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## A 500,000-person study?

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The National Institutes of Health (NIH) is considering undertaking the largest population-based study ever done in the United States. NIH issued a [request for information](#) (RFI) from researchers earlier this month about the questions a large cohort study on the gene-environment interactions involved in common human diseases might ask, and how the study might be constructed.

A project of this kind is "the logical next step beyond the [mapping of the human genome](#) and doing case studies," said Terri Manolio, director of the National Heart, Lung, and Blood Institute's [epidemiology and biometry program](#).

Such a project would try to survey a representative sample of the US population, explained Manolio, and may include as many as 500,000 participants from all geographic, racial, ethnic, and socioeconomic groups defined in the most recent US census. No funds have been appropriated for the project yet, and NIH officials are hesitant to speculate on how much it might cost.

Manolio said that NIH officials hope to find a way to incorporate data from previously conducted studies of individual diseases. "We want to include existing cohorts," she said, "but we have to decide, how feasible is it to add on to these disease studies?"

[Alan Guttmacher](#), deputy director of the National Human Genome Research Institute (NHGRI), said that while there are questions about how the genotyping should be done - for example, whether it should all be done at once or if it should wait until the technology improves - identifying the environmental factors on which the study should focus, such as diet, lifestyle, and geographic area, might be the real challenge. "We don't have the expertise or the imagination to come up with all the hypotheses we want to answer with this data," he told us.

While the project could be likened to the [UK BioBank](#) and Iceland's deCODE Genetics, Guttmacher said, its objectives and approach would not be exactly the same. "The general idea is not dissimilar," he said, "but how we get there... would be different." For example, many of the minority ethnic groups that should be included in a US study are not present at all in the United Kingdom.

So far, the response from the research community has been generally positive, Manolio told us. "People are aware that there is room for something like this," Guttmacher said, adding that he has been "quite impressed" by the fact that scientists involved in similar research seem excited rather than threatened by the idea of this study.

"We know that a lot of genes contribute to [disease] risk, but aren't the only factor involved," said [Terri Beaty](#), an epidemiologist at the Johns Hopkins University Bloomberg School of Public Health. A study of this kind could be "potentially very useful," she said, especially if we ever hope to attain the reality of personalized medicine.

NIH officials are unsure about how long the planning phase will last, how soon the project will get underway, and how soon it will start providing meaningful information. "Ideally, we will get useful data a few years into the study, but still be mining for information decades [later]," said Guttmacher. He said

that a paper by NHGRI director Francis Collins explaining the benefits of such a study would be appearing in a major research journal later this week.

Although the official RFI closes this Friday (May 28), Guttmacher stressed that discussion of the project would be ongoing. Guttmacher said that the project, if initiated, would involve researchers from federal, academic, and private institutes, and that community involvement would also be a large component. NIH hopes to make as much as of the information freely available to the public as possible, which will require strict privacy guidelines.

NIH recognizes that a project of this magnitude would "cost a lot and take a long time," said Guttmacher, "but if you can't do it well, it's not worth doing... We're really trying to have the science design this study [and] drive the budget."

Beaty agreed: "It has a lot of potential, it needs to be done, and it needs to be done well," she told us.

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