A Celebration of the Genome, Part II

During the month of February, we are celebrating the 10th anniversary of the first publications of the human genome sequence. This week, the commentaries explore the impacts of sequence information on our understanding of ourselves, as well as look at future directions for research and medicine.

–Barbara R. Jasny and Laura M. Zahn

My Genome

Desmond Tutu
Archbishop Emeritus of Cape Town, South Africa

This is a monumental month for human genomics as we celebrate 10 years since the publications of the human genome. It is time to reflect on the advances that this endeavor has brought to mankind. The ability for scientists to generate a complete human genome sequence meant that, for the first time, an individual’s entire genetic code could be read from beginning to end. For the first time, these amazing men and women could use the code to study disease, to make sense of inherited risks, and to assess how the body responds to medicines. These advances, however, were biased because the available information provided limited benefit to the African continent and the people of Southern Africa. I have been known to refer fondly to my country as the “Rainbow Nation,” a land of wealth in its many diverse peoples and cultures. The majority of us have experienced many years of oppression, emerging as a free nation only in 1994. As a nation, however, we need to continue to fight against racial inequalities and socioeconomic disparities on a daily basis. My participation in the Southern African Genome Project was a step in this direction, generating the first Southern African genome to be sequenced—exactly 9 years after the publication of the human genomes.

My reasoning was simple. Southern Africans are victims of many devastating diseases whose eradication requires immediate attention and international resources. My hope is that my genetic code may provide a voice for the region and serve as the starting point for a map of DNA variation significant for Southern African peoples, to be used for medical research efforts and effective design of medicines. I implore the scientific community to continue what I hope was just a first step to further medical research within the region.

Many may ask if I learned anything significant from having my genome sequenced. I was certainly not expecting anything dramatic. I have been blessed to be alive for 79 years; we have four beautiful, healthy children and seven gorgeous grandchildren. Wonderfully, I discovered that I was related to my fellow sequenced Southern African in this project, !Gubi, a Kalahari Bushman from Namibia. Meeting !Gubi and his wife Anna in Windhoek in February 2010 was for me a highlight of this project. Anna bore an uncanny resemblance to my mother. It was a truly uplifting experience to discover that I was genetically related to a long line of peaceful and gentle people that have trod the soils of Southern Africa for centuries.

My dream is that by including all peoples in understanding and reading the genetic code we will realize that all of us belong in one global family—that we are all brothers and sisters. Wow!

Genome Literacy

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The transition from knowing small patches of the genome to having whole human genomes available to explore has been a unique experience. The biggest surprise initially came from the number of protein-coding genes—estimates anywhere from 15,000 to 160,000 had appeared in the literature before the publications came out in 2001 and settled at 20,000 to 30,000 genes. Although protein-coding genes were the most identifiable functional elements in the human genome, 10 years ago, the exact location of regulatory regions was unknown, and only a small fraction of the variations existing within the human population had been characterized. Ten years later, the ability to use the complete human genome backbone to map sequence variation and the availability of technologies to interrogate genome function are driving our ability to read the compendium of functional elements and to understand how population variation effects them. The basic components in each genome are largely the same, but the way they are used differs from tissue to tissue and person to person. Understanding the rules of gene regulation, the grammar of the genome, is key to the understanding of the human body. And it is only with the full sequence that we will...
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