Provisional Programme – SASHG 2015

21 May 2015

Sunday, 16 August 2015			
10h00	13h00	Genetic counsellors Meeting	
		Keynote: **Dawn Allain**	Title: TBA
		Genetic Counsellors-South Africa AGM	
14h00	**14h45**	**Opening Session**	
14h00	14h10	Chair SASHG – Amanda Krause	Opening and welcome
14h10	14h30	TBA	Opening Address
14h30	14h40	Chair Organizing Committee	SASHG2015
14h40	15h00	*Comfort break*	
15h00	**18h00**	**SA Human Genome Programme**	
		To be announced	
18h00	**20h00**	*WELCOME COCKTAIL PARTY*	
Monday, 17 August 2015			
8h30	**10h15**	**Mendelian disorders / Molecular Pathology**	
8h30	9h15	**Keynote:Bart Loeys**	Title: TBA
9h15	9h30	Nadia Carstens	Novel mutation in the CHST6 gene causes macular corneal dystrophy in a black South African family
9h30	9h45	Elana Vorster	Determining the molecular basis of spinal muscular atrophy (SMA) in the black South African population
9h45	10h00	ThandiswaNgcungcu	Non-coding variants segregate with disease in South African families with keratolytic winter erythema (KWE)
10h00	10h15	Celia van der Merwe	Curcumin shows protective effects in a PINK1 siRNA-mediated model of Parkinson's disease -
10h15	10h45	*Tea break*	
10h45	**12h15**	**Mendelian disorders/ Complex traits / Genetic epidemiology / Ancestry**	
10h45	11h00	Danielle Smith	Beyond our borders: evidence for a Spinocerebellar ataxia type 7 founder effect in southern Africa
11h00	11h15	Cheryl Stewart	Finger on the pulse – The utility of establishing a national CF registry
11h15	11h30	Jeanne van Rensburg	Cystic fibrosis and rarefaction: Unification through diversity
11h30	11h45	Melissa Nel	The African-specific *-387 C>T TGFB1* Promoter Polymorphism is functional and associates with an 'African complication' in juveniles with Myasthenia Gravis
11h45	12h00	Mario Moller	Population admixture: friend or foe in TB susceptibility
12h00	12h15	Caitlin Uren	Investigating fine-scale population structure between the Nama and ≠Khomani San of South Africa

12h15	13h15	*Lunch Break*	
13h15	**14h15**	**POSTER VIEWING - SESSION 1**	
14h15	14h30	Hadassa Goldfein	Associations between mtDNAhaplogroup variation and sensory neuropathy in South Africans using d4T
14h30	14h45	Nathaniel McGregor	The identification of novel genes in anxiety disorders: a gene x environment correlation and interaction study
14h45	15h00	Melvin AnyasiAmbele	Genome-wide analysis of gene expression during adipogenesis in human adipose-derived mesenchymal stromal cells (ASCs)
15h00	15h30	*Tea break*	
15h30	17h00	SASHG Biennial General Meeting	
18h00	22h00	*DINNER*	*Social*
Tuesday, 18 August 2015			
8h30	**10h15**	**Association Studies**	
8h30	9h15	**Keynote: John Kemp**	**TBA**
9h15	9h30	Tinashe Chikowore	Evaluation of common variants associated with Type 2 Diabetes in the Asian and European ethnicities among the Black South African population of Tswana descent
9h30	9h45	Liesl Hendry	The genetics of blood pressure in black South African individuals from the Birth to Twenty cohort
9h45	10h00	VenessaPillay	The Metabochip as a tool for identifying genetic markers of obesity risk in a South African black population
10h00	10h15	Ellen Ovenden	Investigating the functional significance of genome-wide variants associated with treatment response in schizophrenia
10h15	10h45	*Tea break*	
10h45	**11h15**	**Next Generation sequencing / Bioinformatics / Computational challenges**	
10h45	11h00	Brigitte Glanzmann	Next generation sequencing methods to identify pathogenic mutations implicated in human diseases: Holy Grail or tin cup?
11h00	11h15	Nicole van der Merwe	Functional DNA mismatch repair variant missed in exomes of BRCA-negative breast cancer patients using variant calling with the public human genome reference sequence (hg19)
11h15	11h30	Lisa Roberts	Whole exome sequencing to address the missing heritability of inherited retinal diseases in indigenous Africans
11h30	11h45	ShareefaDalvie	Bipolar Disorder: Whole-genome sequencing in a large Afrikaner family **-**
11h45	12h00	TBA	TBA
12h00	**13h00**	**POSTER VIEWING - SESSION 2**	
12h45	13h45	*Lunch break*	
13h45	**15h30**	**Clinical genetic/Genomic testing**	
13h45	14h30	**Keynote:RalphPfundt**	SNP array analysis - the added value of genotyping information in a spectrum of samples

14h30	14h45	KarenFieggen	Outcome of targeted MLPA CNV testing at a paediatric genetic clinic in South Africa
14h45	15h00	Fahmida Essop	Unique SNPs found in the South African Black population can lead to misdiagnosis of common genetic tests
15h00	15h15	Craig Kinnear	Mutation Screening for Primary Immunodeficiency disorders in a tuberculosis endemic region: a South African perspective
15h15	15h45	*Tea break*	
15h45	16h45	Clinical Genetics Focus Group - Meeting	
19h00	23h00	*GALA DINNER*	*Prize giving*
Wednesday , 19 August 2015			
8h30	**10h15**	**Genetic counselling / Support groups**	
8h30	9h15	**Keynote: Dawn Allain**	TBA
9h15	9h30	ChantelleScott	Development of an informational video regarding prenatal diagnosis for Down syndrome at Tygerberg Hospital
9h30	9h45	MardelleSchoeman	Genetic counselling for Non-Invasive Prenatal Testing: a report on 308 cases
9h45	10h00	Helen Malherbe	The past, present and future of the Southern African Inherited Disorders Association
10h00	10h30	*Tea break*	
10h30	**12h00**	**Future perspectives**	
10h30	11h00	**Keynote: ReinardHiller**	Precision medicine
11h00	11h30	Keynote: TBA	Genome editing
11h30	11h45	Jacqui Greenberg	New technologies, new challenges: guidelines for the ethical oversight of informed consent for the use of induced pluripotent stem cells for future research purposes
11h45	12h00	SASHG Chair	*Closing Remarks*
12h00	13h00	*Lunch break*	