# Genetic causes and stem-cell-based therapeutic strategies in neuromuscular diseases

Propositions belonging to the thesis

# Genetic causes and stem-cell-based therapeutic strategies in neuromuscular diseases

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1. Whole exome sequencing (WES) is the best strategy to identify genetic defects in heterogeneous neuromuscular diseases. (This thesis)
2. The complementation assay with wild-type SLIRP confirmed the pathogenicity of SLIRP mutations identified in our patient with mitochondrial encephalomyopathy. (This thesis)
3. Variation in cellular phenotype exists among mesoangioblasts of myotonic dystrophy type 1 (DM1) patients with comparable repeat expansions. (This thesis)
4. CRISPR/Cas9-based genome editing of DM1 mesoangioblasts is the first-choice strategy to generate corrected mesoangioblasts for treatment. (This thesis)
5. CRISPR/Cas9-edited mesoangioblasts have great potential to be further developed into a stem-cell-based medicine to treat muscle pathology. (Impact paragraph)
6. Exome data should not be limited to identify primary genetic defects, but should also be evaluated to search for actionable, health-related information.
7. The ability to cut DNA where you want has revolutionized the life sciences. --- Pernilla Wittung Stafshede
8. Ethical dilemmas must be dealt with before the patients will benefit from genome editing.
9. Sometimes the darkest challenges, the most difficult lessons, hold the greatest gems of light. --- Barbara Marciniak
10. 吾生也有涯，而知也无涯---庄子 (There is no destination for the pursuit of knowledge in our finite lifetime---Zhuangzi).
11. 千里之行，始于足下---老子 (A journey of a thousand miles begins with a single step---Laozi).