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How Many Copies Is Enough?

By **Andrew Plemmons Pratt** | December 1st, 2008

The advent of bioinformatics has driven home the point that there's a wealth of genetic information that humans will never be able to comprehend without the help of computers. *Science* reports (subscription) from the annual meeting of the American Society of Human Genetics that one of the hottest fields in genomic research leverages computing power to sift through the patterns of copy number variations in gene sequences and determine their connection to phenotype and disease risk.

Copy number variation refers to the fact that the number of copies of a gene, or deletions from sequences within a person's DNA, along with the placement of those copies or deletions, contributes to his or her inherited characteristics. That is, the copies or deletions are themselves genetic information, like the squeaks and pauses in a string of Morse code. Using sequencing methods, researchers can identify the variation in sequence patterns across a population. Spotting those variations is one challenge, but associating them with observable characteristics is another matter altogether. Jennifer Couzin writes:

> The study of CNVs, like any emerging field, is plagued by uncertainty. Often the technology used was not designed to detect CNVs, making results difficult to interpret. And it's not at all clear which CNVs alter the function of genes or influence disease. Last week, scientists at the meeting described links between CNVs and various cancers, schizophrenia, autism, body mass index, and Crohn's disease. But in nearly all these cases, questions remain as to whether CNVs are coincidentally present, are linked to another genetic disease driver, or are themselves causing ill health.

We spoke with Nancy Spinner, a professor at the University of Pennsylvania, earlier this year, about this emerging field of research. She pointed out that CNV studies can be be powerful, but there's a lot more to learn: "The problems that are now facing us are just at the very beginning of a) understanding the variation in the genome, and b) understanding how it relates to health and disease. It is all so new that it is very difficult. The science is just not there yet, to be able to tell us what it means to have something that is extra or missing."
