H3Africa: Human Heredity and Health in Africa

Travel Fellowships

We have the pleasure of announcing the call for applications for the **H3Africa Travel Fellowship Awards*** to the **13th H3Africa Consortium Meeting (8 - 12 April 2019)** to be held in **Tunis, Tunisia**. Successful applicants will be expected to present their work[#] and to take part in consortium meeting activities.

Eligibility: This opportunity is open to early-career stage fellows[ǂ], including postgraduate students, postdoctoral fellows, research fellows, and other trainees. Preference will be given to students and fellows who are attending for the first time. The fellowship is open to all H3Africa projects irrespective of data availability.

Application Deadline: **31 January 2019 11:59PM CAT**

Selection Outcome: The successful applicants will be announced by **Mid February 2019.**

Application Guidelines: Please complete the online application, (note you will be required to register or login on HtrainDB before completing and submitting the application)

Required components of the application include:

- Abstract (max. 350 words)[#]
- Motivation statement (max. 200 words) as to why you should receive this award, with an emphasis on your capacity development and your activities in the broader H3Africa Community.
- Letter of support from PI
- One-page CV
- Passport information (photo) page

Important Dates

- Call Opens: **Friday 22 December 2019**
- Outcome of Application: **mid February 2019**
- Call Closes: **Thursday 31 January 2019**

Cancellation Policy

If awarded this fellowship and travel has been booked and paid for and you **cannot attend**, please provide an explanatory letter of cancellation **as soon as possible** as your H3Africa Project will be held liable for costs incurred.

Contacts

For queries please contact: Rolanda Julius (rolanda.julius@uct.ac.za) or Michelle Skelton (Michelle.Skelton@uct.ac.za), H3Africa Coordinating Centre, University of Cape Town, Anzio Road, Observatory, 7925, Cape Town, South Africa

*The Travel Fellowship is subject to availability of funds.
ǂ Eligibility excludes fellows who have obtained a doctorate degree (MD, PhD, or equivalent degree) more than five years ago.
Abstract selection will subject presenters to either a poster, an oral speed- or formal presentation.

Appendix

GUIDELINES FOR THE ABSTRACT:

Please see examples of abstracts ***below.***

If you **have data** please include the following sections in your abstract:

- Title
- Authors (*next to the applicant's name)
- Introduction
- Objectives
- Methodology
- Preliminary Results
- Next Steps

If you **do not have data** and would like to present your project proposal please include in your abstract:

- Title
- Authors (*next to the applicant's name)
- Introduction
- Objectives
- Methodology
- Next Steps

If you would like to **present a literature review** please include in your abstract:

- Title
- Authors (*next to the applicant's name)
- Introduction
- Objectives
- Methodology
- Next Steps

GUIDELINES FOR THE POSTER, ORAL SPEED AND FORMAL PRESENTATIONS:

Poster dimensions should be size A0 (841 x 1189 mm) and should be in *portrait view*. However please note that if you do not have the resources to print a large laminated poster you may print it per slide in black and white in clear scientific text. Oral presentation guidelines will be communicated upon selection.

EXAMPLES OF ABSTRACTS

Genetic substructure analysis of a sub-Saharan Paediatric HIV Cohort from Botswana.

Gaone Retshabile (1), Sununguko W. Mpoloka (1), Adeodata Kekitiinwa (2), Gabriel Anabwani (3), Graeme Mardon (4), Neil Hanchard (4)

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(2) Baylor College of Medicine Children's Foundation-Uganda, Kampala
(3) Botswana-Baylor Children's Clinical Centre of Excellence, P/Bag BR 129, Gaborone, Botswana
(4) Dept. of Molecular and Human Genetics, Baylor College of Medicine, One Baylor Plaza, Houston Tx 77030

INTRODUCTION: The Southern-African population of Botswana is an amalgam of multiple ancestries based on cultural identity and language. Most speak Benue-Congo Bantu languages and a smaller proportion speaks Khoisan and European languages. However, genetic substructure data on this population is limited. Allele frequency distributions can stratify based on subgroups within a population resulting in genetic substructure. Such stratification may be due to different demographic events between the subgroups in the population and can lead to allelic differences between cases and controls in a genetic association study. This in turn might lead to spurious conclusions in genetic association studies from a population with substructure. We present substructure analysis based on whole exome sequencing data from a paediatric HIV disease-progression cohort from this population, which is part of the Collaborative African Genomics Network (CAfGEN). METHODS: After IRB approvals, DNA from 164 study participants was sequenced on the Illumina HiSeq platform. Variants were then joint genotyped with GATK v.3.50 and filtered for quality metrics with VCFTools v.01.1.12 and PLINK v 1.90b3.36. Principal component analysis (PCA) with SNPRelate v1.2.0 on 61,600 markers was used to infer relationships between Botswana and the 1000 Genomes populations. Then to assess substructure within the Botswana cohort we ran PCA on 1,626,368 independent autosomal markers. After which 590,588 markers shared between Botswana and Uganda were used to assess genetic structure within the CAfGEN cohort. Additional structure analysis was carried out with ADMIXTURE v1.3.0 using unsupervised clustering. RESULTS and FUTURES DIRECTIONS: PCA showed the Botswana cohort to cluster within the African populations of the 1000 Genomes. It also showed that the Botswana population was also distinct from West and East-African populations. Moreover, it also suggested structure between Botswana and Uganda within the CAfGEN cohort. Self-reported ancestry and language within the Botswana cohort suggests minimal substructure based on both PCA and unsupervised cluster analysis. In conclusion, confounding due to substructure from within the Botswana cohort is unlikely but any combined CAfGEN genetic association analysis would need to account for substructure between the Botswana and Ugandan populations. Future directions include additional comparison with other Southern-African populations and haplotype based fine structure analysis.

An expert review of pharmacogenomics of Sickle Cell Disease therapeutics: Not yet ready for global precision medicine.

Khuthala Mnika (1), Gift Pule (1), Collet Dandara (1), Ambroise Wonkam (1, 2)

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(2) Department of Medicine, Faculty of Health Sciences, University of Cape Town, Republic of South Africa.

BACKGROUND: Sickle Cell Disease (SCD) is a blood disease caused by a single nucleotide substitution (T > A) in the beta globin gene on chromosome 11. The single point mutation (Glu6Val) promotes polymerization of hemoglobin S (HbS) and causes sickling of erythrocytes. Vaso-occlusive painful crises are associated with recurrent and long term use of analgesics/opioids and hydroxyurea by people living with SCD. AIM: The present analysis offers a state of the art expert review of the effectiveness of pharmacogenomics/genetics of pain management in SCD, with specific focus on hydroxyurea, and opioids. METHODS: The literature search used the following key words: SCD, pharmacogenomics, pharmacogenetics, pain, antalgics, opioids, morphine and hydroxyurea. The literature was scanned until March 2016, with specific inclusion of targeted landmark and background articles on SCD. RESULTS: Surprisingly, our review identified only a limited number of studies that addressed the genetic/genomic basis of variable responses to pain (e.g., variants in OPRM1, HMOX-1, GCH1, VEGFA COMT genes), and pharmacogenomics of antalgics and opioids (e.g., variants in

OPRM1, STAT6, ABCB1 and COMT genes) in SCD. There has been greater progress made towards identifying the key genomic variants, mainly in BCL11A, HBS1L-MYB or SAR1, that contribute to response to hydroxyurea treatment. However, the complete picture on pharmacogenomics determinants of the above therapeutic phenotypes remains elusive. Strikingly, no study has been conducted in sub-Saharan Africa where majority of the patients with SCD lives. CONCLUSION: This study alerts the broader global life sciences community towards the existing disparities in optimal and ethical targeting of research and innovation investments for SCD specifically, and precision medicine and pharmacology research broadly.

Influence of the Country of Origin on the Microbiome and Fatty Acid Composition Human Milk - Four Countries Study.

Linderborg, Kaisa M (1), du Toit, Elloise (2), Kumar, Himanshu (3), Carmen Collado, Maria (4), Kulkarni, Amruta (1), Zhang, Yumei (5), Isolauri, Erika (6), Salminen, Seppo (3), Yang, Baoru (1)

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(2) Division of Medical Microbiology, Department of Pathology, University of Cape Town
(3) Functional Foods Forum, University of Turku, Finland
(4) Institute of Agrochemistry and Food Technology. National Research Council (IATA-CSIC), Valencia, Spain
(5) Department of Nutrition & Hygiene, School of Public Health, Peking University Health Science Center, Beijing, China
(6) Department of Pediatrics, University of Turku, Finland

Breast feeding results in long term health benefits in the prevention of communicable and non-communicable diseases at both individual and population levels. Geographical location directly impacts the composition of breast milk including microbiota and lipids. The aim of this study was to investigate the influence of geographical location, i.e., Europe (Spain and Finland), Africa (South Africa) and Asia (China), on breast milk microbiota and lipid composition in samples obtained from healthy mothers after the first month of lactation. Altogether, 80 women (20 from each country) participated in the study, with equal number of women who delivered by vaginal or caesarean section from each country. Lipid composition particularly that of polyunsaturated fatty acids differed between the countries, with the highest amount of n-6 PUFA (25.6%) observed in the milk of Chinese women. Milk microbiota composition also differed significantly between the countries ($p=0.002$). Among vaginally delivered women, Spanish women had highest amount of Bacteroidetes (mean relative abundance of 3.75) whereas Chinese women had highest amount of Actinobacteria (mean relative abundance 5.7). Women who had had a caesarean section had higher amount of Proteobacteria as observed in the milk of the Spanish and South African women. Interestingly, the Spanish and South African women had significantly higher bacterial genes mapped to lipid, amino acid and carbohydrate metabolism ($p<0.05$). Association of the lipid profile with the microbiota revealed that monounsaturated fatty acids were negatively associated with Proteobacteria ($r= -0.43$, $p<0.05$), while *Lactobacillus* genus was associated with monounsaturated fatty acids ($r= -0.23$, $p=0.04$). These findings reveal that the milk microbiota and lipid composition exhibit differences based on geographical locations in addition to the differences observed due to the mode of delivery.