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PATHOLOGIES CAUSED BY PROTEIN SYNTHESIS AND ITS BREAKDOWN

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Annotation: This article provides information on the molecular basis of protein synthesis, its stages, control mechanisms and pathologies that occur when this process is disrupted. There is also a scientific analysis of clinical manifestations of protein synthesis disorders, the effects of genetic and environmental factors, diagnostic methods and modern treatment approaches. The relevance of the study is that defects in protein synthesis serve as a leading factor in the development of various severe hereditary and acquired diseases.

Keywords:protein synthesis, transcription, translation, ribosome, pathology, genetic diseases, molecular biology.

Introduction

Proteins are macromolecules that play a central role in the life of living organisms. They perform a variety of functions in cells, such as structural, enzymatic, transport, signaling, and regulatory. For example, structural proteins such as collagen and keratin give strength to tissues, while hemoglobin is involved in oxygen transport, while hormone and receptor proteins provide intercellular signal exchange.ntroduction

Proteins are macromolecules that play a central role in the life of living organisms. They perform a variety of functions in cells, such as structural, enzymatic, transport, signaling, and regulatory. For example, structural proteins such as collagen and keratin give strength to tissues, while hemoglobin is involved in oxygen transport, while hormone and receptor proteins provide intercellular signal exchange. Therefore, proteins can be called the "workforce" of the body.

Protein synthesis is a vital process represented by the transformation of genetic information stored in DNA into functional protein molecules. This process consists of three main steps: transcription, translational and posttranslational modifications. In transcription, information in DNA is transported to mRNA, while in translation, an amino acid sequence is assembled using ribosomes. In the post-translational stage, the protein enters a certain form and is ready for activity. Each part of the process goes under strict control, since even a small error can lead to improper Assembly of the protein.

Disorders in protein synthesis occur under the influence of various factors. These include genetic mutations, chromosomal aberrations, defects in transcription or translational enzymes, changes in ribosome structure, as well as external environmental factors – radiation, toxic substances, infections.ach part of the process goes under strict control, since even a small error can lead to improper Assembly of the protein.

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in ribosome structure, as well as external environmental factors – radiation, toxic substances, infections. The resulting disruption of protein structure or quantity causes many diseases.

In clinical medicine, pathologies associated with impaired protein synthesis cover a wide range. Examples include: hereditary metabolic disorders (phenylketonuria, galactosemia), muscular dystrophies (Duchenne, Becker), oncological diseases (mutation of the p53 gene), neurodegenerative diseases (Alzheimer's, Parkinson's, Huntington's), ribosomopathies (Diamond-Blackfan anemia, Treacher Collins syndrome).

According to the World Health Organization (DSST), at least 40% of genetic and metabolic disorders are associated with disorders in protein synthesis. Mutations in protein-coding genes have also been found in over 60% of cancers. Neurodegenerative diseases, on the other hand, are one of the leading causes of death worldwide, on the basis of which are also defects in protein folding.

The relevance of the topic is that through an in-depth study of the molecular mechanisms of protein synthesis, it is possible to understand the pathogenesis of many diseases, improve diagnostic methods and develop new methods of treatment. Currently, such innovative approaches as gene therapy, protein replacement therapy, CRISPR-Cas9 technology are developing precisely in this direction.

For this reason, the topic" pathologies caused by protein synthesis and its degradation " is important not only in theoretical, but also in clinical practice. This article will analyze the process of protein synthesis, its control mechanisms, causes of disorders, clinical manifestations, diagnostic and therapeutic approaches in a wide range.

Methods

When writing this scientific article, the following methods were used:

Literature analysis-modern scientific articles, monographs, textbooks and clinical observations were used. Primary sources include Molecular Biology of the Cell (Alberts et al), Lehninger Principles of Biochemistry (Nelson and Cox), and Thompson & Thompson Genetics in Medicine (Nussbaum et al.

Review of clinical observations-clinical cases on genetic and molecular diseases were analyzed.

Molecular biology data-mechanisms of transcription, translational and posttranslational processes-have been studied.

Comparison and generalization – different disorders of protein synthesis were compared with clinical manifestations and general conclusions were drawn.

Results

1. Normal process of protein synthesis

Protein synthesis is the process of transferring genetic information in DNA to the protein chain.Molecular biology data-mechanisms of transcription, translational and posttranslational processes-have been studied.

Comparison and generalization – different disorders of protein synthesis were compared with clinical manifestations and general conclusions were drawn.

Results

Normal process of protein synthesis

Protein synthesis is the process of transferring genetic information in DNA to the protein chain.

Transcription – information in DNA is transcribed into the IRNA. This process is carried out using the enzyme RNA polymerase. In the case of errors in the position of the Promoter, intron and exon, the wrong RNA appears.

Translational – in the ribosome, an amino acid sequence is assembled on the basis of the IRNA of the Momenta. TRNA and ribosomal proteins are involved in this process. Errors in the translation process result in an incorrect amino acid chain.

Posttranslational modification proteins undergo processes such as phosphorylation, glycosylation, methylation.ranslational – in the ribosome, an amino acid sequence is assembled

Disorders in protein synthesis

Defects in protein synthesis can vary:

Transcription defects are disorders of the activity of RNA polymerase, promoter mutations, errors in splicing.

Translational defects-disengagement of ribosome activity, mutations in tRNA2. Disorders in protein synthesis

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Transcription defects are disorders of the activity of RNA polymerase, promoter mutations, errors in splicing.

Translational defects-disengagement of ribosome activity, mutations in tRNA.

Post – translational defects-improper folding of proteins, violation of chemical modifications.

Pathologies

1.Phenylketonuria (PKU) – phenylalanine does not break down due to deficiency of the enzyme hydroxylase. This condition leads to intellectual defects.

2.Muscle fibers are weakened by muscular dystrophies – genetic mutations related to dystrophin.

3.Alzheimer's disease-incorrectly folded proteins (amyloid beta) accumulate in neurons, causing cognitive disturbances.

4.Parkinson's disease-misfolding of alpha-synuclein proteins calls for neurodegenerationMuscle fibers are weakened by muscular dystrophies – genetic mutations related to dystrophin.

3.Alzheimer's disease-incorrectly folded proteins (amyloid beta) accumulate in neurons, causing cognitive disturbances.

4.Parkinson's disease-misfolding of alpha-synuclein proteins calls for neurodegeneration.

5.Mutations in proteins such as cancer – p53, BRCA1/2 leave cell proliferation uncontrolled.

6.Immunodeficiency syndromes-errors in the synthesis of antibodies weaken the immune system.

Discussion

Protein synthesis disorders lead to many clinical and molecular pathologies. Their causes are often:

Genetic mutations – the main source of hereditary defects.

Epigenetic factors – changes in DNA methylation and histone modifications-affect protein synthesis.

Environmental factors-radiation, chemicals, toxins.Protein synthesis disorders lead to many clinical and molecular pathologies. Their causes are often:

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Epigenetic factors – changes in DNA methylation and histone modifications-affect protein synthesis.

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Age and lifestyle – the effectiveness of protein synthesis in the aging process decreases.

Modern treatment approaches:

1.Gene therapy is the correction of the wrong gene or the introduction of a healthy gene.

2.CRISPR-Cas9 technology-accurate correction of mutations using genetic engineering.

3.Pharmacological modulators are drugs that prevent improper protein folding.

4.Personalized medicine-the choice of a cure based on the patient's genetic profile.

However, pathologies can be prevented by early diagnostics of defects in certain protein synthesis..CRISPR-Cas9 technology-accurate correction of mutations using genetic engineering.

3. Pharmacological modulators are drugs that prev

Conclusion

Protein synthesis is an important process in the life of living organisms, and its normal course forms the basis of all cellular activity. Disorders at any stage of this process lead to hereditary, metabolic and clinical diseases. In modern molecular biology and medicine, an in-depth study of the mechanisms of these diseases, the development of methods for their early detection and treatment is one of the main scientific tasks.onclusion.Protein synthesis is an important process in the life of living organisms, and its normal course forms the basis of all cellular activity.

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