# MORPHOLOGICAL AND GENETIC BASIS OF STRUCTURAL AND FUNCTIONAL CHANGES IN ORGANS IN BETA THALASSEMIA AND CYSTIC FIBROSIS

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Scientific and Practical Conference: Science and Technology in the Modern World
https://doi.org/10.5281/zenodo.18888296

**Relevance**

Genetic disorders represent a major health challenge worldwide. Among the most important hereditary diseases are Beta Thalassemia and Cystic Fibrosis. These disorders are caused by mutations in specific genes that affect normal physiological processes and lead to severe systemic complications. Beta thalassemia results from mutations in the HBB gene responsible for beta-globin chain production in hemoglobin. The disorder leads to ineffective erythropoiesis, chronic anemia, and structural changes in the bone marrow, liver, and spleen. Patients often require lifelong blood transfusions and medical management. Cystic fibrosis is caused by mutations in the CFTR gene, which regulates chloride ion transport across epithelial cell membranes. The mutation leads to thick mucus accumulation in respiratory and digestive organs, resulting in chronic infections, lung damage, and pancreatic dysfunction.

**Purpose of the Study**

The purpose of this study is to investigate the morphological, genetic, and functional alterations associated with beta thalassemia and cystic fibrosis. By analyzing hematological parameters, tissue structure, and genetic mutations, the research aims to understand how these inherited disorders affect organ function and contribute to disease progression.

**Research Tasks**

Evaluation of morphological alterations in organs affected by beta thalassemia and cystic fibrosis.

Study of erythrocyte morphology and hemoglobin levels in patients with beta thalassemia.

Investigation of epithelial tissue changes in lungs and pancreas in cystic fibrosis.

Identification of genetic mutations using molecular diagnostic techniques.

Analysis of inflammatory markers associated with chronic disease progression.

Correlation between gene mutation severity and structural organ damage.

**Materials and Methods**

The study utilizes clinical samples and laboratory data obtained from individuals diagnosed with beta thalassemia and cystic fibrosis. Biological materials analyzed include peripheral blood samples, bone marrow samples, and tissue samples from lungs and pancreas when available. Advanced laboratory methods were applied to analyze morphological and genetic characteristics. Hematological testing was performed to determine hemoglobin levels, erythrocyte morphology, and other blood indices. Histological examination was conducted using Hematoxylin and Eosin staining to evaluate structural tissue alterations. Molecular genetic analysis was performed using polymerase chain reaction (PCR) and DNA sequencing techniques to identify mutations in the HBB and CFTR genes. Statistical analysis of the collected data was performed using Student's t-test and correlation analysis with significance defined as $p < 0.05$.

**Expected Results and Discussion**

The expected findings of this study include significant structural and functional changes in organs affected by genetic disorders. In beta thalassemia, bone marrow expansion and abnormal erythrocyte development are expected to be observed. Enlargement of the spleen and liver due to increased red blood cell destruction may also be detected. In cystic fibrosis, epithelial damage in respiratory tissues, mucus accumulation in airways, and chronic inflammatory responses are anticipated. These pathological changes are expected to correlate with mutations identified in the CFTR gene. Understanding the relationship between genetic mutations and morphological alterations will contribute to improved diagnostic approaches and therapeutic strategies for managing hereditary diseases.

**References:**

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