Chronic pancreatitis (CP) is a persistent inflammation of the pancreas, that results in irreversible morphological changes and impairment of both exocrine and endocrine functions. The pancreas is an organ that does not regenerate and each subsequent exacerbation impairs the function of the organ. CP develops as a consequence of the interactions between genetic and environmental factors. In children, the most common causes of CP include gene mutations, anatomical defects of the pancreatic duct, lipid disorders and diseases of the biliary tract. However, approximately 25% of CP have undefined cause of CP – idiopathic CP. In a significant proportion of patients pancreatitis also occurs in the family and these are termed familial CP (FCP).

Patients with FCP typically show mutations in a set of known genes associated with CP, but still the genetic basis of a large fraction of this group remains unexplained. In 60% of children with FCP, subjected to genetic diagnostic testing in Department of Medical Genetics, Institute of Mother and Child, no defects in known CP genes were identified. We hypothesized that in these patients, other - not yet described - genetic factors are involved in CP development. Therefore, the goal of this study is to identify novel genetic factors associated with increased risk of CP. To this aim, we plan to apply next-generation sequencing technology (NGS), which enables the simultaneous analysis of all the coding regions of the human genome. Our approach will allow us the identification of new defects in genes previously not linked to the process of CP. This may lead to discovery of new mechanisms underlying pathology of CP. The results of this study may be of value in future establishment of new therapeutic strategies depending on the type of defect and lead to development of a more efficient molecular diagnostic algorithm for CP patients. This last is essential for providing the complex genetic counselling, special medical care of patients and perhaps implementation of preventive measures (e.g. lifestyle modifications) in high-risk CP individuals.