seattle and I also visited Dr Gemma Carvill at the Mefford Laboratory, University of Washington (October 17-18th & 20th). As a " Visiting Thinker" at the Eastside Preparatory School I addressed 11 & 12th grad scholars on "*Ethics and Genetics"* as well as on genetic counselling as a profession and then other scholars, the teachers and some parents and public on : *"Stem cell research; moving from the Hype to Hope"*.

* 24. October 2010: Invited to give a seminar at the University of Surrey. My talk entitled " *The establishment of an ophthalmic genetic service in a developing country in the 21st Century and the search for genes in and out of Africa."* focused on the inherited Retinal Degenerative Disorders research project at UCT and highlighted some of the challenges experienced in the translation of molecular genetic research results to clinical care in South Africa .

23. Ms Danielle Smith (nee Berkowitz) presented a poster entitled *"A method for measuring allele-specific RNAi knock-down in SCA7 patient lymphoblasts"* at the annual conference of the British Society for Human Genetics (BSHG) which took place at the University of Warwick, UK from 6th-8th September, 2010.

* 22. In 28 Feb 2009, I attended the European Huntington Disease Network Physiotherapy Working Group Meeting in Cardiff, Wales

*21. In June 2008, the TAGC group (Transnational Alliance of Genetic Counsellors) which was formed after the Genetic Counselling workshop in Manchester in 2006, met in Barcelona, Spain. I chaired the session on " *Research Collaborations*". I was a one of the 6 member organising committee for this meeting and was elected onto the TAGC Standing Committee where one appointee from each of the fifteen countries represented at Manchester 2006 was nominated and elected. The TAGC is now an independent and autonomous organisation and the principal office of the TAGC is located in the County of Richland, State of South Carolina USA.

20. In May 2008, I presented a poster entitled '*Genetic Counselling challenges with a family with HDL2: From the bedside to the bench and back to the bedside'* at the European Conference of Human Genetics 2008, in Barcelona , Spain.

*19. Following the meeting in Barcelona, I again went to Oxford, UK where I visited Dr Matthew Wood from the Department of Physiology, Anatomy and Genetics with whom I am working on the gene silencing project. We now have two students working on this project which is going very well indeed. A third PG student will hopefully be joining our group in 2009.

*18. In April 2008, I was invited by Dr Jane Kaye from the Ethox centre, University of Oxford to participate at a conference called *Governing Biobanks - What are the Challenges?* in June. The purpose of the conference was to focus on the issues that emerge from the management of DNA and sample collections. Unfortunately I could not attend that late in June, so I visited Dr Jane Kaye at the Wellcome Trust Ethox Centre before I left the UK on the 22nd of June, 2008.

17. In May 2007 I presented a poster at the GeNeMove Symposium that was held in Bonn, Germany entitled *"Post-transcriptional gene silencing of the pathogenic mutation that causes SCA7 in Black African patients with inherited ataxia, using shRNA that distinguishes between alleles that differ by a single nucleotide."* J. Scholefield, M. Weinberg, P. Arbuthnot, M. Wood, J. Greenberg

Following the meeting in Bonn, I went to Oxford, UK where I visited Dr Matthew Wood from the Department of Physiology, Anatomy and Genetics with whom I am working on the above mentioned gene silencing project. This visit was funded by the Oppenheimer Fund (University of Oxford) for collaborative research of this nature.

Later that month, I attended the Annual International Society of Nurses in Genetics (ISONG) Conference in Bristol as an invited guest (of Dr Heather Skiton).

*16. Following the Manchester Genetic counselling workshop in May 2006, I was invited as an academic visitor to St Mary's Hospital, Manchester, UK to meet with Ms Tara Clancy, a genetic counsellor and honorary lecturer at this genetic counselling training facility . Later that week, I traveled to to the Institute of Medical Genetics, Cardiff, Wales to meet with Dr Clara Gaff, Co-director of the MSc in Genetic Counselling Course & a Consultant Genetic Counsellor (Honorary) lecturer at this genetic counselling training facility and Professor Angus Clarke, Professor in Clinical Genetics and Director of the Cardiff MSc Course in Genetic Counselling.

*15. In May 2006, in Manchester UK, at the International Working Conference on "***Genetic Counselling Education: Connecting the Global Community*****":** I chaired 2 sessions entitled : *" Global Perspectives in the Developing Genetic Counseling Profession*" and then "*Developing Capacity for Genetic Counseling In Smaller or Developing Countries.* 72 Participants from 22 different countries attended this workshop and the group/s had very meaningful discussions and exchange of ideas and genetic counselling experiences.

*14. In April 2006, I was one of 12 participants from outside the USA (representing 8 different countries) who were invited to attend a 2 day workshop at the NIH (along with 12 USA Ophthalmologists/researchers). We were specifically selected and invited to bring some perspective and also the interests of the NEI-supported research community to this workshop to address the issue of Collaborative International Research Opportunities for the future. There were fifteen minute presentations from each participant addressing pre-specified and self-identified topics, with significant discussion following each presentation. I was the only non-medical scientist and South African as this workshop and was asked to address the deliver of genetic

results, the needs for genetic counselling and ethics related issues. The final discussion session was focussed on developing actionable recommendations for specific collaborative opportunities that the NEI appears uniquely positioned to address along with mechanisms for future funded research projects.

*13. In November 2004, I was personally invited to attend and presented a poster at the first international symposium of the National Neurovision Research Institute on translation of clinical and molecular research for inherited retinal disease therapies in Washington DC. The symposium was entitled : "International symposium on translational clinical research for inherited and orphan retinal diseases." and my poster *"The importance of counselling in the delivery of genetic results to patients & families with retinal degenerative disorders in southern Africa : from Lab to Life."* **J. Greenberg**, A. Peters, R. Ramesar.

*12. In 2004 I was invited to join leaders in the Genetic Counselling profession from the United States, the United Kingdom, Australia and Japan, who share an interest in genetic counselling education world wide, to address the topic : ***"Genetic Counselling Education: Connecting the Global Community "*** I was subsequently elected onto the advisor board and together, we planned an International Working Conference to take place in the UK in 2006.

*11. Xth International Symposium on Retinal Degeneration. October 2002 BURGENSTOCK, SWITZERLAND. Poster presentations *: Possible germline mosaicism in a family with the Arg120stop mutation in the RP2 gene. Time to address the question of genetic counselling following RD research?* **J Greenberg,** A Voster, G Rebello, L Roberts, L Ehrenreich, D Gama, R Ramesar.

10. *Retinal Degenerative Disorders in Southern Africa : A molecular genetic approach.* R Ramesar, L Roberts, G Rebello, R Goliath, A Voster, A September, L Eherenreich, D Gama, L Bartmann, **J Greenberg.**

9. The 10th International Congress of Human Genetics. MAY 2001 VIENNA, AUSTRIA Poster presentation: *A rare homozygous rhodopsin splice site mutation : To test or not to test, is there a question?* **Jacquie Greenberg,** Lisa Roberts, Raj Ramesar

8. IX International Symposium on Retinal Degeneration OCTOBER, 2000, DURANGO, USA. Poster presentations: *Unusual frequencies of Rhodospin mutations and polymorphisms in Southern African patients with Retinitis Pigmentosa.* **Jacquie Greenberg,** Lisa Roberts and Raj Ramesar .

Migratory history of populations and its use in determining research direction for retinal degenerative disorders. Ramesar, R.,September, A., Rebello, G., **Greenberg, J.**, Goliath, R.

7. Second Degeneration & Functional Restitution satellite symposium prior to the ARVO meeting. MAY 1998, FORT LAUDERDALE, FLORIDA USA
Poster presentation: *Mutations in RPE65 associated with retinal dystrophy as well as Leber's congenital amaurosis in Southern Africa.* **J. Greenberg,** R. Goliath, G. Rebello, A. September, T. Frantz, S. Short, D. Thompson, R. Ramesar.

Data services and software for identifying genes and mutations causing retinal degeneration. Daiger S.P., Rossiter B.F., **Greenberg J**., Christoffels A., Hide W. Investigative Ophthalmology & Visual Science 1998 39: S295.

*6. VII International Symposium on Retinal Degeneration. OCTOBER 1996, SENDAI, JAPAN. Poster presentation: *Growth factors in the retina: pigment epithelium-derived factor (PEDF) now fine mapped to 17p13.3 and tightly linked to the RP13 locus.* **J Greenberg,** R Goliath, J Tombran-Tink, G Chader, R Ramesar.

American Journal of Human Genetics (Conf*).* Poster presentation: *Data services and computational tools for identification of genes and mutations causing retinal degeneration.* SP Daiger, BJF Rossiter, **J Greenberg,** A Christoffels, W Hide (1997)) 61: 4 P A409-A409 Univ Chicago Press

*5. XII International Congress of Eye Research: SEPTEMBER 1996, YOKOHAMA, JAPAN. Poster presentation: *Genetic analysis of the pigment epithelium-derived factor gene in 25 South African families with recessive retinitis pigmentosa and Stargardt's disease.* **J Greenberg,** R Goliath, J Tombran-Tink, G Chader, R Ramesar.

* 4. International Retinitis Pigmentosa Association Congress: JULY 1994, PARIS, FRANCE. Oral Presentation : *A new locus for autosomal dominant retinitis pigmentosa in a large South African family.* **J Greenberg,** R Goliath, P Beighton and R Ramesar.

Poster Presentation: *A molecular and cell biology approach to retinitis pigmentosa in South Africa.* **J Greenberg,** R Goliath, R Ramesar, P Beighton, T Wiggins, S Kidson.

3. American Society of Human Genetics: OCTOBER 1994, MONTREAL, CANADA
Poster Presentation : Localization of a new Autosomal Dominant Retinitis Pigmentosa gene on chromosome 17p and screening of candidate genes. **J Greenberg,** R Goliath, Y Shugart, C Freund, S Bardien, P Beighton, R Ramesar.

2. March of Dimes ; Clinical Applications of Molecular Genetics. JULY 1989, BOSTON, USA. Poster presentation : *Preparation for a Molecular Service for Huntington Disease in Southern Africa.* **LJ Greenberg**

*1. The 7th Biennial meeting of the International Huntington Association & Research Group on Huntington Disease the World Federation of Neurology. OCTOBER 1987, St MARGHERITA LIGURA, ITALY.
Oral Presentation : *Preparation for predictive testing for Huntington Disease in South Africa*. **LJ Schram,** P Beighton, J Goldblatt, G Wallis.

*** Indicates invited contributions**

2.2 Local

*31 Will be attending the MRC Flagship Project meeting in PTA on 6/12/2014 regarding; 2015 on Legislation: A special publication of the SA Journal of Bioethics & Law is planned for July 2015 and I have been tasked with addressing : *"Informed consent Broad vs. narrow Research vs. therapy"* together with Prof A Dhai (Wits) Prof J Greenberg (UCT.

Abstract submitted: *New Technologies, new challenges: guidelines for the ethical oversight of informed consent for future research proposes.* Jacquie Greenberg, Danielle Smith, Richard Burman

The MRC Flagship ELSI group as a whole will be looking at:
1. Development of legislation pertaining to stem cells together with the National Department of Health.
2. Characterization of the phenomenon of stem cell tourism in South Africa and abroad, and how this impacts on patients and their families, as well as on public perception of the stem cell field; development of concrete proposals on how to address the problem in South Africa.
Profs: Michael Pepper (PTA), Melodie Slabbert (PTA/UNISA), Ames Dhai (Wits), Susan Kidson (UCT), Jacquie Greenberg (UCT), Drs: Robea Ballo (UCT) and Safia Mohomed (Wits).

*30. Attended the Stem Cell Flagship Project collaborators workshop at the University of Pretoria, Prinshof Campus 01 September 2014. Did an oral presentation entitled: *New Technologies, new challenges: Implications for the ethical oversight of the informed consent process.* **Prof J Greenberg (UCT)**

* 29. Invited Speaker/Chair at the 3rd Annual ARESA Research Ethics Seminar Southern Sun Newlands, Cape Town 18-19 September 2014 **Prof J Greenberg (UCT),** Prof H Soodyall (Wits), Prof S Rennie (Chapel Hill, North Carolina) Prof W Jaoko (University of Nairobi, Prof A Davis (Chapel Hill, North Carolina).

28 : The fifteenth congress of the SA Society of Human Genetics (SASHG) together with the African Society of Human Genetics (AfSHG) Joint Congress: 2013, Maslow Hotel, Johannesburg.
Oral Presentations:
***Some proposed guidelines for an ethical approach to comprehensive informed consent for patient-derived stem cell use.* **Jacquie Greenberg,** Danielle Smith in the special closing Stem Cells session.

Next Generation Sequencing for Retinal Degenerative Disorders; speedbumps on the road to providing a genetic diagnosis. L Roberts, S Barton, G Black, S Ramsden, R Ramesar, **J Greenberg**

Using induced pluripotent stem cells to model neuronal degeneration in Spinocerebellar Ataxia type 7. Lauren M Watson, Janine Scholefield, Robea Ballo, Sally A Cowley, Danielle C Smith, Marco S Weinberg, Susan H Kidson, **L Jacquie Greenberg,** Matthew JA Wood

Poster Presentations: *A stem cell-derived model of retinal degeneration associated with Spinocerebellar ataxia type 7.* Danielle C Smith, Lauren M Watson, Robea Ballo, Janine Scholefield, Sally Cowley, Matthew Wood, Susan Kidson, **Jacquie Greenberg**

Targetable disease-associated HTT haplotypes in the South African population. Fiona K Baine, Chris Kay, Jennifer Collins, Amanda Krause, Jacquie Greenberg, Michael R Hayden

Poster at 43rd National OSSA congress (March 2013) : *"The "Quick 7": A South African assay for the genetic diagnosis of ABCA4-associated retinopathies"*. Roberts, L; Nossek, C; Hardie, S; Goolam-Hoosen, T; Ramesar, R; Greenberg, J.

Research Group joint presentation at OSSA regional meeting at Vincent Palotti Hospital in September 2014 to raise the project profile and build relationships. Lisa Roberts, Raj Ramesar, **Jacquie Greenberg**

27. Neurological Association of South Africa (NASA) Congress, Stellenbosch, March 2013:

Jacquie Greenberg, Susan Kidson. Stem Cells: *What neurologists should know (*invited oral presentation)

Lauren M Watson, Janine Scholefield, Danielle C Smith, **L Jacquie Greenberg**, Matthew JA Wood. *An induced pluripotent stem cell-derived neuronal model of Spinocerebellar Ataxia type 7* (awarded

NASA/Novartis Award for Best Neuroscience Oral Presentation)

Danielle C Smith, Lauren M Watson, Fiona K Baine, Alina Esterhuizen, Janine Scholefield, Alan Bryer, Matthew JA Wood, **L Jacquie Greenberg.** *The polyglutamine SCAs in South Africa: a 25-year journey* (poster presentation)

F Baine, Chris Kay, Jennifer Collins, Amanda Krause, **L Jacquie Greenberg**, Michael R Hayden. *Distinct HTT haplotypes are associated with Huntington disease in the South African population* (poster presentation)

26. SASBMB in KZN, South Africa. January, 2012 http://www.sasbmb-fasbmb.co.za/
Oral presentation :
RNAi-based silencing therapy restores a polyQ disease phenotype in patient cells
Janine Scholefield, Lauren Watson, Danielle Smith, **Jacquie Greenberg** Matthew Wood

25. The fourteenth congress of the SA Society of Human Genetics (SASHG) together with the African Society of Human Genetics (AfSHG) Joint Congress: 2011, CTICC, Cape Town.

Oral Presentation at the Young Researchers Forum : Cape Town March 2011
Allele-specific silencing of the disease-causing gene in SCA1 patients in South Africa using a SNP linked to the trinucleotide expansion. F Baine, J Scholefield, A Bryer, M Wood, **J Greenberg**

Posters presented at the Joint Conference of the South African and African Societies of Human Genetics 2011 and the Young Researchers Forum, Cape Town, 6-9 March 2011

Investigation of transcriptional dysregulation in an iPSC-derived neuronal model of Spinocerebellar Ataxia 7. Watson, LM, **Greenberg, LJ**, Weinberg, MS, Wood, MJA and Scholefield, J.

A method for measuring allele-specific RNAi knock-down in SCA7 patient lymphoblasts. D Berkowitz, J Scholefield, M Weinberg, M Wood, **J Greenberg.**

Identification of a suitable SNP for allele-specific silencing of the disease-causing gene in SCA1 patients in South Africa. F Baine, J Scholefield, A Bryer, M Wood, **J Greenberg**

Haplotypes associated with Huntington Disease in SA populations. F Baine, M Ketelaar, M Schoeman, A Semaka, M Hayden, **J Greenberg**

Missing mutations: the molecular basis of retinal degenerative disorders in indigenous Africans.
Lisa Roberts, Christel Nossek, **Jacquie Greenberg,** Rajkumar Ramesar.

Common ABCA4 mutations in South Africans: frequencies, pathogenicity and genotype-phenotype correlations. CA Nossek, LJ Roberts, **LJ Greenberg**, RS Ramesar.

*24. Invited speaker at the PathTech 2009 Congress September 6-9 in Durban ICC. 2 Talks : Plenary (1 hour) on "*Ethics and Genetic Testing"* > 100 attendees. Breakaway session : 20 Min talk on "*Are Umbilical Cord Blood Banks the Ultimate Investment? "*

23. The thirteenth congress of the SA Society of Human Genetics SASHG: 2009, SPIER, STELLENBOSCH, CAPE TOWN

Oral Presentations:

Delivery of an ophthalmic genetic service including a telephone counselling model : a bench-to-bedside review. **Prof LJ Greenberg,** Ms LJ Roberts, Ms M Schoeman , Ms Z Bruwer , Ms K Loggenberg, Ms F Loubser

Knockdown of the disease-causing gene in South African patients with SCA7 using allele-specific RNAi. Ms J Scholefield , **Prof J Greenberg**, Prof M Weinberg, Dr P Arbuthnot, Dr A Abdelgany, Dr M Wood

Poster Presentations:
Identification of a SNP for allele-specific silencing of the disease-causing gene in SCA1 patients in South Africa. Ms F Baine, Ms J Scholefield, Prof A Bryer, Dr M Wood, **Prof J Greenberg**

Developing a molecular genetic diagnostic service founded on the use of the ABCR-400 gene chip; Lessons Learnt. Ms LJ Roberts, Prof RS Ramesar, **Prof J Greenberg**

Personal understanding of predictive test results in South African Nonpolyposis Colorectal Cancer Families. Ms Z Bruwer, Prof R Ramesar, **Prof J Greenberg**, Dr M Futter

Genetic Counselling experiences in delivery of genetic research results to patients affected with ABCA4 associated retinal degenerative disorders. Ms F Loubser, Ms K Loggenberg, Ms L Roberts, **Prof LJ Greenberg.**

Level of genetic knowledge and its impact on reproductive choices and risk communication in families with Cystic Fibrosis. Ms M Schoeman, Dr MJ Futter, **Prof LJ Greenberg.**

Basic understanding of genetic concepts amongst Isixhosa-speaking caregivers of Haemophilia patients. Ms G.A.E. Solomon, Prof C Penn, **Prof J Greenberg.** Dr L Vivian.

*25. November, 2008. PAEDIATRIC NEUROMUSCULAR DISEASES MEETING at Boschenmeer Estate, Paarl organised by PANDA (Paediatric Neurology and Neurodevelopmental Association : Oral presentation "*How close are we to a treatment for DMD, what genetic screens should we be doing?* "

* 24. I was an invited speaker at the MRC research day 2008 and talked on ""*Ethics of stem cell research."*

*23. July 2008, at the Communication, Medicine & Ethics 6th International Interdisciplinary Conference (COMET 2008) I presented a talk on "*Genetic Counselling in Africa ; looking at the new roles and new responsibilities for Health Professionals in South Africa today*. I chaired a session/ colloquium on Genetic Counselling at this meeting.

22. The twelfth congress of the SA Society of Human Genetics SASHG: 2007, GOLDENGATE, OFS. *Investigation of the HDL2 gene in the UCT Genetics Laboratory : Past, Present and Future*. **Jacquie Greenberg,** Janet Lyons-Lewis, Janine Scholefield

21. The eleventh congress of the SA Society of Human Genetics SASHG: 2005, MULDERSDRIFT, GAUTENG. *Sorsby Fundus Dystrophy and the importance of counselling in the delivery of genetic results to at-risk individuals : a Lab to Life experience*. **J. Greenberg,** A. Peters, R. Ramesar.

20. Invited to speak at the Royal Society in CAPE TOWN, September 2005 : *My heart still belongs to Daddy but not my DNA*

19. Optometry congress of Southern Africa : November 2005 at Spier, Cape Town and then at Caesars palace in Gauteng : *"Genetic testing & Genethics"* .

18. Ophthalmology Society of Southern Africa 2005, CAPE TOWN. *Genetics in Ophthalmology.*

17. The tenth congress of the SA Society of Human Genetics SASHG: 2003, DURBAN.
Origin of the South African SCA7 gene mutation **J Greenberg,** G Solomon, A Voster

16. The ninth congress of the SASHG: 2001, KRUGER NATIONAL PARK.
Autosomal dominant and autosomal recessive disease-causing mutations in the rhodopsin gene in families with Retinitis Pigmentosa. **J Greenberg,** L Roberts, R Goliath, R Ramesar.

*15. Ophthalmology Society of Southern Africa 2000, SWAZILAND. *Gene mapping : the role of links and luck and most of all, accurate clinical information in linkage analysis*. **J Greenberg,** S Bardien-Kruger, G Rebello, R Ramesar

14. International Southern African Congress on Cataract and Refractive Surgery 1999, DURBAN, *General Genetics Principles and Genetic Analysis***. J. Greenberg.**

13. The eighth congress of the SASHG : 1999 GORDON'S BAY, CAPE TOWN.
Advances in molecular genetic testing for Duchenne/Becker muscular dystrophy and improved molecular management in South African families. **J. Greenberg,** K. Carelse, R. Goliath, R. Ballo, P. Beighton

12. The Muscular Dystrophy Foundation of SA : 25th Anniversary Symposium 1999 CAPE TOWN. The molecular genetic approach to Duchenne/Becker muscular dystrophy in Southern Africa : an updated protocol. **J. Greenberg,** K. Carelse, C Pheiffer, R. Goliath, R. Ballo, P. Beighton

11. Ophthalmology Society of Southern Africa 1999, CAPE TOWN. *Ophthalmic genetics : a review of the molecular genetics of familial retinal dystrophies in Southern Africa.* **J. Greenberg,** M.T. Rebello and R.S. Ramesar

10. The seventh congress of the SASHG: 1997, KWA MARITANE, PILANSBERG. Huntington's Chorea revisited: Increasing knowledge about the expanding repeats in the Huntington Disease gene. **J Greenberg,** A September

Poster Presentation:
Fine mapping of RP17, the locus for Autosomal Dominant Retinitis Pigmentosa on 17q
Bardien S, Tlali M, Bartmann L, Ramesar R, **J Greenberg.**
Chair : Scientific Session 6 : Cancer Genetics V Corfield / **J Greenberg**

9. Mini congress of the SASHG. 1996, CAPE TOWN. *Expanding services with the expanding repeats : Department of Human Genetics, UCT.* **LJ Greenberg**

8. The sixth congress of the SASHG: 1995, MYKONOS, CAPE TOWN. *Allelic and non-allelic heterogeneity in Waardenburg Syndrome.* **J Greenberg,** J Butt, D Viljoen, S Sellars, P Beighton, R Ramesar

Poster Presentation: *A molecular and cell biology investigation into retinitis pigmentosa.* **J Greenberg,** R Goliath, P Beighton, T Wiggins, S Kidson, R Ramesar.

7. The fifth congress of the SASHG: 1993, BLOEMFONTEIN.
Retinitis Pigmentosa in Southern Africa: Linkage study in six ADRP families & mutation analysis. **LJ Greenberg,** R Goliath, R Ramesar, P Beighton.

Poster Presentation:
Comparison of two staining techniques to visualise DNA fragments in gels. **J Greenberg,** R

Goliath, R Ramesar.

6. Mini congress of the SASHG. 1992, CAPE TOWN. *Retinitis Pigmentosa in Southern Africa, a molecular approach.* **LJ Greenberg**

5. The fourth congress of the SASHG: 1991, VEREENIGING, TVL. a. *Non-Allelic Heterogeneity in Waardenburg Syndrome, Type I.* **LJ Greenberg**, I Winship, R Ramesar, P Beighton

b. *A Molecular Approach to Retinitis Pigmentosa in South Africa.* **LJ Greenberg,** I Winship, R Ramesar, P Beighton

4. SASHG, Medical Genetics meeting: 1990, PRETORIA. Report of a new polymorphic DNA variant and recombination within the G8 region in a family of mixed ancestry with Huntington disease. **LJ Greenberg**

3. The third congress of the SASHG: 1989, DURBAN. At-risk persons' knowledge and attitudes towards Huntington disease and predictive testing: A survey in Cape Town. **LJ Greenberg**

2. The second congress of the SASHG: 1988, CAPE TOWN. Participant in Huntington Disease workshop on: Implementation of a genetic service for HD in RSA. Poster presentation *Huntington Disease: Past, Present and Future.***LJ Schram,** P Beighton, J Goldblatt.

1. The first congress of the SASHG: 1987, RUSTENBERG, TVL. *Predictive testing for Huntington Disease in South Africa.* **LJ Schram,** J Goldblatt, P Beighton, G Wallis.

3. Personal scientific/scholarly attendance on Courses/Workshops

3.1. Refresher/educational courses

17. Invited Speaker/Chair at the 3rd Annual ARESA *Research Ethics Seminar* Southern Sun Newlands, Cape Town 18-19 September 2014 Prof J Greenberg (UCT), Prof H Soodyall (Wits), Prof S Rennie (Chapel Hill, North Carolina) Prof W Jaoko (University of Nairobi, Prof A Davis (Chapel Hill, North Carolina).

16. October 2010: Attended a 2 hour seminar/workshop at UCT on Teaching with Technology entitled *" Reconceptualizing conceptual change in science education "* by Professor Sten Ludvigsen from InterMedia, University of Oslo who reported on a study concerning secondary school students' meaning making of socioscientific issues in Information and Communication Technology (ICT) mediated settings. The study considered students working with web-based groupware systems concerned with genetics. The analysis identified how the students oriented their accounts of scientific concepts and how they attempted to understand the socioscientific task in different ways.

15 Attended a 3 hour Supervisory Skills - Training Seminar by Dr Brian Jude and Associates on " *Succeeding as a first time supervisor"* on 10 March 2010 at the Riverside Club.

14 Invited speaker at the 2010 IRENSA MODULE 2 : (17^{th} May — 28^{th} May 2010) at GSH Bioethics center:

SESSION 5 (May 21) ETHICAL ISSUES IN GENETICS RESEARCH

13 Invited speaker at a Harry Crossley Funded Seminar entitled *"The ten things science has taught me"* on Thursday 20th May from 12:00 pm - 13:00 pm together with two senior academics from Chemistry and Physiology.

12. Attended 3 "Ethics in Research" two day courses over the past few years offered by . IRENSA in Cape Town : 2002,2004,2006, 2007. Invited to present a case study for the 2006 IRENSA course.

11. Attended and participated in a two day workshop on Benefit Sharing with Developing countries – The case of Human Genetics Resources. May 2004. Monkey Valley, Cape Town

10. Attended a two day Course on Mentorship for Capacity Building and Empowerment. University of Stellenbosch, March 2004

9. Attended and participated in two 3 hour long workshops. One on Predictive Testing for Huntington Disease and the other : Diagnostic testing for the Inherited Ataxias at the American Society of Human Genetics : 50th Annual meeting. 2000, Philadelphia, USA

8. Attended 2 two hour workshops. One on : Getting published in Investigative Ophthalmology and Vision Science and the other : Conflict resolution in the research environment: Do men have the advantage? as well as a symposium on Advances in positional cloning by international leaders in their fields such as Alan Wright, Ed Stone, Bernard Weber and Jim Lupski at the Annual meeting of the Association for Research in Vision and Ophthalmology (ARVO)
1998, FORT LAUDERDALE, FLORIDA, USA

7. Participated in a series of lectures given by Department of Human Genetics during 1994 and 1995 : CME on Genetics in Medicine to specialists at Groote Schuur hospital; Introductory lecture on molecular genetic at every course; Lecture to the Department of Neurology on expanded repeats; Lecture to the Department of Psychiatry on the molecular basis of Huntington disease.

6. Organised and attended a Genetic Linkage Analysis course : J Ott & MC King. 1995, CAPE TOWN, SA

5. The sixth meeting of the Waardenburg Syndrome Consortium. OCTOBER 1994, MONTREAL, CANADA. Participation and presentation: 6 International research centres on the consortium. 50 participants. Mutation analysis in PAX3 in Southern African Families with Waardenburg syndrome.**LJ Greenberg,** J Butt, R Ramesar, I Winship, D Viljoen, P Beighton

The fourth meeting of the Usher Syndrome Consortium. OCTOBER 1994, MONTREAL, CANADA. Participation and presentation: Collaborative linkage studies of Southern African Families with Usher Syndrome. **LJ Greenberg,** L Bartmann, P Beighton, W Kimberling.

4. The fourth meeting of the Waardenburg Syndrome Consortium. OCTOBER 1992, SAN FRANCISCO, USA. Participation and presentation : 6 International research centres on the consortium. 40 participants. Linkage study using markers at the HUP2 locus in 7 Southern African Families of mixed ancestry with Waardenburg syndrome. **LJ Greenberg,** R Ramesar,
I Winship, P Beighton

The second meeting of the Usher Syndrome Consortium. OCTOBER 1992, SAN FRANCISCO, USA. Participation and presentation: Rod-Cone dystrophy, sensorineural deafness and renal dysfunction; an autosomal recessive syndrome entity? **LJ Greenberg,** L Bartmann, P Beighton.

3. Attended the American Society of Human Genetics Annual meeting and participated in a Retinitis Pigmentosa workshop : 1992, SAN FRANCISCO, USA.

2. The second meeting of the Waardenburg Syndrome Consortium. APRIL 1991, WASHINGTON, USA. Participation and presentation : 6 International research centres on the consortium. 20 participants. Waardenburg syndrome in 5 Southern African Families of Mixed Ancestry.**LJ Greenberg,** R Ramesar, I Winship, P Beighton

1. Enrolled in the Department of Continuing Education of Harvard Medical School for the course entitled : Clinical Applications of Molecular Genetics : 1989, BOSTON, USA. Participated in the March of Dimes workshop on Predictive testing for Huntington Disease: 1989, BOSTON, USA.

3.2 Other eg chairing or organising of conferences:

12. Participated in and chaired a number of 2-3 hour session on ethics and genetic research as part of the International Research Ethics Network for Southern Africa Diploma course at the UCT Bioethics Centre every year in July from 2003 to 2007.

11. Invited by the British Council : Foreign and Commonwealth Office to participate in a panel discussion at the DNA 50 seminar series "Reconstructing Human History Through Genetics" in July 2003.

10. The Xth congress of the SASHG: 2003, DURBAN, SA. Chair : Opening Scientific Session : Molecular Genetics **J Greenberg** / D Donnai

9. Chaired a session on Recent Advances in Genetic Analysis and invited to write a short review of the posters on this topic as well as the session at the Xth International Symposium on Retinal Degeneration in Burgenstock, Switzerland, 2002.

8. The IXth congress of the SASHG: 2001, Kruger National Park, SA. Chair : Scientific closing session : Molecular Genetics **J Greenberg** /A Krause

7. The IXth International Symposium on Retinal Degeneration. DURANGO, USA, 2000. Chair : Animal Model & Treatment Session M LaVail/**J Greenberg.**

6. The IIXth congress of the SASHG: 1999, GORDONS BAY, CAPE TOWN, SA. Chair : Scientific Session 5 : Molecular Genetics **J Greenberg** /A Olkers

5. Organisation of Scientific Programme for the SASHG Congress; 1999, GORDONS BAY, CAPE TOWN, SA, NATIONAL & INTERNATIONAL PARTICIPATION : 150 participants.

4. The VIIth congress of the SASHG: 1997, KWA MARITANE, PILANSBERG, SA. Chair : Scientific Session 6 : Cancer Genetics V Corfield /**J Greenberg**

3. Organisation of a Genetic Linkage Course; UCT, March 1995. Two international world experts invited to run the course: NATIONAL PARTICIPATION : 20 participants.

2. Organisation of the Experimental Biology Group (EBG) Scientific Computer-Software workshop whilst chairperson of the EBG; 1993 ; 100 participants from the WESTERN CAPE.

1. Organisation of the Experimental Biology Group (EBG) Spring symposium on: The future of biological research in the new South Africa ; 1993 130 participants from the WESTERN CAPE.

4 Recognition from peers.

4. 1. Awards:

URC Conference Travel grant	(2008)
Oppenheimer travel fund (University of Oxford)	(2007)
URC Conference Travel grant	(2006)
Visiting Scholars Fund Committee (VSFC) travel award	(2004)
URC Conference Travel Grant	(2002)
The John Sainsbury/Linbury Trust travel grant to visit my Ph.D. student in Oxford, UK. Ms A September was then a Sainsbury/Linbury scholarship recipient.	(2001)
Merrin Travel grant	(2001)
SASF travel grant (USA)	(1999)
Merrin travel grant	(1998)
International Society of Eye research: USA : Travel grant to attend the XII International Congress of Eye Research in Japan	(1996)
Merrin travel grant	(1996)
Merrin travel grant	(1994)
Isaac Greenberg bursary	(1968-1970)

4. 2. Research grants:

See 6.2 Funding for research projects (source and amounts)

MRC SIR, UCT URC & NRF CPRR	:	2009-2014
SANPAD Grant with C Penn	:	2007/8/9
MRC Grant with C Penn	:	2007/8/9
UCT URC project grant	:	2008
UCT URC project grant	:	2007
NRF Grant awarded March and July 2007	:	2007
Ataxia Foundation (UK) (PI's M Wood & J Greenberg).	:	2008
Ataxia Foundation (UK) (PI's M Wood & J Greenberg).	:	2007
NRF Grant with Prof. Claire Penn	:	2006
UCT Stimulation Grant	:	2005
UCT URC project grant	:	2002
Muscular Dystrophy Foundation of SA	:	1998-2004
P.I.s on the project: **J. Greenberg** and R. Goliath		
RP Foundation of South Africa	:	1991-2008
P.I.s on the project : **J. Greenberg** and R. Ramesar		
Foundation Fighting Blindness (USA)	:	1996-1999
P.I.s on the project : **J. Greenberg** and R. Ramesar		
THRIP : National Research Foundation (SA)		
Back-to-back funding for RDD projects	:	1998-2001
P.I.s on the project : **J. Greenberg** and R. Ramesar		
Bridgeman bequest	:	1995
P.I. **J. Greenberg**		
URC grant	:	2002
P.I. **J. Greenberg**		
Goldman Trust	:	1994-1997
P.I.s on the project : **J. Greenberg** and R. Ramesar		
National Institutes of Health (USA)	:	1993

P.I **J. Greenberg**

Actual amounts of these research grants are listed on page 28.

4.3. **International collaborations**

1. Professor Michael Hayden, UBC, Vancouver, CANADA *
2. Waardenburg Consortium: NIH, Washington USA
3. Professor Andrew Read, University of Manchester, UK
4. Professor W Kimberling, Boys Town, Omaha, USA
5. Professor J Ott, New York University, USA
6. Professor Shomi Bhattacharya, Moorefields Eye Hospital, LONDON, UK
7. Dr Chris Ingelhearn, Moorefields Eye Hospital, LONDON, UK
8. Professor Gerald Chader and Dr Joyce Tombran-Tink, National Eye Insititute, USA
9. Professor Anand Swaroop, University of Michigan, USA
10. Professor Bernard Weber, Wurtzburg, GERMANY
11. Professor Debra Thompson, University Michigan, USA.
12. Professor Stephen Daiger, University of Texas, USA.
13. Dr J Kaplan, Paris, FRANCE.
14. Professor Steven Pittler, Alabama, USA.
15. Dr Chris Campbell, USA.
16. Professor Michael Lovette, University of Texas, USA.
17. Dr Sue Chamberlain, St Mary's Hospital, London, UK.
18. Dr Richard Callaghan, Oxford University, UK.
19. Professor Chris Higgins, Hammersmith Hospital, London.
20. Dr Ian Kerr, Oxford University, UK.
21. Dr Kenny Linton, Hammersmith Hospital, London.
22. Professor Frans Cremers. University of Nijmegen, Holland *
23. Professor Janice Edwards. University of South Carolina. USA
24. Dr Matthew Wood. Oxford University, UK. *
25. Dr Janine Scholefield, Oxford University, UK. *

National collaborations

26. Professor Amanda Krause, Wits University, SA *
27. Professor Patrick Arbuthnot, Wits University, SA
28. Dr Marco Weinberg, Wits University, SA *
29. Professor Claire Penn, Wits University, SA *
30. Professor Jo Wilmshurst, RXH/UCT *
31. Professor Jeannine Heckmann, GSH/UCT *
32. Professor Alan Bryer, GSH/UCT *
33. Professor Susan Kidson UCT*
34. Dr Robea Ballo UCT*
35. Professor Raj Ramesar UCT *
36. Dr Rene Goliath UCT
37. Dr Merle Futter UCT*

* = current : 2011

4.4 Other scholarly activities:

34 Ran a one hour workshop on "*Ethics and public engagement "* at a Wellcome Trust-funded International Engagement initiative organised by Professor V Corfield at the MTN Science center on 17 March 2010 entitled :

Catalysing partnerships: the role of science centres as intermediaries between the public and scientists in engagement with biomedical sciences in South Africa : phase 2 which was attended by 25 participants involved in science education from the Western Cape.

33. Helped put together a programme for the Teaching Biology Project for March 31st (2 hours) for 52 teachers as part of the Mendel Group programme organized by Cheryl Douglas/ Melody Williams: TBP Coordinator, Teaching Biology Project, Africa Genome Education Institute

32 Invited speaker (by Ms Margaret Wolff) at the Grahamstown Scifest : Sunday 28 March 2010 13h30 - 14h30 Olive Schreiner Hall (seats 200) and Monday 29 March 2010 14h00 - 15h00 Art Gallery (seats 100*) " Should one clone alone or even at home?"*

31. Was invited to be part of the HPCSA team to evaluate the facilities for the training of the Intern Medical Scientists, Intern Genetic Counsellors and the Intern Physicists in Johannesburg and Pretoria in September, 2008.

30. Was invited by the Scientific Committee of the 2008 MRC Research Day to deliver a Plenary/Invited talk.

29. Chaired a colloquium on Genetic Counselling at the 6th International Communication, Medicine & Ethics Conference (COMET) meeting at the ICTCC in July 2008

28. In May, 2008, I was a plenary speaker at the South African Paediatrics Association (SAPA) in Sun City. My talk was in the session on Ethics and was entitled " *Ethics of stem cell research: The potential use and abuse of stem cells in medical research* " .

27. Invited to give an Ethics and Genetics lecture talk at the National Neurology meeting at Spier, Stellenbosch in March, 2008. Lecture entitled *" To test or not to test. Is there a question?"*

26. Invited to give an Ethics and Genetics lecture talk at the Physicians Conference in February, 2008. Lecture was entitled *" Issues in Genetic Counselling "*

25. Chaired a session on *Neurodegenerative Disorders in Africa* at the SASHG Specialist Genetics Meeting on Neurodegenerative Disorders in May 2008

24. Attended a half day MBChB TEACHING AND LEARNING RETREAT in April 2008.

23. Invited to participate in the evaluation process of the THRIP grant application reviews and renewals at the NRF in November 2007 for the NFR funding cycle for 2008.

22. Reviewed 2 grant proposals for the NHLS RESEARCH TRUST in 2007.

21. April 2007 : Invited to serve on the editorial board of the Journal of Ocular Biology, Diseases, and Informatics (*JOBDI*), which was launched at the beginning of 2008. The initial term as editorial board member will run through December 31, 2010.

20. Participated in a session at the PAEDIATRIC REFRESHER COURSE in NEONATAL MEDICINE in February 2007 which comprised case presentations and a panel discussion of ethical and psychosocial issues.

19. Invited to participate in the evaluation process of the HEALTH AND PHARMACEUTICAL SCIENCES grant application reviews at the NRF in February 2006 for the NFR funding cycle for 2006.

18. Invited to participate in the evaluation process of the University of Stellenbosch where the

Department of Genetics was being evaluated during the 2004-2009 cycle for accreditation together with 4 other academics all from different Universities.

17. Invited to participate in the evaluation process as a reviewer on the MRC grant review panel that visited Professor Himla Soodyall's department/MRC Unit at Wits University in June, 2004,

16. Invited speaker at the HSRC symposium on the Human Genome Project at Spier, Stellenbosch in 2003 where I addressed some issues relating to "THE NEW GENETICS AND GENETIC TESTING."

15. As a Division we were all involved in a series of lectures entitled : Continuing medical education on Genetics in Medicine to specialists at Groote Schuur hospital where I spoke on Molecular Genetics and genetic testing today.

14. Involved in a workshop at UCT Winter school on "Genetics in the Community" as well as 3 Summer School courses on Medical Genetics and the New Genetics over the past 10 years.

13. I regularly attend the National executive meetings of the then Retinitis Pigmentosa Foundation of South Africa, now called Retina South Africa and present current data on research in SA on Retinal Degenerative Disorders. In addition, I also report back on any relevant congresses that I have attended, such as the International Symposia on Retinal Degeneration that I attended and have participated at on 3 occasions.

12. Interviewed on SATV ; Good Morning South Africa, Radio Today and other SABC programmes such as the "In Touch" programme, following the announcement of the finding of the first and second genes for Retinitis Pigmentosa in South Africa

11. Regularly attend the National executive meetings of the Duchenne Muscular Dystrophy Society of South Africa and the Muscular Dystrophy Foundation and present current data on DMD and BMD and plans for research at UCT. I am currently the chair of their Scientific Medical Advisory board and have often served on their grant selection committee.

10. Organised a scholar's/student's information day in Human Genetics and this is now a regular annual event where I present some of our current research projects and introduce the concept of ethics in genetics to the scholar/students.

9. Invited to address lay societies on a regular basis on molecular genetics and research projects and am also invited to lecture at least once a year to the department of health genetic service nursing sisters.

8. Interviewed on Cape Talk Radio on cloning and a number of other Cape Radio stations on the genetics of cloning and the moral and ethical issues related to human cloning. Invited to participate in ethical discussions and debates on the ethics of cloning and have also addressed MENSA, the Cape Natural History Society and Rotary on the Biology of Cloning as well as ethics and genetics.

7. Invited to address school leavers at schools throughout the Western Cape at least once a year and have given talks on Human Genetics at De Kuilen, Herzlia, Rustenberg and Westerford High School.

6. Invited guest speaker at research centres, hospitals and universities in the UK, Ireland, France, Canada, Japan and the USA including the National Institutes of Health in Washington USA as well as genetic counselling training units in the USA. See travel grants page 22 : all funded and

invited visits are listed.

5. Regular reviewer for the Journal of Investigative Ophthalmology and Vision Research (IOVS), Genomics, Clinical Genetics, American Journal of Human Genetics and Ophthalmic Genetics.

4. Regularly asked to review new text books for recommendation for use for post graduate study in Human Genetics.

3. Invited to participate in peer reviews for FRD (now the NRF) which include Evaluation of Research outputs for new applications and re-evaluations for existing grant holders. Regularly asked to review grant applications for funding from the NRF and MRC.

2. Invited to review the grant applications for 3 international research groups for funding by the Foundation Fighting Blindness (USA).

1. Invited to be the (only) Guest Editorial Board Member to assess the reviews of a manuscript under review for publication in the Journal Investigative Ophthalmology & Vision Research.

5 Education and Scholarship

5.1. Undergraduate lectures/tutorials per year :

6. I co-ordinated and lectured in the Cytogenetic Module that was offered to the 2008 Honours students. I routinely give the Gene-Ethics lecture to this course every year.

5. The Human Genetics involvement in the new curriculum is overseen by Dr George Rebello and I assisted him with this new course. I was also involved with the planning of the lectures and tutorials as well as the Problem Based Learning cases which involve genetics in 2003. I supervised a SSM student assigned to Human Genetics for 2003 and 2 in 2005 and participated in the PBL tutoring for semester 3 in 2005. I routinely give the Gene-Ethics lecture to the 6th year MBChB students.

4. The Human Genetics 20 lecture series course for the Human Biology first year course was co-ordinated and run by me for 2 years which included 10 tutorials, marking of 200 essays, mid year tests and final exam scripts of the 200 students.

3. For the past 7 years I am regularly invited as a guest lecturer as part of a 4 day course in Human Genetics run by the Department of Health, for the training of Genetic Nursing sisters throughout SA.

2. Invited special guest lecturer & tutorials instructor in Human Genetics (genetic mapping and linkage analysis) for the UCT Biochemistry course BCH304S.

1. Often invited to address students (3rd years and honours) at the University of Stellenbosch, University of Western Cape as well as the Faculty of Health Sciences at Tygerberg Hospital.

5. 2. Postgraduate lectures/tutorials per year:

5. Following a sabbatical in 2001, when I spent three weeks attending the only genetic counselling course offered in Africa at that time (at the University of the Witwatersrand), I drew up a proposal to establish a Masters course in genetic counselling at UCT. This Masters course was

accepted by the Faculty and UCT in 2003 selected its first intake of students for registration in 2004. In 2005, the first two students qualified, 1 in 2006 and 2 in 2007. 4 of the 5 have completed their Internship at UCT MSc. There are currently only 14 registered genetic counsellors in Southern Africa. With a population of close to 45 million people, there can be no doubt that there is an urgent need for developing qualified professionals in human genetics in Africa.

4. I have been involved with the Human Genetics honours course since 2000 and was the Human Genetics Co-ordinator when we joined forces with Cell Biology and ran a joint Honours course where students did modules with Medical Biochemistry and Medical Microbiology. In 2001 the Biomedical Sciences Honours Course was launched and was the Human Genetics Co-ordinator for this programme till 2005.

3. This has been a very successful merger and joint endeavour and the numbers of registered students has escalated dramatically. In total, there are 36 students registered in the Biomedical Sciences Honours Course in 2006. I supervise 1 of the Human Genetics Honours students this year and assist with other student supervision where necessary.

2. I ran a 3 week module in genetic counselling in 2003 for the Biomedical Sciences Honours Course and taught in the Medical Genetics Module in May 2003 as part of the Biomedical Sciences Honours Course.

1. In 2002, I offered a 2 week module in genetic counselling for the first time at UCT and 5 students from the Biomedical Sciences Honours Course attended this module. Some senior scientific staff member from the Division of Human Genetics at UCT, as well as students from Tygerberg Medical school and senior scientific staff from UWC also attended this module.

5. 3. External examining at undergraduate level:

3. Served as external examiner for the Human Genetics Honours course at Tygerberg Hospital (Faculty of Health Sciences), Pretoria University and Wits on a number of occasions ([past 2 years for Wits).

2. Invited examiner for a genetics course for senior medical technologists at the Cape Technicon.

1. Invited special guest lecturer for a number of courses on gene mapping and linkage analysis; UCT Biochemistry Course (2^{nd} & 3^{rd} years) and examiner.

5. 4. External examining of dissertations/theses :

Masters thesis for University of Witwatersrand.
Masters thesis for University of Stellenbosch.
Masters thesis for University of Western Cape.
Masters thesis for University of Cape Town.

- **Personal supervision of Post Graduate students (43) :**

Co-Supervision of Master's student	: Ms G Dusterwald	(2014-2015)
Co-Supervision of Honour's student	: Ms Lynn Tyers	(2014)
Co-Supervision of Honour's student	: Ms Romy Wood	(2013)
Co-Supervision of Honour's student	: Ms Ansie Wichers	(2012)
Co-Supervision of Honour's student	: Mr Gift Pule	(2012)
Co-Supervision of Honour's student	: Ms Reinette Weiderman	(2011)
Supervision of PhD student	: Ms Danielle Smith	(2011-2014)
Supervision of PhD student	: Ms Fiona Baine	(2011-2015)
Supervision of PhD student	: Ms L Watson	(2009-2011)

Supervision of MSc student	: Ms Danielle Berkowitz	(2009-2011)
Supervision of MSc student	: Ms Fiona Baine	(2008-2010)
Supervisor of Honour's student	: Ms A Foxon	(2010)
Co- Supervision of MSc student	: Ms Christel Nossek	(2009-2010)
Co- Supervision of MSc student	: Ms Alina Esterhuizen	(2008-2010)
Reader of MSc Student in the USA	: Dr Shobana Kubendran	(2006-2007)
Supervision of PhD student	: Ms J Scholefield	(2006-2008)
Supervision of MSc student	: Ms G Solomon	(2006-2009)
Co- Supervision of PhD student	: Ms G Carvill	(2006-2009)
Co-Supervision of MSc student	: Ms Z Bruwer	(2006-2007)
Co-Supervision of MSc student	: Ms M Schoeman	(2006-2007)
Supervision of PhD student	: Dr M Futter	(2005-2007)
Supervision of MSc student	: Ms K Loggenberg	(2005-2006
Supervisor of Honour's student	: Ms J Lyons-Lewis	(2006)
Co-Supervision of MSc student	: Ms L Roberts	(2006)
Supervision of MSc student	: Ms J Scholefield	(2004-2005)
Supervision of MSc student	: Ms F Basson.	(2004-2005)
Supervision of MSc student	: Ms B Kruger.	(2004-2005)
Supervisor of Honour's student	: Ms S Marais.	(2004)
Supervisor of Honour's student	: Ms M Sasindranath	(2003)
Supervision of PhD student	: Mrs A. September.	(1999- 2003)
Supervision of MSc student	: Mr J. Bekker (thesis only)	(2003)
Supervisor of Honour's student	: Ms G. Solomon.	(2003)
Supervisor of Honour's student	: Ms O. Matthysen.	(2002)
Supervisor of Honour's student	: Ms N. Pillay.	(2002)
Supervisor of Honour's student	: Ms J. Scholefield.	(2000)
Supervision of MSc student	: Mrs A. September.	(1999)
Co-Supervisor of PhD student	: Mrs R Goliath.	(1996-2000)
Co-Supervisor of PhD student	: Ms S. Bardien.	(1996-1999)
Co-Supervisor of PhD student	: Dr J. Gardner.	(1999)
Co-Supervisor of Honour's student	: Mrs A. September.	(1996)
Co-Supervisor of Honour's student	: Sister S.Beatty.	(1995)
Co-Supervisor of Ph.D. student	: Dr A.Bryer.	(1994)
Co-Supervisor of M.Sc. student	: Ms J.Butt.	(1994)
Co-Supervisor of Honour's student	: Dr J Gardner.	(1994)

Supervised the research project of Dr. Nathaniel Khaole which was submitted in part fulfilment of the Diploma in International Research Ethics that is presented by the International Research Ethics Network for Southern Africa (IRENSA), Bioethics Centre, FHS, UCT. Awarded a Diploma in 2005

6 Research

6.1. Research projects in progress and output

1. The Genetic basis of Inherited Retinal Degenerative Diseases (RDD) in Southern Africa (SA) : Co-PI : Professor Raj Ramesar. Project Leader: Ms Lisa Roberts

Output: Over the past 15 years our research group has published over 40 articles in international peer-reviewed journals as well as a number in local journals. For most of the more recent publications, I am the last author, indicating that I have been responsible for directing the research presented in the manuscript and for initiating and overseeing the writing of the paper. As co-investigators on this project, Prof Ramesar and I both contribution to concept and design, grant-writing, and the provision of infrastructure and I am solely responsible for compiling the budget report. My main area of responsibility on this project currently being assessing the clinical utility of the ABCR400 Microarray and basing a genetic service on a commercial gene chip which includes our genetic

counseling experiences in the translation of these ABCA4 gene mutation results to clinical care (see the two latest publications in 2009 and 2010 related to the RDD group's activities in this regard). I am last author on the International one and first on the local publication.

Over the past five years I have personally presented results of our research findings at international and local congresses. Students and colleagues working on the project have presented our findings at international congresses and there have been >20 posters and oral presentations at many local congresses.
There have been 4 successfully completed Ph.D. projects on various aspects of this project as well as two MSc (both with distinction) and 12 successful Honours projects have been completed.

2. Huntington Disease (HD) & the Inherited Ataxias (SCA) in Southern Africa (SA).

In collaboration with on HD research:

: Professor Michael Hayden (UBC Vancouver) & Honorary Professor(UCT)
: Professor Amanda Krause (Wits)
: Professor Jeannine Heckmann (GSH/UCT)

In collaboration with on Ataxia research:

: Professor Matthew Wood (Oxford, UK) and Honorary Professor(UCT)
: Professor Marco Weinberg (Wits)
: Professor Susan Kidson (UCT/Human Biology: the iPSC research facility)
: Dr Robea Ballo (UCT/Human Biology: the iPSC research facility)
: Professor Alan Bryer (GSH/UCT)
: Dr Lauren Watson (UCT/NRF: Post Doc on the Ataxai SCA7 project)

In consultation with Dr Janine Scholefield (CSIR) on whose initial research to develop an RNAi-based therapy for the South African SCA7 patient population in patient iPSC-derived cells, much of this current research is now based.

I am the main PI at UCT on this project with the other researchers (co-investigators and or collaborators/associates) who all contribute to the concept and design, grant-writing, and the provision of infrastructure, depending on their areas of expertise (ie clinical and/or technical as well as research methodology support).

My PhD thesis, completed in 1990, was entitled *"A molecular approach to HD in SA"*. Two Masters projects were subsequently undertaken looking at the origin of the HD gene in SA, from a molecular genetic point of view (1999 & 2005). In 2006 an Honours research project was undertaken looking at HDL-2 in SA. In 2007, Dr Merle Futter completed a PhD project on the HD Predictive testing programme in the Western Cape.

This research group has published in international peer-reviewed journals together with research colleagues with whom we collaborate on HD. Our research findings have been presented at a number of international and local congresses. In 2002 an Honours research project was undertaken on SCA7 in SA.

Recent publications have been in the Eur J of Hum Gen (IF3.251), Clin Genet (IF 3.181), J Mov Disorders (IF3.323), as well as the SA Medical Journal (2.042). For most of these I am either first or last author and there are generally two or three authors only per manuscript.

Ms Fiona Baine won third prize at the 3rd MRC Research day in 2008 for her poster entitled "*Identification*

of a suitable SNP for allele-specific silencing of the disease-causing gene in SCA1 patients in South Africa" and Janine Scholefield received a special prize at the 3rd RESEARCH DAY 2008 of the SCHOOL OF BIOMEDICAL SCIENCES at UCT for her oral presentation entitled : *"An allele-specific RNA-based approach to specifically knockdown the disease-causing gene in SCA7."*

PhD student : Ms Lauren Watson was awarded a prestigious NRF Commonwealth Split-Site Doctoral Scholarship which is tenable in the UK in 2010/2011. She spent a year in total in the Oxford laboratory of Professor Matthew Wood working with Dr Janine Scholefield, who completed a postdoctoral fellowship in Oxford from 2008-2011. In September 2012, Lauren Watson submitted her PhD thesis entitled "*Novel cell models for the study of Spinocerebellar Ataxia type 7 pathogenesis and therapy in a South African patient cohort* " for examination. This project is part of a larger collaborative project looking at developing a gene therapy for spinocerebellar ataxia type 7 using a novel RNAi approach. Dr Watson graduated in December 2012 and proceeded to take up a Post Doc position with Profs Sue Kidson and LJ Greenberg at UCT/FHS.

Current SCA/HD research projects (2008-2013)

- PhD : Ms Danielle Smith *"An induced pluripotent stem cell derived model of retinal degeneration associated with Spinocerebellar ataxia type 7."*
- PhD : Ms Fiona Baine *"An investigation of haplotypes associated with Huntington disease in South African populations."*
- Honours Project: Ms Romy Wood: "*A haplotype study of South African SCA2 patients"*

3. Genetic Counselling based-research till 2012

Co-investigator : Professor Claire Penn (Wits)
Research Associate : Dr Merle Futter (UCT)

I am the main PI at UCT on this research with the other researchers (co-investigator and or collaborators/associates) who contribute to the concept and design, grant-writing, and the provision of infrastructure depending on their areas of expertise (ie research methodology support). To date, only one manuscript has been published in an important journal related to this new discipline, which is not yet rated but there are 3 manuscripts currently being prepared for publication on genetic counselling related topics.

Current and completed genetic counselling-based projects :

- Mrs Gillian Dusterwald (2014-2015) MSc (Med) : " *A preliminary investigation into the knowledge, attitudes and opinions of physicians to genomics, genetics and genetic counselling in South Africa. what are the barriers to patient referral to genetics services?*
- Ms Gabriele Solomon (2007-2009) MSc (Med) : "*An investigation into the level of understanding of basic genetics of mothers or caregivers of Xhosa haemophilia patients. "*
- Ms Mardelle Schoeman (2007) MSc mini project *"An investigation into the level of genetic knowledge and family communication about genetic risk in parents of children with Cystic Fibrosis".*
- Ms Zandre Bruwer (2007) MSc mini project : *"Hereditary Nonpolyposis Colorectal Cancer : Comprehension of a cancer risk in conjunction with a genetic risk. "*
- Ms Kelly Loggenberg (2006) MSc mini project : *"An investigation into the level of genetic knowledge of parents of sons with Duchenne Muscular Dystrophy and their satisfaction with the genetic counselling service at Red Cross War Memorial Children's Hospital."*
- Ms Frieda Basson (2005) MSc mini project: *"A pilot study of how individuals with inherited retinal degenerative disorders perceive being part of a molecular genetic research programme".*
- Ms Brenda Kruger (2005) MSc mini project: *" HNPCC: Factors contributing to adherence & non-adherence to surveillance for mutation carriers in rural areas of Northern & Western Cape".*

1. Muscular Dystrophy (MD) : Developing a comprehensive service for DMD and BMD in SA.
Co-PI's : Professor Jo Wilmshurst, Dr Rene Goliath.

Two Honours students have undertaken mini projects on this D/BMD project (2002, 2003).

Recent DMD research project (2008-2010)

Ms Alina Esterhuizen is currently registered to do a MSc project part time : "*Detection of Nonsense Mutations in Duchenne's Muscular Dystrophy Patients: Establishing a Cohort for Potential Treatment with PTC124."* .

6.2 Funding for research projects (source and amounts) :

Source	Amount.	
RDD : P.I.s on the project **J Greenberg** & R Ramesar		
RP Foundation /Retina South Africa	R 15,000.00	1992
	R 46,000.00	1993
	R 200,000.00	1994
	R 155,000.00	1995
	R 205,000.00	1996
	R 205,000.00	1997
	R 225,000.00	1998
	R 315,000.00	1999
	R 327,137.00	2000
	R 384,967.00	2001
	R 441,212.50	2002
	R 441,212.50	2003
	R 500,000.00	2004
	R 500,000.00	2005
	R 660,000.00	2006
	R 620,000.00	2007
	R 500,000.00	2008
	R 450,000.00	2009
	R 450,000.00	2010
	R 500,000.00	2011
	R 500,000.00	2012
	R 500,000.00	2013
	R 509,650.00	2014
Foundation Fighting Blindness (USA)	R 235,000.00 p.a.	1997-1999
THRIP back-to-back funding for RDD project	R 450,000.00 p.a.	1998-1999
THRIP back-to-back funding for RDD project	R 1,200, 000.00	2008
HD : P.I.: **J. Greenberg**		
Bridgeman bequest	R 160,000.00	1995
Interest from this bequest is used for research		
DMD: P.I. **J Greenberg** 1999-2001 with R. Goliath 2002-2004.		
Muscular Dystrophy Foundation	R 50,000.00 p.a.	1999-2004
SCA: P.I. **J Greenberg** 2002-present		
2014 NRF CPRR Grant	R 165,000.00	2014
2013 NRF CPRR Grant	R 300,000.00	2013
2012 NRF CPRR Grant	R 290,000.00	2012
(Competitive Programme For Rated Researchers (CPRR)		
2014 MRC Self-Initiated Research Grant	R137,000.00	2014
2013 MRC Self-Initiated Research Grant	R137,000.00	2013

2012 MRC Self-Initiated Research Grant	R137,000.00	2012
2012 NRF for a rated Scientist	R 40,000.00	2012
2011 UCT top up	R 40,000.00	2011
2011 NRF for a rated Scientist	R 40,000.00	2011
2010 UCT/NRF top up	R 40,000.00	2010
2010 for a rated Scientist	R 40,000.00	2010
2009 UCT/NRF top up	R 40,000.00	2009
2008 Ataxia Foundation (UK) (PI's M Wood & J Greenberg).	R 140,000.00	2008
NRF Grant	R 90,125.00	2008
URC grant for a project on SCA7 & 1	R 20,000.00	2008
URC grant for a project on SCA7	R 20,000.00	2007
NRF Grant	R 90,125.00	2007
Oppenheimer Fund (University of Oxford)	R 43,000.00	2007
The John Fell Fund (Oxford University) (PI's M Wood & J Greenberg).	R 60,000.00	2007
Ataxia Foundation (UK) (PI's M Wood & J Greenberg).	R 140,000.00	2007
URC grant for a project on SCA7 in SA	R 12,000.00	2002

I was rated by the NRF as a C1 rated scientist in 2004 and received a once off grant from UCT for newly rated NRF Scientists in 2005 : **R30,000.00**

GENETIC COUNSELLING :

In 2005 UCT awarded us a Stimulation grant of **R40,000.00** for the Genetic Counselling course that I started 2004 and in 2007, we received a URC Top-up for a genetic counselling-based project on DMD **(R 20,000.00)** .

In 2006, together with Professor Claire Penn from Wits, we were funded **R30,000.00** from the NRF for a genetic counselling project on language and communication at UCT (Haemophilia/genetic communication project).

In 2007, together with Professor Penn, we were awarded **R400,000.00** for 3 years by SANPAD for a project entitled *"Investigating communication and cultural practices in genetic counselling sessions in South Africa*" and **R330,000.00** from the MRC for a project around *"Transcending cross-cultural and cross-linguistic barriers : the challenges of communication practices in genetic counselling in SA"*.

Travel awards from International collaborators & hosts where I was an invited guest speaker or workshop co-ordinator and or participant:

The John Fell Fund **(Oxford University)** and Ataxia UK for the SCA7 project and my visit to Oxford in **May 2007** (collaboration with Dr Matthew Wood from Oxford).
Oppenheimer Fund **(Oxford, UK)** Dr James Tibbet and Dr Matthew Wood. **May 2007**
National Eye Institute, USA : Professor Paul Sieving, **USA/NIH. 2006**
Professor Janice Edwards, University of South Carolina, **USA** to visit her unit in **2004** as well as funded via her **2005** Jane Engelberg Memorial Fellowship to attend the "Genetic Counseling Education: Connecting the Global Community" workshop in Manchester, **UK in May 2006.**
Professor P Humphries : Trinity College, Dublin, Ireland with whom we collaborate on RDD.
Dr W Kimberling : Boys Town Hospital **Omaha Nebraska**
Retinal Preservation Foundation of Southern Africa: Funding to attend the International Retinal Preservation Association meeting in **Paris**.
Professor A Swaroop : Kellogg Eye Center, **Michigan** with whom we collaborate on RDD.
Professor G Chader and Dr J Tombran-Tink : **National Institutes of Health, USA** with who we collaborate on RDD. **1996**

The Waardenburg Syndrome Consortium: **NIH Funded** : To attend meetings of the consortia for Waardenburg and Usher Syndromes. (Official representative for SA group: Washington **1991, San Francisco 1992, Montreal 1994)**.

International Society of Eye research : USA : To attend the XII International Congress of Eye Research in **Japan**. **1997**

Professor J Tombran-Tink : The Children's National Medical Center, Washington D.C ; USA with who we collaborate on RDD. **1999**

Retinal Preservation Foundation of Southern Africa: Funding to attend Annual meeting of the Association for Research in Vision and Ophthalmology (ARVO) in **Florida, USA 1998.**

Society for Alstrom Syndrome Families **(USA**) : Funding to attend the first meeting of the Medical Advisory Board of the SASF. **1999**

Activities such as refereeing for international journals ;

In April 2007 I was invited to serve on the editorial board for the *Journal of Ocular Biology, Diseases, and Informatics* (*JOBDI*), which will be launched at the beginning of 2008. The initial term as editorial board member will run through December 31, 2010. The Journal is an extension of an Ophthalmology Series published by Humana Press.

I have been invited to review articles for publication in Investigative Ophthalmology and Visual Science, Archives in Ophthalmology, Clinical Genetics, European Journal of Human Genetics, American Journal of Human Genetics, Genomics, Ophthalmic Genetics and the South African Medical Journal. I was also invited to be the (only) Guest Editorial Board Member to assess the reviews of a manuscript under review for publication in the Journal of Investigative Ophthalmology and Vision Research (IOVS).

Examination of postgraduate theses from other institutions;

I have examined a total of 9 Master theses from the University of Stellenbosch, Western Cape and Wits and one from UCT.

7 Community and Clinical Service

7.1 Direction of community/clinical services

To provide education and raise awareness about genetics in the community and also to establish and provide diagnostic services for genetic patients and their families throughout SA in the form of :

- molecular genetic tests
- genetic counselling and
- presymptomatic testing programmes for late- onset genetic conditions.

7.2 Participation in clinical services

1. Huntington Disease: Monthly Clinic attendance, regular interaction with patients and HD Foundation of SA, provide diagnostic and presymptomatic testing service, genetic counselling for families as well as for predictive testing.

2. Inherited Ataxia: Monthly Clinic attendance, diagnostic and presymptomatic testing , genetic counselling for families as well as for predictive testing.

3. Retinitis Pigmentosa and Macular Degeneration: Interaction with patients and Retina SA, diagnostic and presymptomatic testing, genetic counselling for families as well as for predictive testing. I head the translational genetic aspect of our joint RDD research project. 4. Duchenne and Becker Muscular Dystrophy: Interaction with Muscular Dystrophy Foundation of SA, diagnostic, carrier detection and prenatal testing.

7.3 Participation in extension services

I have regular contact with the lay organisations (Retina SA and Muscular Dystrophy Foundation) that fund the research projects that I work on and I address their members at meeting throughout the country on genetics and the diagnostic services available to them. I handle all the telephone calls that come through to the molecular genetics laboratory from patients and doctors throughout SA regarding diagnostic testing for genetic disease. I regularly address the Department of National Health genetic nursing sisters on general molecular genetic principals and genetic testing and teach on a regular basis at their 4 day teaching course at Karl Bremner Hospital.

7.4 Clinical supervision of students

Visiting foreign students as well as 4^{th}, 5^{th} and 6^{th} year medical students who attend the monthly Neurogenetic clinic for Huntington and Spinocerebellar Ataxia at GSH are taken through the genetic counselling aspect of the predictive testing programme for late onset genetic disease.

Clinical supervision of genetic counsellors/students

A genetic counselling student from South Carolina, USA spent her summer rotation in 2006 in our training unit and another rotation student from Utah, USA is currently doing her elective with us at UCT.

8 Leadership and Administration

8.1 University- committees etc.
(Administrative contributions at Departmental, Faculty or University level).

I headed up the diagnostic and research laboratory in the Division of Human Genetics till 2007 where there up to 16 researchers/research assistants who are all BSc (Hons), M.Sc. and Ph.D. graduates. Professor Ramesar, the HOD, is the P.I. on some of the research projects however, he has many duties and so all the researchers in the laboratory report to me. I am therefore responsible for driving and sustaining the research and diagnostic laboratory. There is one departmental assistant assigned to the molecular laboratory, who falls directly under me.

There is no secretarial support in the laboratory so all typing and filing and administration that is undertaken in the laboratory is done by the researchers and or myself. Most of the physical ordering of chemicals and stationary is referred to the divisional secretariat, once the order has been ratified by me. In addition, I manage and monitoring all the funds under my FMC's. When

Professor Ramesar is away, I am required to be the Acting Head for the Division of Human Genetics, where Professor Martin is the HOD of the Department of Clinical Laboratory Sciences.

I represent the Division of Human Genetics on a number of the Department of Clinical Laboratory Sciences (D.C.L.S.) committees as well as for the FHS:

- Chair the FHS Masters Committee (part course work and part dissertation) and serve on the FHS Post graduate exco. (from March 2007-2010).
- FHS/UCT Acting Port folio Manager : Postgraduate Affairs. Jan 2006 -Feb 2007.
- The D.C.L.S. Post Graduate committee for 2010 and again 2013-2015
- The D.C.L.S. Research committee 2006: Portfolio = Health and Safety and Radiation control
- The D.C.L.S. Post Graduate Programme committee : Deputy chair to Professor Hall. I was tasked with setting up the Basic Sciences Lecture Course, organising a number of PPC Functions as well as the D.C.L.S. PPC research day in 2002 and compiling the PGS evaluation forms which have just been implemented in April 2003. The controversial seminar series that started this year is also an initiative of the D.C.L.S. PPC.
- the co-ordinator for Human Genetics on the Biomedical Sciences Honours Course.

For the Faculty of Health Sciences (F.H.S.) I :

- a member of the Faculty of Health Sciences Research Ethics Committee (2002-2007) having previously served on the Animal Research Ethics committee (2000-2001).
- Deputy head to Professor Swanepoel on the Post Graduate Programme Committee for scientists registered for higher degrees for 2003-5.

8.2 Hospital

I oversee all the molecular genetic diagnostic testing for the GSH/NHLS genetics laboratory. In 1995 I established and still co-ordinate a predictive testing programme for late onset genetic diseases in Cape Town.

8.3 Professional organisations

1. Medical Scientist registered with the HPCSA
2. Genetic Counsellor registered with the HPCSA
3. Member of National Society of Genetic Counsellors (USA).

8.4 Extension services, or services to industry, government and NGOs, including

As chair of the South Africa Society of Human Genetics (SASHG) I represented the SASHG, the Cape Branch of the Department of Health as well as the Division of Human Genetics at workshops in Pretoria assisting with the drafting of regulations and guidelines for section 57 of the National Health Act 2004, Act no 61 of 2003: Prohibition of reproductive cloning of human beings (2003-06)

8.5 Private work: all work based on the staff member's academic knowledge and skills.

Molecular genetics diagnostic testing for private patients.

9 Societies – National and International

9.1 Membership

Member of the Southern Africa Society of Human Genetics (SASHG)
Member of the African Society of Human Genetics (AfSHG)
Member of Experimental Biology Group of the Western Cape (EBG)
Member of Association of Medical Scientists
Member of National Society of Genetic Counsellors (USA).
Member of Southern African Inherited Disorders Association (SAIDA)

Member of the Royal Society of South Africa
Member of International Huntington Association (Canada).
Member of Retina South Africa
Member of Muscular Dystrophy South Africa
Member of The Society for Alstrom Syndrome (USA)

9.2 Office held (committee member, secretary, president, etc)

Society/Association	From …to	Office held (committee member, secretary, president, etc)
South African Society of Human Genetics (SASHG)	1999-2005	Committee member
SASHG	2011-present	Committee member
SASHG	2003-2005	Chair
SASHG	1997-1999	Secretary
Muscular Dystrophy Foundation of South Africa (MDF)	1999-2000	Grant Committee member
MDF Medical Advisory Board	2002-2006	Chair
International Society for Alstrom Syndrome (IAS)		
IAS Medical and Scientific Advisory Board	1999-present	Committee member
UCT FHS Human Ethics Committee	2002-2007	Committee member
UCT FHS Animal Ethics Committee	2000-2001	Committee member
Medical Alumni Club : UCT MAC club	1997-2005	Committee member
UCT Association of Medical Scientists (AMS)	1995-1998	Committee member
AMS	1996-1997	Secretary
Experimental Biology Group (EBG)	1992-1995	Committee member
EBG	1992-1993	Secretary
EBG	1993-1994	Chair
EBG	1994-1995	Vic-chair
GSH peer review (of specialist scientists) committee	2002-2003	Committee member
UCT/FHS Committee for the research director on Transfer of DNA & the GMO act.	2000-2001	Ad Hoc committee member
Retina South Africa: Medical & Scientific Board Member	2000-2010	Ad Hoc committee member
SA Wise: UCT/Western Cape branch	2000-2003	Committee member
SAWISE	2002	Treasurer
SAWISE	2009-2011	Committee member
SAWISE HOPE Scholarship	2010-present	Committee member (mentorship)
Transnational Alliance of Genetic Counsellors (TAGC)	2004-present	Steering Committee & Founder Member
Genetic Counsellors South Africa (GC-SA)	2012-present	Committee member