Weill Cornell Medicine

Department of Genetic Medicine

Gene Therapy for the Cardiomyopathy of Friedreich's Ataxia

The Department of Genetic Medicine at Weill Cornell Medical College is initiating a Food and Drug Administration approved clinical study of gene therapy to treat the cardiac dysfunctions associated with Friedreich's ataxia. Friedreich's ataxia is caused by mutations in the frataxin (FXN) gene, resulting in decreased levels of FXN in the nervous system and the heart. While the nervous system manifestations are debilitating, the cardiac disease is the major cause of mortality. To treat the cardiac disease, the therapy involves a single, intravenous administration of a modified adeno-associated virus that delivers the normal FXN gene to the heart.

The goal of this initial clinical study is to assess safety and initial estimates of efficacy in treating the cardiac dysfunction. Potential participants will be screened for eligibility with assessment of cardiac and neurologic status. Eligible individuals who participate in the gene therapy study will be assessed periodically over 5 years with studies to assess the safety of the therapy and parameters to evaluate cardiac function. This clinical trial is funded by the National Heart, Lung, and Blood Institute at no cost to you. Travel and accommodation expenses will be compensated.

Study Involvement:

The study is divided into three periods:

1. Initial examination
2. Study drug administration
3. Follow-up.

The initial examination, or "Screening/Baseline Visit" will take place over several days and includes a series of tests to determine your eligibility to receive the study drug. All tests, except for the blood tests related to the study drug and future research, are standard medical tests and procedures.

If, after completing the initial examination, you wish to continue with the study and are eligible to do so, you will be scheduled to receive the study drug. You will initially repeat some of the tests and procedures from the initial examination and begin immunosuppression therapy with prednisone. The immunosuppression therapy reduces the risk that your immune system may recognize the study drug as foreign and try to remove it. You will be given oral prednisone for 14 weeks after receiving the study drug.

A study physician or physician assistant will administer the study drug intravenously by inserting a catheter into a vein in your arm. The study drug will be infused over 1 hour through the catheter using an infusion pump. After infusion, you will be observed for 2 hours. You will then be admitted overnight to New York Presbyterian Hospital (NYPH)/Weill Cornell Medicine (WCM) for observation to monitor any potential complications or side effects of the study drug. The following day after infusion, the catheter will be removed. If everything is stable, you will be discharged the day after the therapy.

For safety purposes, we will ask you to stay at the Hemsley Hotel on the WCM campus for the first week after dosing. You will be asked to come back to WCM at days 1-4, 7, 10, and 14 after the drug infusion to repeat some of the tests and procedures from the initial examination. Visits at weeks 4 -10 after drug infusion may be done remotely via video teleconference, thereby lessening travel.

You will be asked to come back to WCM at 1, 3, 6 and 12 months following the infusion of the study drug. Additional blood and urine tests will be assessed at 5, 6 and 10 weeks at your city of residence. For your safety and effectiveness of the study drug, you will receive the study procedures as described in the Visit Schedule below. After you complete the 12-month on-site visit, you will have an additional assessment at WCM in year 2 at 3, 6 and 12 months, and years 3-5 once yearly.

If you are interested in participation and would like to learn more about the study, please contact the Department of Genetic Medicine at (646) 962-2672 or via email at cora@med.cornell.edu.