Lay Summary

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DNA/RNA instability in spinal muscular atrophy

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Spinal muscular atrophy (SMA) is severely debilitating and ultimately life-limiting conditions that selectively affect motor neurons, the nerve cells that control our voluntary muscles. People affected by motor neuron diseases lose the ability to walk, move, talk, and finally breathe. Currently, there is no cure or therapy to treat these devastating diseases effectively, as the precise mechanisms that cause motor neuron diseases are still poorly understood. The aim of our research therefore is to identify key disease mechanisms and targets to accelerate the development of much-needed treatments for motor neuron diseases.

This research project is based on our compelling results that links nerve cell injury to damage in the molecule that carries the genetic information (DNA) in all the cells. The studies proposed will use a multidisciplinary approach to explore the exact role of the gene causing Spinal muscular atrophy in DNA damage. Using experimental models and human tissue, these studies should create significant insights into the mechanisms behind motor neuron injury and identify key targets and new strategies for the development of effective treatments for motor neuron diseases. Moreover, these insights should also benefit and improve outcomes for other related and currently untreatable disorders caused by the damage and loss of nerve cells.