Immune System Dysfunction in Spinal Muscular Atrophy

- Principal Investigator: Rashmi Kothary
- Awarded $765,000 from the Canadian Institutes of Health Research (CIHR) in January 2018

SMA is a motor neuron disease leading to muscle wasting, and difficulties in controlling movement. This disease is inherited. There has been major progress in identifying the responsible gene, and in generating model systems. Recent work has highlighted defects in non-neuronal cells and tissues in the context of SMA. We now have strong evidence indicating immune system abnormalities in multiple mouse models of the disease. Therefore, in this study we will conduct an investigation to examine immune system function in both mouse and human. Our findings will shed light into the importance of immune dysfunction in SMA pathogenesis and disease burden.

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