The term "Overgrowth syndromes" (OGs) is frequently used by clinical geneticists as well as other medical practitioners. However, no uniform definition has yet been developed. The medical literature on OGs recognizes them as a highly heterogeneous group of disorders, that share a common features of overgrowth. OGs can be divided into two categories based on their phenotype: generalized (complete) or partial (segmental). Generalized OGs are often characterized by high overall parameters as body weight, height and head circumference being + 2 SD (standard deviation). Segmental OGs cause hypertrophy located in a specific part of the body, several organs, or regions. Often, using clinical criteria, congenital hypertrophic defects are classified as *syndromic*, if the clinical diagnostic criteria for any syndrome are met. In such cases, OGs are associated with facial dysmorphic features, large head, enlarged visceral organs, muscular hypotonia, joint laxity, and psychomotor retardation. If the intellectual range is normal, isolated OGs should be considered due to family predisposition, maternal gestational diabetes and / or hormonal disorders.
It is extremely important to make an early diagnosis when OGs is suspected, and to implement an appropriate prophylactic management plan, due to the increased risk of neoplasia in children and adolescents.

Currently, the most attention is paid to the development and possibilities that GWAS (Genome Wide Association Study) or WES (Whole Exome Sequencing) research brings to the evolution of molecular genetics. For this reason, almost every month, new mutations or variants in known genes are discovered, candidate genes for previously unknown entities are described, and new syndromes are defined. Compared to the previous 5 years, the number of new descriptions of "Overgrowth" in the OMIM database (Online Medelian Inheritance in Man) has increased significantly, and it would seem that we already know everything about genes. Although the molecular basis of OGs has been defined and diagnostic steps and molecular techniques are evolving, the cause still remains unknown in approximately 40% of patients.

The main assumption of the proposed project is to determine the genetic basis of OGs in the Polish and Lithuanian populations; to classify the identified genetic variants into known and/or unknown candidate genes related to overgrowth conditions; to evaluate what effects identified pathogenic variants may have on the phenotype of patients with overgrowth conditions; to determine potential structural significance of identified oncogene and tumour suppressor gene mutations; to establish rapid and cost-effective diagnostic algorithms for overgrowth conditions.

We hope that after the development of appropriate diagnostic algorithms, the Medical Technology Assessment and Tariffication Agency in Poland and its Lithuanian counterpart will consider adequate valuation of genetic tests. Furthermore, knowing the molecularly confirmed clinical diagnosis, and the expected symptoms associated with the condition, will let us develop appropriate management protocols for specific overgrowth conditions to prevent or readily detect and successfully treat neoplasia.

To the best of our knowledge, no one in Poland or Lithuania has thus far attempted to thoroughly describe the morphological phenotype of people with isolated or syndromic overgrowth. Currently, there are no reports of ongoing molecular studies on overgrowth in our countries either. This first collaborative study between Poland and Lithuania makes our project particularly innovative and could bring tremendous hope for people struggling with overgrowth and its consequences in our countries. Furthermore, geographical and historical proximity of Poland and Lithuania may suggest a possible migration between our populations. This context presents us with an interesting scientific question and provides further reasoning for collaboration on the study on “Overgrowth syndromes”. The project activities will strengthen international collaboration between the partners and provide a strong basis to develop further collaborative population studies in the future.