Contribution of human genome project to medicine

Human Genome project has been proclaimed by many as “The end of the beginning” and this phrase best describes current accomplishments in genetics. The distribution of genes on human chromosomes can be compared to geographical maps, presenting a world of settlements, which differ from each other by population, size and function. Some coding genes are condensed in big and small cities and they are separated form each other by lifeless ‘deserts’ of intervening sequences. For example, 19th chromosome represents “capital city” as it is abundant with coding genes.

Any occurring phenomenon can be expressed with the combination of 33 letters of Georgian alphabet. In the same manner, 1,5% of human genes can code such a big variety of life in all of its forms. Even though the majority of genes are mapped, the analysis of the gained information, its implementation and the application of the basic theoretical knowledge to everyday medical service are ahead. Despite the fact that the human genome project’s success can lead us to several ethical and social problems, in my opinion, the numerous positive effects that it will bring about will far outweigh them.

Human genome project can have an advantageous effect on extending our understanding of complex diseases. Oncology is a good example of how understanding our genome and its intricate workings could greatly facilitate diagnosis and treatment of diseases. In case of chronic myeloid leukemia, the mutated gene is BCR-ABL; knowing this fact gives us the opportunity to diagnose CML by simple sensitive blood testing, which gives us the possibility of identifying the CML on early stages and makes it possible to treat it.[1]

In the future medicine will become completely patient-oriented. For example, patients suffering from acute myeloid leukemia with mutation in FLT3 gene require intense treatment, including stem cell transplantation. Meanwhile, patients who have mutation in NPM1 gene should be given mild course of treatment. Therefore, identification of mutation enables us to make treatment unique for each patient. Therefore, the success of human genome project can dramatically improve therapy in individual cases. Another example of how genomic studies improved medical diagnosis is the identification of causes of Kabuki and Miller syndromes. Dramatic decline in costs of sequencing genomes facilitated discovery of the fact that a loss-of-function mutation in gene MLL2 might cause Kabuki syndrome, while Miller syndrome may be caused by an anomaly in DHODH gene.

Based on the knowledge gained from the studies of the human genome, many new controversial and revolutionary projects have arisen. For example, since 2002, SNPs (single nucleotide polymorphism) were characterized in different ethnical groups. Identification of SNPs among individuals and ethnic groups can make predisposition testing toward disease much more accurate. [2]

Human genome project is, in a way, a breakthrough in the field of pharmacogenomics. Better understanding of genes that influence drugs’ pharmacodynamics and pharmacokinetics will be a big step forward in prescribing more specific drugs and their doses. Clopidogrel, anti-platelet aggregation, blood thinner drug, is widely used to prevent strokes and heart attacks. 30 percent of population has a gene, which abolishes activation of Clopidogrel in liver; therefore the drug cannot achieve desirable effect on some patients. Testing patients genome will save the time and decrease the risk of taking the inadequate dose.

Despite numerous advantages of Human Genome Project, we cannot neglect its debatable side effects. One of the drawbacks of so called “personalized” medicine, which was made possible by the advancements in HGP, is the risk of discrimination on the basis of human genome. For instance, individuals with predisposition towards some disease are likely to face problems in getting hired. Employers might discriminately choose potentially healthier people for their vacant jobs. At the same time, insurance companies may refuse to provide insurance to potentially ‘ill people’. These causes biased division of society into groups.

With all its positive contributions to our life the human genome project has put forward new challenges for humanity to overcome. Personalization of medicine is a revolutionary step, however, for it to be successful it is necessary to find out answers to ethical and social problems that it will bring about and this is only possible through high level of public education, for which scientists hold great responsibility.