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Biotech claims comprehensive SNP map

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The Mountain View, California-based company Perlegen recently made public the completion of its search for single nucleotide polymorphisms (SNPs) in the human genome. Following the November 2001 publication in Scienceof a SNP map of human chromosome 21, Perlegen scientists say they sequenced 50 complete haploid genomes, yielding more than 1.7 million SNPs.

More important, according to Perlegen CEO Brad Margus, was their demonstration that they could interrogate all 1.7 million SNPs in massively parallel fashion, opening the door for genome-wide association studies, which previously could not be attempted because of the prohibitive cost of genotyping.

Key to this work were high-density oligonucleotide arrays, which Perlegen scientists designed and had fabricated by Affymetrix, Perlegen's parent company. Amplifying the entire human genome required nearly 250,000 PCR assays, and more than 12 billion oligonucleotide probes were used to sequence the 50 haploid chromosomes. The company claims their system can complete a genome in 10 days.

Using the specially designed SNP-reading oligonucleotide arrays to genotype all 1.7 million SNPs, Margus estimates the cost of genotyping 1,000 individuals to be roughly $2 million. In contrast with the estimated $300 million it would cost using current SNP genotyping technologies, Margus told *The Scientist*, "It's still not chump change, but a do-able project if the question is serious and important."

In an e-mail interview David Altshuler, Director of Medical and Population Genetics at the Whitehead/MIT Center for Genome Research, said that he believes large-scale SNP databases and genotyping methods will be important tools to understand human disease. He cautioned that significant and unanswered questions remain about the success of such large-scale association approaches, however.

"By placing information in the public domain, a broad set of perspectives, from both academic and for-profit researchers, can be applied, and an open forum encourages broad input on the course of research. Information that is in private hands can certainly add value, but the history of genomics is that the greatest impact is achieved when such information is freely released for all to use," Altshuler wrote.

Perlegen does not plan to publish the rest of its SNP collection. The company has already announced a collaboration with GlaxoSmithKline to scan the genome for markers associated with drug response, according to Margus, who said that three more collaborations will be announced shortly. In addition, Margus said, Perlegen is working with academic researchers with expertise in complex human diseases, such as diabetes and Alzheimer's disease.

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