

Genome at 10: Scientists urge patience for medical breakthroughs

April 1 2010, by Richard Ingham



This undated illustration shows the DNA double helix. The genome has yet to deliver on promises it would usher in a golden age of medicine, experts say in a debate unleashed by the looming anniversary of the first draft of the human genetic code.

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On June 26 2000, two teams -- one a public consortium, the other a private venture -- tied in a frenzied, multi-billion-dollar race to be first to publish a working blueprint of <a href="https://doi.org/10.2016/jnan.

Tens of billions of dollars have been pumped into genomics research



since this landmark event.

The drive is the allure of <u>new drugs</u> to cure cancer, heart and <u>brain</u> <u>disease</u> and other curses.

Just as compelling: diagnostic tools to pinpoint an individual's inherited vulnerability to disease and steer the patient to hand-tailored treatments.

Yet as the anniversary nears, opinions as to what the decade has achieved coalesce around the conclusion that so far the outcome has fallen way short of a medical revolution.

"The consequences for clinical medicine... have thus far been modest," Francis Collins, who led the public-sector Human Genome Project (HGP), admits in this week's issue of Nature, the British science journal.

"Some powerful new drugs have been developed for some cancers; genetic tests can predict whether people with breast cancer need chemotherapy; the major risk factors of macular degeneration [an eye disease] have been identified; and drug response can be predicted for more than a dozen drugs.

"But it is fair to say that the Human Genome Project has not yet directly affected the health care of most individuals."

US bio-entrepreneur Craig Venter, who jousted with Collins in that epic battle 10 years ago, urges patience.

"The genome revolution is only just beginning," Venter believes.

For one thing, the cost of cracking -- "sequencing" -- a genome has fallen astonishingly, by 14,000 times between 1999 and 2009.



The <u>Human Genome Project</u> (HGP) was funded with three billion dollars, or roughly a dollar for every base pair, or rung in the genetic ladder, and took 10 years to produce the rough draft, which was followed by a "polished" version in 2003.

Today, Californian companies Illumina and Life Technologies say they can do the same job in a day for less than 6,000 dollars, while rival Complete Genomics is reportedly offering a price of 5,000 dollars.

This plunge in costs has enabled scientists to prise open the DNA of a whole range of non-human species: more than 3,800 organisms, in fact, including the mouse, the fruitfly and other vital tools in lab research.

In the next decade, writes Venter, we can expect the DNA of individual sperm, egg cells, early-stage embryos, pre-tumour cells and stem cells, to see whether they are healthy or harbingers of disease.

Why does genomics seem to be taking so long to bear fruit?

One reason is that having a goldmine of data is not the same as understanding what the data mean.

And the meaning turns out to be far more complex than thought: the number of genes in an organism is not automatically the sign of its complexity, for genes and the proteins they control can have multiple roles.

With the information pool now the size of an ocean, the more urgently we need the means to be able to manipulate these data coherently -- and figuring it out.

"Now that the human genome has been sequenced almost completely, there is still little understanding of how genes actually work," say



Monika Gisler and colleagues from ETH Zurich, a Swiss research and technology university, in the open-access journal Arxiv.org

"(...) It is now widely recognized that it will take decades to exploit the fruits of the HGP, via a slow and arduous process aiming at disentangling the extraordinary complexity of the problem."

Those waiting for a financial return on genomics will have to wait a little bit more in what has already turned out to be a long-term bet.

As Arthur Levinson, then the chief executive of Genentech, said in March 2006: "Investors have been very patient with the biotech industry, which has been one of the biggest money-losing industries in the history of mankind."

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