Proper functioning beta cells in humans is necessary to maintain blood glucose levels within normal values. In the case of defects of genes involved in beta-cell function we can be observed symptoms of diabetes. Form of the disease associated with severe, rare gene mutations β cells can take clinical picture of MODY (ang. Maturity onset diabetes of the young) or neonatal diabetes (PNDM - called. Permanent neonatal diabetes mellitus). Some report suggest that observed in children, idiopathic form of diabetes type 1 (no autoantibodies present) may be caused by a single gene defect. So far, according to a database cataloging inherited human disease (OMIM - called. Online Mendelian Inheritance in Man) distinguished 11 different genes responsible for the formation of phenotypic clinical MODY: HNF4α (MODY1), GCK (MODY2), HNF1α (MODY3), PDX1 (MODY4), HNF1β (MODY5), NEUROD1 (MODY6), KLF11 (MODY7), CEL (MODY8), PAX4 (MODY9), INS (MODY10) and BLK (MODY11). The aim of the project is to search for mutations in the sequence of known genes involved in the development of neonatal diabetes, MODY, and the identification of new not fully characterized genes responsible for the development of diabetes using the above technique NextGeneraton Sequencing (target sequencing and exome sequencing). This will allow you to define the clinical picture of these diabetes-induced genetic defects in the system hetero- and homozygous.

To date, no publications have appeared clearly identifying the genes involved in the development of monogenic forms of diabetes. Publications based on sequencing technique called chromosome entire areas. exosome sequencing are difficult to make due to the large amount of data for analysis, and are currently available bioinformatics tools are not able to consolidate the large number of variables in regard to the clinical parameters. Publications describing the above problem mainly identify areas for potential location of the gene responsible for the development of monogenic diabetes, forcing undertake the next stage of the project focused on a specific region of the chromosome.

Analysis of candidate genes originating, inter alia, the GWA - Genome-wide Association Study, in patients with MODYx and PNDx would take weeks would consume large financial outlays and its effect would not be adequate for the intended purpose. Therefore, the technique of sequencing exon of the whole genome (exome sequencing) appears to be the most effective solution because of the specificity, narrowing the analyzed areas, precision and speed with as many treated groups is a technology of choice and in terms of economic and time while a trial in the future, adjusting the optimal treatment protocols in accordance with a defined pathogenesis