



FOR IMMEDIATE RELEASE

Promising Pre-clinical Results for Muscle Disorder That Took Life of NFL Player's Son *Joshua Frase Foundation Savors Breakthrough After 17-year Crusade*

PONTE VEDRA BEACH, FL. (January 23, 2014) – It was three years ago Christmas Eve that Paul and Alison Frase were forced to say goodbye to their 15-year-old son, Joshua, but the Frases and the foundation that bears their son's name finally have good reason to rejoice: promising pre-clinical data results from a five-year study on myotubular myopathy (MTM) – the muscle disorder that took Joshua's life. Before now, there has been no hope for survival for these children with MTM. And while this groundbreaking advancement comes too late to spare the Frases from tragedy, they are proud to know their commitment to the cause may, in fact, one day spare others the same pain.

"My son's life was a struggle between life and death, and ours was a fight to keep him alive. This journey started with a mother's vision of hope, and has turned into a global collaborative effort that will one day potentially save the lives of many, just like our Joshua ." said Alison Rockett Frase. "We are one step closer to realizing that dream."

This week, researchers released a study showing that gene therapy can improve muscle function in pre-clinical trials. Alan Beggs, Ph.D. of Boston Children's, Martin Childers, D.O., Ph.D., of University of Washington and Anna Buj-Bello, Ph.D., of Genethon were co-principal investigators on the study. Having isolated the cause of this disorder, scientists utilized an innovative form of gene replacement therapy in two animal models using a harmless virus serving as a vehicle to carry a healthy copy of the gene. The results were remarkable.

In the most recent stage of this research, scientists attempted to address symptoms in large animals born with the disease. A dog-named "Nibs" became a glimmer of hope for this desperate community. Nibs was found to be a rare large-animal-carrier of MTM, resulting in early deaths for her affected pups. To show the treatment was successful, three of Nibs' lineage – all suffering from MTM – were given one intravascular injection of this unique gene replacement therapy. With just one dose, their strength doubled after three weeks. By the end of six weeks, muscle strength in these "affected" dogs was entirely normal.

The Joshua Frase Foundation (JFF) was a major funding source for this research early on when there was very little support in the US. The initial research team was organized at Boston Children's Hospital in 1997, where the journey began for the Frases. The work has since grown to include the groups in Seattle and France (where much of the seminal work on MTM and gene therapy began) as well as many other laboratories around the world. To help support this, Alison

and Paul have used the NFL as a platform to raise money and awareness for MTM; Paul played in the NFL for 11 years.

“Our dream has been that one day all the Joshuas of the world will take our scientists by the hand and say thank you.” said Paul Frase, President of JFF.

For more information on this groundbreaking research, you may find the manuscript here: [January 22, 2014 issue of *Science Translational Medicine*](#). You may find a video about the research here: <http://www.youtube.com/watch?v=kBPYSNIJOXc>

About The Joshua Frase Foundation

The Joshua Frase Foundation is an organization crusading to find a cure for myotubular myopathy and other congenital myopathies while supporting affected families that struggle from the day-to-day existence of caring for these critical children. The foundation was established in 1996 by Paul and Alison Frase whose son, Joshua was born with MTM. At the age of 15, Joshua lost his life-long battle with MTM, but his legacy lives on in the promising search for a cure. To learn more about MTM research and to invest in saving the lives of many, please visit www.joshuafrase.org.

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