

Healthy genome: a myth or a paradigm shift in bioinformatics research?

Opinion

The human genome holds the clues to diverse diseases and improved quality of life. The majority of the bioinformatics and genomics research revolves around finding the genetic fingerprint for diseases and using that knowledge to improve diagnosis and treatment.¹⁻¹² These approaches while very effective look at the genome in the context of particular diseases rather than healthiness. Provocatively, one can raise a question whether in the genome resides a signature for healthy disease-free life. What would it take to approach wellness of human beings from a health rather than disease perspective? Among the 22,000 proteins and other products of the human genome such as the non-coding RNAs, pseudogenes etc., could there be a signature for normal health?

In the last decade since the completion of the human genome project, academia, government funding agencies and industry have focused on the disease aspect of the genome. Is it time to think outside the box and begin to look at the power of the human genome from the perspective of healthiness? Google X recently announced an ambitious project termed 'Baseline Study' to develop baseline information using the body fluids (urine, blood, saliva and tears) from normal volunteers.¹³ Inclusion of other body fluids such as semen, ascites, cerebrospinal and amniotic fluids would greatly augment such a study. The outcome of such a study could in theory lead to prevention rather than treatment of diverse diseases. Proteomic datasets from normal patients are increasingly available for mining the human genome for the healthy proteome.¹⁴⁻¹⁶

This is a major paradigm shift of changing the focus from the disease genome to the normal genome. Government and industry spend vast amount of money in combatting illnesses. However, until we understand what constitutes normal, disease-free health, efforts to combat diseases will always be at a disadvantage. We will at best be treating illnesses rather than attacking the root cause and preventing the disease.

The Healthy Genome approach is the next dimension in the human genome project and is likely to hold surprises. Similar to the Thousand Genome and Genomics England's Hundred Thousand Genome projects,^{17,18} genome projects around the world ought to be created to clearly establish a baseline data of normalness. A clear metric for normalness needs to be established prior to massive data collection, where as it is attractive to start with body fluids as does Google X's "Baseline study", numerous proteins and non-coding RNAs, which play a crucial role in normal cell function are not readily detected in secreted form. Normal health baseline cannot be solely defined on the basis of certain known genes, although they are very often implicated in numerous diseases. The baseline information must involve the entire transcriptome and proteome, if it is to provide a rationale for defining what constitutes health.

A meaningful outcome of the Healthy Genome efforts would depend on the right choice of volunteers around the globe to account for epigenetic effects, genome diversity, individual variations, and

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environmental and nutritional factors. Further, the Ethical, Legal and Social Implications (ELSI) issues need to be addressed with the experience that has been gained from the genome project.

The completion of the human genome project was made possible with major funding commitment from government agencies across US, UK, Europe, Japan, Canada and Australia. As health issues are global, the Healthy Genome project requires a strong funding commitment from western countries and emerging economies. Further, industry-government partnership around the globe is going to be critical for the Healthy Genome project to succeed. Google X is already setting the stage for an industry-academic partnership to develop a baseline fingerprint of the genome from healthy individuals. We can anticipate other industries (computer, pharmaceutical, biotechnology, diagnostic and insurance) joining forces in the future to accomplish the Healthy Genome objectives. Private foundations such as Gates Foundation and other healthcare foundations can also help with these efforts.

Whether it is a myth or a complete bioinformatics paradigm shift toward normalness, these bioinformatics efforts can greatly enhance the worldwide quality of life. These normal health-oriented Healthy Genome approaches are likely to modernize the future of medicine and healthcare.

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Conflict of interest

The author declares no conflict of interest.

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