Despite what you've heard, human genome is not completely sequenced

The feat made headlines around the world: "Scientists Say Human Genome is Complete," the New York Times announced in 2003...There was one little problem.

"As a matter of truth in advertising, the 'finished' sequence isn't finished," said Eric Lander, the director of the Broad Institute of MIT and Harvard.

. . .

"The human genome has not been completely sequenced and neither has any other mammalian genome as far as I'm aware," said Harvard Medical School bioengineer George Church....

. . .

Perhaps nobody paid much attention because the missing sequences didn't seem to matter. But now it appears they may play a role in conditions such as cancer and autism.

Most of the unsequenced regions, [Church] said, "have some connection to <u>aging</u> and <u>aneuploidy</u>" (an abnormal number of chromosomes such as what occurs in Down syndrome). Church estimates 4 percent to 9 percent of the human genome hasn't been sequenced.

. . .

"As new sequencing technology has begun allowing scientists to peek into unsequenced territory, however, they have seen that "these tough-to-sequence regions frequently have important genes," said Michael Hunkapiller, chairman and CEO of Pacific Biosciences, which makes DNA sequencers.

. . .

"Finish the sequence!" hasn't become a rallying cry, but maybe it should be, genomics luminary Craig Venter said.

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post: Psst, the human genome was never completely sequenced. Some scientists say it should be