An upsurge in genetic research and genetic counseling in the last two decades has raised hopes and caused anxiety alike in researchers, bioethicists, biomedical practitioners, politicians and last but not least society in general. Along with genetic research and new genetic technologies new discourses and practices emerged that have been identified and analyzed by anthropologists and social scientists. Nonetheless, there still are some “uncharted” and “underdeveloped” domains such as rare, “orphan” diseases that need scrutinizing. Rare diseases are characterized by a low prevalence in the population (no more than 5 in 10 000 in the European Union). It is estimated that they may affect 6-8% of the population; according to the Polish Ministry of Health around 2,3-3 million individuals may be afflicted with rare disorders. Rare diseases are often severe, chronic, disabling, and possibly lethal conditions that constitute challenge to patients, their families, and the healthcare system.

This project focuses on rare genetic diseases in general and LCHAD deficiency (Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase deficiency) from the perspective of medical anthropology. First described in 1989, LCHAD deficiency is a rare, albeit life-threatening inherited metabolic disorder. It may lead to sudden death in infancy, severe handicap, and blindness. The disease seems to be extremely rare in Australia and Northern America; it is however relatively frequent in Europe, around the Baltic Sea in particular. The project aims at comparing two regions that have been singled out by genetic researchers due to their biological similarity, i.e. high frequency of LCHAD deficiency in population: (1) Pomerania region in Poland and (2) southern Finland. By utilizing ethnographic methods (participant observation and interviews in particular) and by emphasizing comparative perspective inherent to anthropology, the project will illuminate experiences of patients and/or families with children with rare diseases (especially LCHAD deficiency) in a broader socio-political and healthcare context. Simultaneously, it will analyze biomedical and genetic approaches to rare diseases that tend to essentialize ethnicity and bypass, among others, socio-cultural differences.

Despite a recent upsurge in anthropological studies of the cultural embeddedness of genes, there are no anthropological and social science studies that analyze experiences of patients (and their families) living with rare disorders such as LCHAD deficiency and/or their entanglements with physicians and healthcare system(s) in the Baltic region in general and Poland or Finland in particular. Thus, this project would not only scrutinize an underdeveloped research domain but also introduce data from and analysis of this region into the international body of anthropological scholarship. The results of this project could facilitate a better understanding of rare diseases in a broader society as well as their cultural, socio-political, and ethical impact among the body of medicine practitioners.