Autism affects up to 1/150 children and represents therefore a serious social problem. It is a complex disorder with a clearly documented genetic component, but so far unexplained aetiology, which is currently a subject of intensive research. In the field of genetics various gene and chromosome defects are examined, as well as other mechanisms, including epigenetics, which could play a role in pathogenesis.

In our work we tried to replicate the finding of association between the ADA*2 risk allele and autism in a sample of 385 Czech children. Our sample was larger than those originally published. We also focused on individual endophenotypes (types of autism, degree of mental retardation and co-morbidity). Our results did not confirm the association of autism with the ADA*2 allele in the complete sample or in any of the subsets.

Chromosomal changes represent another finding in autistic patients. We performed the analysis of a ring chromosome 17 and a chromosome X deletion in two patients. Our studies represent an up to now unimaginable link between classical cytogenetics and molecular genetics at the DNA sequence level. As the first in the world, we described the structure of a human ring chromosome. Characterization of the defects allowed us to speculate on the impact of the genes involved in the phenotype of our patients. Both of these pilot analyses focused on rather larger aberrations affecting dozens of genes, which did not allow us to show causal links with individual genes. However, the analyses revealed a huge and unexpected complexity of both cases, with involvement of mosaicism or biased X-inactivation. Our results also allow us to speculate on the mechanisms of the aberrations.