

Maintaining the Health of Organism at Molecular Level Based on Informative Genomics

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Received date: August 07, 2024; **Accepted date:** August 14, 2024; **Published date:** August 21, 2024

Citation: Evgeniy Bryndin, (2025), Maintaining the Health of Organism at Molecular Level Based on Informative Genomics, *J. Clinical Case Reports and Studies*, 6(7); DOI:10.31579/2690-8808/269

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Abstract

In the context of the molecular life of organisms, genomics helps to understand how genetic information is encoded and how it is implemented at the molecular level, ensuring the vital activity and characteristics of each organism. In the structure of genomes, DNA sequences, the arrangement of genes, regulatory elements and other molecules that make up the genome are studied. Gene functions determine how genes encode proteins and other molecules, their role in metabolism, growth, development and interaction with the environment. Studying changes in the genomes of different species, which helps to understand the origin and adaptation of organisms. The use of modern sequencing, analytics and bioinformatics methods is important for the rapid and accurate study of the molecular basis of life. Molecular mechanisms help to uncover processes such as replication, transcription and translation that underlie the functioning of cells. Overall, informative genomics provides a deep understanding of the molecular basis of life, allowing us to trace how genetic information controls all aspects of the existence of organisms - from the simplest bacteria to complex multicellular creatures. Informative genomics is the foundation for maintaining the life of an organism at the molecular level, the development of biotechnology, genomic medicine, ecology and evolutionary research.

Key Words: molecular level; informative genomics; translational genomic medicine

Introduction

Modern medicine is constantly evolving, introducing new technologies and approaches that change the way we diagnose, treat and prevent diseases. Here are some of the most relevant new areas in medicine today:

1. Bioethics and regulation of new technologies:

- Ethical issues when using genetic technologies and genome editing (e.g., CRISPR).

2. Tissue and cell engineering:

- Creating bioengineered organs and tissues for transplantation.
- Using stem cells to regenerate damaged tissue.

3. Artificial intelligence and machine learning:

- Processing big data to diagnose and predict diseases.
- Automation of image analysis (e.g. X-rays, MRI).

4. Nanomedicine technologies:

- Using nanoparticles to deliver drugs directly to affected cells.
- Improving diagnostics and therapy through nanotechnology.

5. Telemedicine and digital health platforms:

- Online consultations and remote patient monitoring.
- Development of mobile applications for health management.

6. Robotic surgical systems:

- Minimally invasive surgeries using robots (e.g. da Vinci).
- Increasing precision and reducing the risk of complications.

7. Immune therapy and biotechnology:

- Development of vaccines (including mRNA vaccines).
- Treatment of cancer and autoimmune diseases using immunotherapy.

8. Personalized medicine:

- Using genetic data to develop individualized treatment plans.
- Targeted therapy for oncological and hereditary diseases.

9. Informative Genomics:

- Informative genomics is the study and use of all the genetic information of organisms—their genomes—to understand the structural, functional, and evolutionary aspects of life at the molecular level. This field relies on extensive data on nucleotide sequences, genetic variations, regulatory elements, and the relationships between them. Key components of informatic genomics include:

9.1. Genome sequence data: complete or partial DNA sequences obtained using sequencing techniques.

9.2. Genetic annotation: identification and description of genes, regulatory elements, structural features, and functions of genomic regions.

9.3. Databases and bioinformatics resources: organized systems for storing, processing, and analyzing genomic information, such as GenBank, Ensembl, UCSC Genome Browser.

9.4. Analytical algorithms and models: software tools for comparison, search, prediction of functions and interactions based on genomic data.

9.5. Data integration: combining genomic information with transcriptomics, proteomics, metabolomics and other levels of biological data for a comprehensive understanding of the functioning of organisms.

The informative nature of genomics allows not only to store and process huge amounts of data, but also to extract knowledge from them about the molecular mechanisms of life, evolution and interactions of organisms with the environment [1,2]. This is the foundation for the development of personalized medicine, biotechnology and systems biology.

2. Theoretical aspects of genomics

Genomics studies the structure, function, evolution, and relationships of genes and genomes of organisms. It combines knowledge from molecular biology, genetics, bioinformatics, and other sciences to understand the full picture of genetic information and its role in the life of organisms. The main concepts of theoretical aspects of genomics include:

1. Genome structure:

- Study of the organization of DNA, chromosomes, and genes.
- Analysis of repeats, regulatory elements, and non-coding regions.

2. Genetic information:

- Protein coding and regulation of gene expression.
- Mechanisms of transcription, translation, and post-translational modifications.

3. Genomics and its methods:

- DNA sequencing and sequence analysis.
- Comparative genomics to identify evolutionary relationships.
- Genomic mapping and gene annotation.

4. **Genome evolution:

- Study of gene mutations, genomic duplications, horizontal gene transfer.
- Analysis of genome changes during species evolution.

5. Functional genomics:

- Relationship between genotype and phenotype.
- Study of regulatory networks and interactions between genes.

6. Models and algorithms:

- Use of mathematical and statistical methods for analyzing big genomic data.
- Building models to predict the functions of genes and regulatory elements.

7. The importance of the theoretical foundations of genomics:

- Allow us to understand the fundamental mechanisms of genome operation.
- Provide a basis for developing new diagnostic methods, therapy, and biotechnological applications.
- Contribute to the study of evolutionary processes and biological diversity.

Thus, the theoretical foundations of genomics serve as the foundation for all modern genomic science and technology, allowing us to interpret huge amounts of data and gain new knowledge about life at the molecular level.

3. Genomics about the causes of diseases

In the context of the causes of diseases, genomics plays an important role in understanding how changes in DNA affect the development of various diseases. The main aspects of genomics in the study of the causes of diseases include:

1. Identification of genetic risk factors: By analyzing the genomes of patients, scientists identify genetic variations associated with certain diseases, for example, mutations associated with hereditary forms of cancer or cardiovascular diseases.

2. Understanding molecular mechanisms: Genomics helps to understand how certain changes in genes cause pathological processes, which contributes to the development of new diagnostic and therapeutic methods.

3. Personalized medicine: Based on genomic data, it is possible to develop individualized treatment plans that take into account the unique genetic profile of the patient.

4. Study of hereditary diseases: Genomics allows us to identify mutations that are inherited, which is important for prevention and early diagnosis.

5. Drug Development: Understanding the genetic basis of diseases helps to create targeted drugs that attack specific molecular targets.

6. Genomic Diseases: These are inherited diseases caused by abnormalities in the number or structure of chromosomes, in particular, changes in the entire genomic set (for example, aneuploidy). These diseases occur as a result of errors in cell division, leading to an increase or decrease in the number of chromosome sets. The main types of genomic diseases:

6.1. Trisomies - the presence of a third copy of one of the chromosomes:

- Down syndrome (trisomy 21) is the most common genomic disease, characterized by mental retardation, characteristic facial features, heart defects and other disorders.

- Edwards syndrome (trisomy 18) is a severe condition with multiple abnormalities, high mortality at an early age.

- Patau syndrome (trisomy 13) - severe malformations, low survival.

6.2. Tetraploidy and polyploidy - an increase in the full set of chromosomes:

- Usually incompatible with life in humans, with the exception of some cases in the embryonic stage.

6.3. Aneuploidy - the presence of an extra or missing chromosome: - For example, monosomy X (Turner syndrome) — in women, characterized by underdevelopment of sexual characteristics, short stature and other features.

- Multifollicular syndromes and others.

6.4 Causes: - Errors in meiosis or mitosis, leading to incorrect distribution of chromosomes. - The age of the parents (especially the age of the mother during pregnancy) increases the risk of such errors.

6.5 Diagnostics and treatment: - Genetic counseling.

- Karyotyping, molecular diagnostic methods.

- Symptomatic and rehabilitative treatment, since genomic diseases are in most cases chronic and incurable.

It is important to understand that genomic diseases require a comprehensive approach to diagnostics, support and correction of patients' condition. In general, genomics is a powerful tool for revealing the causes of many diseases and developing effective methods for their prevention, diagnosis and treatment.

4. Genomic research of malignant diseases

4.1 The process of transition of normal cell into malignant cell

The process of transition of a normal cell into a malignant cell is a complex and multi-stage biological process, including sequential genetic and epigenetic changes. Below is a description of the main stages and mechanisms of this transformation:

1. Genetic and epigenetic changes:

- Mutations and chromosomal abnormalities: ** occurrence of point mutations, deletions, duplications, translocations, inversions leading to activation of oncogenes or inactivation of tumor suppressor genes.

- Epigenetic changes: hyper- or hypomethylation of DNA, histone changes, which affect the expression of genes associated with the control of growth and proliferation.

2. Activation of oncogenes and inactivation of suppressor genes:

- Oncogenes: genes that stimulate cell growth and division (e.g., *RAS*, *MYC*, *EGFR*). Their activation leads to uncontrolled proliferation.

- Suppressor genes: genes that suppress growth and prevent tumor transformation (e.g., *TP53*, *RB*). Their inactivation removes control over cell division.

3. Changes in cell cycle regulation:

- Dysregulation of cell cycle checkpoints, which leads to uncontrolled division.

4. Formation of abnormal cellular characteristics:

- Eutolization of apoptosis: decreased sensitivity to programmed cell death.

Migration and invasiveness: acquisition of the ability to penetrate into surrounding tissues and metastasize. - Angiogenesis: stimulation of the formation of new blood vessels to feed the tumor. 5. Mechanisms of resistance and survival:

- Providing resistance to stresses such as hypoxia, nutrient deficiency, therapy.

6. Mechanisms and factors involved in transformation:

- Genetic instability: increased probability of new mutations.

- Environmental influences: carcinogens, radiation, toxic substances, viruses.

- Viral infections: e.g. human papillomavirus (HPV), hepatitis B and C viruses, which can integrate into the genome and activate oncogenes or inactivate suppressor genes.

7. Summary model:

- The process of cell transformation involves the accumulation of mutations, changes in gene regulation, activation of oncogenes and inactivation of suppressor genes, which leads to loss of control over growth, invasion and metastasis. This process is often multifactorial, and its gradual nature allows for early detection and potential interruption.

4.2 Molecular pathways and genes of malignant tumor formation

The formation of malignant tumors (cancer) is a complex multi-stage process that includes the activation of oncogenes and the inactivation of tumor suppressor genes. The main molecular pathways and genes involved in oncogenesis are listed below:

1. Rig-casting pathway (RAS/MAPK pathway):

- Genes: RAS (KRAS, NRAS, HRAS), RAF (RAF1, BRAF), MEK (MAP2K1, MAP2K2), ERK (MAPK3, MAPK1)

- Role: Regulates cell growth, division, and differentiation. Mutations lead to constant activation of the signal, stimulating proliferation.

2. PI3K/AKT/mTOR pathway:

- Genes: PIK3CA, AKT1-3, PTEN (inhibits the pathway), mTOR

- Role: Controls cell growth, survival and metabolism. Activation promotes oncogenesis.

3. Tumor suppressor genes (growth inhibition signals):

- Genes: TP53, RB1, CDKN2A (p16INK4a, p14ARF), PTEN

- Role: Provides control over division and prevents uncontrolled proliferation. Their inactivation leads to tumor development.

4. Nuclear factors and the Wnt/ β -catenin pathway:

- Genes: APC, CTNNB1 (β -catenin), TCF7L2

- Role: Regulates cell differentiation and growth. Mutations lead to constant activation of the Wnt signal and tumor growth.

5. Growth factors and their receptors:

- Genes: HER2 (ERBB2), EGFR, MET

- Role: Mutations or expression enhance signaling, stimulating tumor growth.

6. Involvement in angiogenesis:

- Genes: VEGFA, VEGFR

- Role: Provide blood supply to the tumor, promoting its growth and metastasis.

7. General mechanism:

- Mutations and changes in these pathways lead to disruption of cell cycle regulation, increased proliferation, resistance to apoptosis and promote metastasis.

- Modern therapies are aimed at these molecular targets.

4.3 Genomic oncology

Genomic oncology is a field in medicine and oncology that studies the role of genomic changes in the development, progression, and treatment of cancer. The basic idea is that cancer is the result of genetic and genomic mutations that disrupt the normal processes of cell growth and division. Key aspects of genomic oncology:

1. Genomic cancer research:

- Analysis of changes in the DNA of tumor cells, such as mutations, translocations, deletions, amplifications.

- Use of sequencing methods (e.g., next-generation sequencing) to identify characteristic genomic abnormalities.

2. Personalized therapy:

- Determination of genomic tumor profiles to select the most effective drugs.

- Development of targeted drugs aimed at specific genomic mutations (e.g., EGFR, ALK, BRAF inhibitors).

3. Prognosis and diagnostics:

- Using genomic data to assess the prognosis of the disease.

- Monitoring mutations during treatment to assess the effectiveness and possibility of resistance development.

4. Biomarkers:

- Identification of genomic biomarkers that help predict the response to therapy or the risk of relapse.

5. Therapeutic strategies:

- Development of new treatments based on the genomic features of the tumor, such as gene therapy or immunotherapy.

6. Hormonal genomic suppression of prostate cancer cells combines hormonal therapy with genomic technologies to more effectively treat prostate cancer. The main aspects of this approach are presented below:

6.1. Rationale for the approach:

- Prostate cancer often depends on androgens (male hormones) such as testosterone, which stimulate the growth of tumor cells.
- Hormonal therapy aims to reduce testosterone levels or block its action, which slows the growth of cancer.

6.2. Genomic intervention:

- Use of gene therapy technologies such as CRISPR-Cas9 or other gene editing methods to suppress or disable genes responsible for the growth and survival of cancer cells.
- Target genes may include androgen receptors, genes involved in proliferation signaling pathways and anti-inflammatory response.

6.3. Combination of hormonal and genomic therapy:

- Hormonal drugs (e.g., testosterone antagonists, luteinizing hormone-releasing factor) reduce androgen levels.
- Genomic methods allow additional switching off of internal mechanisms of cancer, making treatment more precise and effective.

6.4. Personalized approach:

- Analysis of genomic profiles of tumors to identify the most vulnerable genes and pathways.
- Development of individual suppression strategies that minimize side effects and increase efficiency.

6.5. Prospects and challenges:

- Possibility to slow down or stop cancer progression, making treatment more targeted.
- Need to develop safe methods of delivering genomic agents and minimizing the risk of unwanted changes.
- Importance of monitoring genomic changes in the tumor during treatment dynamics.
- Hormonal genomic suppression of prostate cancer cells is a promising direction that combines hormonal therapy with advanced genomic technologies to achieve more effective and personalized treatments for this oncological disease.

7. Advantages of genomic oncology:

- Increasing the effectiveness of treatment due to its individualization.
- Reducing side effects due to more precise therapy.
- The ability to detect resistance mutations and adjust the treatment strategy.

In general, genomic oncology is a promising area that promises to significantly improve the results of cancer treatment and make it more accurate and personalized [3-9].

5. Genomic diagnostics

Genomic diagnostics is a method of determining and analyzing genetic information in order to identify hereditary and acquired genetic changes associated with various diseases and conditions of the body. It allows obtaining accurate information about the presence of mutations, variations or structural changes in the genomes of patients, which contributes to more accurate diagnostics, prognosis and selection of therapy. The main aspects and possibilities of genomic diagnostics:

1. Detection of hereditary diseases: detection of mutations that cause genetic diseases (for example, cystic fibrosis, hereditary cancer, hemophilia).

2. Diagnostics of oncological diseases: detection of genetic changes in tumor cells, which helps to determine the type of cancer and choose the most effective treatment.

3. Personalized medicine: selection of therapy based on the patient's genetic profile.

4. Genetic predisposition: identification of variants that increase the risk of developing certain diseases.

5. Psychiatric and neurological diagnostics: search for genetic factors associated with diseases of the nervous system.

6. Career and reproductive consulting: determination of carriage of genetic diseases, pregnancy planning.

7. Main methods of genomic diagnostics:

- Next-generation sequencing (NGS) - allows you to quickly and accurately determine the DNA sequence and identify mutations and variations.

- Genetic microarrays (arrays) - identification of genome variants, such as SNPs, copy numbers and structural changes.

- Multigene panels - analysis of several genes associated with a specific disease.

- PCR methods - to determine the presence or absence of specific mutations.

- Fluorescent hybridization (FISH) - to detect structural changes in chromosomes.

8. Advantages of genomic diagnostics:

- High accuracy and sensitivity.

- Possibility of early detection of diseases. - Individualized approach to treatment.

- Improving the prognosis and quality of life of patients.

Thus, genomic diagnostics is the most important tool of modern medicine, allowing us to move towards a more accurate and personalized approach to the diagnosis and treatment of diseases [10-13].

6. Genomic Therapy

Genomic therapy is a modern medical technique aimed at correcting or changing genetic material in order to treat or prevent diseases. This approach uses various technologies to introduce, remove, replace, or edit genes in a patient's cells to eliminate the cause of the disease or reduce its symptoms. The main types of genomic therapy include:

1. Gene replacement - introducing a corrected gene to replace a defective or missing one.

2. Gene editing - using technologies such as CRISPR-Cas9 to precisely edit DNA in specific areas.

3. Gene inactivation - suppressing the activity of harmful genes.

4. Transgene therapy - introducing genetic material from foreign organisms to achieve a therapeutic effect.

5. The use of genomic therapy covers a wide range of diseases:

- Rare hereditary diseases such as cystic fibrosis, hemophilia, muscular dystrophy.
- Oncological diseases, where genes responsible for tumor growth can be suppressed or corrected.
- Infectious diseases, such as treatment for HIV or hepatitis B/C.
- Some forms of inherited eye diseases and cardiovascular diseases.

6. Advantages of genomic therapy:

- Ability to eliminate the cause of the disease at the genetic level.
- Long-term or even permanent effect.
- Personalized approach to treatment.

7. Disadvantages and challenges:

- High cost of development and implementation of procedures.
- Possible side effects, including unpredictable mutations. - Ethical and legal issues associated with genome editing.
- Limited availability and the need for further research to confirm safety and effectiveness.

Genomic therapy is a promising area of medicine that can radically change approaches to the treatment of many diseases and provide more effective and personalized medical care [14-15].

7. Genomics-based Translational Medicine

Genomics-based translational medicine is a field of medical science and practice that aims to rapidly translate basic research findings into clinical practice to improve diagnosis, treatment, and prevention of diseases. This field combines genomic data with other biomarkers and technologies to create a more personalized and effective approach to healthcare. Key aspects of genomics-based translational medicine include:

1. Personalized treatment:

- Using genomic data to select the most effective drugs and dosages.
- Developing individualized therapies based on a patient's genetic profile.

2. Rapid diagnostics:

- Implementing genomic data sequencing and analysis methods for early detection of diseases.
- Detecting predispositions to diseases long before symptoms appear.

3. Biomarkers and molecular diagnostics:

- Identifying molecular markers associated with disease progression and response to treatment.
- Monitoring the effectiveness of therapy and timely adjustment of treatment strategies.

4. Development of new therapeutic approaches:

- Gene therapy and genome editing technologies to eliminate the causes of hereditary diseases.
- Targeted drugs aimed at molecular targets identified during genomic research.

5. Clinical research and implementation:

- Transfer of research results into practice through clinical protocols and guidelines.
- Training of medical professionals in new methods and technologies.

6. Ethical, legal and social aspects:

- Ensuring the protection of personal genetic data.
- Regulation of the use of genomic information and technologies.

7. Benefits of translational medicine based on genomics:

- Increasing the accuracy of diagnosis and the effectiveness of treatment.
- Reducing side effects due to more accurate therapy methods.
- Improving the prognosis and quality of life of patients.
- Minimizing unnecessary procedures and costs.

8. Challenges and Prospects:

- Need for large-scale investments in infrastructure and training of specialists.
- Addressing ethical issues related to the storage and use of genetic information.
- Developing standards and protocols for the implementation of genomic technologies in clinical practice.
- Continuing research to expand the knowledge base and improve the accuracy of methods.

Overall, translational medicine based on genomics is a powerful tool for transforming healthcare, making it more personalized, preventive and effective [16].

9. Conclusion

The development of medicine based on genomics is becoming one of the most promising and revolutionary areas of modern science and healthcare. Its use in medicine allows us to move from mass treatment methods to personalized medicine based on the individual genetic characteristics of the patient. Assessment of the risk of developing diseases based on genetic data. Monitoring and early intervention to prevent complications.

Genomics continues to develop, opening up new opportunities in medicine and biotechnology. Genomics of viruses, bacteria and other microorganisms for the development of vaccines and antibiotics. Detection of new strains and their characterization. More effective and less toxic treatment. Development of new methods of prevention.

The development of genomic medicine based on artificial intelligence is one of the most promising areas of modern medical science [17-23]. It allows us to significantly improve the accuracy of diagnosis, personalize treatment and accelerate the discovery of new therapeutic agents. The key aspects of this development are presented below:

1. Big data analysis and genome sequencing:

- Using machine learning algorithms to process and interpret huge amounts of genomic data.
- Identifying significant mutations, associations, and patterns that may indicate the risk of developing certain diseases.

2. Personalized Medicine:

- Creating individual health profiles based on genetic information.
- Developing targeted therapies and preventive measures that take into account the unique genetic characteristics of each patient.

3. Predictive Analytics and Early Diagnostics:

- Predicting the likelihood of developing diseases based on genetic markers.
- Optimizing preventive strategies and monitoring health status.

4. Automating the interpretation of genetic data:

- Using artificial intelligence to automatically evaluate sequencing results and determine the clinical significance of detected variations.

5. Drug Development:

- Using artificial intelligence to quickly find potential targets and develop new drugs based on genomic data.

6. Ethical and Legal Aspects:

- Ensuring the security and privacy of genomic information.

- Creating a regulatory framework for the use of AI in genomic medicine.

7. Development prospects:

- Improving the efficiency of diagnostics and therapy.

- Reducing the cost of medical services.

- Expanding the possibilities of disease prevention.

- Providing a more accurate and personalized approach to treatment.

The integration of artificial intelligence into genomic medicine opens up new horizons for the medicine of the future, making it more accurate, individualized and effective. In general, the integration of genomic data into clinical practice promises to significantly improve the quality of medical care, make it more accurate and effective, and open up new horizons in the fight against diseases [24].

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