

African genomics project takes shape at Cape Town meeting

CAPE TOWN — More than 200 medical researchers met under sunny skies here on 4 and 5 March to discuss practical ways for the African continent to start benefiting from advances in genomics.

The meeting's aim was to inform the design of the \$37 million, five-year Human Heredity and Health in Africa (H3Africa) initiative. The initiative, funded by the Wellcome Trust, a UK medical research charity, and the US National Institutes of Health (NIH), aims to bring modern medical technology to bear on Africa's heavy disease burden.

Africans are extremely genetically diverse—yet little is known about this variance and its health impact. To date, three quarters of the thousands of genetics studies completed worldwide have been conducted on populations of European descent. Africans are also poorly represented in international genetics projects such as the HapMap and 1,000 Genomes projects.

This gap presents both an opportunity and a challenge for Africa, NIH director Francis Collins said at the meeting. The rapidly falling cost of sequencing and genetic analysis has put the technology within reach for cash-strapped African researchers. Moreover, the importance of Africa as the birthplace of humanity makes African genetics an important and intriguing area of study, he said.

But the continent's poor health systems—including scant medical data, low research capability and a lack of trained health professionals—means this research effort risks being managed outside the continent, he continued. “We need capacity building in areas where disease occurs.”

The H3Africa initiative plans to help



Ready for a close-up: Sudanese children.

plug Africa's genomics knowledge gap by addressing shortfalls in equipment, training and regulations. Although the exact remit for H3Africa remains flexible, a white paper presented at the Cape Town meeting set out a number of possible activities for the initiative. These include the development of regional centers of excellence in genotyping and sequencing and a continent-wide bioinformatics network.

Gauging reactions

Research suggests that reactions to commonly used drugs vary greatly between African

populations. For instance, the presence of a duplication within *CYP2D6*, a gene associated with adverse reaction to codeine, a common painkiller, varies widely between African populations, from being totally absent among South Africa's Bantu population to being present in 29% of Ethiopians and nearly 40% of Algerians.

Other studies hint to the danger of using studies on Europeans as a basis for clinical care in Africa. One paper found that African-Americans had a higher frequency of a variant in the gene *TPMT* associated with deafness induced by the cancer drug cisplatin than did people of European ancestry (*Nat. Genet.* 41, 1345–1349, 2009). With cancer cases a “looming epidemic” in Africa, such insights are increasingly valuable, says Michael Hayden, a geneticist at the University of British Columbia in Vancouver, Canada who attended the meeting.

The H3Africa initiative will build on and link local research elements, such as the South African National Bioinformatics Institute based near Cape Town and the Biosciences eastern and central Africa Hub based in Nairobi, Kenya.

“African researchers talk to people in Europe; they talk to people in the US. But they don't talk to each other,” Charles Rotimi said at the meeting. “I don't think this is because they don't like each other. They are just following the money,” added Rotimi, director of the NIH's Center for Research on Genomics and Global Health and one of the founders of the H3Africa initiative.

The white paper presented at the meeting set out some ambitious projects that may or may not be funded as part of the initiative. One is a plan to collect samples from 10,000 people—100 individuals from 100 ethnic African populations—to map genetic diversity on the continent. Another is a continental repository to store all the biological samples gathered by the H3Africa project in one place.

The initial funding will not stretch to pay for all these projects, but the funders want H3Africa to act as an umbrella that will enable others to step in and sponsor projects, making use of H3Africa infrastructure. “I think lots of funders will be interested in using the H3Africa network in specific disease areas,” said Collins.

H3Africa's first call for proposals is due in early summer, and the first awards are expected to be announced next year.

Linda Nordling

Moreover, 69 of the primary trials disclosed at least one author with financial ties to industry, yet none of the meta-analyses reported these disclosures. And whereas 126 of the trials reported at least one author being employed by the pharmaceutical industry, none of the meta-analyses made mention of these ties.

To create more transparency, Thombs and his colleagues suggest developing disclosure guidelines for financial conflicts for meta-analyses. According to Ciprian Jauca, a managing editor at the Cochrane Hypertension Review Group in Vancouver, Canada, discussions about

this idea are ongoing within the Cochrane Collaboration, the parent organization that coordinates the publication of many health-related reviews.

But not all investigators agree that more guidelines are needed. “Meta-analyses are already so onerous to read,” says Colin Baigent, an expert in clinical trial design at the University of Oxford, UK. Instead, Baigent argues that judgments about disclosing conflicts should rest with authors and reviewers. “There is a whole industry generating rules about meta-analyses and it is already so challenging,” he says.

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