

ALL PUBLICATIONS ARISING FROM THE IRD PROJECT

56. [Whole-Exome Sequencing Identifies Biallelic IDH3A Variants as a Cause of Retinitis Pigmentosa Accompanied by Pseudo-coloboma](#). Pierrache LHM, Kimchi A, Ratnapriya R, Roberts L, Astuti GDN, Obolensky A, Beryozkin A, Tjon-Fo-Sang MJH, Schuil J, Klaver CCW, Bongers EMHF, Haer-Wigman L, Schaliq N, Breuning MH, Fischer GM, Banin E, Ramesar RS, Swaroop A, van den Born LI, Sharon D, Cremers FPM. *Ophthalmology*. 2017; pii: S0161-6420(16): 32230-32238.
55. [Molecular Diagnosis of Inherited Retinal Diseases in Indigenous African Populations by Whole-Exome Sequencing](#). Roberts L, Ratnapriya R, du Plessis M, Chaitankar V, Ramesar RS, Swaroop A. *Invest Ophthalmol Vis Sci*. 2016; 57:6374–6381.
54. [Inherited retinal disorders in South Africa and the clinical impact of evolving technologies](#). Roberts L, Goliath R, Rebello G, Bardien S, September AV, Bartmann L, Loubser F, Greenberg LJ, Ramesar RS. *South African Medical Journal* 2016; 106(6):10988 -10992.
53. [A Founder Mutation in MYO7A Underlies a Significant Proportion of Usher Syndrome in Indigenous South Africans: Implications for the African Diaspora](#). Roberts L, George S, Greenberg, J, Ramesar RS. *Invest Ophthalmol Vis Sci*. 2015;56:6671–6678.
52. [A mutation in a splicing factor that causes retinitis pigmentosa has a transcriptome- wide effect on mRNA splicing](#). Korir PK, Roberts L, Ramesar R, Soeighe C. *BMC Research Notes* 2014; 7(1):401
51. “The value of genetic testing for inherited retinal disease caused by mutations in the ABCA4 gene in South Africans.” Roberts LJ, Hardie S, Goolam Hoosen T, Ramesar R, Greenberg LJ. *South African Medical Journal* 2013; 103(10): 702-703.
50. [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, Jacquie Greenberg, 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 Aug 14. doi: 10.1002/humu.22398. [Epub ahead of print]
49. [Stargardt disease: towards developing a model to predict phenotype](#). Heathfield L, Lacerda M, Nossek C, Roberts L, Ramesar R. *Eur J Hum Genet*. 2013; 21(10):1173-1176. [Corrigendum](#) in 21(10);1190.
48. “The parent trap: Unexpected revelations that influenced the genetic management of a family with Stargardt disease.” Greenberg J, Roberts L. *SA Ophthalmology Journal* Summer 2012; 7(1): 18-21.

47. [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.
46. "Great Expectations: RPE65 Mutations in South Africa." Lisa Roberts, George Rebello, Jacquie Greenberg and Raj Ramesar. Published in "Retinitis Pigmentosa: Causes, Diagnosis and Treatment" (Nova Publishers). 2010; 89-110.
45. [Prognosis for splicing factor PRPF8 retinitis pigmentosa, novel mutations and correlation between human and yeast phenotypes.](#) Towns KV, Kipioti A, Long V, McKibbin M, Maubaret C, Vaclavik V, Ehsani P, Springell K, Kamal M, Ramesar RS, Mackey DA, Moore AT, Mukhopadhyay R, Webster AR, Black GC, O'Sullivan J, Bhattacharya SS, Pierce EA, Beggs JD, Inglehearn CF. Hum Mutat. 2010 May;31(5):E1361-76.
44. [Cell-specific differences in the processing of the R14W CAIV mutant associated with retinitis pigmentosa 17.](#) Pandor A, Ramesar R, Prince S. J Cell Biochem. 2010 Oct 15;111(3):735-41.
43. "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 2010; 5(2): 14-20.
42. [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.
41. [Management of a South African family with retinitis pigmentosa-should potential therapy influence translational research protocols?](#) Roberts L, Rebello G, Ramesar R, Greenberg J. J Ocul Biol Dis Infor. 2008 Mar;1(1):55-8.
40. [Evaluation of splicing efficiency in lymphoblastoid cell lines from patients with splicing-factor retinitis pigmentosa.](#) Ivings L, Towns KV, Matin MA, Taylor C, Ponchel F, Grainger RJ, Ramesar RS, Mackey DA, Inglehearn CF. Mol Vis. 2008;14:2357-66.
39. [Qualitative research methodology in the exploration of patients' perceptions of participating in a genetic research program.](#) Basson F, Futter MJ, Greenberg J. Ophthalmic Genet. 2007 Sep;28(3):143-9.
38. [Novel variants in the hotspot region of RP1 in South African patients with retinitis pigmentosa.](#) Roberts L, Bartmann L, Ramesar R, Greenberg J. Mol Vis. 2006 Mar 15;12:177-83.
37. [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. 2004 Jan;65(1):7-10.
36. [Apoptosis-inducing signal sequence mutation in carbonic anhydrase IV identified in patients with the RP17 form of retinitis pigmentosa.](#) Rebello G, Ramesar R, Vorster A, Roberts L, Ehrenreich L, Oppon E, Gama D, Bardien S, Greenberg J, Bonapace G, Waheed A, Shah GN, Sly WS. Proc Natl Acad Sci U S A. 2004 Apr 27;101(17):6617-22.

35. [Mutation spectrum and founder chromosomes for the ABCA4 gene in South African patients with Stargardt disease.](#) September AV, Vorster AA, Ramesar RS, Greenberg LJ. Invest Ophthalmol Vis Sci. 2004 Jun;45(6):1705-11.
34. [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.
33. [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. 2003 Aug;64(2):137-41.
32. [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 Dec;24(4):225-32.
31. [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):364-72.
30. "Unusual frequencies of Rhodopsin mutations and polymorphisms in South African patients with Retinitis Pigmentosa." Greenberg J, Roberts L, Ramesar R. Published in "Insights into Retinal Degenerative diseases", a book on the proceedings of the IXth International Symposium on Retinal Degeneration. 2002; 329-333
29. "Migratory History of Populations and its use in Determining Research Direction for Retina Degenerative Disorders." Ramesar R, September A, Rebello MT, Greenberg J, Goliath R. Published in "Insights into Retinal Degenerative diseases", a book on the proceedings of the IXth International Symposium on Retinal Degeneration. 2002; 335-338.
28. [A computer-based register for inherited retinal dystrophies in Southern Africa.](#) Rebello MT, Greenberg LJ, Ramesar RS. Ophthalmic Genet. 2002 Mar;23(1):61-5.
27. [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, Reardon W, Bitoun P, Millan J, Legge R, Friedman TB, Kimberling WJ. Am J Hum Genet. 2002 Aug;71(2):262-75.
26. [Mutations in the pre-mRNA splicing factor gene PRPC8 in autosomal dominant retinitis pigmentosa \(RP13\).](#) McKie AB, McHale JC, Keen TJ, Tarttelin EE, Goliath R, van Lith-Verhoeven JJ, Greenberg J, Ramesar RS, Hoyng CB, Cremers FP, Mackey DA, Bhattacharya SS, Bird AC, Markham AF, Inglehearn CF. Hum Mol Genet. 2001 Jul 15;10(15):1555-62.
25. [Genomic structure and 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, Greenberg J, Ramesar R, Martini A, Moller C, Smith RJ, Sumegi J, Kimberling WJ. Am J Hum Genet. 2000 Apr;66(4):1199-210
24. [Low frequency of rhodopsin mutations in South African patients with autosomal dominant retinitis pigmentosa.](#) Roberts L, Ramesar R, Greenberg J. Clin Genet. 2000 Jul;58(1):77-8.

23. [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, Jacobson SG, Ayuso C, Heckenlively JR, Tamayo M, Gorin MB, Reardon W, Kimberling WJ. *Am J Hum Genet.* 2000 Dec;67(6):1569-74.
22. [Refinement of the RP17 locus for autosomal dominant retinitis pigmentosa, construction of a YAC contig and investigation of the candidate gene retinal fascin](#). Bardien-Kruger S, Greenberg J, Tubb B, Bryan J, Queimado L, Lovett M, Ramesar RS. *Eur J Hum Genet.* 1999 Apr;7(3):332-8.
21. "Ophthalmic Genetics: A review of the molecular genetics of familial retinal dystrophies in Southern Africa." Greenberg J, Rebello G, Ramesar R. *Specialist Medicine.* 1999 Vol. XXI No.2 108-112.
20. [A photoreceptor gene mutation in an indigenous black African family with retinitis pigmentosa identified using a rapid screening approach for common rhodopsin mutations](#). Greenberg J, Franz T, Goliath R, Ramesar R. *S Afr Med J.* 1999 Aug;89(8):877-8.
19. [Alström syndrome: further evidence for linkage to human chromosome 2p13](#). Collin GB, Marshall JD, Boerkoel CF, Levin AV, Weksberg R, Greenberg J, Michaud JL, Naggert JK, Nishina PM. *Hum Genet.* 1999 Nov;105(5):474-9.
18. [Retinitis pigmentosa locus on 17q \(RP17\): fine localization to 17q22 and exclusion of the PDEG and TIMP2 genes](#). Bardien S, Ramesar R, Bhattacharya S, Greenberg J. *Hum Genet.* 1997 Nov;101(1):13-7.
17. [Rhodopsin mutation G109R in a family with autosomal dominant retinitis pigmentosa](#). Goliath R, Bardien S, September A, Martin R, Ramesar R, Greenberg J. *Hum Mutat.* 1998;Suppl 1:S40-1.
16. [Sorsby fundus dystrophy: reevaluation of variable expressivity in patients carrying a TIMP3 founder mutation](#). Felbor U, Benkwitz C, Klein ML, Greenberg J, Gregory CY, Weber BH. *Arch Ophthalmol.* 1997 Dec;115(12):1569-71.
15. "Growth factors in the retina: Pigment epithelium-derived factor (PEDF) now fine mapped to 17p13.3 and tightly linked to the RP13 locus." Greenberg J, Goliath R, Tombran-Tink J, Chader G, Ramesar R. Published in "Degenerative Retinal Diseases", edited by La Vail et al. Plenum Press, New York, 1997; 291-294.
14. "Retinal Blindness in South Africa." Bartmann L. *Nursing News* (1997)
13. [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](#). Goliath R, Tombran-Tink J, Rodriguez IR, Chader G, Ramesar R, Greenberg J. *Mol Vis.* 1996 Jun 19;2:5.
12. [Gene mapping of Usher syndrome type IIa: localization of the gene to a 2.1-cM segment on chromosome 1q41](#). Kimberling WJ, Weston MD, Möller C, van Aarem A, Cremers CW, Sumegi J, Ing PS, Connolly C, Martini A, Milani M, Tamayo ML, Bernal J, Greenberg J, Ayuso C. *Am J Hum Genet.* 1995 Jan;56(1):216-23.
11. [Genetic blindness--macular dystrophies and retinitis pigmentosa](#). Greenberg J, Peters AL. *S Afr Med J.* 1995 Jun;85(6):492-3.

10. [An eighth locus for autosomal dominant retinitis pigmentosa is linked to chromosome 17q.](#) Bardien S, Ebenezer N, Greenberg J, Inglehearn CF, Bartmann L, Goliath R, Beighton P, Ramesar R, Bhattacharya SS. Hum Mol Genet. 1995 Aug;4(8):1459-62.
9. [Fine localization of the locus for autosomal dominant retinitis pigmentosa on chromosome 17p.](#) Goliath R, Shugart Y, Janssens P, Weissenbach J, Beighton P, Ramasar R, Greenberg J. Am J Hum Genet. 1995 Oct;57(4):962-5.
8. [Sorsby's fundus dystrophy. A South African family with a point mutation on the tissue inhibitor of metalloproteinases-3 gene on chromosome 22.](#) Peters AL, Greenberg J. Retina. 1995;15(6):480-5.
7. [Genetic mapping of retinitis pigmentosa--implications for South African patients.](#) Greenberg J, Ramesar R, Beighton P. S Afr Med J. 1994 Jul;84(7):410-2.
6. [A new locus for autosomal dominant retinitis pigmentosa on the short arm of chromosome 17.](#) Greenberg J, Goliath R, Beighton P, Ramesar R. Hum Mol Genet. 1994 Jun;3(6):915-8.
5. [Rod-cone dystrophy, sensorineural deafness, and renal dysfunction: an autosomal recessive syndrome?](#) Beighton P, Bartmann L, Bingham G, Sellars S. Am J Med Genet. 1993 Nov 1;47(6):832-6.
4. [Retinitis pigmentosa in southern Africa.](#) Greenberg J, Bartmann L, Ramesar R, Beighton P. Clin Genet. 1993 Nov;44(5):232-5.
3. [Linkage of Usher syndrome type I gene \(USH1B\) to the long arm of chromosome 11.](#) Kimberling WJ, Möller CG, Davenport S, Priluck IA, Beighton PH, Greenberg J, Reardon W, Weston MD, Kenyon JB, Grunkemeyer JA, Pieke Dahl, Overbeck LD, Blackwood DJ, Brower AM, Hoover DM, Rowland P, Smith RJH. Genomics. 1992 Dec;14(4):988-94.
2. [Retinitis pigmentosa, AD type I: exclusion of linkage to D3S47 \(C17\) in a large South African family of British origin.](#) Greenberg J, Babaya M, Ramesar R, Beighton P. Clin Genet. 1992 Jun;41(6):322-5.
1. [Retinitis pigmentosa in South Africa.](#) Oswald AH, Goldblatt J, Sampson G, Clokie R, Beighton P. S Afr Med J. 1985 Dec 7;68(12):863-6.

ALL POSTGRADUATE STUDENTS & RESEARCH ON THE IRD PROJECT:

1. PhD 1999:

Soraya Bardien Kruger: A molecular investigation of the novel gene underlying autosomal dominant retinitis pigmentosa in a South African family

2. PhD 2000:

Rene Goliath: Towards identifying the ADRP gene in a large South African family with retinitis pigmentosa

3, 4. Honours 2000:

Janine Scholefield: Detection of the Underlying Defect caused by a mutation in a South African Choroideremia family

Maia Matshikiza: Usher Syndrome: Identification of disease-causing loci in some South African families

5. Honours 2001:

Natalie Coutts: Hotspot screening for X-linked Retinitis Pigmentosa

6. Honours 2002:

Dvora Balkin: Determination of the gene responsible for the phenotype in a family with Retinitis Pigmentosa and Friedreich Ataxia

7. Honours 2003:

Maithili Sashindranath: Investigation of mutations in the *USH2A* gene in autosomal recessive Retinitis Pigmentosa

8. PhD 2004:

Alison September: The molecular investigation of Stargardt disease in South Africa

9. Honours 2004:

Surita Marais: Screening of the *Sans* (USH1G) gene for mutations that cause Usher Syndrome type 1G

10. MSc (Genetic Counselling) 2005:

Frieda Basson: A pilot study of how individuals with inherited retinal degenerative disorders perceived being part of a genetic research programme

11. Honours 2005:

Paola Ghignone: The molecular analysis of *GUCY2D* in patients with Leber Congenital Amaurosis

12. MSc 2006:

Lisa Roberts: Mutation analysis of important retinal candidate genes: progressing from research to diagnostic service

13. Honours 2006:

Aisha-Bibi Pandor: Molecular analysis of the *GUCY2D* gene in patients with recessive Retinitis Pigmentosa

14. Honours 2007:

Fiona Baine: Deletion screening of major genes in autosomal dominant Retinitis Pigmentosa

15. Msc 2010:

Christel Nossek: Common ABCA4 mutations in South Africans: frequencies, pathogenicity and genotype-phenotype correlations

16. Honours 2010:

Stephni Venter: Investigation of the clinical spectrum of retinal disease caused by ABCA4 founder mutations

17. Msc 2011:

Maureen Akinyi: Investigation of a genetic variant in the Rhodopsin gene that may act as a modifier of the RDD phenotype

18. Honours 2011:

Iyaloo Mbodo: Genetic susceptibility factors in Age Related Macular Degeneration

19. PhD 2012:

Aisha- Bibi Pandor: An investigation into the molecular mechanisms underlying retinitis pigmentosa 17 with the view to developing novel gene-based therapies

20. Honours 2012:

Taahira Goolam Hoosen: Frequent retinal gene mutations in South Africa and their roles in autosomal recessive retinitis pigmentosa

21. MSc 2016:

Dr. Johann Baard: Genetics of Age-related Macular Degeneration and Stargardt disease in South African populations

22. PhD 2017:

Lisa Roberts: Genetic analysis of inherited retinal diseases in indigenous Southern African populations

23. Honours 2017:

Akshay Vanmali: Analysis of novel candidate genes for inherited retinal diseases

24. MSc Genetic Counselling 2017:

Kalinka Popel: Young adults' perceptions of the implications of their hereditary visual impairment/blindness: A Cape Town based study.

25. MSc Genetic Counselling 2017:

Dr Rene Goliath: Exploration of the impact of genetic counselling and patient support group involvement on retinal degenerative disorders (RDD) patients: A qualitative study

26. MSc Genetic Counselling 2017:

Dr Nicole van der Merwe: Exome sequencing in South Africa: Exploring stakeholder views on feedback of individual research results and incidental findings

TOTAL:

13 Honours Students

4 MSc (Med); 4 MSc (Genetic Counselling)

5 PhD Students