

# Genomics: the words gene and chromosome. [4] .

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**GENOMICS:** Genomics is an academic discipline of science.

It categorizes within the molecular biology. It focuses on the structure, function, evolution, and mapping of genomes. Genome is an entire set of genes or DNA within the organism's cell at individual level or whole organism level. The purpose of Genomics is focusing on collective characterization and quantification of genes, which related directly with the production of proteins with the support of enzymes and messenger particles. Genomics also includes the sequencing and exploration of genome. 1

**HISTORY:** .

In 1920, Hans Winkler invented the term genome which means, all the genes in an organism by joining the words gene and chromosome. 4 . In 1953, James Watson and Francis Crick with the aid of Rosalind Franklin, revealed the double helix structure of DNA. 4 . In 1966, Marshall Nirenberg was attributed with cracking the genetic code and relating how it works to create proteins. 4 .

In 1978, biotechnology company Genentech Inc, produce genetically engineered bacteria for making human insulin for handling of diabetes. 4 . In 1983, researchers recognized the gene answerable for Huntington's disease, which directed to the first genetic test for this disease. 4 .

In 1984, Sir Alec John Jeffreys established DNA profiling or fingerprinting, may be used for parenthood testing and in forensics. 4 . In 1986, Richard Buckland was the first one, which determined that criminals can be detected on the basis of DNA evidence. **TYPES OF GENOMICS:** • **FUNCTIONAL**

**GENOMICS:** It is the type of genomics that focuses on the function of genome

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that is functions include translation and transcription etc. Different molecular techniques that are used in functional genomics are microarray analysis, real time PCR etc.

1 "• **STRUCTURAL GENOMICS:** It is the type of genomics that defines the 3-dimensional structure of all proteins coded by a specified genome. 1 •

**EPIGENOMICS:** It is the type of genomics that study epigenetic variations (variations due to environmental factors) of genome. Most common epigenetic variations include histone modification and DNA methylation. 1 •

**METAGENOMICS:** The type of genomics which is related with the study of genetic material collected from the environmental samples. 1 **GENOME**

**ANALYSIS:** In order to study the genome of individual, there are three main mechanisms: • The sequencing of DNA • The assembly of that sequence to produce a demonstration of the creative chromosome • Explanation and investigation of that demonstration. 1 1.

**SEQUENCING:** With the help of sequencing it is possible to analyze genome because sequencing allows us to determine the nucleotides sequence of a particular gene by using some molecular approaches. Sequencing helps us in different ways e. g., it allows us to detect crime in forensics, it allow us to diagnose a particular disease before its treatment etc. Basically, there are many types of sequencing but two of them are: • **SHOTGUN SEQUENCING:** In this sequencing, DNA is broken down into small pieces that is typically around one thousand base pairs. Then these pieces are cloned into cloning vectors e.

g. M13. After this sequence are assembled by matching overlaps. • HIGH-THROUGHPUT SEQUENCING This technique is mostly used because it reduces the cost for DNA sequencing. Its read per run capacity is up to 6 billion reads per length (in illumine).

It has potential for high sequence yield. "2. ASSEMBLY: Assembly of sequence means to align and merge fragments of a very long DNA sequence in order to rebuild the original sequence. 1 3. ANNOTATION: It is the method in which biological information is attached to the sequences. It has three steps: 1 • Recognizing part of the genome that do not code for proteins • Classifying elements on genome.

(genome prediction) • Assigning biological information to these elements  
APPLICATIONS OF GENOMICS: There are many fields in which genomics have various applications like in medicine, biotechnology, anthropology and other social sciences. 1 • GENOMIC MEDICINE: Next-generation technologies in genomics permit investigators and biomedical scientists to significantly enhance the quantity of genomic data gathered on big study populations. 1 • BIOTECHNOLOGY AND BIOENGINEERING: In biotechnology, genomics has various applications like with the help of molecular approaches of genomics we are able to sequence given DNA sample then we also get information about protein conformation. • CONSERVATION GENOMICS: In order to get better evaluation of genetic factors, conservationists can utilize the data collected through genomic sequencing. For example: in order to get information about the genetic diversity of a population and to know whether person is heterozygous for a recessive inherited genetic disorder,

conservationists can utilize the data collected through genome sequencing.

1 THE HUMAN GENOME PROJECT: When this project began, sequencing technologies of DNA were not able to work with three billion base pairs of human genome. But now there are many ways by which we sequenced the whole genome like by pyrosequencing and by illumine (sequence by synthesis) by using "1. DIAGNOSIS AND TREATMENT OF DISEASE: illumine, whole human genome has been sequenced in 8 weeks to an average depth of ~40X.

this project has been an international struggle and the countries that have contribution in this project are Britain, France, Germany, Japan, China, and Canada. 2 IMPLICATION OF GENOMICS FOR MEDICAL SCIENCES: In 2010, it is anticipated that the prophetic genetic tests for common conditions allowing the individuals to know about their individual vulnerabilities and to revenue steps which reduce the interventions and risks. These interventions accept the form of medical scrutiny, modifications, diet, lifestyle and drug therapy. 2 In the recent years, there has been enormous research in genetics and genomics. And these emerging research and technologies revolutionized the practice and applications of medical sciences including the treatment and diagnosis of a disease. 4 Genomic medicine implies the idea that virtually most of the diseases are caused, regulated and influenced by genes. The genomics determine where the defected gene is present in the genome as well as the other environmental factors that affect the gene, consequently causing the disease.

5 Nowadays, genomic biomarkers are used, for example whole-genome gene expression, that help in the diagnosis and study of progression of the disease. Such biomarkers are used and the disease can be diagnosed at an early stage of onset. For example, whole-genome gene expression is used to diagnose and identify subclass of cancer, including acute myeloid leukemia subclass.

Along with genomic biomarkers, genomic signatures; the characteristic frequency of oligonucleotides in a genome, are also used that can identify the projection of non-small cell lung cancer as well as study the response of drugs in an individual. 4 By diagnosis of certain diseases, such as diabetes, heart disease, cancer disease, scientists will be able to identify the genes responsible for common illnesses. 2 Pharmacogenomics is the study of how a person's gene is involved in the response towards a drug. This field clearly describes how genomics can be used in the targeted treatment of diseases. 4 Recent research and work has been done for predicting and identifying graft rejection in the area where an organ has or going to be transplanted by studying the expression of genes in peripheral blood mononuclear cells in order to monitor a disease. 4 "2. PREDISTORTION OF RESPONSE TO A DRUG: 3. DETECTION OF GENES INVOLVED IN THE RISK OF DISEASES: Fig.

Steps involved in a genetic approach to the diagnosis and treatment of the disease. 4 Genomics provide opportunities with the design of new drugs and to predict the responsiveness of drug intrusions and variation in these responses is mostly attributable to the endowment of genetics of the individual. There are some common examples identified in which variants in

genes are involved in metabolism or action of drugs and are allied with the good and bad responses. Over next 10 years the expectation of these associations with many drugs, including the agents that act on the mechanism of drugs are available to the market and this revolution of pharmacogenomics aptitudes the individualize prescribing and practices.

The promising reports and the application of severe combined immunodeficiency syndromes and the gene therapy for hemophilia B23 have already been allied. 3 Evidences of data about the sequence of human genome and its variants must be pragmatic to categorize the particular genes which play a crucial role in the contribution of hereditary disease and this will be unnerving challenge. 3 In case of diseases such as diabetes mellitus (5 to 10) genes are involved and each of them docks a variant and conferring degree of increased risk.

These modifications intermingle with each "4. DESIGNER DRUGS: 5. PERSONALIZED MEDICINE: other and with the environment in intricate ways and rendering their credentials orders and their single gene defects. With the combinations of careful phenotyping in which different disorders will not involuntarily endured together and the sampling of genetic variants at a high density of the genome become able to identify gene association diseases for the common illnesses in next 5 to 7 years. 3 The need for advances in the technology of efficient genotyping for example the use of DNA chips and mass spectrometry.

The understanding of the foremost pathways that involved in the homeostasis of the human and also the development of pathways which are

unhinged in diseases. Identification of the gene that docks the high-risk of variant and the critical pathways. 3 Efficient and high-volume approaches are developed and applied to design the drugs and to modulate the pathways which are disease-related in specific direction. The gearing up of pharmaceutical industry considering the future of drug development in relation to the arena of genomics. The systematically applicable methods combine all chemical components into high volume assays and drugs for efficacy.

It is anticipated to identify the compounds that stimulate or block the particular pathway. The recent example of the drug STI-571 is developed to slab the kinase activity of the Bcr-Abl kinase. New revolution of gene-based (designer drugs) soon introduced to the souk for hypertension, diabetes mellitus, mental illness, and many like these. 3 Personalized medicine is a vast concept that focusses on an individual's genetic and genomic makeup, clinical and environmental information and consequently help in disease prevention and treatment.

4 Pharmacogenomics objective is to develop a drug, medicine that target an individual and more effective according to that person's genome.

Information from an individual's genome is retrieved and studied and a drug is developed that lessens the adverse effects and increase the beneficial effects. Recent advancements in technology has made the research more accurate, quick 6.

TARGETED THERAPY: 7. GENE THERAPY: and cost-effective. According to FDA, each year over 100, 000 people die due to adverse effect of the drug.

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Hence, such drugs need to be developed that has less harmful effects. 6 Personalized medicines are widely used in the treatment of cancers such as melanoma, thyroid, colorectal, lung and pancreatic as well as breast cancer. Scientists are also looking forward to the treatment of HIV/AIDS by personalized medicine. 8 Fig.

Idea of Personalized medicine 6 The improved diagnostic treatment of cancer is the most advanced form of clinical genetics and provides vast molecular information which has already been gathered about genetic basis malevolence. In 2020, it is assured that every tumor will have a specific molecular fingerprint which is determined and cataloging the genes which was gone skewed and individually targeted therapy to that fingerprint. 3 Gene therapy is a technique that involves genes for treatment or prevention of a disease. This technique might help the researchers to treat a patient by inserting a gene rather drug or surgery.

There are many approaches in gene therapy: 1. Replace a defected gene with healthy gene 2. Stop the improper functioning of a mutated gene by inactivating or knocking out a gene "3. Insert a new gene in order to combat the disease 7 Image result for gene therapy8. STUDY OF DISTURBANCES IN MOLECULAR PATHWAYS DURING ILLNESS: Apart from all the research, this field has faced little disappointment in the past few years, when a volunteer died during gene therapy trial in 1999. Hence, there is a need to work upon the development of such vectors that are more safe and effective to play the significant role in treatment of a disease.

However, it is being used for the treatment of hemophilia B23 and severe immunodeficiency. 3 Fig. Gene Therapy The study of metabolic pathways involved under normal conditions in an individual can be used to detect the change and imbalance in these pathways during the disease. By detecting the gene that harbors a high risk and affect the metabolic pathway at a critical point, we can increase our knowledge about the disease and consequently treat the disease in a better way. 3 During pathway analysis, certain genes sets that corresponds to biological pathways are tested to check if they have a relationship with the phenotype.

Data for this analysis is primarily obtained from gene expression assays and genotyping, as well as from data elements in theory. Genome-wide data sets are now used for discovering pathways and networks related to phenotypes.

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