Infertility is a rising problem for many couples and male factor is present in approx. 50% of diagnosed patients. Despite the development both diagnostic and therapeutic new techniques significant portion causes of infertility remains unknown. Next-generation sequencing technology enabled the discovery of the genetic basis of many diseases so it to discover the causes of male infertility is fully justified.

The aim of the project is to use the analysis of WGS (*Whole Genome Sequencing*) to define new critical genetic variants responsible spermatogenesis impairment. A key and innovative aspect of the proposed approach is to focus on sequences which are localized in DNA regulatory fragments such as promoters, enhancers or silencers and sequences coding RNA regulatory molecules. These sequences described earlier as "junk DNA" appear to be extremely important especially in the cell differentiation regulation and pathogenesis of diseases. The integral and key part of proposed project is functional verification of the novel causative variants in the non-coding sequences in unique *in vitro* model of male gonad.