

## Abstract

Rare diseases, as their name indicates, individually affect only a low number of people around the world. Due to their low prevalence, finding appropriate therapy is very difficult. Insufficient understanding of the molecular causalities and mechanisms accompanying these disorders and the inability to conduct clinical studies to the usual extent due to the low occurrence of rare diseases belong to the main problems hindering the development of proper treatment. The creation of mouse models is a promising way to solve these difficulties since mice have numerous qualities necessary for modelling human diseases. CRISPR/Cas9 enables scientists to make precise changes in the genome by employing Cas9 nuclease which creates double-strand breaks in the DNA after the specifically designed guide RNA leads it to the site of interest. If the sequence of the chosen site is known, the possible edits can be installed almost anywhere in the genome and, moreover, their repertoire is practically endless. The use of the CRISPR/Cas9 technology proved to be perfect for creating mouse models of rare diseases as most of these disorders are caused by genetic mutations that this method is fully capable of mimicking. This thesis focuses on strategies used in creating such mouse models with the CRISPR/Cas9 system and summarizes their detailed mechanisms.

**Keywords:** rare diseases, mouse model, CRISPR/Cas9, knock-out, conditional knock-out, knock-in, transgenic mice, base editing, prime editing