ALL PUBLICATIONS ARISING FROM THE IRD PROJECT

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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.

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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

27. Honours 2018:
Nicole Midgley: Investigation of a hypomorphic variant of the ABCA4 gene in South Africans with inherited retinal disease

28. Honours 2019:
Buhle Ntozini: Investigation of causal mutations in the ABCA4 gene in a cohort of South Africans with Stargardt Disease

29. PhD 2019:
Stacey Moses: Towards identifying the precise molecular pathogenesis underlying the disease phenotype in individuals carrying a mutation in the spliceosomal factor PRPf8

30. Honours 2020:
Lara Holtes: An investigation of deep intronic variants in ABCA4, CEP290 and USH2A underlying Inherited Retinal Diseases in a cohort of South African patients

31. Honours 2021:
Indiana van Rensburg: An investigation of specific mutations underlying autosomal recessive inherited retinal disease in cohorts of South African patients

TOTAL:
17 Honours Students
4 MSc (Med); 4 MSc (Genetic Counselling)
6 PhD Students