ALL PUBLICATIONS ARISING FROM THE IRD PROJECT


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


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:
Taahirra 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