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10 June 2021

Pan-African study identifies genetic component to rheumatic heart disease risk in Black Africans

From left to right: PHRI lab manager Reina Ditta, Tafadzwa Machipisa, Prof Bongani Mayosi, Babu Muhamed and senior author, Gui Paré.

A study published in JAMA Cardiology is the largest of its kind to provide insights into the genetic determinants of rheumatic heart disease (RHD) in Africa. RHD is a sequela (effect/result) of rheumatic fever, characterised by permanent heart valve damage. Despite

being completely preventable, RHD remains rampant in poor communities where there is widespread overcrowding and poor access to health care.

Globally, RHD claims as many as 350 000 lives annually and constitutes one of the leading causes to need cardiac surgery in sub-Saharan Africa (SSA), which carries a quarter of the global disease burden of approximately 39 million people.

Coordinated by Dr Mark Engel, an epidemiologist in the Department of Medicine at the University of Cape Town (UCT) and researchers at UCT, The Genetics of Rheumatic Heart Disease (RHDGen), the study was inspired by late renowned UCT cardiologist Professor Bongani Mayosi and conceptualised in partnership with colleagues at the universities of Manchester and Newcastle in the United Kingdom and Population Health Research Institute (PHRI)/McMaster University in Canada. Mayosi, a pioneer and global leader in his field, had over the years translated his research on rheumatic fever into policies adopted by the African Union, the World Heart Federation, the World Health Organization and the World Health Assembly.

"Our findings may provide insight as to why RHD is even more common in Black Africans, than in previously studied populations. Our results will further allow interstudy analyses to get the bigger picture on a global population scale, to find global solutions to the RHD epidemic," says Tafadzwa Machipisa, a final-year UCT PhD student who led the complex genetic analysis under the guidance of Guillaume Paré, joint senior author on the paper, and director of the genetics and molecular epidemiology laboratory where the DNA for the RHDGen study was analysed.

The genome-wide association study has discovered a 'locus' (genetic location on a chromosome) associated with RHD susceptibility in Black Africans, among the 4809 individual participants (made up of ethnically matched cases and controls) and 348 participants in families, in Kenya, Mozambique, Namibia, Nigeria, South Africa, Sudan, Uganda, and Zambia. "The next step is to further investigate our top candidate genes, using functional analyses and explore biomarker-based methods to understand the process of disease development," said Paré.

"The RHDGen Study, led by UCT, provides a striking example of continental collaboration and international partnership at tackling a much-neglected disease, despite its clinical prominence, while contributing to building research capacity on the African continent for the next big project," said Professor Liesl Zühlke, acting dean of research at UCT's Faculty of Health Sciences, paediatric cardiologist and co-principal investigator.

"We are grateful for the generous funding of the Wellcome Trust who helped realise the vision of our mentor, friend and colleague, Bongani Mayosi," commented Engel.

He added: "We are grateful to all participants for taking part in this study, as well as the members of the Mayosi Research Group, notably the late Veronica Francis, and RHDGen consortium across the collaborating sites."

The RHDGen Network is a project within the Human Heredity and Health Africa Consortium (H3Africa).

Read the full study.

Photos of Mark Engel, Liesl Zühlke, Bongani Mayosi, Tafadzwa Machipisa, and Guillaume Pare found at: https://drive.google.com/drive/u/1/folders/0ANy6F4ZtO2f5Uk9PVA

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