Protocol Registration Receipt
12/17/2010

Mechanisms of Mitochondrial Defects in Gulf War Syndrome

This study is currently recruiting participants.
Verified by Medical Neurogenetics, LLC, December 2010

Sponsor:	Medical Neurogenetics, LLC
Collaborators:	
Information provided by:	Medical Neurogenetics, LLC
ClinicalTrials.gov Identifier:	

Purpose

The purpose of the study is to investigate possible causes for Gulf War Syndrome. Gulf War Syndrome is associated with increased incidences of amyotrophic lateral sclerosis (Lou Gehrig's Disease), pain syndromes, muscle complaints that include fatigue and myalgias (muscle pain), as well as other neurological symptoms. Abnormalities in the part of the cell known as mitochondria have been delineated in Gulf War Syndrome. Mitochondria are the "power plants" of the body. Mitochondria take the food you eat and break the food down into a form of energy that the body can use. We propose that Gulf War Syndrome is determined by a complex interaction of factors that interfere with mitochondrial function. This study will be the first investigation of mitochondrial function in Gulf War Syndrome. Our objective is to establish the cause for symptoms in affected veterans, develop testing that can more easily identify Gulf War Syndrome, and ultimately develop treatment protocols for Gulf War Syndrome.

Condition	Intervention
Gulf War Syndrome Mitochondrial Disease	Procedure/Surgery: Skin biopsy Procedure/Surgery: Blood Collection

Study Type: Observational
Study Design: Case-Only, Cross-Sectional

Official Title: Mechanisms of Mitochondrial Defects in Gulf War Syndrome

Further study details as provided by Medical Neurogenetics, LLC:

Biospecimen Retention: Samples With DNA

whole blood and tissue

Primary Outcome Measure:

- Characterize mitochondrial cellular energetics in GWS patients [Time Frame: At enrollment] [Designated as safety issue: No]

  After collecting a skin and blood sample, mitochondrial cellular energetics in GWS patients will be characterized by: 1. high resolution respirometry of intact cells, 2. quantitative analysis of individual mitochondrial proteins, 3. analysis of intact OXPHOS enzyme complexes and supercomplexes, 4. in gel enzyme activity assessment of intact OXPHOS enzyme complexes and supercomplexes, 5. mtDNA copy number quantitation to assess for defects in regulation mtDNA replication and 6. cellular coenzyme Q10 quantitation.

Secondary Outcome Measures:

- Mitochondrial DNA [Time Frame: at enrollment] [Designated as safety issue: No]

  Assess the mitochondrial DNA (mtDNA) from each patient with GWS for mtDNA mutations by whole genome sequencing of leukocyte and skin cell mtDNA.

Estimated Enrollment: 30
Study Start Date: May 2009

Groups/Cohorts	Interventions
Gulf War Syndrome patients Gulf War veterans who have been diagnosed with Gulf War Syndrome.	Procedure/Surgery: Skin biopsy A small skin sample will be obtained from the patients arm which is approximately the size of the top of a thumbtack (a small circle no more than a 1/4 inch across) Procedure/Surgery: Blood Collection Approximately 45ml or 3 tablespoons for blood will be drawn from a vein in the patient's forearm.

Eligibility

Gulf War Veterans who have been diagnosed with Gulf War Syndrome

Sampling Method: Non-Probability Sample
Ages Eligible for Study: 18 Years and older
Genders Eligible for Study: Both

Inclusion Criteria:

- Short-term memory loss or a severe inability to concentrate that affects work, school or other normal activities
- Muscle Pain, myalgias
- Pain without redness or swelling in a number of joints
- Intense or changing patterns of headaches
- Unrefreshing sleep
- After any exertion, weariness that lasts for more than a day

Exclusion Criteria:

- Organ failure (e.g. emphysema, cirrhosis, cardiac failure, chronic renal failure)
- Chronic infections (e.g. HIV/AIDS, hepatitis B or C)
- Rheumatic and chronic inflammatory diseases (e.g. systemic lupus erythematosis, Sjogren's syndrome, rheumatoid arthritis, inflammatory bowel disease, chronic pancreatitis.)
- Major neurologic diseases (e.g. multiple sclerosis, neuromuscular diseases, epilepsy or other disease requiring ongoing medication that could cause fatigue, stroke, head injury with residual neurologic deficits)
- Diseases requiring systemic treatment (e.g. organ or bone marrow transplantation; systemic chemotherapy; radiation of brain, thorax, abdomen, or pelvis)
- Major endocrine diseases (e.g. hypopituitarism, adrenal insufficiency)
- Myocardial infarction, heart failure
- Morbid obesity (body mass index >40)
- Permanent psychiatric exclusions: Lifetime diagnoses of bipolar affective disorders, schizophrenia or any subtype, delusional disorders of any subtype, dementias of any subtype, organic brain disorders, and alcohol or substance abuse within 2 years before onset of the fatiguing illness.
- History of allergic reaction to lidocaine
- History of keloid formation with skin incisions.

Contacts and Locations

Contacts

Emily E Voigt, RN, BSN	678-597-5667	evoigt@mnglab.com
Maureen E Starnes, CPNP	678-225-0222	mstarnes@mnglab.com

Locations

United States, Georgia

Medical Neurogenetics, LLC Recruiting

Atlanta, Georgia, United States, 30338

Contact: Emily E Voigt, RN, BSN 678-597-5667 evoigt@mnglab.com

Principal Investigator: John M Shoffner, MD

Investigators

Principal Investigator:	John M Shoffner, MD	Medical Neurogenetics

More Information

Responsible Party:	Medical Neurogenetics (John M. Shoffner, MD)
Study ID Numbers:	H09378 GW080138 [Grant/Funding Number: Department of Defense CDMRP]
Health Authority:	United States: Federal Government