GENETICS

NOTHING MAKES SENSE EXCEPT IN THE LIGHT OF A HYPOTHESIS

24.11.2006

Looking again at human variation it seems that my recent estimate of 99,9% sequence identity is wrong as shown in an nature editorial yesterday and of course the new paper with the first copy number map of the human genome

> 3,080 million 'letters' of DNA in the human genome
> 22,205 genes, by one recent estimate
> 10 million single-letter changes (SNPs) â€"
> that's only 0.3% of the genome
> 1,447 copy-number variants (CNV),
> covering a surprisingly large 12% of the genome
> About 99.5% similarity between two random people's DNA

I am organizing my literature in folders where the CNV section is still very thin but labelled as high priority – this seems to be adequate as the new study shows that the CNV emcompass hundreds of genes and functional elements.

> Notably, the CNVRs encompassed more nucleotide content per genome than SNPs, underscoring the importance of CNV in genetic diversity and evolution

The Wellcome Trust has a nice website about copy number variants. If you want to read more, you will find information about the methods (array-based comparative genome hybridisation, cytogenetics, population genetics, comparative genomics and bioinformatics) as well as the questions that drive CNV research.
Again it seems that disease genetics is not only about