Newborn screening for cystic fibrosis allows diagnosing patients with cystic fibrosis during asymptomatic stage of their disease or when the symptoms had not fully developed. Due to early diagnosis, patients with cystic fibrosis have the possibility to be treated prior to the occurrence of irreversible changes in the relevant organs, which leads to significantly improved quality of life and patient survival.
Commented version of the doctoral thesis presents issues concerning the selection of a suitable newborn screening programme for cystic fibrosis in neonates born in the Czech Republic and establishes requirements for particular analytical and molecular genetics tiers in the tested screening schemes.
The aim of this thesis is to nominate newborn screening protocol for cystic fibrosis that leads to optimal parameters in terms of its high sensitivity and specificity, including acceptable costs in the conditions of the Czech Republic health care system.