

Abstract

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Title: Examination of MPL in myeloproliferative diseases

This bachelor thesis is focused on the myeloproliferative diseases related to mutations in the MPL gene. By identifying a specific mutation, it is possible to determine the diagnosis of patients with myeloproliferative diseases and thus set appropriate treatment. This thesis, therefore, aimed to introduce new diagnostic procedure at the Department of Molecular Genetics at the Regional Hospital Liberec.

The aims of the work:

The first objective was to get acquainted with the myeloproliferative diseases and their essential characteristics. The second objective was to study the role of mutations in the MPL gene in myeloproliferative diseases. The main aim was then to introduce a method for the determination of MPL gene mutations at the Department of Molecular Genetics at the Regional Hospital Liberec.

Methods:

The methodological part describes the protocol that I established for the diagnosis of mutations in the MPL gene:

1. Isolation of granulocytes from whole non-coagulated blood by gradient centrifugation
2. DNA column isolation
3. Real-time PCR using hybridization FRET probes

Conclusion:

The bachelor thesis summarizes the role of the MPL gene mutations in myeloproliferative diseases and the methods for their detection. The protocol for the

determination of mutations in the MPL gene was optimized and successfully implemented into laboratory praxis in the hospital.

Key words: MPL gene, MPL mutations, hematopoiesis, thrombopoietin, thrombopoietin receptor, myeloproliferative neoplasms, real-time PCR