MEDICAL BREAKTHROUGHS
RESEARCH SUMMARY

TOPIC: A LITTLE COWBOY SAVED BY GROUNDBREAKING GENE REPLACEMENT THERAPY
REPORT: **MB #4814**

BACKGROUND: Spinal muscular atrophy, or SMA, is a genetic disease. SMA affects the part of the nervous system that controls voluntary muscle movement. Most of the nerve cells that control muscles are in the spinal cord. SMA is muscular because its primary effect is on muscles which don't receive signals from these nerve cells. Atrophy is what happens to muscles when they are not active. It is also the medical term for reduction of the muscles. SMA involves the loss of nerve cells called motor neurons in the spinal cord and is classified as a motor neuron disease. It can have a great deal of variation in the scope and severity in different people.
(Source: https://www.mda.org/sites/default/files/publications/Facts_SMA_P-181.pdf)

GENE THERAPY: Gene therapy involves altering the genes inside your body's cells to treat or stop disease. Genes contain your DNA which is the code that controls much of your body's form and function, from making you grow taller to regulating your body systems. Therefore, genes that don't work properly can cause disease. What gene therapy attempts to do is replace a faulty gene or add a new gene to cure disease or improve the body's ability to fight disease. It has been used in treating a wide range of diseases, such as cancer, cystic fibrosis, heart disease, diabetes, hemophilia, and AIDS. There are risks when it comes to this treatment. A gene can't easily be inserted directly into your cells, so it usually has to be delivered using a carrier called a vector. The most common vectors are viruses because they can recognize certain cells and carry genetic material into the cells' genes. Researchers remove the original disease-causing genes from the viruses, replacing them with the genes needed to stop disease.
(Source: https://www.mayoclinic.org/tests-procedures/gene-therapy/about/pac-20384619)

NEW TREATMENT OPTION FOR SMA: Gene replacement therapy for SMA is called onasemnogene abeparvovec-xioi (brand name Zolgensma). Zolgensma is a new, working copy of a human SMN gene, and makes up for the missing or nonworking survival motor neuron 1 (SMN1) gene, which helps motor neurons work properly. The new gene tells motor neuron cells to produce more survival motor neuron (SMN) protein, which motor neuron cells need to survive and support muscle functions. This therapy is given as a one-time infusion into the vein using an IV and runs for 60 minutes. The child's vital signs will be checked every 15 minutes and then hourly for two hours following the infusion. Clinical trials for SMA gene therapy have shown clear efficacy in young children with SMA type 1, resulting in a decreased need for respiratory support as well as improvement in motor skills. Research trials have also shown that the earlier children receive gene therapy for SMA, the better the results.
(Source: https://www.chop.edu/treatments/gene-therapy-spinal-muscular-atrophy-sma)

FOR MORE INFORMATION ON THIS REPORT, PLEASE CONTACT:

JULIE KIEFER
JULIE.KIEFER@HSC.UTAH.EDU

If this story or any other Ivanhoe story has impacted your life or prompted you or someone you know to seek or change treatments, please let us know by contacting Marjorie Bekaert Thomas at mthomas@ivanhoe.com