Role of Human Genome Project in Medical Science

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DOI: 10.38177/AJAST.2020.4202

Article Received: 21 January 2020

Article Accepted: 23 March 2020

Article Published: 26 April 2020

ABSTRACT

Human Genome Project is a worldwide scientific achievement. It was a thirteen-year project initiated in 1990 and completed in 2003. Human Genome Project helped a lot in the identification of diseased genes as DNA is very significant for understanding the diseased gene and their functions. It helped in the identification of disease loci for many diseases and presented their treatment through preventive measures. It identified the gene loci for many diseases like cancer, asthma, high blood pressure, diabetes type 2, obesity, Alzheimer's disease, Down's syndrome, Turner's syndrome, depression and many types of heart diseases including cardiovascular disease and coronary artery disease. This project does not directly treat the diseases but it helps in the identification of disease gene loci and then allows the treatment of disease through its preventive measures before the appearance of symptoms or at the initial stages of the disease through many techniques like gene therapy, pharmacogenomics, and targeted drug therapy. These are the helpful techniques in the diagnoses of the human disease gene locus.

Introduction

Human Genome Project is a worldwide collaborative biological project initiated in 1990 by the Department of Energy collaboration with the National Institutes of Health to sequence three billion nucleotide bases of 80,000 estimated genes of Human Genome. Up till now, **Whole Genome Sequence** is considered as an encoding of the blueprint of life but the function of nearly 3 billion nucleotide bases is still unknown (Elements, 2012).

Human Genome Project helped a lot in the identification of diseased genes as DNA is very significant for understanding diseased genes and their functions. Except for some cases of trauma, every human disease has some genetic component. The large-scale genome research has driven the technology advancement in genetic testing, drug design, gene therapy, and other genetic related areas such as pharmacogenetics (Innovation & Wattanapitayakul, 2016).

This project does not directly treat the diseases but it helps in the identification of disease gene loci and then allows the treatment of disease through its preventive measures before the appearance of symptoms or at the initial stages of the disease through many techniques like gene therapy, pharmacogenomics, and targeted drug therapy (Collins & Mckusick, 2001).

Disease Names	Locus identified for the disease	Techniques used for the identification	Findings of the author	Citations
Asthma	A locus on chromosome 17q12-21	single nucleotide polymorphisms (SNPs)	The strongest evidence was found	(Wan et al., 2012)
Diabetes type 2	Locus on 12q15	Microsatellite markers	The strongest evidence was found for this disease on chromosome 12q15	(Bektas, Hughes, Warram, Krolewski, & Doria, 2001)

Depression	locus in the human IL6 promoter	Single nucleotide polymorphism (SNP)		(Cole et al., 2010)
Cancer	Locus identified between chromosome 9 and 12	DNA sequencing	Mutated- cancer genes were identified	(Stratton, Campbell, & Futreal, 2009)
Cardiovascular Disease	Chromosome 9p21	Single nucleotide polymorphism (SNP)	Association of chromosome locus 9p21 with coronary heart disease (CAD) and myocardial infection (MI) and represents the most replicated locus.	(Disease, 2007)
Hypertension	A locus on chromosome 17	Solar software was used	The strongest evidence was found on chromosome 17 for systolic B.p. and for diastolic B.P. on chromosome 17 and 18.	(Levy et al., 2000)
Obesity	Gene locus was identified on TNF (Tumor necrosis factor) on alpha gene	Gender-pooled quantitative sib-pair analysis	A remarkable effect of the gene locus on 3 global and 7 regional measures of obesity	(Pausova et al., 2000)
Alzheimer's disease	The locus on chromosome 14	Markers assisted technology	The strongest evidence was found for Alzheimer's disease on chromosome 14	(Schellenberg et al., 2019)
Down's syndrome	Due to the presence of an extra chromosome 21	Southern blotting and FISH (fluorescent in-situ hybridization) was used	Strong evidence was found for the locus present on chromosome 21 for Down's syndrome	(Daumer et al., n.d.)
Turner's syndrome	X-linked chromosome (45X, 46X).	Cytogenetic analysis was performed	Neuropsychological effects indicated that short arm of chromosome 46X was missing in the Turner's patient.	(Skuse, Creswell, & McGurk, 1997)

Conclusion

Human Genome Project is a worldwide scientific achievement. It was a thirteen-year project initiated in 1990 and was completed in 2003. Human Genome Project helped a lot in the identification of diseased genes as DNA is very

important for understanding diseased genes and their functions. It helped in the identification of disease loci for many diseases and presented their treatment through preventive measures. It identified the gene loci for many diseases like cancer, asthma, high blood pressure, diabetes type 2, obesity, Alzheimer's disease, depression and many types of heart diseases. This project does not directly treat the diseases but it helps in the identification of disease gene loci and then allows the treatment of disease through its preventive measures before the appearance of symptoms or at the initial stages of the disease through many techniques like gene therapy, pharmacogenomics, and targeted drug therapy.

Acknowledgments

We thank Dr. Muhammad Baber, **Dr. Ghulam Shabbir** and members of the Institute of Molecular Biology and Biotechnology (IMBB), Bahauddin Zakariya University, Multan Pakistan for helpful discussion.

Conflict of Interest

There is no conflict of Interest.

Funding

No source of funding is reported by the author.

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