SUMMARY

INTRODUCTION: Newborn laboratory screening is a process used for early detection and treatment of selected rare diseases which leads to improvement in patient quality of life. All diseases included in newborn laboratory screening are classified as rare diseases, defined by a population frequency less than 1:2 000. The evaluation of newborn laboratory screening is an important tool for its improvements. The main aim of this doctoral thesis was to evaluate clinical and population-wide efficacy and balance detection rate and impact on healthy part of population.

METHODS: The doctoral thesis was based on results from screening laboratories in period 2002–2017 in the Czech Republic. Dried blood spots from newborns were analyzed using fluorescence *immuno*-assay, tandem mass spectrometry and fluorimetry.

RESULTS: The outcomes of this doctoral thesis led (1) to objectify prevalence of rare diseases in the Czech Republic, (2) to objectify association between prevalence of screened diseases and newborn birthweight, (3) to propose the change of decision limits of screening of 21-hydroxylase deficiency with aim to decrease high false positivity and negative impact on health part of population, (4) to define recommendations for managing of patients screened as positive in the 21-hydroxylase deficiency newborn laboratory screening.

CONCLUSION: Newborn laboratory screening in the Czech Republic detects patients with rare diseases in the early preclinical stages and the level of this system corresponds with the standard used by many states of the European Union. The above mentioned outcomes will be used in laboratory and clinical practice.