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A cheap personal genome?

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BOSTON - Step right up and have your genome sequenced in a single afternoon. Or, why wait all afternoon? How about having it done within minutes - better yet, seconds? Such was the tone of predictions made during a panel discussion titled "The Future of Sequencing Technology: Advancing Toward the $1,000 Genome," held Wednesday evening, October 2, at the 14th International Genome Sequencing and Analysis Conference.

Not all of the speakers promised such fast turnaround times, but Susan Hardin of Houston, Texas based VisiGen Biotechnologies forecast that in two to four years her company will have developed the technology to sequence an entire human genome within a couple minutes. Not to be outdone, Eugene Chen told the audience that US Genomics' goal "is to be able to read your genome instantaneously," and that he expects the technology to make it possible to be up and running in about three or four years.

The pursuit of quick, affordable personal genome sequencing has been gaining momentum, amid growing expectations surrounding the healthcare potential of personal genetic testing. The ultimate personalized medicine tool, a personal genome sequence conveniently saved to a CD, is currently a luxury item. Former Celera chief, Craig Venter, is taking orders for full personal sequences at a price of $620,000 and UK-based Solexahas said it will soon offer consumers just-SNPs for considerably less. But the $1,000 genome is considered the point at which widespread personal sequencing will become economically feasible because both consumers and health insurers might be willing to pay for it.

Wednesday evening's discussion of technological progress toward that mark was co-moderated by Craig Venter, representing one of his three non-profit foundations, The Center for Advancement of Genomics, and Gerald Rubin of Howard Hughes Medical Institute. Participants also included George Church of Harvard University's Lipper Center for Computational Genetics, Trevor Hawkins of Amersham Biosciences, Tony Smith of Solexa, and Michael Weiner of 454 Corporation.

Both VisiGen and US Genomics' sequencing technologies, along with Solexa's, are single-molecule approaches, and single molecule detection is "absolutely a doable thing," Hardin told the audience. However, some questioned its feasibility. It would be wonderful if it worked, Weiner said after the session, but "show me the data." The technique has been around for 15 years, he told us and there are still major limitations.

454's picotiter plate technology, which is not single-molecule based, currently achieves a rate of 2.4 million bases per day per machine. It may not be as fast as the others, but it is the closest to actually being available as a usable product, Church told us. Of all the new technologies discussed, Weiner agreed, only 454's picotiter plate and Church's 'polony sequencing' are currently totally functional.

Single-molecule or otherwise, the realistic price of mass personal genome sequencing was also raised. Reagents are particularly expensive and comprise about half the cost of sequencing, panelists noted. Increasing miniaturization and reducing reagent volumes are major challenges to reaching the $1000 genome, Hawkins told the audience. The ultimate goal of a $1000 genome is challenging but achievable, said Smith, although he would not say when.

The bottom line, Hawkins said, is that there are technologies that will be made available that can drop the cost down to around $30,000 per genome within the next two or three years. To get past that point, he said, we need a "new technology." In the meantime, $30,000 may not be within the means of the average consumer, said Hardin, but still it is considerable progress. "After all," she observed, "for a scientist, a thousand dollars is practically pennies."

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