## Abstract

The study of rare genetic diseases presents unique opportunity to uncover the genetic and molecular basis of human traits and greatly helped to the identification of genes, to the elucidation of their function and to the characterization of metabolic pathways and cellular processes. Over the past decades, linkage analysis has been appropriate approach to search for the genes causing Mendelian diseases and contributed to the identification of many genes, but the genetic cause of many diseases remains unknown. New methods of studying the human genome, microarray technology and massively parallel sequencing (next generation sequencing), represent a way to efficiently identify the cause of genetically determined diseases, based on direct observation of mutations in the genome of affected individuals. These techniques replaced the traditional method of disease gene identification represented by linkage analysis and sequencing of candidate genes and have become the standard approach to elucidate the molecular basis of diseases. In this work, i describe the the results achieved by using these methods - identification of the genes underlying mucopolysacharidosis type IIIC, isolated defect of ATP synthase, Rotor syndrome, autosomal dominat ANCL and GAPO syndrome.