A mitochondrion is a organelle under nuclear and mitochondrial genetic control and its maintence requires a nucleo-mitochnodrial cross-talk. Better understanding of mitochondrial pahologies using various gene expression techniques. This thesis presents the implementation and application of microarray technology for identification of disease causing genes in respiratory chain disorders, expecially in patients with ATP synthase deficiency of nuclear origin. The introduction to this thesis provides an overview of mitochondrial structure and biogenesis, respiratory chain complexes and their disorders as well as different types of microarrays. A study of mitochondrial ATP synthase deficiency represents the major experimental part of this thesis and is accordingly that were used in further studies centered on characterization of inherited metabolic disorders. This is documented in positional cloning of patientes with lysosomal storage disorders – mucopolysaccharidosis IIIC.