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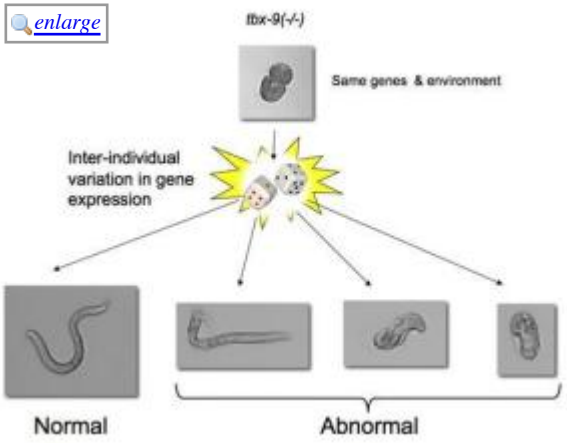
Why Does the Same Mutation Kill One Person but Not Another?

ScienceDaily (Dec. 7, 2011) — The vast majority of genetic disorders (schizophrenia or breast cancer, for example) have different effects in different people. Moreover, an individual carrying certain mutations can develop a disease, whereas another one with the same mutations may not. This holds true even when comparing two identical twins who have identical genomes. But why does the same mutation have different effects in different individuals?



The figure shows how the same mutation can differently affect each individual from C. elegans. The dice represent the stochastic component in the gene expression. (Credit: Image courtesy of Centre for Genomic Regulation)

Since the early twentieth century researchers have studied the role that genetic variability (mutations) and the environment (consumption habits, lifestyle, etc.) have in the development of diseases. "However, genetic and environmental differences are not enough" said Alejandro Burga, one of the authors of the article. "In the last decade we have learned by studying very simple organisms such as bacteria that gene expression -- the extent to which a gene is turned on or off -- varies greatly among individuals, even in the absence of genetic and environmental variation. Two cells are not completely identical and sometimes these differences have their origin in random or stochastic processes. The results of our study show that this type of variation can be an important influence the phenotype of animals, and that its measurement can help to reliably predict the chance of developing an abnormal phenotype such as a disease ."

The researchers conducted their study using the roundworm *Caenorhabditis elegans* as a model. Due to its simplicity, this microscopic worm is one of the most widely studied organisms in biology, and was the first animal to have its genome sequenced. Recently three different Nobel Prizes have been awarded for research using *C. elegans*.

Since the genetic composition and the environment are insufficient to determine whether or not a mutation will affect an individual, they developed a methodology to measure small differences in gene expression in vivo. "The challenge was not only to develop a method to quantify these small differences among individuals, but also to predict which genes are relevant for a particular mutation," adds Ben Lehner, coordinator of the study and ICREA Research Professor in the European Molecular Biology Laboratory-Centre for Genomic Regulation Systems Biology

Research Unit. "In both round worms and humans, genes cooperate and help each other to perform functions within the cell. A few genes are very "generous" and help hundreds of others to perform many different processes, whereas others only help a few other genes to perform more specific functions. The key to predict what will happen in each individual is to measure variation in the expression of both types of gene."

The work suggests that, even if we completely understand all of the genes important for a particular human disease, we may never be able to predict what will happen to each person from their genome sequence alone. Rather, to develop personalised and predictive medicine it will also be necessary to consider the varying extent to which genes are turned on or off in each person.

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1. Alejandro Burga, M. Olivia Casanueva, Ben Lehner. **Predicting mutation outcome from early stochastic variation in genetic interaction partners**. *Nature*, 2011; 480 (7376): 250 DOI: 10.1038/nature10665

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