Pathogenesis of mucopolysaccharidosis type IIIC (MPS IIIC) and Gaucher disease has not been yet fully clarified, and the causes of phenotypical variability between the patients with the same genotype in Gaucher disease remain obscure. Because the variants in the regulatory regions of genes can cause phenotypical differences mentioned above, I have studied promoter regions of \textit{HGSNAT} and \textit{GBA} genes mutated in these lysosomal disorders. I have shown that there is an alternative promoter of \textit{GBA} (P2). Additional studies were aimed to elucidate possible physiological functions of P2, and its possible role in the pathogenesis of Gaucher disease. I have found that P2 is not tissue specific, and that its variants do not influence the variability of phenotype in Gaucher patients with the same genotype. P2 is used differentially neither during the differentiation of monocytes to macrophages nor in macrophages from controls and Gaucher patients, in whom there is a prominent storage only in cells of macrophage origin. We have thus not found any changes that would suggest a role for P2 in the pathogenesis of Gaucher disease.

I have characterized the promoter region of \textit{HGSNAT} and shown that the binding of Sp1 transcription factor is important for its expression. Sequence variants found in \textit{HGSNAT} promoter in patients did not influence its expression. Both promoters have features common with the majority of housekeeping gene promoters, as they do not contain TATA box, have multiple transcription initiation sites, contain an unmethylated CpG island and have multiple binding sites for transcription factor Sp1. They share the same features with many promoters of genes encoding other lysosomal enzymes.

I have studied the MPS IIIC mouse model and detected increased autophagy in the brains of HGSNAT deficient mice, which apparently plays a role in the pathogenesis of MPS IIIC.

I have also participated in the optimization of a method for isolation of lysosomal membranes, which will be used for biochemical study of N-acetyltransferase, the enzyme deficient in MPS IIIC, which has not been completely characterized.