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Writing Assignment 3: Citing a Primary Article

In researching the uses of gene therapy and the mechanisms for delivery I stumbled on a study that highlights the real-world consequences of these tools wherein children with a debilitating and fatal genetic defect (Spinal Muscular atrophy type 1) were given a second chance at life via an infusion of an adeno-associated virus that carried the instructions for producing survival motor neuron (SMN) protein (1).

With that in mind I think this type of directed gene therapy will prove to be a tool of enormous importance going forward to deal with other similar genetic diseases which affect our loved ones. My own mother-in-law suffers from a type of muscular dystrophy that has confined her to a wheelchair for the rest of her life and the idea that we can start to reverse some of these maladies is part of the incentive for focusing on the biological sciences. Currently there isn’t any long-term effective treatments for muscular dystrophy since this is ultimately a genetic disorder rather than an acquired illness. However, with muscular dystrophy (Specifically Duchenne muscular dystrophy) in mind, we can see a promising study (2) where CRISPR was leveraged to help reframe exon 51 and deletion of exon 44 that in turn provided about 90% restoration of dystrophin protein expression in all muscles and the heart of affected mice. The long term benefits for such a study and treatment could lead to a reversal of an otherwise cruel genetic abnormality.

Works Cited

1. Mendell, J. M.D. et al. Single-Dose Gene-Replacement Therapy for Spinal Muscular Atrophy. *N Engl J Med,* 10.1056/NEJMoa1706198 (November 2nd 2017)
2. [Min](https://www.science.org/doi/10.1126/sciadv.aav4324#con1), Y. et al. CRISPR-Cas9 corrects Duchenne muscular dystrophy exon 44 deletion mutations in mice and human cells. *Science Advances,* https://doi.org/10.1126%2Fsciadv.aav4324 (March 6th 2019)