

SMA Research Update – Providing Hope!

Biomaterial Collection and Banking for Translational Research in Spinal Muscular Atrophy

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SMA is an often severely disabling neuromuscular disease primarily affecting children. It causes muscle atrophy and weakness as well as frequent pulmonary, nutritional, and orthopedic complications. Many children with SMA eventually undergo surgery, most commonly for placement of feeding tubes and for scoliosis repair. These surgical procedures provide an opportunity to collect and bank small amounts of muscle and skin tissue to be used for research in SMA and related disorders. Similarly, children with disease other than SMA may undergo similar surgical procedures, providing an opportunity to collect muscle and skin samples from non-SMA control subjects. SMA is caused by a defect in the "Survival of Motor Neurons" (SMN1) gene. Researchers are hopeful to find a cure, because nature has provided humans with a second gene, almost a copy of the SMN1 gene. Normally, the second gene does not contribute much, but researchers think that its function can be increased by medications.

To learn more about the steps that lead to muscle weakness in SMA, skin and muscle samples are helpful to researchers working in the laboratory. This research aims to better understand the disease and to find and test potential treatments first in the laboratory and then ultimately in patients through clinical trials. Collecting samples from SMA and control subjects who undergo surgeries for clinical reasons provides an opportunity to gather important material for SMA research without more than minimal additional risk for the research participant undergoing surgery.

If you are interested in participating, please contact Nicole Holuba LaMarca at nh2282@columbia.edu or 212-305-5205