ABSTRACT

This bachelor thesis deals with the topic of congenital anomalies. The sorting of the congenital anomalies is discussed in the first two chapters. The third chapter concerns the mutations of the short arm of chromosome 17. I have chosen to describe the Smith – Magenis syndrome, which is caused by the deletion of 17p11.2, and the Potocki – Lupski syndrome, which is its reciprocal duplication. The gene *RAI1* is specified in this part. The other topic mentioned in this thesis are RASopathies. The research of this section deals with the Noonan syndrome and focuses on mutations in genes *PTPN11*, *SOS1*, *RAF1*, *KRAS* and *NRAS*. In the fifth chapter the issue of thalidomide, its history and the mechanisms of action discovered up to now are described. The critical period and the mechanisms of limb reduction defects related to the timing of exposure are also defined here. New methods and findings related to these topics are discussed in the end of this work.

Key words: congenital anomalies, teratology, ontogenesis, Potocki – Lupski syndrome, Smith – Magenis syndrome, RASopathies, Noonan syndrome, thalidomide