



RESEARCH AND ACADEMIC PROGRESS IN GENETIC COUNSELLING AT THE UNIVERSITY OF CAPE TOWN

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# RESEARCH PROJECTS

# <u>2011:</u>

• Zandre Bruwer (now Laing) received her PhD for her study entitled: "An investigation into the psychosocial factors which have an impact on access to and utilisation of the mobile endoscopic surveillance service offered to high-risk members of known Lynch families identified through predictive testing."

### <u>2010:</u>

- Nakita Verkijk (now Laing) graduated <u>with distinction</u> with a MSc in Human Genetics following her research project entitled: "An investigation into the reasons for non-uptake of carrier testing in a family affected by alpha thalassaemia X-linked mental retardation (ATR-X) syndrome."
- Chantelle Scott received her MSc in Genetic Counselling with the research project entitled: "An investigation into the experiences of parents with a preschool child with Down syndrome"

### <u>2009:</u>

• Gabriele Solomon graduated <u>with distinction</u> with a MSc in Human Genetics following her study on *"Develop strategies to explain genetic terminology to Xhosa speaking lay persons."* This study was part of a national NRF funded study on linguistics (Health communication project) (PI: C Penn and J Greenberg listed as a collaborator).

## <u>2007:</u>

- UCT awarded funding for 3 years by SANPAD for a project entitled "Investigating communication and cultural practices in genetic counselling sessions in SA"
- The MRC awarded UCT funding for a project around "Transcending cross-cultural and crosslinguistic barriers: the challenges of communication practices in genetic counselling in SA."
- Mardelle Schoeman was awarded her MSc in genetic counselling for the project addressed "An investigation into the level of genetic knowledge and family communication about genetic risk in parents of children with Cystic Fibrosis".
- Zandre Bruwer/Laing was awarded her MSc in genetic counselling with the project addressed: "Hereditary Nonpolyposis Colorectal Cancer: Comprehension of a cancer risk in conjunction with a genetic risk."
- Dr Shobana Kubendran was a MSc student at the University of South Carolina, USA who undertook a qualitative research project entitled "Genetic Counseling Professionals in Eleven Countries: Emergence, Practice & Current Issues." Together with Professor Janice Edwards from the University of South Carolina, USA and Dr Shobana Kubendran, Jacquie Greenberg was involved with this qualitative research.

### 2006:

• Together with **Professor Claire Penn** from Wits, UCT was funded from the NRF for a genetic counselling project on *"language and communication at UCT"* (Haemophilia genetic communication

project).

• Kelly Loggenberg received her MSc in Genetic counselling with the project entitled: "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."

### <u>2005:</u>

- Brenda Kruger received her MSc in Genetic counselling with the project entitled: *"Hereditary Nonpolyposis Colorectal Cancer: Factors Contributing to Adherence and Non–adherence to Surveillance for Mutation Carriers in Rural Areas of the Northern and Western Cape."*
- Frieda Basson/Loubser received her MSc in Genetic counselling with the project entitled: "*How individuals and families with Inherited Retinal Degenerative Disorders perceive being part of a genetic research programme.*"

# PRESENTATIONS

## <u>2011:</u>

<u>Oral Presentation</u> at the Joint Conference of the South African and African Societies of Human Genetics: 6-9 March 2011, CAPE TOWN.

• Regular colonoscopic surveillance reduces risk of colon cancer by fifty percent in Lynch Syndrome: Why patients do not heed this advice? <u>Z Bruwer</u>, M Futter, R Ramesar.

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

- BRCA1 and BRCA2 predictive testing: Case report of two double heterozygotes present in an Afrikaner family and implications for diagnostic testing. <u>F Loubser</u>, JNP de Villiers, NC van der Merwe.
- Albinism: Attitudes and Stigmatisation in a Local South African Population. <u>C Lewis</u>, F Loubser, L Stakes.
- Experiences of parents with a child with Down syndrome in the Western Cape. <u>C Scott</u>, M Futter, A Wonkam.
- Determining the effect of Modifier Genes on the survival period until diagnosis of Lynch syndrome. <u>N</u> <u>Verkijk</u>, A Vorster, S Owens and R Ramesar.
- The Phenotype of Juvenile Huntington Disease in the Western Cape. <u>M Schoeman</u>, K Fieggen, J Heckman, R van Toorn, G Riordan, M Urban.

<u>Poster presented</u> at the third Biennial meeting of the Transnational Alliance of Genetic Counsellors (TAGC): 11 October, 2011, MCGILL UNIVERSITY, MONTREAL, CANADA. (<u>http://tagc.med.sc.edu/montreal11.asp</u>)

• Tina Wessels (Wits), Jennifer Kromberg(Wits) and <u>Jacquie Greenberg</u>(UCT) *Genetic Counselling Training in South Africa.* 

### <u>2009:</u>

<u>Oral presentation</u> at the thirteenth congress of the SA Society of Human Genetics SASHG: April 2009, SPIER, STELLENBOSCH, WESTERN CAPE:

• Delivery of an ophthalmic genetic service including a telephone counselling model: a bench-to-bedside

review. J Greenberg, LJ Roberts, M Schoeman, Z Bruwer, K Loggenberg, F Loubser.

Poster presentations at the thirteenth congress of the SA Society of Human Genetics SASHG: April 2009, SPIER, STELLENBOSCH, WESTERN CAPE:

- Personal understanding of predictive test result in South African nonpolyposis colorectal cancer families. <u>Z Bruwer</u>. R Ramesar. J Greenberg. M Futter
- Genetic counselling experiences in delivery of genetic research results to patients affected with ABCA4 associated retinal degenerative disorders. <u>F Loubser</u>, K Loggenberg, L Roberts, LJ Greenberg

### 2008:

Poster presentation at the School of Child & Adolescent Health Annual Research Day: 21st & 22nd October, 2008 RED CROSS CHILDREN'S HOSPITAL, CAPE TOWN

The Impact of level of Genetic Knowledge on Reproductive Choices and Risk Communication in Cystic Fibrosis Families." <u>M Schoeman</u>, M Futter, J Greenberg

### <u>2007:</u>

<u>Oral presentation</u> at the third International Conference on Birth Defects and Disabilities in the Developing World: June 17-21, 2007, RIO DE JANEIRO, BRAZIL

• Exploring Patient Perceptions of Participating in a Genetic Research Programme in South Africa using Qualitative Methods. <u>F Basson</u>, M Futter , J Greenberg

<u>Oral presentation</u> at the twelfth congress of the South African Society of Human Genetics: March 2007, GOLDEN GATE HIGHLANDS NATIONAL PARK

• An Investigation into the Level of Genetic Knowledge of Parents of Sons with Duchenne Muscular Dystrophy. <u>K Loggenberg</u>, M Futter, J Greenberg.

Poster presentation at the annual Congress of the European Society of Human Genetics: June 2007, NICE, FRANCE

• A qualitative investigation of the impact of DMD on South African families. <u>K Loggenberg</u>, M Futter, J Greenberg.

## <u>2005:</u>

<u>Presentation</u> at the eleventh congress of the SA Society of Human Genetics SASHG: <u>2005</u>, 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.

### <u>2004:</u>

Poster presentation at the twenty-fifth Annual Meeting of the Society of Genetic Counsellors: October 2004, WASHINGTON

• THE GREAT DIVIDE: The challenge to genetic counsellors of maintaining ethical standards of practice in previously disadvantaged and under-resourced communities in South Africa. <u>PA</u> Craig, U Algar, R Felix, G Pietersen, A Vorster, R Hendriks, PA Goldberg, RS Ramesar.

<u>Poster presentation</u> at the first International Symposium of the National Neurovision Research Institute on translation of clinical and molecular research for inherited retinal disease therapies: November 2004,

• The importance of counselling in the delivery of genetic results to patients and families with retinal degenerative disorders in southern Africa : from lab to life. J. Greenberg, A. Peters, R. Ramesar.

Seminar given for Professor Janice Edwards, Director, Genetic Counselling Program, University of South Carolina School of Medicine and for Professor Barbara Biesecker at the NIH in Washington: November 2004

• Championing change and challenges facing genetic counsellors in Southern Africa. J Greenberg.