Monogenic conditions are based on a single gene mutation and therefore a „rare“ allele formation. Such a mutation leads to a phenotypic abnormity with autosomal dominant, autosomal recessive, or gonosmal mode of inheritance. According to mutation severity, monogenic disorders manifest either in infancy or later in life. Phenotype of the disease is caused by a total or partial absence of the gene transcript (protein product). The protein product may represent either a structural molecule or an enzyme or receptor involved in regulation of physiological processes in organism and maintenance of homeostasis. In addition, the protein may act as a signal molecule or transcription factor regulating adequate organ development and function in embryogenesis or also later in life. Monogenic conditions represent a substantial number of paediatric endocrine diseases. Exact recognition of their etiopathogenesis allows understanding of the physiological processes in human body. The phenotype-genotype correlation supports to elucidate the complex physiology of endocrine regulations.

The first part is devoted to the transcription factor PROP1 which regulates embryonic differentiation of anterior pituitary. We present a Czech study investigating the frequency of PROP1 gene mutations and its functional effect on hormonal status and pituitary morphology during the life-span. The author personally conducted the laboratory work (molecular genetic analysis) and a phenotype assessment (evaluation of MRI scans).

The second topic is addressed to the physiology and patophysiology of calcium-sensing receptor regulating the calcium metabolism. We present a case report of a baby boy with severe form of neonatal hyperparathyreoidism due to heterozygous missense mutation in CASR gene. The author personally performed the molecular genetic analysis of the calcium-sensing receptor and participated in diagnosis and treatment of the patient.