Research on understanding the etiology of miscarriages has been conducted for decades now. It might seem we know a lot, yet it is not enough. The reason for 50% of miscarriages still remains unknown. With such an extensive progress in the field of molecular biology and genetics that we have witnessed over the past years, there's more and more focus on the genetic factors which may be connected to the pregnancy lost. Since the 60s, when the first data was released on karyotype's analysis in a still-born fetus, we have known that the chromosomal aberration are the most common changes in the material samples from a spontaneous miscarriage. The anomalies in the number of chromosomes are identified with more than 50% miscarried embryos.

Along with the introduction of new techniques from the molecular biology field into the diagnostics of couples suffering from spontaneous miscarriages, we discover new genetic factors in the etiology of miscarriages. Applying the array comparative genomic hybridization (aCGH) led to identifying structural changes in the embryo' genome. These microdeletions and microduplications turned out to be the second most reoccurring change detected in the product of conception. Introducing the next generation sequencing (NGS) revolutionized the genetic diagnostics, opening the way to the extensive analysis of single-gene causes of miscarriages. This innovative technique allows a parallel analysis of tens, thousands of genes or even the entire genome over the course of one single experiment. To this day, polymorphisms in over 100 genes were connected to the etiology of recurrent miscarriages.

As a clinical geneticist, I encounter the problem of recurrent miscarriages among my patients on a regular basis. Seeing their heroic fight to become parents, I decided to identify the least recognized causes of pregnancy lost, these being single-gene mutations. The main objective of my research is identifying candidate genes related to the etiology of recurring miscarriages within the population of Polish patients. Applying the NGS technique will allow to analyze in parallel changes in dozens of genes, named based on the literature data. The experimental group will consist of couples suffering from recurrent pregnancy lost, but with excluded selected well-known genetic and non-genetic causes of miscarriages. The chorionic villi sample, in which the chromosomal aberration and microaberrations had been excluded during preliminary research, will also be analyzed. This will help determine if the genetic changes in fetus related to the recurring miscarriages are more often a new mutation or are of a paternal origin.

I believe that the results of my research will lead to a more extensive understanding of the mechanism of human reproduction and will also translate into higher efficiency of genetic diagnostics for couples suffering from recurrent pregnancy lost.