THE YEAR IN REVIEW

- Amanda Krause

As we approach the end of another year, it is important to reflect on the year that has passed and look forward to what lies ahead for the Southern African Human Genetics community. Firstly, I would like to extend my thanks to the entire SASHG committee for their hard work and dedication through the year.

In 2014, human genetics research has continued to flourish, and the South African Human Genome project (SAHGP) has made significant strides (p4). We continue to have challenges in our service areas, and remain desperately short of medical geneticists, genetic counsellors and laboratory personnel across the country. In addition, we need to be training at all levels to ensure continuity of our professions and our field.

There is some light at the end of the tunnel, in that after a year we have made some progress in engaging the National Department of Health towards developing a National Genetics plan.

In 2014 we sadly also lost our dear friend and colleague, Dr Elaine Beckh Arnold (p2).

The SASHG committee has initiated an expertise database for members. We encourage all our members to register on the database and populate it. The success of the database is dependent on investment form all the members.

In addition, we have made some efforts to try to make the Society (and its website) more useful to its members. A number of new management guidelines for Lysosomal Storage Diseases are now available on the website. We are working on a booklet of available diagnostic tests around the country. This should be available in early 2015 via the website. It is important for members to inform us of omissions or errors so that this can be updated. Our focus groups continue to develop their particular areas of interests (p3; p5). A group for medical scientists would be a welcome addition to SASHG.

We look forward to 2015 as an SASHG Congress year when we all have opportunities to interact face-to face. Look forward to seeing you all there.
IN MEMORIAM:

DR ELAINE BECKH ARNOLD

- Amanda Krause

Dr Elaine Beckh Arnold our friend and colleague sadly passed away on 22 June 2014, after being unwell for some months. In 2006, Dr Beckh Arnold joined the Division of Human Genetics, of the National Health Laboratory Service and the University of the Witwatersrand, as a fellow, training in the sub-specialty of Medical Genetics. She obtained her Certificate in Medical Genetics, and was then appointed to a medical genetics consultant post in the Division in 2008, a post she occupied until her untimely passing.

Dr Elaine Beckh Arnold was born on 10 November 1967 in Lusaka (Zambia), where she grew up for the most part on the family farm. She was so fond of the farm, a love which she passed on to her children. They would spend many holidays there. She completed her schooling in Zambia, and went on to study at The University of Zambia, fondly referred to as "UNZA," where she qualified as a doctor in 1992, having won the prize for best graduate in Paediatrics. At this time Elaine and Francis became engaged and she joined him at Shongwe Hospital in Mpumalanga, where she completed her internship. On 24th April 1993 they were married.

Dr Beckh Arnold joined the Department of Paediatrics, University of the Witwatersrand, in 1994 and qualified as a Paediatrician in 1998.

She worked in the Neonatology Unit at Chris Hani Baragwanath Hospital until 2005, during which time she qualified as a neonatologist. She then moved to the Division of Human Genetics, where she worked until she became unwell. She continued to retain strong links with her paediatric colleagues.

Dr Beckh Arnold served as an examiner for a number of specialist examinations of the College of Medical Genetics. She also served as a Senator for the College of Medical Genetics from 2011 until 2014, and in this capacity also served on the Exams and Credentialing Sub-Committee of the Colleges of Medicine of South Africa.

Dr Beckh Arnold is remembered by her work associates as determined, efficient and extremely diligent. She was quiet and dignified and well respected by her peers. She shared her academic knowledge with her colleagues, and showed her leadership skills as a dedicated teacher and a supervisor at clinics, but never sought praise or attention for her work. Her sense of humour was shared with everyone.

She was extremely dedicated to her patients, for whom she often went the extra mile. They, in turn, always showed, and continue to show their appreciation for her.

Dr Beckh Arnold’s devotion to her family- her husband, Francis, her children, Gabi and Graham, her mother and sisters was always evident.

Dr Elaine Beckh Arnold will be remembered and sadly missed by all of her colleagues and family, and particularly those of us who worked closely with her in the Clinical Section of the Division of Human Genetics in Johannesburg. We have lost not only an exceptional colleague, but also a friend.
The GC-SA embarked on a process of developing standards of practice as additional detailed documentation for the scope of practice published on 22 May 2009. The standards of practice was finalised in 2013 and has now been formally approved by the Medical and Dental Board of the HPCSA. The document can be viewed at: www.hpcsa.co.za/PBMedicalDental/Education

Having a clearly laid out description of the practice and training (competencies, reciprocity agreements, continuing professional development and supervision) of an independently registered genetic counsellor is pivotal in the process of creating posts in South Africa. The group has been eagerly awaiting the approval and with this now officially in place all efforts can be directed at creating positions for genetic counsellors.

The profession has been through a difficult time in that there were no positions available for qualified genetic counsellors. This resulted in the two training centres not being able to take on new genetic counselling students. This year started off on a very positive note, which seems be a turnaround for the profession. The Western Cape managed to secure one full-time genetic counsellor post in the province and a full-time lecturer post at the Division of Human Genetics at UCT. Both of these were filled and as a result, UCT were able to take in three genetic counselling students with a commitment to take in three students yearly. In addition, UCT started offering the theoretical aspects of the Masters degree as semester courses for those interested in becoming genetic counsellors and medical geneticists as well as other health care professionals who want to up-skill.

A positive change has also occurred in the Division of Human Genetics at Wits and the NHLS as two full-time permanent posts were made available, with plans to open more posts in 2015. Further posts may also become a reality in KZN.

There are a number of genetic counsellors who practice in the private sector. They are successfully establishing the profession in this sector and in so doing increasing the general awareness of genetic counselling.

The future of the profession is looking very positive. With combined efforts, the group is lobbying to convert contract posts into permanent positions, create additional posts, build private sector and look at ways of expanding the role of the genetic counsellor.

**Important Dates for 2015**

- World Rare Disease Day | 28 Feb
- World Down Syndrome Day | 21 Mar
- World Haemophilia Day | 17 Apr
- AfSHG Meeting | 9 – 13 May
- Cystic Fibrosis Awareness Month | May
- **SASHG 2015 Congress** | 16-19 Aug
- Birth Defects Conference | 21-24 Sept
- HBOC Week | 28 Sep – 2 Oct
- ASHG Meeting | 20 – 24 Oct

Want to find out what other members of SASHG are up to?

Check out the members’ database for research interests and areas of expertise:

http://www.searchsashg.co.za

Don’t forget to update your user profile while you’re there!
The Southern African Human Genome Programme (SAHGP) has made significant and exciting progress over the past 18 months. The SAHGP aims to develop capacity for genomic research in southern Africa, to establish sustainable resources for genomic research in the region and, with time, to translate knowledge and information into improvements in human health in the region. The SAHGP core group met regularly over the past year to promote these aims.

By the end of 2013 samples for the pilot study had been collected and by May 2014, 24 whole human genome sequences were completed.

Three workshops took place in the latter half of 2014 contributing to the original aims of the Programme. Access to the data will be governed by a policy document that is currently under review. The objective is to have a managed access process to ensure that the data are, in the first instance, analysed within South Africa and published, before making it more widely accessible.

In order to streamline the analysis of the genomes, three bioinformaticists joined the core group of the SAHGP: Scott Hazelhurst (Wits), Fourie Joubert (UP) and Nicky Mulder (UCT). In addition a technical team was assembled to do the analysis in preparation for the first publication.

**Workshop 1 - Training in NGS analysis:** Twenty-two participants attended the course from 9 South African Institutions and the course content included the development of UNIX skills, handling NGS data, PLINK and an introduction to human population structure.

**Workshop 2 – Understanding and characterising Southern African genomes:** The thirty three participants had several theory presentations on the SAHGP and genome analysis and also deliberated on the types of analyses that could be performed on the 24 South African genomes. The Technical Team produced an excellent report at short notice and this was a great opportunity to examine the preliminary analyses. There were two interactive sessions aimed at generating ideas for further analysis and for the next phase of the SAHGP.

**Workshop 3 - Ethical, Legal, Social Implications (ELSI) and Educational aspects of the SAHGP:** The focus was on the various stakeholders of the SAHGP: researchers, study participants, the communities, members at various levels of governance, ethical review boards, data management centres, funders, publishers and the general public. ELSI-type issues are the ‘glue’ that holds the project together so that it is fair, respectful, and transparent and communicated in ways to ensure that people are informed about the research. The ELSI workshop was structured in two parts and about 60 people attended. The first was a scientific meeting including invited speakers and researchers across disciplines interested in this discourse, and the second session was more relevant to the general public, media and interested stakeholders to advance the SAHGP mission of public engagement and education.

The objectives for 2015 are to finalise the data access policy and to publish several papers on the data produced by the SAHGP in collaboration with research groups in South Africa. Once the policy has been finalized it will be advertised to the Human Genetics community so that they may apply. Furthermore, there will be additional public outreach and education events. This will be an important year to deliberate on the nature of the future expansion of the SAHGP, and to ensure that it moves toward its vision of improving the quality of life of South Africans through an understanding of the genetic diversity of its peoples.
2014 has been a rather static year with regard to the medical geneticists. Whilst work continues in the background, and at various organisational levels, to raise awareness of our ongoing challenges, the SASHG has been highly supportive as we advocate for our specialty.

We are still far behind the curve in terms of numbers, remaining at about 10 fulltime equivalent medical geneticists in South Africa, 46 short of the World Health Organisation recommendations for our population.

There are currently 6 registrars in training, and we hope to increase this number in the coming year.

A sincere thank you to our focus group chairperson, Dr Bertram Henderson, who takes on much of the regulation, scope of practice and essential administration of our discipline.

We look forward to rebuilding much of what has been lost in the past few years, and enter 2015 on a positive note.

Have you visited the SASHG website recently? You can find information pertaining to:

- Treatment guidelines for specific genetic conditions;
- HPCSA regulations for medical scientists, genetic counsellors, and medical geneticists;
- SASHG membership guidelines and forms; and so much more!

www.sashg.org
**SAIDA – A WAY FORWARD**  
- Jo-Anne Richards & Helen Malherbe

The last couple of years have been an extremely challenging period for the South African Inherited Disorders Association (SAIDA). In 2013, the SAIDA office closed due to a financial crisis and critically low funds. Judy Christie, who has worked as SAIDA secretary for 14 years, retired and Sarah Walters, the SAIDA trainer, was forced to find other employment.

At a meeting of independent stakeholders in Pretoria in April 2014 and closely followed by a meeting of the SAIDA committee in May, it was decided that SAIDA’s activities should take a different direction in the current circumstances. Due to personal commitments making it impossible to continue in a lead role at SAIDA despite her continued enthusiasm, Jo-Anne Richards resigned as National Chair and nominated Helen Malherbe, who was elected as the new chair.

Historically, SAIDA has focused on providing support for those affected by congenital disorders; education and public awareness; and supporting research into the causes, treatment and prevention of congenital disorders in South Africa. In the current economic climate it is no longer possible to continue with this same approach.

For the immediate future, SAIDA’s efforts will therefore be centered on advocacy to help garner the necessary political will and recognition of inherited disorders, to develop genetic services for their care and prevention. Public awareness activities and support will continue wherever possible, as enquiries continue to be received on a weekly basis from across the country. Updating the SAIDA website will help facilitate this role, so that where possible, patients can directly contact the relevant support groups. Google has radically changed the role of support groups for those with internet connectivity – and we need to be mindful of the changing needs of our members.

A major part of SAIDA’s work has been education and training. SAIDA has implemented the Medical Genetic Education Programme (MGEP) courses since 2004, training primary healthcare, labour ward, neonatal and paediatric nurses to provide basic medical expertise to poorly serviced, under-resourced areas in some of the poorest regions of the country. MGEP was originally developed by SAIDA with funding from the March of Dimes Foundation followed by the Department of Health (DOH).

Between 2004-2011, over 300 nurses were trained countrywide and the courses implemented on very limited funding in partnership with the Wits University Department of Human Genetics, with DOH funding and support. In 2011, MGEP training ceased due to a lack of funds.

Since an even greater need exists for education and training in medical genetics, the SAIDA MGEP is currently being revised to make it more relevant and self-sustainable. The new content will be piloted in 2015 and is being developed to ensure SETA compliance and to better meet the health needs of the country.

SAIDA is functioning on a voluntary basis, with zero funding, but with a lot of passion and enthusiasm in the face of many challenges. Udo Hellburg has joined SAIDA as a volunteer for a six-month period and is assisting with the current refocus of SAIDA, especially the website.

It is hoped that 2015 will bring many opportunities along with its many challenges. By pulling together as a community of stakeholders, we can help bring about change in the medical genetic services our country needs so desperately needs.
Save the dates
SASHG 2015 Conference

The 16th Biennial Congress will take place in Centurion, 16 - 19 August 2015.
The theme will be “The Next Generation”
Further information will follow early in the New Year

Please remember to renew your SASHG membership and encourage new students to join the Society.

Students must be members of the Society in order to apply for a scholarship to attend next year’s Congress!

Contact Zané Lombard for details:
sashgtreasurer@gmail.com

Wishing you well over the festive season & looking forward to an excellent 2015!