WHY DO WE DO THIS AGAIN?

I follow a popular Twitter account called Sh*t Academics say (@AcademicsSay) – it’s a humorous/satirical view of the matters most academics encounter on a regular basis from the mind of Nathan Hall, an associate professor at McGill University. This week I saw a line from this account come across my screen that really resonated with me – it read: “Yes, I’m a doctor. Just not the kind that helps people.” This quote is obviously poking fun at the conundrum of explaining the difference between an MD and a PhD, but it made me think about our motivations for becoming involved (and staying) in the field of Human Genetics. I think most folks who end up in this field gravitated to it partly due to their strong sense of wanting to make a positive contribution to society – whether it is through helping patients suffering from a particular genetic condition, working towards finding the cure for a debilitating illness or training the next generation of scientists. However, the daily struggles of being an academic in South Africa can become overwhelming, and it is easy to forget this novel goal when one is drowning in competing for grant funding in a pool that is too small to support all (and dwindling by the day), wading through red tape from bureaucratic administrations, and trying to finish the academic year despite student protests. It is important that we remind ourselves daily why it is that we chose this career path in the first place, and to find that gleaming beacon in the workday that makes our soul buzz and reinvigorates our scientific altruism. Some of our colleagues from SASHG are great examples of doing just that, and we celebrate some of the achievements and accolades they have achieved this year in this newsletter. One I have to highlight is the hard work and tremendous effort our SASHG Secretary, Dr Careni Spencer, has put into revamping the SASHG website to ensure that the Society is more accessible and that we can reach more patients, stakeholders and young members. You can view the great work she and our web developer did at http://www.sashg.org. I hope you enjoy reading the 2016 newsletter, and that all will have a peaceful and invigorating festive season. Until we meet again in 2017!

- Zané Lombard
SASHG Chair
New SASHG website launched!

http://sashg.org/

In the 2015/2016 year the SASHG committee undertook to revamp our current SASHG website. This was completed and officially launched at the beginning of October.

The website now features our new logo, a brief history of our organization, information about local and international events and also membership details along with our newsletter and many other features.

This was the first phase of our website re-launch and in the second phase (2017) we will tackle membership specific login and privatizing certain areas and information to members only. Membership applications and renewals have also been streamlined and can now be done via the online submission form.

The website is now managed by studiovene (www.studiovene.co.za) who also designed it. This site has the additional benefit that certain members on the SASHG committee can now also make interim and small edits to the website. This means that we can do regular updates of events, news and also vacancies.

Please visit the site and send us your comments! Also, if you have a local or international event or a vacancy relevant to our community please use the online submission form to send us your information.

Please also have a look at our new social media presence via our Facebook page and our twitter handle
Medical Genetics Group

Feedback for the Second Medical Genetics registrar workshop

The Clinical Geneticists decided that it was time to arrange a second Medical Genetics Registrar Workshop. This was to follow the very successful workshop hosted by UCT in 2013. Bloemfontein arranged and hosted this workshop at the end of August 2016. All the registrars from across the country were invited and the workshop was very well attended by all those in posts at the time. This enabled new registrars to meet the registrars from elsewhere in the country and become incorporated into the group. The workshop was partially funded by the SASHG with each department contributing towards their registrars travel and accommodation costs. Catering costs were covered by the hosting institution and a special thanks to Genzyme for their sponsorship to cover part of the catering costs. A variety of specialists outside of Genetics were asked to present talks on topics relevant to our training and work.

The opening presentation was by an ophthalmologist who demonstrated the optimal use of an ophthalmoscope and then discussed a few common inherited eye disorders. The rest of the first day was devoted to neurodevelopment, loss of milestones, autism and speech delay.

The second day opened with the registrars presenting cases and quizzing each other. The rest of the day revolved around metabolic and endocrine disorders with talks renal tubular disorders, approach to hypoglycaemia, metabolic muscles disease, CDG’s, porphyria and other inherited endocrine disorders.

The third day included talks about inherited skin diseases, the neurological examination and approach to movement disorders. Again the registrars presented patients. The highlight of that day was Dr Spencer giving a quiz and presentation on skeletal disorders in which she shared the wealth of knowledge she acquired at a recently ESHG course on skeletal dysplasias.

The final day had a session on the cardiac examination and cardiomyopathies. This was followed by a session on Down syndrome with a talk on the principles of early intervention and closed by Sheri Brynhard talking about her life with Down syndrome and her role as international ambassador for Down Syndrome (see photo).

The organizers wish to thank all presenters and attendees for well-prepared talks and case presentation as well as the fantastic interactive atmosphere throughout the workshop. The slides from most of the talks and video recordings of the presentations will available of the Clinical Geneticists’ Dropbox shortly.

From left to right: Maureen Conradie, Ayman Hussain, Cedrik Ngongang, Sheri Brynard (front), Dorothy Russel en Liani Smit. (with permission)

- Bertram Henderson
  Chair: Medical Genetics Group
The SASHG Young Researchers’ Forum

The YRF is a focus group of the SASHG, first set up in 2011 prior to the Joint African and Southern African Societies’ Meeting. The recognition that postgraduate students, junior academics and researchers in the field have a unique set of challenges and interests led to the establishment of the group; the aim of which is to bring together younger members in the field of human genetics to connect and share their experiences. However, there is no restriction to membership and we welcome the participation of our more experienced colleagues, senior scientists, clinicians and professors.

This year (2016) has been refreshing for all involved in the SASHG. We have a brand spanking new website which can be accessed at: http://sashg.org/. Please check it out if you haven’t already. The site is open to the public BUT there are benefits to your membership, including exclusive access to a monthly newsletter profiling YRF members and their research interests. Our FaceBook activities have also been reviewed and all YRF-related communications are via: https://www.facebook.com/Postdocs.SASHG.YRF Posts regarding funding opportunities, studentships and research positions are welcome and moderated by the administrators.

Importantly, the YRF has a representative on the Executive Committee of the SASHG to voice the concerns of the next generation of human geneticists and to feedback appropriately to the group members. The current representative (2016/17) is Dr Fiona Baine, a postdoctoral fellow at the University of the Witwatersrand. Fiona is readily available to address any questions and or comments related to YRF members, please contact her on fiona.baine@wits.ac.za or via the FaceBook page above.

- Fiona Baine
  Chair: YRF

Please remember to renew your annual SASHG membership on our website: http://sashg.org
What a Bumper Year for Genetic Counselling

Genetic counselling is one of the top healthcare jobs for 2030 (BRICS Business report [https://extranet.ampath.co.za/mobipdf/?batchid=2108881&email=noelene@geneticcounselling.co.za](https://extranet.ampath.co.za/mobipdf/?batchid=2108881&email=noelene@geneticcounselling.co.za) and details in the Atlas of Future jobs [http://atlas100.ru/en/](http://atlas100.ru/en/)). In South Africa genetic counselling is an established profession and the GC-SA strives to grow the profession into the future. Following is a summary of the happenings in 2015/16.

It all started with a BIG – discovery – when an insurer proposed to offer whole exome sequencing (WES) to its members. This certainly presented the genetics community with a challenge. The insurer in collaboration with SASHG members have worked towards and continue to establish processes to ensure that the product is delivered in a manner that is appropriate and benefits the South African community as a whole. Discussions and open debate has exposed the public and commerce to clinical genetic services and highlighted the value of genetic counselling in the delivery of genetic tests. Affording genetic counsellors (GCs) an opportunity to showcase our ability to recognise, adapt and provide innovative solutions to new challenges. GC-SA members have established unique genetic counselling services to meet the need for access and education to support the healthcare profession in the introduction of WES. A challenge highlighted in this process is capacity and the need to skill GCs and grow the profession.

Nine students entered the Masters programmes this year, five at UCT and four at Wits. Wits introduced a new course structure – the degree is now offered as a two-year part-time Masters – with an option to continue with two years of internship training thereafter, or combining the second year of Masters with a first year internship. This affords the opportunity to increase the footprint of genetic counselling in the academic sector should they not wish to proceed into clinical practice.

Three interns qualify at the end of 2016 - two at UCT and one at Stellenbosch. They were funded through the NRF and research funds. Funding of internships remains a concern and risks the loss of further skilling. Various solutions are being considered.

On the employment front, NHLS in Johannesburg employed a full time GC and a 6/8th post was made available through the Department of Health at Groote Schuur Hospital. Even with the discussion of capacity there are still limited vacancies. Initiatives to address employment requirements include recommendations to the Department of Health to include genetic counselling in healthcare services and the GC-SA’s formal comment on the NHI White Paper. Training centres will approach faculties that benefit from genetic counselling service for funding.

GCs are being employed in different roles. Two of the major private labs in South Africa have employed GCs on a full-time basis. Their interaction with the healthcare community has bolstered the profile of genetic counselling skills and knowledge. GCs in private practice continue to grow their practices providing support to various healthcare providers and offering unique access to overcome the limited national representation of GCs. Funding of genetic counselling consultations by health insurers has assisted with this process. Although part of a GCs scope of practice, the issue of payment by medical aids for genetic tests requested by GCs still needs to be addressed.

The standard of genetic counselling qualification and practice is equal to that of overseas countries. This year Wits got to welcome the first GC from the UK. Reciprocity agreements had previously been established with the UK, Canada and Australia. This year the European Board of Medical Genetics confirmed a reciprocity agreement between Europe and South Africa based on our training programmes. Although this creates an alternate work option we do not want to lose our highly skilled counsellors to the world out there and continue to seek alternatives.
Tina-Marie Wessels was invited to be the co-chair of TAGC (Transnational Alliance for Genetic Counselling). Both Tina and Shelley Macaulay were invited to represent the South Africa training institutes at the TAGC conference in Barcelona this year. Feedback is that the challenges experienced by GCs are similar and relate mainly to WES and WGS, big data and communication of large amounts of complex information, and how to address these through appropriate training.

The GC-SA committee established committee member profiles to address ongoing challenges including education, awareness, and professional standards.

New initiatives for continued education include the ongoing use of the professions intranet for case discussions and the introduction of online Genetic Counselling Clinical Meetings. We hope to establish an online Journal Club.

Awareness of genetic counselling continues to grow. Other opportunities include interaction with support groups, profile at the RareX Congress, media interviews and a banner competition for Genetic Counselling Awareness Day.

On a professional front GC-SA worked together with the Medical Geneticists Group (MGG), SASHG, GA-SA and SAHGP and published an editorial comment in the SAMJ which includes recommended guidelines for ethical provision of WES. Collaboration between the GC-SA, MGG and the South African Society for Ultrasound in Obstetrics and Gynaecology (SASUOG) has developed guidelines for the provision of Non-Invasive Prenatal Testing (NIPT). This is a work in progress and has not been published. Inclusion of a MGG representative on the GC-SA committee has proven insightful.

With the re-establishment of the HPCSA Medical Science Committee the Scope of Practice and Internship Assessments and Accreditation Guidelines for Genetic Counselling have been reviewed to ensure the standard aligns with the guidelines of the HPCSA and the GC-SA committee took the opportunity to update these documents and await approval.

Hopefully this illustrates the GC-SA’s commitment to the ongoing development of the profession of genetic counselling in South Africa. For the upcoming year the committee hopes to address internship funding, posts, ethical standards, reciprocity processes and agreement, awareness campaigns, direct to consumer testing, counselling protocols, testing guidelines and establish a workshop for the SASHG Congress with an international guest. Thanks to all that have contributed to such a great year!

- Noelene Kinsley
Chair: GC-SA
9th Conference of the African Society of Human Genetics (AfSHG)

The AfSHG met for the ninth time as a Society on the African continent, this time in the historic city of Dakar in Senegal, the most westerly city in Africa. A total of 210 delegates gathered and represented 26 nationalities including Senegal, Mali, South Africa, Congo, DR Congo, Benin, Cameroon, Burkina Faso, Zimbabwe, Emirates, Morocco, Ethiopia, Nigeria, Uganda, Tanzania, Sudan, Tunisia, Switzerland, Rwanda, Ghana, Egypt, Botswana, USA, UK, Canada and France.

The theme of the conference was “Strengthening Human Genetics Research in Africa” and is central to the mission of the African Society of Human Genetics. In the absence of extensive research on the genetic contribution to diseases in diverse African populations, it would not be possible to offer appropriate genetics services or to offer the hope of a precision medicine approach on the continent. This meeting brought together members from country-specific Societies of Human Genetics from Cameroon, Democratic Republic of Congo, Mali and Southern Africa. Many African countries do not have Departments for Human Genetics as a stand-alone discipline and therefore we need to be creative in nested our discipline and activities in existing structures in our tertiary institutions, hospitals and health care sectors and to promote its contribution to patient care.

The first day of the meeting was jointly hosted as a collaborative venture with the 8th Human Heredity and Health in Africa (H3Africa) Consortium meeting (http://www.h3africa.org) and included a panel discussion on “Funding sustainable genomic research in Africa” that provided much food for thought. The breaks brought opportunities for rigorous debate about the state of genetic services across the continent and sharing ideas about advancing the discipline and expanding services for the benefit of African patients and their families. The ethics sessions and talks on community engagement highlighted the ingenuity of the researchers in conveying the content of their research to communities that do not have a vocabulary for explaining genetic and genomic concepts.

Since the conference brought together the Human Genetics Communities from across Africa to a Francophone country in order to invigorate the discipline and to provide a platform to present and share research findings, we spared no cost on simultaneous translation for French and English to ensure that everyone would benefit in both languages, this proved to be a great success. The Conference was opened by the Chancellor of University Cheikh Anta Diop of Dakar, Professor Ibrahima Thioub. Dr Charles Rotimi (founding President of the AfSHG) gave a thoughtful keynote address titled: The African Society of Human Genetics: looking back to shape the future. It was an inspiration to be reminded of the beginnings of our Society and the considerable challenges we have faced along the way. The Scientific sessions included cancer genetics and genomics, medical genetic services, genetics of infectious diseases, human population genetic diversity and health, genomic medicine and the ethics of genomics approaches to patient care and in research. Two mini-symposia were held during the meeting one on Human non-human system studies: TB genomics and one on Disorders of sex development. In addition, there were two training workshops on Research leadership (hosted by Nature Genetics) and Next generation sequence data analysis in complex traits. The formal deliberations were concluded by the Director General of Research, Ministry of Higher Education and Research Senegal, Professor Cheikh Becaye Gaye. It was a wonderful opportunity for participants to engage, participate and learn from one another as peers, mentors, colleagues and friends. We were united in our quest to explore the treasure trove of African genetic diversity and to promote an understanding of the genetic contribution to health and disease in African populations with a view to improving health on the continent.

- Michele Ramsay
  President: AfSHG
African scientists should strongly engage with policy makers and African communities, and communicate with them in direct and accessible language that would spur them to positive action and support for scientific research on the continent. This was the message of the President of Mauritius, Dr Ameenah Gurib-Fakim (see photograph), in her opening address to the 9th Biannual meeting of the Human Heredity and Health in Africa (H3Africa) conference at the La Meridien Resort in Mauritius, this past Friday, 29th of October 2016. Dr. Gurib-Fakim opined that too often scientists resort to technical language that is accessible only to members of their community and very little of their considerably beneficial output is communicated clearly to governments and the people.

The Mauritius meeting was historic because this was the first time that H3Africa meeting is being attended and formally declared open by the Head of an African state. President Gurib-Fakim, herself an internationally renown scientist who works in the field of biodiversity and is one of the very few heads of state to hold a formal scientific qualification, highlighted the role that the H3Africa consortium can have on Africa’s development agenda. She counselled the young and old scientists gathered at the exciting location in Mauritius on the need to conduct scientifically relevant research and build capacity on the continent. She commended the efforts of the H3Africa program in this regard and pledged to strongly advocate for the program locally in Mauritius, among her fellow Heads of State in the African Union and in NEPAD as well as at all international fora.

H3Africa is an initiative of the African Society of Human Genetics and a partnership with the funding agencies, National Institutes of Health (USA) and the Wellcome Trust (UK) which started in earnest in 2012. The primary aims of H3Africa are: (i) explore the rich human genetic diversity on the continent to better understand the role of genomics in human health and diseases, (ii) human capacity development, (iii) research infrastructure development, (iv) biobanking, (v) knowledge (data) generation and sharing, and (vi) community engagement. To achieve these aims, H3Africa has funded projects in common chronic and infectious diseases on the continent including TB, cardiovascular disease (heart disease and stroke), psychiatric and neurological disease, cancer and viral diseases in 28 African countries. More information about the organization of H3Africa and its projects can be found on the website of the Consortium - www.h3africa.org.

One of the objectives of H3Africa is to reduce and possibly reverse the trend of scientific collaborations which has often seen Africans scientists working preferentially with European, American and other international research groups to the detriment of collaborations within and between scientists in Africa. H3Africa has encouraged research projects to be developed and led by African scientists in partnership with a network of their African colleagues – with the option of engaging with foreign researchers who may add special skills or technologies as part of the development programme of the research group. Funding is made available directly to the African institutions that meet stringent administrative/financial management criteria.

H3Africa has developed a very vibrant network of researchers among research institutions in Africa and is already developing the next generation of researchers/scientists on the continent. It provides access to the state of the art research facilities on the continent which is complemented by resources in the UK and the USA thereby ensuring greater retention of brains on the continent. The Consortium meets every 6 months in different African countries in order to spread the gospel of genomics research and engage the local scientific resources in the host countries.

The Funding agencies, NIH and Wellcome Trust have been impressed at the progress in all of the H3A projects to the extent they have both put out calls for a further 5 year cycle funding.

- Press release
The Southern African Human Genome Programme

The Southern African Human Genome Programme (SAHGP) is a national initiative emanating from South Africa. It aims to unlock the unique genetic character of southern African populations with a vision to improving quality of life through understanding human genetic diversity. The SAHGP was launched in January 2011 with seed funding from the National Department of Science and Technology (DST) of South Africa. This funding was used to develop and interim proposal, the aim of which was to establish a pilot sequencing project.

The SAHGP pilot sequencing project has now been completed and includes deep whole genome sequencing and characterization of 24 genomes (8 Coloured [admixed] and 16 black south-eastern Bantu-speakers [9 Nguni-speakers and 7 Sotho-Tswana speakers]). The results of this project have now been submitted for publication. Variant calling identified a total of ~16 million unique variants, including over 0.8 million novel SNVs (defined as absent from dbSNP, KGP Phase 3 and the AGVP). PCA and STRUCTURE analysis revealed differentiation between the two south-eastern Bantu-speaking groups and $F_{ST}$ measures revealed regions with high divergence. The Coloured group showed high admixture with Khoesan, Bantu-speakers, Europeans and populations from the Indian sub-continent. With this work we highlight the need to study different African populations to understand their unique patterns of genetic diversity, to discover novel genetic variation and to promote biomedical research on the sub-continent. These genomes have also been used together with 350 African high coverage genomes for the development of the H3Africa SNP array to ensure enrichment for common African SNPs.

The SAHGP is an initiative that has developed in parallel with the H3Africa initiative, and has contributed significantly to capacity development in South Africa, particularly in the area of bioinformatics. The objective is to respond to the need for data on genomes from the African continent and in particular in our case from southern Africa. Although the project has until now been limited to South Africa, and is small in scope relative to many genome initiatives currently underway internationally, it is our intention to extend this to other countries in the region in the near future.

As the SAHGP enters its next phase, it is important to note that it has now been incorporated into the South African Precision Medicine Initiative to which it will make a significant contribution. The South African Precision Medicine Initiative is led by the DST together with the South African Medical Research Council. Separate funding instruments are being created in this Initiative which will allow for future funding of SAHGP projects.

- Michael Pepper
Important dates 2017

27-30 May 2017 – European Human Genetics Conference, Copenhagen, Denmark

13-16 August 2017 - 17th Biennial Congress of the Southern African Society for Human Genetics, Durban, South Africa

17-21 October – American Society Human Genetics, Annual meeting, Orlando, United State of America
Dr Ayman Hussein has passed his Part 2 FCMG examinations making him the newest member to join the Clinical Genetic community as a consultant – well done!

Congratulations to Dr Venesa Sahibdeen for receiving her PhD entitled “The Identification of Genetic Markers of Obesity Risk in a South African Black Population”.

Well done to Dr Jacqueline Frost for completing her PhD entitled “The Wnt pathway in Systemic sclerosis”

Dr Manogari Chetty has also completed and received her PhD, her thesis title was “Dental Implications of Connective Tissue Disorders in South Africa”

Dr Matshane Masemola also obtained her PHD entitled “Epigenetic modification at imprinting loci following alcohol exposure during prenatal development”. Congratulations!

Prof Lizette Jansen van Rensburg has been awarded the Oettle Memorial Medal for 2016 from CANSA – congratulations (http://www.cansa.org.za/oettle-memorial-award-2016-to-prof-lizette-jansen-van-rensburg/)

Well done to Mr Shaun Aron at the Sydney Brenner Institute for Molecular Bioscience for being awarded the H3A-Harvard Internship for 2016!

Congratulations also to Thandiswa Ngcungcu who was nominated for the Young Investigator Award at the ESHG 2016 conference and also received the CRG-Novartis mobility award to spend 6 months in Barcelona