



PUBLICATIONS RELATED TO GENETIC COUNSELLING FROM THE UNIVERSITY OF CAPE TOWN



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PUBLICATIONS IN PEER REVIEWED JOURNALS

37. A call for policy action in sub-Saharan Africa to rethink diagnostics for pregnancy affected by sickle cell disease: differential views of medical doctors, parents and adult patients predict value conflicts in Cameroon. Wonkam A, Hurst S. *OMICS*. 2014 Jul 18(7):472-80. doi: 10.1089/omi.2013.0167. Epub 2014 Apr 22

36. Psychosocial Stressors of Sickle Cell Disease on Adult Patients in Cameroon. Wonkam A, Mba CZ, Mbanya D, Ngogang J, Ramesar R, Angwafo FF 3rd. *J Genet Couns*. 2014 Feb 21. [Epub ahead of print]

35. Genomic medicine in Africa: promise, problems and prospects. Wonkam A, Mayosi BM. *Genome Med*. 2014 Feb 24 6(2):11

34. Predictive Genetic Testing in Children: Constitutional Mismatch Repair Deficiency Cancer Predisposing Syndrome. Bruwer Z, Algar U, Vorster A, Fieggen K, Davidson A, Goldberg P, Wainwright H, Ramesar R. *J Genet Couns*. 2014 23(2):147-55

33. Psychosocial Burden of Sickle Cell Disease on Parents with an Affected Child in Cameroon. Wonkam A, Mba CZ, Mbanya D, Ngogang J, Ramesar R, Angwafo FF 3rd. *J Genet Couns*. 24 Jul 2013; 23(2):192-201. doi: 10.1007/s10897-013-9630-2

32. Would you terminate a pregnancy affected by Sickle cell disease? Analysis of views of patients in Cameroon. Wonkam A, J de Vries J, Royal CD, Ramesar R, Angwafo FF III. *J Med Ethics*. 1 July 2013; 0:1-6. Doi: 10.1136/medethics-2013-101392

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

30. 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.

[29.](#) 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

[28.](#) Communicating cancer risk within an African context: Experiences, disclosure patterns and uptake rates following genetic testing for Lynch syndrome. Bruwer Z, Futter M, Ramesar R. Patient Educ Couns. 28 Feb 2013; 92(1):53-60

[27.](#) A mobile colonoscopic unit for lynch syndrome: trends in surveillance uptake and patient experiences of screening in a developing country. Bruwer Z, Futter M, Ramesar R. J Genet Couns. 2013; 22(1):125-37

[26.](#) Prenatal diagnosis and termination of pregnancy: perspectives of South African parents of children with Down syndrome. Scott CJ, Futter M, Wonkam A. J Community Genet. Jan 2013; 4(1):87-97. doi: 10.1007/s12687-012-0122-0. Epub 2012 Oct 25

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

[24.](#) Two double heterozygotes in a South African Afrikaner family: implications for BRCA1 and BRCA2 predictive testing. Loubser F, de Villiers J, van der Merwe N. Clin Genet. 9 Apr 2012; doi: 10.1111/j.1399-0004.2012.01878.x

[23.](#) Understanding of Genetic Inheritance among Xhosa-Speaking Caretakers of Children with Hemophilia. Solomon G, Greenberg J, Futter M, Vivian L & Penn C. J Genet Couns. Oct 2012; 21(5):726-40

[22.](#) 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 1.19

[21.](#) The parent trap: Unexpected revelations that influenced the genetic management of a family with Stargardt disease. Greenberg J, Roberts L. SA Ophthalmology Journal, Summer Edition 2012; 17(1):18-21

[20.](#) Delivery of an ophthalmic genetic service in South Africa. Greenberg J, Roberts L, Bruwer Z, Schoeman M, Loggenberg K, Loubser F. SA Ophthalmology Journal, Autumn Edition 2010; 5(2):14-1

[19.](#) Predictive testing for Huntington disease in a developing country. Futter MJ, Heckmann JM, Greenberg JL. Clin Genet. Jan 2009; 75(1):92-7

[18.](#) Genetic testing for Huntington's disease in South Africa. Krause A, Greenberg J. Editorial S Afr Med J. March 2008; 98(3):193-194

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

[16.](#) Global Awakening in Genetic Counseling. Available from Nature Precedings. Edwards J, Greenberg J, & Sahhar M. <<http://hdl.handle.net/10101/npre.2008.1574.1>> (2008)

- [15.](#) Epidemiology, risk factors and genetics of breast cancer. Loubser F, Edge J, Fieggen K. SA Journal of CPD: Breast cancer. 2008; 26(10);497-498,500-501
- [14.](#) Management of a South African family with retinitis pigmentosa — should potential therapy influence translational research protocols? L Roberts L, Rebello G, Ramesar R, Greenberg J. J Ocul Biol Dis & Inf (JOBDI). 2008; 1(1);55-58
- [13.](#) Exploring Patient Perceptions of Participating in a Genetic Research Programme in South Africa using Qualitative Methods. Basson F, Futter M, Greenberg J. Ophthalmic Genetics. 2007; 28(3);143-149 (doi:10.1080/13816810701356627)
- [12.](#) 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; 22(14);2083-2089
- [11.](#) Qualitative Research Methodology in the exploration of Patients' Perceptions of Participating in a Genetic Research Programme. Basson F, Futter M, Greenberg J. Ophthalmic Genetics. 2007; 28:143-149
- [10.](#) Genetic testing and related ethical issues. Greenberg J. CME. January 2005; 23(1);18-21
- [9.](#) Arg120stop nonsense mutation in the RP2 gene: mutational hotspot and germ line mosaicism? Vorster AA, Rebello MT, Coutts N, Ehrenreich L, Gama AD, Roberts LJ, Goliath R, Ramesar R, Greenberg LJ. Clin Genet. Jan 2004; 65(1);7-11
- [8.](#) 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
- [7.](#) A rare homozygous rhodopsin splice site mutation: The issue of when and whether to offer presymptomatic testing. Greenberg J, Roberts L, Ramesar R. Ophthalmic Genet. 2003; 24(4):225-32
- [6.](#) Analysis of RPGR in a South African Family with X-linked Retinitis Pigmentosa; research and diagnostic implications. Rebello G, Vorster A, Greenberg J, Coutts N, Roberts L, Ehrenreich L, Gama D, Ramesar R. Clin Genet. Aug 2003; 64(2);137-41
- [5.](#) Ophthalmic Genetics: A review of the Molecular Genetics of Familial Retinal Dystrophies in Southern Africa. Greenberg J, Rebello G & Ramesar R. Specialist Medicine. February 1999; 108-112
- [4.](#) A predictive testing service for Huntington Disease (HD) and late onset Spinocerebellar Ataxia (SCA1) in Cape Town. Greenberg J, Beatty S, Soltau H, Bryer A. CME Review. 1996; 14:1364-1367
- [3.](#) Genetic Mapping of Retinitis Pigmentosa - Implications for South African Patients. Greenberg J, Ramesar R & Beighton P. SAMJ. 1994; 84:410-412
- [2.](#) Huntington Disease: Prenatal Screening for late onset disease. Greenberg J: J Med Ethics, Letter to the Editor 1993; 19:121-122
- [1.](#) A molecular service for Huntington Disease in Southern Africa. Greenberg LJ. SAMJ Editorial. 1989; 76:135-136

PUBLISHED CHAPTERS IN BOOKS

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

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