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Diet may treat some gene mutations

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Scientists have moved a step closer to correcting some unhealthy gene mutations with diet, according to a new research report appearing in the April 2012 issue of the journal *Genetics*. Researchers from the University of California, Berkeley, determined variations and responses to vitamin treatment in the human cystathionine beta synthase gene, which when defective, causes the disease homocystinuria, an inherited metabolic disorder sometimes treatable with vitamin B6. After the analysis, scientists correlated specific gene mutations with severity of the disease, ranging from perfectly healthy and functional to severe and untreatable. Although the current study focused on homocystinuria, testing the effects of naturally occurring gene variations using surrogate organism genetics can be applied to other inherited disorders, such as neural tube defect, cleft palate, and blindness.

"The era of personal genome sequences is upon us, but there is a growing gap between the ability to sequence human genomes and the ability to understand the significance of variation in genome sequences," said Jasper Rine, Ph.D., the principal investigator of this research in the Department of Molecular and Cell Biology at the California Institute of Quantitative Biosciences at the University of California, Berkeley. "This study demonstrates one way to close the gap; the data separate gene variants into distinct classes, including a group amenable to dietary intervention."

To make their determination, scientists "swapped" the cystathionine beta synthase gene of baker's yeast with the gene from humans to test which variants were healthy, treatable, or untreatable with additional vitamin

B6. As a result, the study clarified the function of 84 DNA sequence variants in this gene, which will help physicians more effectively treat patients based on their particular genotypes. In addition, this approach opens doors for future studies examining other human genes that similarly cross over between humans and yeast.

"We may have the DNA sequence of the human genome, but we're still trying to figure out what it means," said Mark Johnston, Editor-in-Chief of the journal *GENETICS*. "This study moves us a step closer toward better understanding the genetic variability among people. More immediately, knowledge of these gene mutations will help physicians prescribe treatment based on genotype rather than outward symptoms or trial and error."

More information: Jacob A. Mayfield, Meara W. Davies, Dago Dimster-Denk, Nick Pleskac, Sean McCarthy, Elizabeth A. Boydston, Logan Fink, Xin Xin Lin, Ankur S. Narain, Michael Meighan, and Jasper Rine. Surrogate Genetics and Metabolic Profiling for Characterization of Human Disease Alleles, *Genetics*, April 2012 190:1309-1323. http://www.genetics.org/

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