This has been a wonderful year for our society, affirming the advances and vibrant activity in human genetics in South Africa and worldwide! The year started with a triple bill of meetings in Cape Town in March. The first was the Human Heredity and Health in Africa (H3Africa) Congress hosted by the NIH and the Wellcome Trust and which brought with it leaders in the field of Human Genetics. This meeting complemented the Joint AfSHG and SASHG Congress which followed. Congratulations to Raj Ramesar and his team for ensuring an international array of scientists who presented truly inspiring research! The day we shared with bioinformatics and computational biology brought a new dimension to the deliberations of our Society, creating a partnership that I hope we can build on into the future. The Young Researchers Forum provided a platform for our talented students and postdocs to showcase their research. The ten days of events was exciting and energizing and a truly wonderful opportunity for South African researchers to meet their counterparts from across the continent.

On a national level, we continue to be concerned about the commitment to providing human genetics services and the dwindling number of state-funded positions in Clinical Genetics and Genetic Counselling, as well as for Medical Scientists. Despite the challenges, our Clinical Geneticists have formed a group who meet every six months to discuss interesting cases and to share knowledge and strategies with one another. I would like to congratulate our Genetic Counsellors who have been flying the flag internationally and who have secured reciprocity for South African Genetic Counsellors with Australia and the UK.

It was a privilege to elect three members of our society as Life Members and to convey to them the enormous pride we have in celebrating their exceptional contributions to Human Genetics in South Africa. We congratulate Geertjie de Yong (University of Stellenbosch), Valerie Corfield (University of Stellenbosch) and Tony Lane (Wits and NHLS). We also had great pleasure in congratulating Bill Winship on being awarded the Oder of the Baobab by the President, for his services in Human Genetics to the people of our country. Bill passed away earlier this year and that is a great sadness to all of us who got to know him so well over many years. We celebrate his optimism, amazing work ethic, selfless service, great and inspiring teaching sessions and his wonderful humour. He is sadly missed.

We are looking forward to making a difference in Human Genetics in Southern Africa in 2012. The SASHG committee has some great ideas which we will soon share with you. Please have a look at the profiles of the committee and we hope to soon have much more information on our members on the webpage. Looking even further to the future – the 15th SASHG Congress is planned for 2013, and will be hosted in Johannesburg. We will be posting updates on our website as the plans progress.

I wish you a joyous festive season and time to rest and then gather your energy into galvanizing your human genetics activities and research agendas for 2012.

Yours in genetics

Michèle Ramsay
Division of Human Genetics, NHLS & Wits
Meet your new SASHG committee

The Biennial General Meeting was held this year at the Joint AfSHG/SASHG Conference in Cape Town, where the new SASHG committee for the 2011-2013 term was announced.

The committee meets (via teleconference) three to four times a year, to discuss matters that are pertinent to Human Genetics practice and research in South Africa. Please do not hesitate to contact any of the committee members if there are matters you would want raised at these meetings.

The new committee members are:

Prof Michèle Ramsay – Chair
Division of Human Genetics, NHLS & Wits
Email: michele.ramsay@nhls.ac.za

Research interests:
- Genetic basis and molecular epidemiology of single gene disorders in SA populations.
- The role of genetic and epigenetic variation in the molecular developmental aetiology of foetal alcohol spectrum disorders (FASD) and other diseases exacerbated by adverse lifestyle choices (including obesity as a risk factor for diabetes and cardiovascular disease).

Skills/areas of expertise:
- Molecular Genetics and Genomics (with a bit of Bioinformatics).

Dr Zané Lombard – Secretary and Treasurer
Division of Human Genetics, NHLS & Wits
Email: zane.lombard@wits.ac.za

Research interests:
- Genetics of complex disease, in particular non-communicable disease.
- Trans-generational epigenetic effects.
- Ethical issues in human genomics research in developing countries.
- Genetic epidemiology.

Skills/areas of expertise:
- Genotyping, study design and association testing.
- Bioinformatics.

Dr Collet Dandara
Division of Human Genetics, UCT
Email: Collet.Dandara@uct.ac.za

Research interests:
- Pharmacogenetics, –genomics and drug metabolism;
- Cancer genetics;
- Human genetic variation.

Skills/areas of expertise:
- Molecular genetics techniques.
- Statistics to medium level.
- Gene sequence analyses.
- Phenotyping assays.

Prof Jacquie Greenberg
– Representative: Genetic Counselling
Division of Human Genetics, UCT.
Email: jacquie.greenberg@uct.ac.za

Research interests:
- The inherited Ataxias.
- Huntington Disease in SA.
- Inherited Retinal Degenerative Disorders in SA.
- Genetic Counselling in and for Africa.
- The development of gene-based therapies for the above: especially allele-specific gene-silencing and siRNA.
- Establishing induced pluripotent stem cells as a "disease in a dish" model.

Skills/areas of expertise:
- Linkage and Gene mapping.
- Translational research for Mendelian Genetic Disorders; all of the above which have already moved from “bench to bedside”.
- Ethics and Genetic Testing.
- Programme co-coordinator for one of only 2 MSc in Genetic Counselling Programmes in SA.
- Student and Staff Mentorship.

Dr Lindsay Lambie
– Representative: Clinical Genetics
Division of Human Genetics, NHLS & Wits
Email: lindsay.lambie@nhls.ac.za

Research interests:
- ARPKD in the Afrikaans population.
- Constitutional submicroscopic chromosome imbalances.

Skills/areas of expertise:
- Clinician.
- Main interests dysmorphology, cytogenetics (specifically molecular cytogenetics in the clinical setting) and undergraduate education.

Ms Lisa Roberts
Division of Human Genetics, UCT
Email: lisa.roberts@uct.ac.za

Research interests:
- Inherited retinal degenerative diseases.

Skills/areas of expertise:
- Mutation detection and pathogenicity analysis, translational research.

Prof Magda Theron
Division of Human Genetics, NHLS & UOFS
Email: TheronM1@ufs.ac.za
Prof Philip Venter  
Department of Medical Sciences, UL  
Email: philip.venter@ul.ac.za  
**Research interests:**  
- Community genetic services in rural areas.  
- Develop genomics/bioinformatics at the University of Limpopo and make this technology available and accessible to disadvantaged communities.  
- The genetics/genomics of prostate cancer in several South African ethnic groups.  
**Skills/ areas of expertise:**  
- Trained cytogeneticist.  
- Director of School of Health Sciences, UL for 8 ½ years.  
- Part of the group who sequenced the full genome of the first African, Archbishop Desmond Tutu.

Dr Soraya Bardien  
Molecular Biology and Human Genetics, US  
Email: sbardien@sun.ac.za  
**Research interests:**  
- Parkinson’s disease & other inherited neurodegenerative disorders.  
**Skills/ areas of expertise:**  
- Mutation screening  
- MLPA  
- Will be involved in whole-exome sequencing.

Prof Raj Ramesar  
– Representative: African Society of Human Genetics  
Email: Raj.Ramesar@uct.ac.za  
**Research interests:**  
- Using the exciting developments in the field of genomic sciences to investigate human biodiversity.  
- Genetics of familial colorectal cancers  
**Skills/ areas of expertise:**  
- Head of the Division of Human Genetics, UCT.  
- Large scale research into human diversity and health.  
- Teaching and training.

Dr Thirona Naicker  
Division of Medical Genetics, Inkosi Albert Luthuli Central Hospital, UKZN  
Email: thironanaicker@gmail.com  
**Research interests:**  
- Haemophilia carrier mothers.  
**Skills/ areas of expertise:**  
- Paediatrician with interest in Genetics.

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**REPORT BACK: 2011 AfSHG/SASHG Joint International Conference**

**Raj Ramesar**  
Chair of the local organizing committee  
Division of Human Genetics, UCT

This year the African Society for Human Genetics (AfSHG) and the Southern African Society for Human Genetics (SASHG) co-hosted the 2011 joint AfSHG and SASHG Conference in Cape Town from the 6th to the 9th of March, 2011. The objective was to bring together foreign scientists working in Africa, institutions with an interest in Africa and African investigators to South Africa to present information about patient populations, databases and technologies that would generate collaborative research.

The meeting was the culmination of a lengthy process involving bidding by various countries in Africa, the awarding of the bid to Cape Town (through Professor Ramesar at the University of Cape Town), extensive planning, fund raising and then materialising the conference at the Cape Town International Convention Centre, Cape Town. Although the Convention Centre was a relatively expensive option, the local organising committee chose this venue because it showcased Africa at its best in terms of coordinating a large event.

The Conference was further enhanced by the Planning meeting of the Human Heredity and Health: Africa (H3Africa) conference held on the 4th and 5th of March, also in Cape Town. H3Africa is a joint initiative of the African Society of Human Genetics, the National Institutes of Health (USA), and the Wellcome Trust, UK (http://www.h3africa.org/). Our organisation of this conference also attracted the African Bioinformatics Society to organise their Conference in a back to back fashion with us, also at the convention Centre. This series of events meant that African (and other) researchers could maximise their ‘returns’ out of investing in a single trip to Cape Town.

The meeting attracted an unprecedented 463 delegates, and also a spectacular list of Invited and Keynote speakers, including Nobel Laureate, Dr Sydney Brenner, Dr David Bentley Scientific Director of Illumina (UK) and Dr Vanessa Hayes (author of the recent paper published on the first complete DNA sequences of peoples from Southern Africa), to name but a few.
Possibly most importantly was the organisation of the Young Researchers Forum, which was organised by two postgraduate students from the Division of Human Genetics at UCT, Ms Aisha-bibi Pandor, and Ms Kusha Kalideen. The objective was to create a vibrant network of young researchers and an opportunity for them to interact for a full day together. Aisha-bibi and Kusha’s report on this day can be read below.

**REPORT BACK: 2011 Young Researchers Forum (YRF)**

_Aisha Pandor & Kusha Kalideen_

_Co-chairs, YRF organizing committee_

**Division of Human Genetics, UCT**

The Young Researchers’ Forum (YRF) was originally borne from the Division of Human Genetics at the University of Cape Town, under the auspices of the African Society of Human Genetics (AfSHG) and our first meeting was held as a satellite of the Joint International Conference of the African and South African Societies of Human Genetics (Cape Town, March 2011).

As doctoral students ourselves, we recognised the crucial stage in their careers that post-graduate and post-doctoral students are at, and we wanted to create a space and a place where students could voice our opinions, share ideas and network among our peers. We believed that these relationships and networks developed amongst young African researchers would be important for sustained collaborative research in the genomics era, particularly on the African continent.

Organising the YRF was interesting, stressful and challenging, but most of all rewarding. We were privileged to have surrounded ourselves with an incredible team who performed their duties with aplomb. Fundraising and logistics of the event became our “new” projects – from which we learnt enormously. In total, thanks to our generous sponsors (The University of Cape Town, Whitehead Scientific, Inqaba Biotech, Roche, Tullow Oil, IAPO, Anatech, Lancet Laboratories, and MDS) the YRF managed to raise substantial funds, which were spent on catering, transport and prizes to ensure we could host a free event for all of our attendees.

After sleepless nights hoping for the event to be a success, on Saturday the 5th March 2011 the inaugural YRF hosted 154 attendees at the forum representing 10 countries worldwide (South Africa, Zimbabwe, Sudan, Egypt, Cameroon, United Kingdom, Italy, United States of America, Kenya, Malawi).

Our programme consisted of a full day of activities for Human Genetics and Bioinformatics students from Africa and beyond, including distinguished plenary speakers, student presentations and posters. The first of three plenary speakers, Prof Charles Rotimi (Center for Research on Genomics and Global Health at the National Institutes of Health) presented his personal story, from Africa to the world, highlighting the challenges that have faced African researchers in the past. He also spoke of a shift, with improving academic institutions, collaborations between African and international institutions, and training programmes between African and external institutions making remaining to study on this continent more and more attractive.
all to aim high. Rounding up the plenary speakers, Dr Roger Glass (Director: Fogarty International Centre) provided inspirational leadership in addressing how circumstances have changed in the networked world and mentioning opportunities for strengthening the relationship of emerging countries as players on the global stage. Dr Glass spoke of the challenges that have faced Global Health researchers in the past, gave an outline of the Fogarty program, and spoke of their commitment to investing in researchers from emerging countries through various fellowships internationally. He capped off his presentation with some great tips for young scientists in gaining international exposure. A spitbraai and social concluded our inaugural YRF, where we handed out prizes for the best presenter (Ms Fiona Baine, UCT, South Africa), and the top three posters (1: Mr JJ Noubiap, Cameroon; 2. Dr Jibril Hirbo, USA, 3. Dr Shahida Moosa, Wits, South Africa).

We have received excellent feedback from students who attended the event, as well as senior researchers who had students attending, or popped in to see first-hand what had been organised. We believe that this first event hosted by the YRF meeting was a huge success and are planning more events for 2012. We hope that this initiative will continue to forge bonds between young researchers on the continent, and look forward to seeing the YRF grow from strength to strength and become a mainstay at all AfSHG and SASHG meetings in the future.

**SASHG members receive bursaries to attend AfSHG/SASHG conference**

*Zané Lombard*  
*Division of Human Genetics, NHLS & Wits*

One of the benefits of the SASHG membership for full-time students is being eligible for a bursary to attend the biennial SASHG meeting. This year the SASHG committee was fortunate enough to have sufficient funds to sponsor 20 students to attend the conference.

The recipients were:

- Chanell Herfurth (North West University).
- Natalie Strickland, Brit Drogemoller, Carola Rohrich, Enid Nieuwoudt, Galen Wright, Marika Bosman, Martmari Botha and Nadia Carstens (Stellenbosch University).
- Danielle Berkowitz, Lauren Watson, Marelize Swart and Nakita Verkijk (University of Cape Town).
- Tyren Dodgen (University of Pretoria).
- Andrew May, Kenneth Davison, Sanam Patel, Shatadi Masemola, Tasha Wainstein and Pareen Patel (University of the Witwatersrand).

We hope these students had a wonderful and stimulating time at the conference!

**International Congress of Human Genetics – Montreal, Canada**

*Shahida Moosa*  
*Division of Human Genetics, NHLS & Wits*

The ICHG takes place only every five years, and I could thus not let the opportunity pass me by to attend the congress in Montreal! It was my first trip to Canada, and I could find no better way to attend the largest meeting of the international Human Genetics community, and simultaneously be afforded the opportunity to explore an exciting city and meet up with old friends.

**Shahida and a cosmopolitan view of Montreal.**
The congress provided an excellent platform to learn more about how the newest molecular techniques and intricate basic biological constructs are instrumental to our increasing understanding of clinical genetics. I came away from the congress with a greater appreciation for the importance of close collaborative interaction between the laboratory and the clinic: a relationship, which I view as vital to the improvement of our practice of medical genetics and ultimately, the service we provide to our patients.

Many people came to view my poster specifically because I was from Africa! People are interested in Africa, and what we do here. It is important to use the plentiful opportunities to network with peers and seniors, so that avenues for closer interaction and collaboration can be explored. I was fortunate to be able to take part in many of the trainee events, which catered specifically to the fostering of bonds between young researchers and clinicians from all over the world.

Attending the ICHG in Montreal was a wonderful experience, full of intense learning opportunities in a beautiful and vibrant environment. The social highlight of the trip was climbing Mount Royal with my friend Isabelle, who is a clinical geneticist from Montreal! The picture shows us at the top of the mountain, overlooking the wonderful city. Au revoir Montreal, I hope to visit again soon!

The Transnational alliance for Genetic Counselling (TAGC)

Jacquie Greenberg
Division of Human Genetics, UCT
Tina-Marie Wessels
Division of Human Genetics, NHLS & Wits

The Transnational alliance for Genetic Counselling (TAGC) is a global partnership of genetic counselling practitioners and educators whose mission is to enhance international communication, collaboration, and educational program development. The TAGC met in Montreal on October 10-11, 2011 immediately preceding the 12th International Congress of Human Genetics (ICHG). This was the third Biennial meeting of TAGC which was chaired once again by Professor Janice Edwards, the founding chair. The programme included a plenary and poster session as well as workshops.

As this was a relatively small meeting (46 registrants from 12 countries) there was much interaction and discussion both during the meeting and, in particular, around the ~15 posters and our SA programmes are similar to and align favourably with many others from around the world.

One of the main topics for discussion was the credentialing and certification process in different countries. Currently there are six countries with well-established certification for Genetic Counsellors - USA, UK, Australia, Canada, Japan and South Africa. The TAGC has been instrumental in facilitating reciprocity agreements regarding certification between three of these countries - the UK, Australia, and after the 2011 meeting, South Africa is now also included. We believe that this is a major achievement for our fledgling profession both nationally and internationally.

Update on the Southern African Human Genome Programme (SAHGP)

Michael Pepper
Department of Immunology, UP

As 2011 draws rapidly to a close, we can look back on an excellent start for the SAHGP. The SAHGP was successfully launched on 27 and 28 January at the Irene Country Lodge. Almost 60 people attended from multiple disciplines and institutions. A commitment was made by all present at the launch to work together towards the preparation of a detailed report for the Dept. of Science and Technology (DST). A 78 page report (with several attachments) was submitted to the DST in July, and was the culmination of a tremendous effort from 5 national writing groups and the skilful piecing together the information by Hugh Napier. After several months of reflection and discussion, the DST requested the preparation of an interim proposal, submitted in October, the purpose of which is to ensure interim funding which will (a) allow the project to maintain its current momentum while the final business plan is being prepared (deadline April 2012), and (b) allow the SAHGP to start sequencing its first full southern African genomes in the first half of 2012.

This year’s SAHGP activities culminated in November with a highly successful SASTA/PUB Critical Thinkers Forum held at the Baxter Theatre in Cape Town. The event attracted people from across the country from many disciplines, and was the first occasion at which ethical, legal, social/community and educational issues were discussed in an open forum. The take-home messages from the forum were (a) the necessity to obtain buy-in from a broad base which includes both the scientific and non-
scientific communities; (b) the unique opportunity that the SAHGP has created to address educational issues from primary school upwards; (c) the strong desire from religious and other groups to be intimately involved in the roll-out of the SAHGP; and (d) the willingness of everyone concerned to become involved in ensuring the success of this unique opportunity. In this context it would perhaps be worthwhile to recall the words of Brutus in Shakespeare’s Julius Caesar:

There is a tide in the affairs of men.
Which, taken at the flood, leads on to fortune;
Omitted, all the voyage of their life
Is bound in shallows and in miseries.
On such a full sea are we now afloat,
And we must take the current when it serves,
Or lose our ventures.

Several funding proposals were submitted this year on behalf of the SAHGP and these include:

1. The SAHGP report which will form the basis of the SAHGP business plan to be finalized by April 2012
2. An interim proposal (detailed above) on which a decision from the DST is imminent
3. An application to SANBio (part of NEPAD) on which a decision has been deferred to September 2012
4. A submission by the DST on behalf of the SAHGP for additional funding.

2012 will be a critical and exciting year for the Programme, in which, as mentioned above, we will sequence our first full southern African human genomes. The data derived therefrom will be used to host a national event at which interested parties will have an opportunity to learn how to approach and analyze the large amount of information generated. It is perhaps pertinent to recall the aims of the SAHGP, all of which will receive attention during the course of the New Year:

1. The development of capacity for genomic research in southern Africa
2. The establishment of sustainable resources for genomic research (including a regional biorepository and a genomics database)
3. The translation of knowledge and information into improvements in human health in the region

Chapter 8 of the NHA, which deals with what can broadly be classified as human tissues, has to date not been enacted, despite the fact that the NHA itself was promulgated in 2003. The consequences of this regulatory vacuum are (a) the existence of legislation that dates back to 1983 (Human Tissues Act) that covers a limited number of aspects pertaining to human tissues, and (b) a complete lack of legislation in other critical areas which include assisted reproductive technology (including IVF), cell-based therapy (including stem cells), transplantation, tissue banks and genetic services.

A national working group has, in response to a request from the Dept. of Health (DoH), submitted a revised Chapter 8 which addresses most of the unlegislated areas listed above. The revisions are currently being finalized and it is anticipated that a revised Chapter 8 will be submitted to Parliament in the course of 2012. Several sets of regulations pertaining to Chapter 8 were published for public comment in the Government Gazette on 01 April 2011. The comments received by the DoH are being integrated into the draft regulations, and once the revised Chapter 8 has been accepted the regulations will be aligned thereto.

2012 should therefore see the promulgation of a revised and updated Chapter 8 and its regulations. This will provide a great deal of relief to all who are currently affected by the legislative vacuum.

A fond farewell: Prof Bill Winship

We remember and celebrate the life of Professor Bill Winship. It is an honour and a privilege to have known and worked with him – he was a man of great integrity and had a phenomenal work ethic. Prof Winship was a most gracious and humble individual with a deepest respect for people –he did not box’ individuals into categories such as race religion and creed. He saw the good in all and valued the input of others. An
example of his humility and graciousness is when I became the Head of Paediatrics: here, a man who was my father’s age would introduce me as ‘my head of Department’!

He was always kind and gentle. Whatever he undertook, he made an impact, was passionate about it and made it work!! He was an astute clinician and academic. He made a complex problem simple and was an outstanding teacher.

He was appointed to run the children’s outpatients at King Edward VIIIth Hospital in 1963, and established the first children’s heart clinic at King Edward and at Wentworth Hospital, and commenced what we call interventional cardiology. As the team developed at Medical School and KEH in 1972 -1987 Prof Bill was requested to take over the services for children at the Addington Children’s Hospital. He established it as an academic unit, developed the newborn services and once more I had the privilege of working with him.

He undertook many other aspects of child care but then developed the Genetic services with the national Human Genetics Group. He developed and trained a consultant to undertake what was required in this field of care. Despite a number of obstacles we carried on with the services. In the last few years of his life he worked with me to establish what we call step down care for children. The Addington Children Hospital has been ear-marked for that service. One team of health professionals will continue working with genetics, although it will be much more difficult without the guidance he provided. Another team will work for the children’s hospital and hopefully realize that dream.

He recently received, from the President, the Order of the Baobab for his contribution to development of services for children.

Sonia and the Winship family, we cherish the time we have had with Professor Winship. He was a man of great warmth and character. He was a man that respected everybody, and held his own with people irrespective of age. We mourn the passing of this wonderful being that has left a huge void in a personal and professional capacity for those that had the privilege of meeting Prof Winship.

PLEASE MARK YOUR CALENDARS!
**SASHG CONFERENCE 2013**

The 2013 SASHG Conference will take place in: **Johannesburg, October 6-9, 2013**.

The theme will be "Genetworking: Bases building bridges".
Further information, including the sub-themes and pre-conference workshops, will follow early in the New Year.

Best Wishes for a well-deserved break over the Festive Season!

Please travel safely.
Looking forward to an exciting 2012!