Phenylketonuria (PKU) is one of the most common inherited metabolic diseases. Untreated PKU can result in brain damage due to the toxicity of phenylalanine – an amino acid, which in PKU patients cannot be removed as it happens in healthy people. To avoid brain damage, patients with PKU have to follow a very strenuous diet for their entire lives, which is very difficult. However, the biggest challenge of PKU treatment is probably prevention of brain damage in children of mothers with PKU. High amounts of phenylalanine in mother's blood during pregnancy can irreversibly block normal growing process of the child's brain and head (which is referred to as maternal PKU syndrome). Unfortunately, such dramatic situations are very common. Interestingly, some children still develop normally even in case of poor diet control in their mothers. Our preliminary findings suggest the existence of a not yet known genetic variant that controls the mother's capacity to remove the excess of phenylalanine and could be responsible for the observed differences in risk of brain damage. In this project we intend to identify this variant.

We will invite all Polish patients with PKU to our study, and will use the most advanced genetic techniques (so-called next generation sequencing and microarrays), which allow for reading of hundreds of thousands "genetic letters" within a short time and can help us to solve the investigated problem. We hope that this project can add to our knowledge on PKU and in the future can help to save some of the children at risk of maternal PKU syndrome.