Eddy Padgett
01139031

Writing Assignment #4

This article is a peer-reviewed, primary source article implemented in a scientific journal. It implemented a hypothesis with first hand methods and results sourced from the original authors. This article relates to genetics as it carries out gene replacement therapy for patients compromised with spinal muscular atrophy.

Spinal muscular atrophy type 1 is a progressive, monogenic motor neuron disease with onset occurring during infancy that concludes with death or diminished motor skills. This experiment involved the study of a replacement of a mutated gene encoding survival motor neuron 1 in the disease. Of the 15 patients treated, three received a low dose therapy while the other 12 received a high dose. Primary outcome was safety while the secondary outcome was time until death or needed permanent ventilation.

Results for all 15 patients were a success and alive at 20 months of age as compared to the 8% survival rate. In the high dose party, rapid increase in baseline score following gene therapy, increased a score of 9.8 points at 1 month then 15.4 months after 3 months. A total of 11 of 12 patients were able to sit unassisted and achieved head control, while only 9 could roll over, and 2 were able to crawl, stand and walk. 11 patients were able to speak.

Patients with SMA1 (spinal muscular atrophy type 1), a single intravenous infusion of viral vector containing gene therapy properties, resulted in longer survival, improved motor function, and achievements of motor milestones.

Citation

Mendell, Jerry R. M.D., et al. Single-Dose Gene-Replacement Therapy for Spinal Muscular Atrophy. New England Journal of Medicine 377, 1713-1722 (2017).