# Johns Hopkins to participate in 1000 Genomes Project

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Researchers at the McKusick-Nathans Institute of Genetic Medicine (IGM) at Johns Hopkins will join other national and international scientists in the 1000 Genomes Project, an ambitious effort that will involve sequencing the genomes of numerous people from around the world to create the most detailed and medically useful picture to date of human genetic variation.

“We have made considerable headway in creating a map of common DNA variation in humans, the HapMap Project, and applied it with great success toward understanding many common diseases,” says Aravinda Chakravarti, Ph.D., professor of medicine, pediatrics and molecular biology and genetics at Hopkins and a member of the IGM. “It is apparent that more detailed maps will speed up disease gene discovery for the many illnesses we still fail to understand. It’s crucial to complete the catalog of common variation in DNA, the small fraction of genetic material that varies among people and leads to differences in susceptibility to disease, drug response or reaction to environmental factors, and the 1000 Genomes project will home in on these differences.”

By sequencing the genomes of at least 1,000 people, the project will produce a catalog of genetic variants that are present at 1 percent or greater frequency in the human population across most the genome. The 1000 Genomes Project builds on the human haplotype map - a map of genetic variants - developed by the International HapMap Project. The HapMap already has helped researchers discover more than 100 regions of the genome containing genetic variants associated with risk of common human diseases, such as diabetes, age-related macular degeneration, prostate and breast cancer, and coronary artery disease, including work at Johns Hopkins on sudden cardiac death.

The new map will give researchers important clues to which genetic variants might be causal, including more precise information on where to search for causal variants.

“This new project will increase the sensitivity of disease discovery efforts across the genome fivefold and within gene regions at least tenfold,” said Francis S. Collins, M.D., Ph.D., director of the National Human Genome Research Institute. “By harnessing the power of new sequencing technologies and novel computational methods, we hope to give biomedical researchers a genome-wide map of variation down to the 1 percent level. This will change the way we carry out studies of genetic disease.”

“Most common diseases, such as diabetes and heart disease, are influenced by many common genetic variants, each with a weak effect but cumulatively a strong effect on risk,” says Chakravarti. “The new catalog of gene variants will follow up our genome-wide disease hunts so that we can read off almost all the variants in that region and pursue functional studies to nail down the direct contributors to disease.”

As with other major human genome reference projects, data from the 1000 Genomes Project will be made swiftly available to the worldwide scientific community through freely accessible public databases.

The detailed map of human genetic variation will be used by many researchers seeking to relate genetic variation to particular diseases. In turn, such research will lay the groundwork for the personal genomics era of medicine, in which people routinely will have their genomes sequenced to predict their individual risks of disease and response to drugs.

Source: Johns Hopkins Medical Institutions