Dear SASHG members

As we dive into the second half of the year, it really is with disbelief that I say the time has come for the final newsletter from the current YRF team.

Our initial aim for the newsletters was to feature as many different Southern African universities as possible. Looking back over the last two years, we are pleased to have featured the Systems Genetics Research Group (University of Stellenbosch), the Clinical Genetics Unit (University of KwaZulu-Natal), and the Mitochondria Research Laboratory (North-West University). We would like to say a special thank you to all the supervisors and students who have made such excellent contributions. It really has been fantastic to work with each of you.

In this edition of the newsletter, we are excited to give you a feature on the Human Genetics Unit headed by Professor Kathrine Scholtz, based at the University of Limpopo. On page 2, Prof. Scholtz gives us an overview of the unit, followed by project summaries and some personal Q&As from the 4th year (Honours equivalent) students, and an MSc student in this group.

In other news, the SASHG 2019 Congress and the SASBi-SC/SASHG Young Researchers’ Symposium are fast approaching. Registration for both events has now closed. We are excited to welcome all delegates to Cape Town in August! Full programmes can be downloaded from the SASHG Congress website: https://sashg2019.co.za/programme/.

In light of the recent controversy surrounding race in research, a panel discussion will be held at the SASHG 2019 Congress on Tuesday 6th August. The session titled ‘Getting Beyond Race’ will be chaired by Professor Keymanthri Moodley, the Director of the Centre for Medical Ethics & Law. Congress delegates are encouraged to voice their opinions in this interactive event.

The elections for the new SASHG committee (2019 – 2021) are now open. Thank you for all the nominations that were received. Please remember to cast your vote by Sunday 21st July, via the personal voting link that was emailed to you. The new SASHG committee will take over in August, following their announcement at the Biennial General Meeting that will take place on Monday 5th August at the SASHG Congress. We look forward to welcoming our new committee!

As a note from the outgoing SASHG committee, here is a summary of the committee’s achievements:

1. Organising of the SASHG2019 conference in Cape Town.
2. Provided partial sponsorship of >40 students/Postdocs to attend the SASHG conference.
3. Provided partial sponsorship to members to attend various human genetics meetings on behalf of SASHG.
4. Produced a series of newsletters and tried to represent organisations from around South Africa.
5. Produced a statement on the use of racial labels/categories in human genetics research.
6. Prepared a direct-to-consumer genetic testing article, which will appear in the magazine Specialist Forum.
7. Launched a Marketing Awareness campaign in the Specialist Forum.
8. Appointed a social media liaison person.
9. Cleaned up our membership database and removed all dormant members.
10. Conducted a survey to determine the needs of our members.

Finally, as the outgoing YRF representative, I would like to say thank you to everyone in the SASHG community. It was an honour to have been given the opportunity to work with such an incredible team of scientists and people that are the SASHG committee, as well as those in the SASHG community who I have met or corresponded with along the way. I would also like to invite any SASHG members, especially the young researchers, to drop me an email with any comments, questions, ideas, or suggestions for the YRF focus group moving forward.

We hope you enjoy reading this issue of the newsletter, and we wish you all the best for the remainder of 2019.

Warm regards,

Emma Frickel
Email: 17651166@sun.ac.za
Human Genetics Unit, University of Limpopo

The Human Genetics Unit at the University of Limpopo was revived in 2013 within the Department of Medical Sciences, Public Health and Health Promotion. After a number of institutional reshuffles, the Human Genetics Unit was moved under a new Department within the School of Health Care Sciences, the Department of Pre-Clinical Sciences. This department is made up of three units: the Anatomy Unit, the Medical Biochemistry Unit and the Human Genetics Unit. The Human Genetics Unit is a very small unit headed by Professor Kathrine Scholtz, made up of two lecturers and one lab technician (besides Prof. Scholtz). For the past few years, the Human Genetics Unit has focused on reviving the Human Genetics modules offered at the 3rd and 4th year levels within the BSc Medical Sciences program offered by the Department of Pathology and Medical Sciences. Now that the program is running efficiently, Professor Scholtz is changing her focus to community engagement and research, with the main aim of determining the burden of genetic conditions within the Limpopo Province and the establishment of medical genetic services. Prof. Scholtz is currently registered for a MSc (Med) in Genetic Counselling at the University of Cape Town and aims to become an HPCSA accredited intern training site for genetic counsellors in the future as an important step towards bringing genetic services to the province.

The Human Genetics Unit offers a 4th year (Honours equivalent) course in Advanced Human Genetics, as well as MSc and PhD research opportunities. The 4th year level course includes both a coursework and a research component and at the end of the 4th year, the students graduate with a BSc Medical Sciences, majoring in Human Genetics. There are 5 major electives at the 4th year level, including Chemical Pathology, Medical Microbiology, Immunology, Haematology and Human Genetics. In 2019, four male and four female students registered for the Human Genetics major. Each student has to conduct a research project as part of their course requirements. There are currently 3 projects being conducted with this group of students. Additionally, there are 3 MSc students, and Prof. Scholtz is in the process of trying to recruit a PhD candidate for a larger Haemophilia A project.
4th year students feature: Project summaries and Q&As

**Project 1 Title:** Determination of the performance of sequencing and immunohistochemistry techniques in the differential diagnosis of Fibrous Dysplasia and Ossifying Fibroma in archived histopathology samples from the Pietersburg Hospital in the Limpopo Province.

Our research focuses on testing if molecular techniques can be used to diagnose fibro-osseous lesions, specifically ossifying fibroma and fibrous dysplasia. Fibro-osseous lesions are lesions in the jaw characterised by the replacement of bone with benign connective tissue. Ossifying fibroma and fibrous dysplasia have overlapping clinical, radiographic and histological features, which makes it hard to correctly distinguish one from the other, making it challenging to correctly diagnose. Assignment of correct treatment and management is crucial because their aetiology and pathogenesis are different.

Our project aims to evaluate molecular techniques in the diagnoses of these conditions. The molecular techniques that will be evaluated are DNA sequencing of the GNAS gene and immunohistochemical analysis of osteocalcin. Mutations in the GNAS gene have been identified in individuals with fibrous dysplasia, while no mutations within this gene have been reported in individuals with ossifying fibroma. Osteocalcin is a protein responsible for the binding of calcium to the bone tissue. The expression of this protein is reported to be higher in fibrous dysplasia lesions as compared to ossifying fibroma. The results of this research project may contribute to the proper diagnosis and management of fibrous dysplasia and ossifying fibroma.

- Redia Masemola and Ronewa Mudjadji

**Q&A:**

**What is your background and how did you become interested in your field of research?**

In the rural areas, a lot of genetic conditions do not receive the attention and treatment that they require due to the myths associated with them. This made me realise that there is a need to find a way to identify these individuals within the community and provide them with appropriate information and treatment.

- Redia Masemola

**What piece of advice would you give to somebody beginning their career in global health?**

Nothing worth it comes easy, the field of health is broad and a little difficult but if you put your all in your studies there is no way that you won’t make it. Make it your priority to come up with new research ideas to improve the health of people.

- Ronewa Mudjadji

**Project 2 Title:** The determination of the presence of intron 1 and intron 22 inversion mutations in the F8 gene of severe Haemophilia A patients treated at the Pietersburg Hospital in the Limpopo Province.

Our research focuses on determining the presence of intron 1 and intron 22 inversion mutations in the F8 gene of severe Haemophilia A patients treated at the Pietersburg Hospital in the Limpopo province. Haemophilia A is an X-linked recessive bleeding disorder caused by heterogenous mutations in the F8 gene. The severity of the disorder depends on the amount of factor VIII in the blood. Effects can range from mild, moderate or severe depending on the FVIII levels in the blood. This disorder is more common than Haemophilia B, another bleeding disorder involving the F9 gene. Our research project aims to screen severe Haemophilia A patients for the more common intron 1 and intron 22 inversions in the F8 gene with the main purpose of being able to identify at-risk individuals within families as well as for pre-natal testing and carrier testing in families with familial Haemophilia A.

- Andrew Mpe, Dipolelo Mokaila and Thabo Semenya

**Q&A:**

**How do you like to relax?**

I lock myself up in my room, get into bed, listen to electronic music and Google rare diseases and try to understand what causes them and their manifestations. I’m interested in the “therapeutic approach” aspect of research in genetics. I love reading up on gene and chromosome therapies as these are the biggest hope of correcting most (if not all) genetic disorders. This is one of the reasons why I chose to major in Human Genetics. Another thing I really love doing is reading up on Philosophy. There’s a Podcast on Spotify called “Philosophize This!” by Stephen West, in which he talks about the Pre-Socratic Philosophy (as in ancient philosophy) all the way up to Modern Philosophy and how each known
Philosopher and their ideas contributed and revolutionised Philosophy as we know it today. I also love reading wisdom-inspired novels. I am currently reading one called “The Power of Your Subconscious Mind” by Joseph Murphy.

- Andrew Mpe

Do you have any secret talents?

I love and enjoy singing. I started singing when I was young. I would take a piece of paper and a pencil and try to write down my own lyrics and sing to my family. I still sing and if I hear a song no matter what I am doing I’ll stop and sing along. I was always in the choir in my high school and primary years.

- Dipolelo Mokaila

What piece of advice would you give to somebody beginning their career in global health?

Choosing a career in the health field is one of the most important decisions that one makes because whether you are at the forefront or behind the scenes, the work you do contributes to the advancement of human knowledge and improving the quality of life. The best way to love what you do is give it your all, and I feel like one can achieve this by being up-to-date with the latest developments in the field, reading published journals and engaging with giants within your field on social media platforms like LinkedIn and Research Gate.

- Thabo Semenya

Project 3 Title: Pilot study: The association of deoxyribose nucleic acid methylation with obesity in black females from the Dikgale Health and Demographic Surveillance System centre in the Limpopo Province.

Our research project focuses on the epigenetics of obesity in a black female population. Epigenetics refers to the field of genetics that focuses on factors that affect gene expression rather than the nucleotide sequence in the gene. There are several epigenetic modifications, including histone modifications, DNA methylation and non-coding RNAs. Our study is an associative study analysing the methylation patterns in normal weight, overweight and obese middle-aged female individuals. Obesity is a multifactorial condition accompanied by a multitude of co-morbidities such as metabolic syndrome, type 2 diabetes mellitus and cardiovascular diseases, and is thus detrimental to an individual’s health. The prevalence of obesity is alarming and still rising, yet the molecular basis of obesity is still unknown. The proposed research may assist to determine whether differential methylation patterns of the epigenome is associated with obesity. The identification of target genetic regions could assist with therapeutic interventions aimed at reversing methylation profiles associated with obesity.

- Royce Mabale, Dolphin Mogajane and Rudolph Serage

Q&A:

How do you like to relax?

I like to take long walks or listen to music (electro or tropical house is a preference).

- Dolphin Mogajane

What piece of advice would you give to somebody beginning their career in global health?

Don’t pursue it if you’re not passionate about it. The journey will most definitely be a long one but if you’re passionate enough no obstacle will hinder your success and you’ll definitely enjoy yourself.

- Rudolph Serage

MSc student feature: Project summary and Q&A

Project Title: A Retrospective Study Profiling Congenital Malformations in the Neonatal Unit of the Mankweng Provincial Hospital, Limpopo Province.

A review of the literature indicates that there have been no studies to establish the incidence and prevalence of congenital anomalies in the Limpopo Province in the past two decades. The aim of my study is thus to profile congenital malformations in the Neonatal ward at the Mankweng Tertiary Hospital in order to provide baseline data and a platform for spatial and temporal comparisons. My study may assist in evaluating the potential burden of congenital anomalies in the province and unveil possible determinants with a view to introduce preventive measures and interventions at an appropriate time. Most importantly, the results from this study will also guide future molecular studies that will attempt to answer questions surrounding
congenital anomalies and the how best to prevent
and manage such birth defects in the province.

- Lloyd Magunde

Q&A:
What is your background and how did you become interested in your field of research?

I have been fascinated with science from a young age and it is my desire to follow-on from previous researchers so that science progression never stops. I am a Limpopo resident and following the death of a close friend’s child, I was interested in understanding the scientific reason for his death, as opposed to the reports of human witchcraft. This sparked a desire to better understand unknown deaths and congenital anomalies within this province.

- Lloyd Magunde

The below photo is of the Human Genetics Unit at one of their community engagement visits to a local school.

Photo: Front from left: Dolphin Mogajane, Dipolelo Mokaila, Ronewa Mufadji, Redia Masemola
Back from right: Andrew Mpe, Thabo Semenya, Rudolph Serage, Royce Mabale, Prof. Katie Scholtz, Dima Mohlabe (laboratory technician), and 3 community members