Letter from the SASHG Chair

- Amanda Krause

I am honoured to have been elected as the new Chairperson of SASHG. As can be seen from the newsletter, the previous Chairperson and Committee have been extremely active. I will do my best, with the new Committee, to continue to keep SASHG moving forwards at the rate that it has been.

Our biggest immediate challenge for 2014 will be to try to ensure the continuity of medical genetics service in the country. Medical geneticist and counsellor posts are desperately needed in all the provinces, so that we can serve patients as well as continue to train. Efforts are being made to meet with the Director-General of Health as soon a possible to strengthen our communication links.

The new committee plans to meet in early 2014 and will then set goals for our two-year term. I would appeal to the SASHG members to e-mail me or any members of the Committee with issues that they feel SASHG should be looking at.

Best wishes for 2014!

Contact details:

**SASHG Chair: Prof Amanda Krause**

✉ amanda.krause@nhls.ac.za

📞 011 489 9219
Letter from the outgoing Chair

- Michèle Ramsay

It has been an honour and pleasure to serve as Chair of the Society from April 2009 to October 2013. I extend my sincere gratitude to the committee of 2011-2013 (Zane Lombard, Lindsay Lambie, Jacquie Greenberg, Collet Dandara, Lisa Roberts, Soraya Bardien, Philip Venter, Raj Ramesar, Elizna Schoeman, Magda Theron and Thirona Naicker for their dedication, commitment and hard work on behalf of SASHG. I have handed over the reins to Amanda Krause and wish her and the next Committee strength during their term. I know that they will embrace the opportunities and challenges to make a difference for the Human Genetics community in South Africa. The SASHG is a small society but it plays an important role to promote and advance the practice and science of human genetics in Southern Africa.

The Committee has been busy, focusing on topics such as direct to consumer marketing of genetics tests in South Africa (see page 15) and the DNA project and related legislation (see page 13). Some other highlights include:

What is in a name?

We have revisited the issue of nomenclature of South African populations in scientific publications and have developed a guideline document. Over the past 12 months, while we have debated the subject and requested expert opinion, we have also got new insight. We are mindful to guard against emphasizing a political perspective (white, black, Coloured and Indian), but rather to promote an ethno-linguistic and cultural sensitivity and a biological reality. With high-throughput genomic studies, we are able to examine biological ancestry with greater accuracy and also to use this in our analyses to get better insight. The principle should always be to provide an accurate and detailed description of the study participant groups in the context of the research question.

SASHG Focus groups

The SASHG Committee has three sub-groups who are represented on the committee and provide regular updates on their activities. They are the Clinical Geneticists, The Genetic Counsellors and the Human Genetics postdoctoral fellows. The young members of the Society represent our future and we wish to encourage and nurture their careers in Human Genetics. Please do not hesitate to contact the new committee to take an active part in SASHG.

We all remain extremely concerned about the state of Medical Genetics Services in South Africa. The WHO suggests that a country needs at least one Medical Geneticist and two Genetic Counsellors for every million of the population and clearly we fall far short of this recommendation. Over the last two years our numbers have dwindled yet further. We now have closer communication with the Department of Health through our contact with the Director General, Ms Precious Matsoso, and Dr Nat Khoale and hope to make a positive impact soon.

SASHG membership has increased, with 193 members in good standing. We distribute information to over 300 people through our mailing list and I would like to thank Lisa Roberts and Jacquie Greenberg for managing the process. I would like to thank Zané Lombard for putting together the annual newsletter to the SASHG community. It serves also as a Chronicle of our Society’s History.

Thank you to everyone who contributed to the 15th Biennial SASHG Conference! Modern science is about collaboration and it is about working together effectively. My advice to you is this: Think carefully and strategically about who you wish to collaborate with, how you wish to do it but most importantly DO IT! Opportunities abound.

Wishing you a replenishing break over the festive season!
At the recent Biennial General Meeting of the Southern African Society for Human Genetics (SASHG), a new committee was voted in to serve for the 2013 -2015 term. The committee is tasked with guiding the practice of Human Genetics in southern Africa in both research and service provision. The committee intends to build upon the momentum of the previous committee with regards to putting southern Africa on the Human Genetics world map while at the same time ensuring the current service provision crisis locally, comes to an end.

The committee meets three times a year and is happy to accommodate input from all SASHG members, so please feel free to make contact if you wish to have any matters raised at these meetings.

If you would like to circulate information to the SASHG community, please email Lisa Roberts (lisa.roberts@uct.ac.za). The new committee members are:
**Professor Amanda Krause | Chairperson | Amanda.Krause@nhls.ac.za**

Position: Head of the Clinical & Counselling Section, Division of Human Genetics, NHLS & Wits

Research Interests: Understanding genetic disease in South Africa in order to provide patients with appropriate genetic testing and care; Huntington’s disease; Fanconi anaemia; spinal muscular atrophy; haemoglobinopathies; aspects of service delivery for black South Africans, particularly in cancer genetics

**Professor Soraya Bardien-Kruger | sbardien@sun.ac.za**

Position: Associate Professor, Molecular Biology and Human Genetics Department, Stellenbosch University

Research Interests: Genetic aetiology and disease mechanisms underlying Parkinson's disease

**Dr Engela Honey | SASHG2015 Representative | engela.honey@up.ac.za**

Position: Senior specialist and lecturer, Department of Genetics, University of Pretoria

Research Interests: Clinical genetics; dysmorphology; cleft lip and palate

**Mrs Noeline Kinsley | Genetic Counselling Representative | noelene@geneticcounselling.co.za**

Position: Genetic Counsellor in private practice; co-owner of www.geneticcounselling.co.za

Research Interests: Genetic counselling; genetic counselling services in South Africa; prenatal testing; insurance and genetics; direct-to-consumer genetic testing; ophthalmology and genetics

**Dr Lindsay Lambie | Clinical Genetics Representative | Lindsay.Lambie@nhls.ac.za**

Position: Clinical Geneticist, Clinical & Counselling Section, Division of Human Genetics, NHLS & Wits

Research Interests: Clinical research

**Dr Zané Lombard | Treasurer | zane.lombard@wits.ac.za**

Position: Senior Lecturer, Wits Bioinformatics, University of the Witwatersrand

Research Interests: Complex disease genetics; epigenetics

**Dr Munro Marx | mpm@sun.ac.za**

**Professor Raj Ramesar | African Society of Human Genetics Representative | raj.ramesar@nhls.ac.za**

Position: Head of Division, Human Genetics, UCT

Research Interests: Human diversity and health; teaching and training

**Ms Tasha Wainstein | Secretary | Tasha.Wainstein@nhls.ac.za**

Position: Genetic Counsellor & Associate Lecturer, Division of Human Genetics, University of the Witwatersrand

Research Interests: Inherited cancers in black South African populations; genetic counseling

**Professor Louise Warnich | lw@sun.ac.za**

Position: Professor, Department of Genetics, Stellenbosch University

Research Interests: Pharmacogenetics and – genomics with specific application in antipsychotic treatment of schizophrenia patients and anti-retroviral treatment of HIV positive patients in South Africa

**Dr Ambroise Wonkam | Ambroise.wonkam@uct.ac.za**

Position: Medical Geneticist & Associate Professor, Division of Human Genetics, UCT

Research Interests: Laboratory, clinical, educational and ethical aspects of medical genetics
The SASHG2013 Conference, held in Johannesburg, at the Maslow Hotel, from the 6-9 October, was a showcase for the latest work and interesting findings in the field of Human Genetics from southern Africa. The focus was on genomic research, clinical applications and translational medicine. The theme of “Genetworking: Building bridges base by base” was aimed at promoting networking across groups, across sub-disciplines and across research programs. We had over 200 delegates that included a sprinkling from other parts of the world, who literally placed their footprint on our map.

The Conference was opened by Ms Malebona Precious Matsoso (pictured), Director General of the National Department of Health, who referred to the challenges faced by the discipline and the need for action to ensure wider benefit to patients with genetic conditions.

The SASHG2013 committee was able to support almost 50 young scientists and students through their fundraising. On the opening day there was an excellent session on The Southern African Human Genome Programme, an initiative funded by the National Department for Science and Technology, which is a national project aimed at mining African genome diversity for improved health of the people of southern Africa. The presentations and posters were of outstanding quality and our international speakers (shown below) expressed their interest and wish to be involved with the SASHG community as collaborators and advisors. There were three concurrent workshops on topical issues: Stem Cells, Pharmacogenomics and Next Generation Sequencing. We wish to thank all the speakers, poster presenters, adjudicators and delegates for making it a memorable congress and for sharing their work and their experiences.

Genetworking is vital to the future of Human Genetics, which is relevant across the many different disciplines touched by the insights brought about by our field of research and work. The full abstracts and photographs from the SASHG2013 Conference can be found at: http://www.sashg2013.co.za/. Do visit our SASHG website for news of the next congress (http://www.sashg.org/).
Southern African Human Genome Programme (SAHGP)

- Michèle Ramsay

The SAHGP continues to capture the imagination of the public and our young and not so young scientists and researchers. A session was devoted to highlighting the SAHGP and reaffirming its goals and vision. It is unique on the continent as the first genome project supported by an African government and Ben Durham opened the session with the perspective from the National Department of Science and Technology and their support for the Programme. Michael Pepper, the joint principal investigator, gave an historical perspective and provided local socioeconomic and international context outlining the objectives of the SAHGP and its future medical relevance on the subcontinent. To realise the objectives of the program we will need to enhance our ability to analyse large data sets of genomic, phenotype and epidemiological data and Nicola Mulder outlined a model for skills development and highlighted synergies with the objectives of Human Health and Heredity in Africa (H3Africa) initiative. Himla Soodyall spoke about the importance of understanding population history, one of the objectives of the SAHGP and Raj Ramesar outlined the role that the SAHGP could and should play in education. A lively debate followed and the involvement and commitment of the young participants was especially relevant and encouraging, auguring well for the future of the SAHGP.

Brief update: The SAHGP has received additional funding from the DST to sequence 24 whole human genomes. After much discussion and deliberation, it was decided to sequence 8 Sotho-Tswana-speakers from the Free state, 8 Nguni-speakers from the Eastern Cape and 8 Coloured individuals from the Western Cape. These data will provide valuable insight into the histories of our peoples, give us a deeper understanding of the similarities and differences between the groups and will be of sufficient depth to allow the robust identification of novel genetic variants. A workshop is planned for mid-2014 to involve many groups in the analysis of the data and to develop an approach to showcase the data and to use the SAHGP to make an impact on public awareness and primary and secondary education.

SASHG members who are interested in taking part in these initiatives are invited to write to Michele Ramsay (michele.ramsay@nhls.ac.za or michele.ramsay@wits.ac.za) and to Michael Pepper (Michael.Pepper@up.ac.za).

Raj Ramesar, Michèlè Ramsay, Nat Khaole (DoH), Michael Pepper, Himla Soodyall and Ben Durham
Young Researchers Forum 2013

- Jacqueline Frost and Jayesen Knezovich
YRF Organising Committee

As a forerunner to the main SASHG Congress, a Young Researchers Forum (YRF) was held on 5th October 2013, at the School of Public Health at the University of the Witwatersrand. The Forum provides a means for young researchers to present and evaluate research, and allows them to explore collaborative and career opportunities. The 2013 YRF organising committee comprised of PhD students and Post-Docs from the Division of Human Genetics at the University of the Witwatersrand.

A total of 68 young researchers from Honours to Post-doc level representing institutions across the country attended this years’ YRF. There were 9 oral and 40 poster presentations on the day, which allowed for peer interaction and true “genet-working” with one another. After much deliberation, the YRF adjudicators awarded the following prizes: 1st Prize Oral – Danielle Smith (UCT), 2nd Prize Oral – Marelie Swart (UCT), 1st Prize Poster – Dr Lauren Watson (UCT), 2nd Prize Poster – Naseeha Hassen (UCT). Congratulations, once again!

In addition to the student and post-doc oral presentations, three plenary speakers, Professor Himla Soodyall, Dr Penny Moore and Dr Bavesh Kana, showcased their research activities and insights at Wits. The latter part of the Forum included an interactive Science Communication workshop, organised with, and hosted by Marina Joubert from Southern Science and Erna van Wyk from Wits Communications. The workshop aimed to give researchers a better understanding about how to effectively communicate a scientific research message to the general public. The workshop included cool and crazy ways to share science. This included presentations on social media and how scientists can use it effectively to communicate their research, FameLab, Dance-your-PhD and also a segment on infographics and how to design them. The workshop was a huge success, with Marina reporting back an evaluation score of 87% from attendees. Overall the 2013 YRF was a resounding success. The Committee would like to once again thank everyone who helped make the day such a success – the researchers, generous sponsorship and plenary speakers, who were instrumental in realising the most successful YRF to date!

More news from SASHG 2013 elsewhere in the newsletter...
New Honorary SASHG members – 2013

Prof Philip Venter

Philip started his career in 1970 at Tygerberg Hospital, Parow, Bellville as a cytogeneticist. He was one of the first 6 students ever to complete a Hons Degree in Human Genetics (Stell) in SA in 1972. He also completed his Master’s degree (Stell) in 1978.

For nine years (1978-1988), Philip was employed as Deputy Director by the Genetic Services Division at the Department of Health and Welfare. At that time he and his colleagues undertook extensive surveys of the people with intellectual disabilities in institutions for the disabled in the then Western Transvaal and elsewhere. As a result he identified and diagnosed the first SA many males with the so-called Martin-Bell syndrome, now known as Fragile X syndrome. Philip then investigated the families, completed his PhD (UOFS 1988) project on this syndrome, and became the South African authority on the condition. He made a unique and extensive contribution to the knowledge available on the syndrome in the early days of genetic research in South Africa and participated in several international meetings on the topic. He moved into academics and started (May 1988) as Professor and Head of the Department of Medical Sciences at the University of the North (now University of Limpopo). He served a term of two years as Dean of the Faculty of Health Sciences 1993-94) and later eight (8) years as Director and Coordinator of the School and Faculty of Health Sciences. He maintained his interest in human genetics throughout, becoming active in genetic counselling (having counselled many family members while completing his research on Fragile X syndrome) and registering as a genetic counsellor (under the grandfather clause with the HPCSA) in the 1990s. He was also active in the SASHG Society for many years and organised an excellent biennial congress at a Kruger Park venue in 2001.

He continued his interest in research and community development. Together with Prof Christianson he managed a clinical genetics outreach programme in what is now Limpopo. This programme documented the first epidemiology of birth defects in rural South Africa and from it originated many of the principles for establishing services for the care and prevention of birth defects in primary health care in developing nations.

For his contribution to Human Genetics and Science, he was awarded the Ernest Oppenheimer Memorial Travel Award, the MRC Travel Award and the University Senate Travel Grant and spends his Sabbatical year (1998) in the USA (three months), Cuba (6 weeks) and Erasmus University in the Netherlands (8 months).

In the last few years of his career, he became interested and involved in the genetics of prostate cancer in African men. He teamed-up with Prof Vanessa Hayes who is affiliated with the Craig Venter Institute in San Diego, USA. They started a still ongoing project, with several grants. In cooperation with Prof Hayes, he was involved in the full genome sequencing of Emeritus Arch Bishop Desmond Tutu and two Namibian Bushmen. Arch Bishop Tutu is the first named African whose full genome was published and made available in the public domain. The research was published as a front page Nature publication on the 18th February 2010.

Since Philip has worked in the field of human genetics for more than four decades and served it well, we would like to support the proposal that he be made an Honorary Life Member of the SASHG.
Professor Jenny Kromberg is a truly unique and outstanding person in the history and fellowship of South African human and medical genetics. Her contribution to South Africa, in this regard, is simply too great to do justice to in the limited space available. A medical social worker by training and experience, on the inspiration of Professor Trefor Jenkins, she was appointed in 1971 in the Division of Human Genetics, SA Institute of Medical Research and University of the Witwatersrand. Her brief was to undertake research into the psychological, social and cultural aspects of medical genetics and assist in developing medical genetic counselling clinic services in Johannesburg. All this before medical genetic counselling was a global reality. Between 1971 and her retirement in 1999 Jennifer's academic contribution to medical genetics in South Africa and internationally was immense. Her research for her MA and PhD was on oculocutaneous albinism, a topic on which she is an international expert. She expanded her research to include childhood disability, particularly intellectual disability, a field in which she led a research programme in Bushbuckridge that resulted in the documentation of largest and most comprehensive study on childhood disability in Africa, and one of only 3 such studies in the developing world. Jennifer has written 112 publications, including papers, books chapters, and research reports and has edited one book. Her list of presentations invited, and otherwise, spans the globe. The list of memberships of committees and the roles Jennifer fulfilled thereon is too long to record. Of particular significance is her founding membership of our own Southern African Society of Human Genetics (SASHG), to which she gave unstinting service from its inception in 1986. Jennifer was also a founding member and stalwart of the Southern African Inherited Disorders Association (SAIDA), chairing the organization through most of the 1990s. Jennifer initiated, in 1989, the first Masters in medical genetic counseling in South Africa, a process that ultimately led to the recognition and registration of the profession of medical genetic counseling with the Health Professions Council of South Africa. It is through her teaching, training and tutoring of medical genetic counselors, scientists and doctors, including myself, that I believe Jennifer's most enduring achievement continues to be attained. Indelibly marked by her 'red pen' she has mentored generations of nascent human and medical genetic researchers and the publications continue to flow as testimony to her consummate skill. After her retirement Jennifer spent four years working in Australia. Fortunately she is back in South Africa where she is doing what she does so well mentoring young medical geneticists through their research to the point of publication. Jennifer Kromberg was central to the initiation and development of human and medical genetics in South Africa and with her vibrant energy continues to promote its progress. Truly, she deserves the accolade of Honorary Life Membership of the Southern African Society of Human Genetics in recognition of her outstanding contribution to our Society and human and medical genetics nationally and internationally.

Promotions, Graduations & Accolades

- Tina-Marie Wessels graduated with her PhD (Genetic Counseling) on 10 December 2013
- Careni Pretorius and Anneline Lochan passed their College of Medicine of South Africa exams.
- Zané Lombard was promoted to Senior Lecturer in February this year.

Congratulations!
Registrar Training Workshop 2013

- Karen Fieggen

This year Dr Karen Fieggen and Prof Ambroise Wonkam convened a five-day long training workshop for registrars at UCT. It was attended by Medical Genetic Registrars and the two newly qualified Medical geneticists together with other colleagues who run genetic services, such as Dr Naicker from KZN and Dr Honey from Pretoria, as well as Dr Ngongang from Cameroon who joined UCT as a supernumerary registrar in June this year. There were about 15 medical genetic attendees at each session with other colleagues from diverse disciplines such as Chemical Pathology, Paediatrics, Cell Biology and Anatomical pathology who came in for sessions of interest. We also had scientists form our own environment as well as two medical science interns based at the Pathcare Molecular Genetics laboratory for a day. Dr Henderson travelled from UFS to teach on the course and a number of local “experts” in their field kindly gave up their time, as did our colleagues from US, Dr Urban and Professor de Jongh.

Those attending were treated to a wide range of excellent talks. The feedback both informal and as formally assessed was very positive, both for knowledge gained and for creating an opportunity for the next generation of medical genetics to get together and strengthen ties. The support of the SASHG was much appreciated.

SASHG support students to attend 2013 congress

This year the SASHG was in the fortunate position to offer travel support to 48 postgraduate students and interns to attend the 15th Biennial SASHG congress in Johannesburg. All supported students presented their work at the conference (either as a poster or oral presentation) and enjoyed the interaction with fellow colleagues tremendously. Feedback on their experiences was overwhelmingly positive and encouraging both in terms of the general running of the conference itself, as well as the science and research presented.

The bursary recipients are pictured on the next page with Professor Michele Ramsay (SASHG chair 2011-2013) and Dr Zané Lombard (SASHG treasurer).

Feedback from bursary students on next page!
**Feedback Report: Bursary recipients attending SASHG 2013**

“I feel that the SASHG highlighted the importance of communication across disciplines as well as the collaboration between scientists so that projects can progress and not stagnate. This was a great experience and an eye opening opportunity that granted me the insight that I needed during this time of my scientific development.”

- **HMVE Combrink, M.Med.Sci (Human Genetics) Student, UFS**

“Both the presentations and the posters at the conference were varied, intriguing and of a high standard. It was great to see that both experts in the field as well as fellow students delivered presentations.”

- **Avani Baruthram, MSc Student, NICD**

“I was inspired and touched by the passion of the organizers in their many efforts to motivate young researchers like me to find their way in this interesting field of human genetics. I was also amazed by the ground-breaking research on epigenetics and stem cells which is being done locally. We also got a chance to meet potential collaborators, which we will build up on in the future.”

- **Tinashe Chikowore, MSc Student, NWU**

“The conference kicked off with a sensational young researcher’s forum (YRF). What was breathtaking about the main conference was the “genetworking”. I got an opportunity to meet some of the great scientists in this field. I would like to thank the SASHG committee for giving me this opportunity”.

- **Sibusiso Malindisa, BHSc (Hons) student, Wits**

“For me, one of the most exciting topics discussed at the SASHG conference is the advent of the SAHGP. This conference has helped to bridge the divide between researchers and clinicians around the country and other parts of the world. It has allowed scientists to realise the tasks we face and the goals we must aim to achieve in order to develop the genetics field in southern Africa. The knowledge I gained is not something that can be read in a journal or found over the internet.”

- **Rupal Jivan, PhD candidate, Wits**
Festschrift in honour of Prof Trefor Jenkins

- Jennifer Kromberg

Prof Trefor Jenkins, pictured with (from left to right): Jennifer Kromberg, Himla Soodyall, Amanda Krause and Michèle Ramsay

Trefor Jenkins was a member of staff at the University of the Witwatersrand from 1965 to his retirement in 1998. He worked in the field of Human Genetics for 30 of those years and as Professor and Head of the Division of Human Genetics (Wits) from 1975 onwards. He was also the first Chairman of the SASHG, which was founded in 1986.

He was awarded an MD (Lond) in 1973 and honorary degrees from the Universities of Cape Town (DSc), Witwatersrand (MD) and South Wales (an honorary doctorate), as well as many other academic distinctions and prizes.

Due to his prestigious international reputation and his extensive contribution to the field of human genetics in South Africa a festschrift will be published in his honour. Profs Jennifer Kromberg, Michèle Ramsay, Amanda Krause and Himla Soodyall have edited this festschrift, which is funded by the Wits Strategic Planning and Allocation of Resources Committee (SPARC).

It contains 18 articles, contributed by colleagues, ex-students and collaborators, on topics reflecting Professor Jenkins’ wide range of interests. The festschrift will be published by the South African Medical Journal and will be distributed with the December 2013 issue of the journal.
Update on the DNA Bill

- Lisa Roberts

After a long journey, the DNA Bill is awaiting Presidential assent, the final ‘home stretch’ of actually becoming law. This law will formally establish the National forensic DNA database (NFDD) for crime-fighting purposes, and provides a regulatory framework with severe penalties for abuse.

The SASHG committee has made recommendations to the various drafts of the Bill since 2009. This May, we made a written submission to Parliament. We were happy to note that issues we had raised in the past have been addressed, namely:

i. lack of ‘informed consent’,
ii. an elimination index to detect possible contamination of samples by those processing them,
iii. concerns regarding unclear retention, storage and expungement policies
iv. the need for regular review of the legislation, and
v. the need for an oversight body to be established.

At the same time, we made several additional recommendations for the oversight body to consider when moving forward, including data security, access policies, and quality management. We also suggested that a member of the SASHG be included in the oversight body.

We were thrilled to receive an invitation to address the Portfolio Committee on Police about our written submission, and on 11th June Prof. Louise Warnich presented the Society’s concerns and suggestions. Subsequently, we were invited to submit specific recommendations on how to ensure the integrity of the database, which was done in July.

On 12th November, the DNA Bill was passed by National Assembly and National Council of Provinces. A call for public nominations to the NFOEB will go out shortly and we believe the SASHG has an important role to play on that board, ensuring that the complex ethical, legal and social issues surrounding the Database are considered.

Salient facts about the new DNA Bill:

• It will be compulsory for all arrestees and convicted offenders to provide a DNA sample. This collection will be retrospective (i.e. all current convicted offenders will provide a sample before release from prison).
• Trained, certified police officers will be allowed to collect buccal samples. Training of Police in how to collect these buccal samples will be provided by the NHLS. The collection of blood samples still requires a medical professional. A person may request to collect their own sample, but this process will be supervised by a trained police official.
• Distinction has been made between a DNA sample and a DNA profile. Reference samples (from suspects, convicted offenders, etc) will be destroyed after a profile has been generated. Crime scene samples (blood, semen, weapons, etc.) will be stored indefinitely. Profiles will be stored for different retention periods, depending on the database index they fall under. For example, a profile will remain on the Convicted offender index indefinitely, however a profile will be removed from the Arrestee index if the person is acquitted at trial. Provisions have also
Illumina (UK) sponsored a competition at SASHG2013 to support capacity development for African genomic science by providing exposure to NGS and data analysis. The prize for the competition was four whole human genome sequences. To be eligible, applicants had to be registered participants at the SASHG2013 Conference, researchers or groups working on the African continent with some experience in analysing human genomic data, based at a recognised tertiary institution, science council or recognised research organization and have access to four human genome samples, with all necessary authorizations and consents.

A bumper crop of ten applications was received from groups around the country, including proposals on pharmacogenomics, cancer, HIV susceptibility, Parkinson’s disease and bipolar disorder, monogenic traits and modifiers, and population studies and capacity development. A panel of independent adjudicators from Illumina and SASHG had a challenging task choosing a winner. The applications were evaluated based on scientific merit, relevance to Africa and contribution to capacity development, motivation and impact, intent to publish, and researcher’s experience. The winning project was a unanimous choice by the reviewers.

Our congratulations go to Dr Nadia Carstens (Wits Bioinformatics, University of the Witwatersrand) for the project entitled:

“Using next-generation sequencing to investigate the cause of congenital cataracts in a South African family”.

The reviewers all agreed that this project has high scientific merit. The proposal to use whole genome sequencing to identify mutations in a familial disease has a strong likelihood of success. The study design was very well presented and is well matched to the scope of the technology. Appropriate samples and clinical information are available. The reviewers noted the additional value of making available the genome sequence and the ability
to derive haplotype information. This will provide valuable information about genetic variation in Southern African human genomes. The project outcome will therefore positively impact on a clinical problem and the study of population genetics in South Africa. The applicants are well-placed to make good use of the training opportunity, to develop capacity for self-sufficiency in South Africa both in the short and long term time-frame, and to assist wider dissemination by data release and through training via future conferences and workshops.

We wish to thank Anne Bowcock and Valerie Corfield (representing SASHG) and David Bentley from Illumina (UK) for rising to the challenge of choosing the SASHG2013 Illumina Competition winner. We wish Dr Carstens a most successful research study and look forward to hearing about the impact of this prize on their research at the next SASHG Conference.

**Tackling direct-to-consumer genetic testing**

* - Soraya Bardien & Collet Dandara

The SASHG Committee 2011-2013 started a discussion on tackling the contentious issue of direct-to-consumer (DTC) genetic testing. On this very important topic for discussion the aim was to raise awareness of the potential risks in an attempt to protect SA consumers and healthcare providers from possible exploitation. Thus, as part of the discussion, and contribution, we published an article in the South African Medical Journal, which can be accessed through the link: [http://www.samj.org.za/index.php/samj/article/view/7049/5295](http://www.samj.org.za/index.php/samj/article/view/7049/5295). The article includes a set of recommendations, particularly, that DTC genetic tests should have appropriate analytical and clinical validity and well-established clinical utility. We also implored the reviewing of existing local legislation to prevent possible stigmatisation and discrimination.

Our stand has been recently vindicated by the recent steps taken by the Food and Drug Administration (FDA) in the USA which has ordered 23andMe (a DTC genetic testing company) to stop sales of its personalized DNA test kits on the basis that the tests it has been offering have not been properly validated and cleared by the US federal government to make the claims they do. In addition, recently an individual filed a class action lawsuit against 23andMe claiming that it ‘falsely and misleadingly advertises their tests as providing health reports on 240+ conditions and traits, drug response, carrier status among other things when there is no analytical or clinical validation’.

With all these developments, and without wanting to hamper development of genetic tests, it is anticipated that the newly appointed SASHG committee will take this matter further by updating the Genetics and health care community on the advances in the genetic testing field. The committee should try to be as inclusive as possible in order to capture the diversity of views.
The year 2013 has been something of a roller-coaster ride for the GC-SA group. It began with a new logo being designed to represent the group’s image as a professional body. In our attempts to have the GC-SA recognized as a professional body, the group launched a website hosted by the SASHG. The website can be accessed on:


Our major achievement this year was finalizing the “Standard of Practice for Genetic Counsellors” document that was originally drafted by the GC-SA in 2012. The document is a supplement to the HPCSA Scope of Profession Document, which was published in May 2009. The Standard of Practice document specifies: the scope of practice; guidelines for intern training including core competencies; the requirements for CPD, HPCSA and supervision. The document is awaiting final approval from the Medical and Dental Board of the HPCSA.

Other achievements include the first long awaited national intern exchange. Ms Chanelle le Roux, a Wits student, carried out part of her internship at Tygerberg. The exchange was very beneficial and greatly enhanced the students learning and experience. NHLS/Wits also had two international exchanges. Mrs Kara Stoler spent a month at the Hospital for Sick Children in Toronto, Canada and Ms Prue Dunstan from Melbourne spent a month in the Division of Human Genetics at NHLS/Wits.

The group raised funds to host an international genetic counsellor for the biennial SASHG conference held recently in Johannesburg. We were delighted to have Dr Christine Patch, a consultant genetic counsellor (manager) and reader in clinical genetics from Guys hospital in London, present a talk and workshop at the conference. She also spent time at UCT, Stellenbosch, and Wits, where she gave several excellent talks and workshops. The group benefitted a great deal from her visit and hopes to follow up on possible collaborations.

The lack of posts is the most serious and
stressful situation that faced us in 2013 and remains a National area of concern. Sadly, the number of genetic counsellors in the country remains low. As of December 2013, there are 13 Genetic Counsellors (GCs) with an MSc in Genetic Counselling, who are registered with the HPCSA and actively practicing in SA. We are happy to report that the year ended on a high note, with UCT being awarded a full-time Genetic Counsellor/Lecturer post for 2014 and Groote Schuur Hospital a full-time post for a GC as from April 2014. At the NHLS/Wits, there is a possibility of employing the interns that will qualify in April and September 2014.

The training of genetic counsellors is crucial and both UCT and Wits have not taken in new students since 2011. However, there has once again been some positive movement in this regard with four students being accepted into the MSc programme at UCT in 2014. The unit at NHLS/Wits will be accepting new students in 2015.

There can be no doubt that this year has been turbulent and that there are still numerous obstacles that need to be overcome by the GC-SA, especially with regards to career opportunities for genetic counsellors. This turbulence has fortunately been tempered by the great strides that have been made this year. There is a general feeling of encouragement and possibility as we enter 2014. We wish all members of the SASHG community well over the festive season.

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**Clinical Genetics report back**

* Lindsay Lambie

In the two and a half years the state of medical genetics in SA has declined. I speak on behalf of the clinical subgroup of the SASHG, but would like to point out that the issues extend equally to the Genetic Counsellors and the diagnostic laboratory service. Despite efforts with respect to training of registrars – resulting in 4 registrars completing their Part II examinations in the last year, the number of employed and active clinicians has declined. This is in large part due to restructuring within the National Health Laboratory Service, resulting in a lack of consultant posts for newly qualified registrars, and the freezing of existing posts as people leave. At present, in Gauteng there are 4 ½ employed Medical Geneticists, with two in Cape Town, one in Stellenbosch and one in Bloemfontein: A total of 8 ½ Medical Geneticists. In addition, three retired colleagues assist us, and there are two Paediatricians working predominantly in the field. Of the four registrars who have completed their exams, one has left the country and the remainder does not have Consultant posts for 2014. The total of 8 ½ Medical Geneticists not only falls far short of recommendations, but also is down by 25% on our 2011 numbers.

Aside from one registrar and two supernumerary registrars from the African continent, training has essentially been halted. We have debated the ethics of starting to train new registrars when our own country will not employ them once they are trained. The result is that even if the situation changes, we will now have a 4-year hiatus before our service can improve or expand. As a society, and an informal subgroup, attempts have been made to address these issues, over the 2 years, at various levels, including through the NHLS, Universities, CMSA and HPCSA, but to little effect. Talks between the NHLS and Department of Health are reported to be in progress.

On a more positive note, the Medical Geneticists have continued to organize National training (most recently organized by UCT), hold National Academic and Dysmorphology teleconferences and set and
facilitate College of Medicine Specialist exams. We hope to see increased awareness following the SASHG conference and we have plans to create a more cohesive approach to advocate for and raise awareness of the state of Medical Genetic Services in South Africa.

In Memory of Nelson Mandela

When a man has done what he considers to be his duty to his people and his country, he can rest in peace. I believe I have made that effort and that is, therefore, why I will sleep for the eternity.

Nelson Mandela, 1918-2013