

The next horizon in proteomics and genomics research

Editorial

The completion of the human genome project a decade ago opened new avenues to discover cures to diverse diseases. The potential to discover new genes raised high hopes for many discoveries that would make it possible to cure or even prevent human diseases. The outcome has not kept up with the excitement. Of the predicted proteins in the human genome, a vast number are of unknown nature and their relevance remains a mystery.¹ These novel proteins together with the noncoding RNAs constitute the ‘dark matter’ of the genome.^{2–7} Hence a problem remains about the path that future genomic research should follow: should institutions and researchers continue on their current course of studying the known genes, or should they take the higher-risk route of working to demystifying the dark matter of the human genome?

Currently, researchers face fewer challenges and a higher likelihood of bringing their projects to fruition by continuing to study the known genes. Nonetheless, studying the novel uncharacterized proteins offers numerous advantages. Only through such study can a broad understanding of the complexity of the human genome emerge; this should provide a basis for better pharmaceuticals and diagnostic markers for diverse diseases.^{8–10} Further, the mystery surrounding the genome can disappear as more research goes into the dark matter area of the genome.

Powerful bioinformatics and proteomics tools, which offer ways to harness the vast amount of genomic information, are increasingly available.^{6,7} Genome-wide association databases such as the Genetic Association Database (GAD),¹¹ the Phenotype-Genotype Integrator (PheGenI),¹² the database of Clinical Variations (ClinVar)¹³ and the International HapMap Project (HapMap)¹⁴ allow a user to rapidly establish phenotype-associated traits for a large number of genes in a single experiment. High-throughput gene expression analysis tools for both mRNAs and proteins can provide an initial hint on diverse tissue expression of the genes to be analyzed. Going from the genome to phenome to function and finally to clinical relevance has never before been easier.

A key issue resides in convincing researchers of the value of following the unknown genes. Funding agencies and industry have tended to favor projects that offer the highest probability of success; consequently, not much research has delved into the dark matter of the genome. Recently the US National Cancer Institute (NCI) announced a new initiative to address the paucity of attention that has been given to this basic research: “Illuminating the Dark Matter of the Genome for Druggability”.¹⁵ This is a welcome step in the right direction. More funding by so influential an institution as NCI will undoubtedly lead other institutions around the world to foster study of the unknown elements in the human genome.

Only by an understanding of the entire human genome, encompassing the known and the unknown proteins as well as the noncoding RNAs, can the full potential of the Human Genome Project be realized. The challenges of illuminating the dark matter may be

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daunting, but the potential rewards for the diagnosis and treatment of human diseases are immense.

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Conflict of interest

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