‘Looking back to move forward’

In the ever busy life of a researcher, where failed experiments and writer's block are common occurrences, it is important to take a step back and refocus on the bigger picture, to remember the worth of the work that we do. In times of discouragement, always remember to reflect on your personal accomplishments, however big or small, as well as our progress in the field as a community.

As 2018 draws to a rapid close, this edition of the newsletter will reflect on the past year in anticipation of the year to come, providing highlights from our SASHG community and some updates on the activities of our committee.

We start with an overview of some noteworthy SASHG committee updates, including the social media revamp to watch out for, as well as the 18th SASHG Biennial Congress 'save the date', for 2019.

On page 3 we have a feature on the UKZN Clinical Genetics Unit (CGU) headed by Prof. Colleen Aldous, who gives a brief overview of the focus and ongoing research of this group. We then have a student feature on Ms Mpoi Makhetha, a PhD student in this research group, comprising her personal profile and an article summary.

This is followed by some achievements and research highlights from members of our community on page 4, all of whom deserve recognition and a heartfelt congratulations!

We end off the newsletter by ‘looking back to move forward’ where some results are summarised from the survey that was sent out to SASHG members earlier this year, in the interest of gaining insight into the opinions and needs of the SASHG community, to better direct our goals and actions going forward.

We hope that you enjoy reading this edition of the newsletter. Indeed, this has been an exciting year for Science and Human Genetics all round, and we look forward to all the opportunities and discoveries that the new year will bring!

Ms Emma Frickel
Representative: Young Researchers' Forum

Note from SASHG Chair, Prof. Soraya Bardien:

We appreciate your honest responses to our survey, and will work towards addressing the points raised.

But for now, let me take this opportunity to wish you well for the coming end of year break and Festive Season. We hope that that this holiday will recharge those 'pap' batteries, allowing you to take on the new year with enthusiasm and vigour!

Regards,
Soraya
Some ‘behind the scenes’ SASHG committee updates

Membership recap

SASHG membership includes access to the newsletters, membership to an appropriate focus group, access to job adverts and student opportunities, news alerts, interaction with peers in the field of human or medical genetics, and a special rate at the SASHG Biennial Congress. We recently did a cleanup of our mailing list and now have 228 fully paid-up members. Membership can be renewed on our website: http://sashg.org/membership/. Let’s celebrate our community!

Social media revamp

We now have a new ‘social media representative’ who will be responsible for all SASHG social media platforms going forward. This has started off with a bang, with the Facebook page publishing daily updates on compelling research in the field of Human Genetics. Numerous career opportunities relating to studies, funding, job vacancies, workshops and more, are also published here as they arise. We are in the process of merging the two current Facebook pages – so be sure to watch this space!

https://www.facebook.com/HumanGeneticsSASHG/
https://www.facebook.com/PostDocs.SASHG.YRF/

Member survey 2018

This year, we launched a survey to gain direct feedback from our current members, to better shape our goals as a society moving forward. To increase transparency and communication between the SASHG committee and members, future newsletters will include different focus areas of the society. The survey results are summarised on pages 5 to 7.

SASHG Biennial Congress

The preparations for the 18th Biennial Congress are well underway, so don’t forget to save the date! This congress, with the theme ‘Our Genes, Our Environment’, will be held at the Century City Convention Centre, Cape Town, from 3rd – 6th August 2019. For the first time, the South African Society for Bioinformatics Student Council (SASBi-SC) will collaborate with the SASHG Young Researchers’ Forum on the 2019 Young Researchers’ Symposium, which will be held at Intaka Island Eco-Centre, Cape Town, on the 3rd of August 2019 (preceding the Biennial Congress) with the theme ‘The Bigger Picture’. Visit https://sashg2019.co.za/ for more information.

SAVE THE DATE!
3 – 6 AUGUST 2019

18th Biennial Congress of the Southern African Society for Human Genetics

CENTURY CITY CONFERENCE CENTRE

IMPORTANT CLOSING DATES:
Submission of abstracts:
12 April 2019
Early conference registration:
14 May 2019
Accommodation reservations:
3 June 2019

Visit www.sashg2019.co.za for more details
UKZN Clinical Genetics Unit (CGU)
Prof. Colleen Aldous, UKZN CGU leader

The Clinical Genetics Unit (CGU) in KwaZulu-Natal has consisted of scientists, clinicians and students who have looked at service, research and teaching in clinical genetics for a number of years. The research has been epidemiology and service based up to this point but the recent developments of collaborations within and outside of UKZN has given impetus to new ideas and exciting new ventures are in the pipeline. We look forward to elevating our research in epidemiology with more accurate figures to give to government through the Modell Global Database and also into genetics of cancers and rare diseases as a next chapter in our growth.

Student feature: Mpoi Makhetha, PhD Medical Genetics

1. What is your area of research?
The genetics of breast cancer in South African women who have a family history of cancer. Among many genes, PALB2, which is a newly identified high risk gene, is proving to shed some light into the genetic causes of the disease, and in SA, this has not been widely studied.

2. What is your background and how did you become interested in your field of research?
Other than the fact that breast cancer is becoming increasingly common in SA, the topic was the focus of my study in my Masters and the results of that study provided an insight that made me even more curious as well as hopeful.

3. What piece of advice would you give to somebody beginning their career in global health?
Network and make use of those connections, you learn a lot from others than you do from just reading articles.

4. Do you have any secret talents?
Cooking; how else can we be kind to ourselves?

5. How do you like to relax?
Watch an action movie at the cinema while enjoying butter-salt flavoured popcorn mixed with Astros and Smarties.

6. Has anyone in particular inspired you in your field or science in general?
It was my high school science teacher (Mrs Mohlererepe) who took her time to meticulously listen to our “childish” not-so-new-discoveries. In that manner, she made me comfortable exploring new horizons to quench my curiosity.

7. What would your superpower be?
Time manipulation, because time flies when the thesis is going well or experiments are finally working out.

8. If you could go back in time where would you go (and why)?
To when I was 6 months old and sleeping was classified as good behaviour.

Article summary compiled by Mpoi Makhetha

Using genetics to understand the influence of higher BMI on depression

The question of whether obesity causes depression or vice versa has been a complex one to answer. To investigate this, Tyrell et al (2018) used Mendelian randomization on 48,791 obese and depressed individuals, and 291,995 controls. The study separated the psychological component of obesity from the metabolic consequences, and men from women. The genetic aspect of the study included obesity-associated single nucleotide polymorphisms in genes such as FTO. Higher BMI was observationally associated with higher likelihood of depression. This association was stronger in women than it was in men. Even when looking at cases of higher childhood BMI, and individuals who perceived themselves as plump at age 10, researchers found that there was a great association of childhood adiposity with adult depression. The study basically associates both childhood and adult obesity with an increased risk of adult depression, however, Tyrell et al (2018) also reported that even individuals who perceived themselves as underweight in childhood, are more likely to be depressed adults.

Members’ achievements and research highlights

Breast cancer surgeon and charity runner Dr Karin Baatjes has yet again lived up to her “Wonder Woman” status at Tygerberg Hospital. While completing her PhD study at Stellenbosch University for graduation in December 2018, she was appointed as the new Head of Clinical Anatomy at Stellenbosch University. Her PhD study on bone health in postmenopausal breast cancer patients led to the development of a three-pronged clinical management approach, incorporating whole exome sequencing (WES). With this approach, moderate- to high-risk genes and low-penetrance variants in key disease pathways could simultaneously be assessed for clinical relevance to: 1) familial risk, 2) modifiable lifestyle and environmental triggers, and therapy-induced co-morbidities. Her study supervisors were Prof. Maritha Kotze, Dr Magda Conradie and Prof. Justus Apffelstaedt.

Daniel Tebogo recently obtained his BSc(Hons) degree in human genetics and Good clinical practice certificate at the University of the Free State, which are steps on his plan to becoming a medical scientist.

Vinet Coetzee from the University of Pretoria received the following awards this year:

- Special Recognition prize for the 2018 GAP-Medical competition, Innovation Hub.
- Selected as a member of the World Economic Forum Young Scientist Community, World Economic Forum.

Dr Helen Malherbe (Postdoctoral Fellow in the University of KwaZulu Natal’s School of Clinical Medicine and Chair: Governing Board, Genetic Alliance South Africa) was honoured to be the first winner for the Rare Diamonds Award for Advocacy.

Dr Marlo Möller (from the Tuberculosis Host Genetics group at Stellenbosch University) reported the following achievements from their group:

Postdoctoral Fellowships awarded to:
- Brigitte Glanzmann – Crick African Network African Career Accelerator Award
- Caitlin Uren – Claude Leon Foundation Postdoctoral Fellowship for 2019
- Haiko Schurz - Claude Leon Foundation Postdoctoral Fellowship for 2019

Student graduations:
- Talani van Schalkwyk – MSc (Human Genetics), December 2018
- Haiko Schurz – PhD (Human Genetics), March 2019
- Stephanie Pitts – PhD (Human Genetics, March 2019

Prof. Michael Pepper (University of Pretoria) reports on the ASSAf consensus study: Human genetics and genomics in South Africa: ethical, legal and social implications.

On 04 December 2018 the Academy of Science of South Africa (ASSAf) will launch the report of a consensus study which addresses the ethical, legal and social implications (ELSI) of genetics and genomics work, as it relates to research, health service provision and forensic applications (medical and legal) in South Africa. The study was undertaken by a 13-member panel appointed by ASSAf.

The broad philosophical approach of Ubuntu, a philosophical notion that refers to the essence or quality of being human, has been central to the deliberations and writing of the report. The report describes the benefits to be derived from genetic and genomics work, the need for boundaries to be clearly defined and adherence monitored to ensure that benefits are shared by all and that no harm is done. The report is divided into three thematic areas: Building Relationships, Respect for Persons and Good Stewardship. Each section is followed by recommendations which are ethically and legally sound, culturally appropriate, feasible, enforceable and sustainable, given the resources within the country, and balanced against competing national priorities.

The consensus report should inform the drafting of one or more policy documents, which in turn could guide the drafting of legislation, regulations and guidelines/standards to regulate genetics and genomics research, health care provision, forensic applications and associated areas in South Africa.

http://research.assaf.org.za/handle/20.500.11911/106
Summary of survey results

Q: Is there sufficient transparency in the leadership/management of SASHG?
A: “Yes” (92%, n=33) and “No” (8%, n=3)
Summary of survey results continued...

“What is your current affiliation?” (n=42)

Q: Do you think the SASHG fulfils its aims as stated below? If not, why?
Our aims are:
- To promote and advance the practice of science and human and medical genetics in Southern Africa
- To facilitate contact between persons in the different branches of human and medical genetics
- To maintain the highest ethical and professional standards in all its affairs and activities
- To arrange conferences and symposia
- To initiate contact with similar organisations in other parts of the world
- To practice its affairs in a non-profit making and non-discriminatory manner

A: “Yes” (83%, n=30), “No” (17%, n=6)

Comments from members:
- Can do more to facilitate contact with others in the field and with similar organization in other parts of the world.
- The only thing I see, is arranging conferences; don’t have much else from SASHG for the rest of the time. Unless it all happens, but is not shared.
- For most of the above aims my answer would be YES. However, both SASHG and its members can do more to rid our field of the “fly by nights”. E.g. people practicing human genetics but without human genetics training at all. It is not just an SASHG issue, the members should all report this and SASHG should have an effective strategy to deal with it.
- I think more can be done in terms of promotion and advocacy especially related to governmental service delivery.
Summary of survey results continued...

Q: What are your particular needs or requirements from the SASHG?
A:
- Information is key, regular updates on science breakthroughs opportunities, meetings.
- Keeping updated about news, conferences and opportunities in the South African genetics community.
- Job adverts, relevant conference/training/workshop notifications.
- To provide network opportunities with other South African human geneticists. I think we are not aware enough of each other's activities to form sensible collaborations.
- To host a conference and to create a platform of communication.
- More interaction between academic/public sector and private sector.
- That SASHG must advocate strongly for the ethical practice in our field, and take effective measures to root out unethical practice.

Q: What would you like to see more of?
A:
- I think you are doing a great job!
- Proactive engagement with issues - issuing position statements/papers on controversial issues.
- I think the conferences only every 2 years is not enough. We fortunately have dysmorphology teleconferences as well as I’m on the Clinical genetics group.
- Research outputs and job opportunities as well as opportunities to collaborate.
- The correct way of handling NGS results and how it should be reported to doctors.
- Engagement with the media and commentary from the Society in newspapers, radio and TV.
- I think we are a bit quiet during non-conference years.
- Opportunities for student interaction and networking.
- SASHG organised events. Doesn't necessarily need to be a conference, but creating a space for sub-committees, lectures, courses etc.
- Updates on relevant SA genetics publications or notable articles published by SA geneticists.
- I have enjoyed the newsletters and updates.

Q: What, according to you, should be the main purpose of the SASHG?
A:
- Promotion of genetics and related knowledge both to academics and the general public.
- Education, collaboration, establishing standards and guidelines.
- Promote genetics research in Africa to improve on health delivery through cutting edge science, capacity building, collaboration. Embracing the African genetic diversity for the betterment of Africans through African genetic solutions to African health and environmental problems.
- To keep in touch and liaise with all members informing them of all activities and updates; not only with the connections in SA, but internationally as well; as I said, perhaps it does, but we don't get notification.
- To uphold the values of providing access to genetics to as many people as possible, in order to help patients as a long term goal.
- Be the over-arching body dealing with medical genetics in South Africa - promoting research, but also trying to ensure ethical practice. Needs to be more proactive rather than reactive.
- To connect all individuals in the field of Human Genetics in South Africa in as many possible ways (symposia, conferences, e-mails etc).
- Connecting the global and African human genetic groups to the South African group through conferences/workshops/newsletters/job adverts which informs us about the current work and possible collaborations with other human genetics groups.
- I think the aims of the society is trying to promise too much. Stick to conferences and creating a platform from which members can seek advice, guidance and opportunities to contact other researchers.
- Be responsible stewards of human genetics on behalf of the citizens of South Africa.