DNA-DIAGNOSTICS OF CYSTIC FIBROSIS

Description
Cystic fibrosis is one of the most spread inherited diseases. On the average one per 2000 is born with cystic fibrosis in Russia.
Small deletions in the CFTR gene (ΔF508, ΔI507 and del1677) can be detected by direct electrophoretic separation of polymerase chain reaction (PCR) products in polyacrylamide gel. The method of allele-specific PCR is proposed for differential diagnostics of the ΔF508 mutation, when the mutation is revealed by the presence or absence of PCR-product under amplification with the primer with or without the deletion.

An example of delF508 identification by PCR subsequent analysis of the products by PAGE

1. Pedigree of the family with affected child (black square - affected child, white circle - unaffected child)
2. Electrophoregram of DNA fragments of the members of the family (upper band - normal fragment, lower band - fragment with deletion)

Technical appraisal and economic benefits
Annual costs of medical treatment of one child are about 10 000$. The methods being developed will allow prenatal diagnostics of cystic fibrosis.

Application areas
Medicine.

Development stage
Clinic laboratories of hospitals, polyclinics, medical-genetic consultations, maternity houses, centers for family planning, etc.
Regional Perinatal Center (Novokusnetsk).

Patent situation
Authors’ rights for the diagnostics technique of cystic fibrosis using allele-specific PCR are protected by the RF patent No. 2151188.

Commercial offers
The Institute of Cytology and Genetics concludes contracts with healthcare institutions on development of the methods of analyzing the complete range of mutations in the CFTR gene for diagnostics of cystic fibrosis and on clinical tests, as well as on training of personnel.
**Estimated cost**
One DNA analysis costs about 10$.

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