

FSH Society Research, Fellows and Small Grants Funded

Bronfman Class

Grant: FSHS-MB-001
Researcher: Silvere M. van der Maarel, Ph.D.
Institution: Leiden University Medical Center
Dept. of Human Genetics
Wassenaarseweg 72
PO Box 9503
2300 RA Leiden
The Netherlands

Project Title: "Generation of Transgenic Mouse Models for FSHD."

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|----------|-----------------------|--------|
| \$30,000 | 7/1/1998 - 6/30/1999 | Year 1 |
| \$30,000 | 7/1/1999 - 6/30/2000 | Year 2 |
| \$30,000 | 3/31/2001 - 2/28/2002 | Year 3 |

Goal: To initiate groundbreaking research to create FSHD animal models.

Grant: FSHS-MB-002
Researcher: Sara T. Winokur, Ph.D.
Institution: 240 D, Medical Sciences I
Department of Biological Chemistry
University of California
Irvine, CA 92697-1700 USA

Project Title: "Analysis of Chromatin Structure and Skeletal Muscle-Specific Gene Expression in Facioscapulohumeral Muscular Dystrophy."

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|----------|----------------------|--------|
| \$30,000 | 6/1/1998 - 5/31/1999 | Year 1 |
| \$30,000 | 6/1/1999 - 5/31/2000 | Year 2 |
| \$30,000 | 6/1/2000 - 5/31/2001 | Year 3 |

Goal: To initiate novel research using Genechip and gene expression technologies to gain insight into FSHD. Prior to this project, gene expression studies had never been done in FSHD.

Grant: FSHS-MB-003
Researcher: Denise Figlewicz, Ph.D.
Institution: University of Rochester School of Medicine
Department of Neurology
601 Elmwood Avenue
P.O. Box 673
Rochester, New York 14642 USA

Project Title: "Expression of genes proximal to the D4Z4 deletions: a quantitative study in FSHD patients and controls. "

\$30,000 1/1/1999 - 12/31/1999 Year 1

\$30,000 1/1/2000 - 12/31/2000 Year 2

Goal: To initiate novel research on gene expression and models to study gene expression in FSHD.

Grant: FSHS-MB-004
Researcher: David J. Picketts, Ph.D.
Institution: Ottawa General Hospital
Research Institute
501 Smyth Road
Ottawa, Ontario, K1H 8L6
Canada

Project Title: "Utilizing an epigenetic approach to identify the FSHD gene."

\$30,000 5/1/1999 - 4/30/2000 Year 1

\$30,000 5/1/2000 - 4/30/2001 Year 2

Goal: To initiate novel research on epigenetic features and hyper sensitive sites to understand chromosomal aspects and models of gene expression on FSHD. Prior to this project, no work had been done internationally on elucidating the epigenetic aspects of FSHD.

Grant: FSHS-MB-005
Researcher: Davide Gabellini, Ph.D.
Institution: University of Massachusetts Medical Center
Howard Hughes Medical Institute
373 Plantation Street
Worcester, MA 01605 USA

Project Title: "Identification and characterization of a protein interacting with the DNA repetitive element causally related to facioscapulohumeral muscular dystrophy."

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|----------|-----------------------|--------|
| \$30,000 | 1/1/2000 - 12/31/2000 | Year 1 |
| \$30,000 | 1/1/2001 - 12/31/2001 | Year 2 |
| \$30,000 | 1/1/2002 - 12/31/2002 | Year 3 |

Goal: To initiate novel research on gene expression/repression, disease models and to gain insight into D4Z4 functionality and role in FSHD. This work has led to several landmark publications on gene mis-regulation, gene silencing and repression complexes.

Grant: FSHS-MB-006
Researcher: Fern Tsien, Ph.D. / Melanie Ehrlich, Ph.D.
Institution: Tulane Cancer Center
Human Genetics/SL31
Tulane Medical School
1430 Tulane Avenue
New Orleans, LA 70112 USA

Project Title: "DNA Methylation and Chromatin Structure of FSHD-linked Sequences in FSHD Cells, Normal Cells, and Cells from Patients

with the ICF Syndrome."

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|----------|----------------------|--------|
| \$35,000 | 5/1/2001 - 4/30/2002 | Year 1 |
| \$35,000 | 5/1/2002 - 4/30/2003 | Year 2 |

Goal: To initiate novel research on the role of methylation, chromatin structure and other epigenetic features in FSHD.

Grant: FSHS-MB-007

Researcher: Tonnie Rijkers, Ph.D.

Institution: Leiden University Medical Center
Center for Human and Clinical Genetics
Wassenaarseweg 72
PO Box 9503
2300 RA Leiden
The Netherlands

Project Title: "Mouse models to study candidate genes and epigenetic causes of FSHD."
\$30,000 2/1/2003 – 1/31/2004 Year 1
\$30,000 2/1/2004 – 1/31/2005 Year 2

Goal: To initiate research on genotype/phenotype correlations in successfully created new lines of animal models of FSHD.

Grant: FSHS-MB-008

Researcher: Cecilia Ostlund, Ph.D. / Howard Worman, Ph.D.

Institution: Columbia University
Departments of Medicine and Anatomy and Cell Biology
P & S 10-518
630 W 168th St
New York, NY 10032 USA

Project Title: "The role of DUX4 in facioscapulohumeral muscular dystrophy."
\$30,000 2/1/2003 – 1/31/2004 Year 1
\$30,000 2/1/2004 – 1/31/2005 Year 2

Goal: To initiate research on the role of DUX4, DUX4C and to examine the role of the nuclear envelope, nuclear lamina and nuclear organization in FSHD.

Grant: FSHS-MB-009

Researcher: Alberto Luis Rosa, M.D., Ph.D.

Institution(1): Washington State University -- Spokane
 WSU Spokane Health Science
 PO Box 1495
 Spokane, WA 99210-1495 USA

Institution(2): Laboratory of Neurogenetics
 Institute for Medical Research "Mercedes y Martín Ferreyra"
 INIMEC-CONICET, National Research Council of Argentina
 Friuli 2434,
 B Col. Velez Sarsfield,
 5016 – Córdoba, Argentina

Project Title: "Role of nuclear localization signal (NLS) and H1/H2 motifs in DUX4-mediated cell Death. "

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|----------|----------------------|--------|
| \$43,750 | 8/1/2004 – 7/31/2005 | Year 1 |
| \$14,690 | 8/1/2005 – 7/31/2006 | Year 2 |

Goal: To gain understanding on the molecular and cellular mechanism underlying the pathogenesis of human facioscapulohumeral muscular dystrophy (SHD). To study DUX4, a putative double homeobox-containing protein encoded by a 3.3 kb polymorphic tandem repeat (D4Z4), at the locus FSHD1A on the human chromosomal region 4Q35. It is hypothesized that abnormal temporal or spatial expression of DUX4 has a toxic effect for muscle cells causing FSHD. The study will help identify the mechanism(s) by which DUX4 causes cell death.

Delta Railroad Construction Class

Grant: FSHS-DR-001

Researcher: Alexandra Belayew, Ph.D. / Stephane Plaisance, Ph.D.

Institution: Lab. Biologie Moléculaire

Université de Mons-Hainaut

Pentagone, avenue du Champ de Mars 6

B - 7000 - Mons

BELGIUM

Project Title: "Characterization of a protein expressed from a 3.3 kb element not linked to FSHD."

\$15,000 6/1/1998 - 12/31/1998 Year 1

"Small laboratory equipment for research on FSHD."

\$15,000 2/15/2001 - open Year 1

Goal: To initiate research on the role of DUX, DUX1, DUX4, DUX4C and to elucidate the role of DUX in FSHD and within the D4Z4 region.

Grant: FSHS-DR-002

Researcher: Rossella Tupler, M.D., Ph.D.

Institution: Howard Hughes Medical Institute,

University of Massachusetts Medical Center,

Worcester, Massachusetts 01605 USA

Project Title: "Characterization of differentially expressed genes in facioscapulohumeral muscular dystrophy affected muscles."

\$30,000 6/1/1998 - 5/31/1999 Year 1

Goal: To initiate research into differentially expressed genes involved in FSHD. This groundbreaking research has led to major advances in our understanding of FSHD.

Grant: FSHS-DR-003
Researcher: Robert Bloch, Ph.D.
Institution: University of Maryland School of Medicine
660 W. Redwood St.
Baltimore, MD 21201 USA
Project Title: "Sarcolemmal organization in FSHD and the MYD mouse."
\$30,000 7/1/1999 - 4/30/2001 Year 1
(interrupt/extend)

Goal: To gain insight into structural aspects and patho-physiology of FSHD using the latest techniques as well as revisiting standard methodologies. To examine the structure of FSHD muscle and the sarcolemma for insights into the disease.

Grant: FSHS-DR-004
Researcher: Jane Hewitt, Ph.D.
Institution: Nottingham University
Division of Genetics
Queen's Medical Centre
Nottingham
NG7 2UH, England
Project Title: "Fugu rubripes as a model organism for FSHD gene identification."
\$30,000 7/1/2000 - 6/30/2001 Year 1

Goal: To sequence the analogous 4q35 region in puffer fish for insight into genomic organization of FSHD. To use data to help with mapping, assembly and finishing of the 4q35 human region. Based on this research, we were able to assist the Human Genome Project at Washington University to complete the map and sequence this very difficult and recalcitrant region of 4q35.

Grant: FSHS-DR-005

Researcher: Marcy Speer, Ph.D.

Institution: Duke University Medical Center
 Box 3445
 Durham, NC 27710 USA

Project Title: "Genetic Linkage Studies in Non-chromosome 4 FSHD."
 \$30,000 2/1/2002 - 1/31/2003 Year 2 (See Year 1
 under Tides)

Goal: To examine and find the genetic locus of the non-chromosome 4 families through genome wide search/scan. This project is a high priority for the research community. It aims to register non-chromosome 4 pedigrees with the researchers and clinicians at Duke. The FSH Society plays an important role in identifying such families for this project.

Grant: FSHS-DR-006A

Researcher: Emma Ciafaloni, M.D

Institution: University of Rochester School of Medicine
 Department of Neurology
 601 Elmwood Avenue
 P.O. Box 673
 Rochester, New York 14642 USA

Project Title: "The Course and Outcome of Pregnancy and Delivery in Women with FSH Muscular Dystrophy."
 \$13,074 1/1/2004 - 12/31/2004 Year 1
 (interrupt/extend)

\$1,926 1/1/2005 - 12/31/2005 Year 2 (\$12,973 total
 see under Lewis)

\$0 1/1/2006 - 12/31/2006 Year 3 (\$13,363 total
 see under Lewis)

Goal: Very little is known about the course and outcome of pregnancy and delivery in women with muscular dystrophies. Our current ability to efficiently counsel women with muscular dystrophies when pregnant or planning a pregnancy is very limited due to the lack of studies addressing the issue of pregnancy and delivery outcome in this group. No specific attention has been paid to the possible interaction between gestation and progression of the myopathy. Objectives are: to increase our knowledge about the course and outcome of pregnancy and delivery in women with FSH muscular dystrophy; to assess the effect of pregnancy, delivery and post-partum on the progression of muscle weakness and muscle pain and on quality of life in women with FSH muscular dystrophy; and, to ultimately improve counseling, family planning and obstetric management of women with FSH muscular dystrophy.

Grant: FSHS-DR-006B Honoraria

Researcher1: Wendy M. King, PT

Institution1: Ohio State University
389 McCampbell Hall
1581 Dodd Drive
Columbus, Ohio 43210-1205 USA

Researcher2: Shree Pandya, MS, PT

Institution2: University of Rochester School of Medicine
Physical Medicine and Rehabilitation
University of Rochester
Rochester, NY, 14642 USA

Project Title: "Facioscapulohumeral muscular dystrophy Physical Therapy Booklet/Brochure and Article for Physical Therapy Journal."

\$15,000 5/1/2004 – 4/30/2005 Year 1

Goal: Gather and review of literature/information related to FSHD natural history, surgical options, orthotics, rehabilitation, physical therapy interventions, role of exercise, hydrotherapy, pain, etc. Review scientific literature, brochures and web sites of various organizations from English speaking countries to assess the type and format of information already available. Draft, peer- review and publish booklet/brochure on FSHD and Physical Therapy and submit journal article to Physical Therapy journal on P.T. and FSHD.

FSH Society, Inc. Grants (Named)

FSH Society Tides Foundation

Grant: FSHS-TF-001

Researcher: Marcy Speer, Ph.D.

Institution: Duke University Medical Center
Box 3445
Durham, NC 27710 USA

Project Title: "Genetic Linkage Studies in Non-chromosome 4 FSHD."

\$30,000 2/1/2001 - 1/31/2002 Year 1 (See Year 2
under Delta RR)

Goal: To ensure that the work to examine and find the genetic locus of the non-chromosome 4 families continues. This project is of very high priority to the FSH Scientific Advisory Board.

FSH Society Vicki and Mark Ray

Grant: FSHS-VR-001

Researcher: Robert Bloch, Ph.D.

Institution: University of Maryland School of Medicine
660 W. Redwood St.
Baltimore, MD 21201 USA

Project 1 Title: "Sarcolemmal organization in FSHD and the MYD mouse."

\$15,000 5/1//2001 - 11/30/2001 Year 1

Project 2 Title: "To investigate the "proteome" in FSHD and to compare it to the "proteome" in control muscles and in other common myopathies and muscular dystrophies

using Two-dimensional gel electrophoresis"

\$15,000 2/15//2002 - 8/15/2002 Year 1

Goal: To gain insight into structural aspects and patho-physiology of FSHD using the latest techniques as well as revisiting standard methodologies. To newly examine the structure of FSHD muscle and the sarcolemma and to examine more closely the proteins involved in FSHD using proteomic approaches.

FSH Society Thelma Green Memorial
Grant: FSHS-TG-001

Researcher: Jeanne Lawrence, Ph.D. / Y. Polly Xing, M.D., Ph.D.

Institution: University of Massachusetts Medical Center
55 Lake Avenue North
Worcester, MA 01655 USA

Project Title: "Higher level chromatin packaging and nuclear organization of FSHD cell with an emphasis on its 3.3 kb deletion involving high

resolution transcript mapping by mRNA in situ and direct visualization of this region of the chromosome via In situ hybridization

with loop halo DNA preparations."

(interrupt/extend) \$30,000 1/1/2002 - 12/31/2002 Year 1

Goal: To gain insight into nuclear organization, scaffolding, structure and chromatin packaging involved in FSHD. To examine epigenetic features, nuclear compartmentalization and aspects of D4Z4 and telomere organization.

FSH Society Sam E. and Mary F. Roberts Foundation Grant for Nutrition Research

Grant: FSHS-SMRF-001

Researcher: Graham J Kemp, M.D.

Institution: Faculty of Medicine
University of Liverpool
Liverpool L69 3GA, UK

Project Title: "Muscle damage by reactive oxygen species, muscle atrophy and effects of creatine supplementation in facioscapulohumeral muscular dystrophy."

(interrupt/extend) \$35,000 1/1/2003 - 5/01/2005 Year 1.5

balance under Lewis) (\$48,650 total see

Goal: This a pilot study designed to test the following hypotheses: (1 that muscle in FSHD shows evidence of damage by ROS in vivo;

2) that this is at least partly due to reduced anti-ROS protection; 3) that this is ameliorated by 6 months creatine treatment; and,

4) that this also partially alleviates muscle atrophy, even in the absence of training, and;

5) that this results in an increase in muscle strength and clinical indices.

This is an open label pre-post protocol examining the effects of 6 months creatin supplementation in 10 patients with proven FSHD. ROS protection and damage will be studied in conchotome biopsies of deltoid. Muscle atrophy and its effect on body composition will be measured by whole-body quantitative magnetic resonance imaging (MRI). Muscle strength and effects on symptomatology will be be quantified.

We will compare pre-creatine results with those of control subjects, and examine differences between post- and pre-creatine values.

This study has several possible benefits: it will contribute evidence of the therapeutic usefulness of creatine over a longer time span than earlier studies; it will throw light on mechanisms of muscle damage in FSHD; if ROS are indeed important then other compounds that reduce oxidative stress in muscle may be useful; lastly, the results will help in the design and interpretation of future placebo-control trials.

Grant: FSHS-SMRF-002

Researcher: Sara Winokur, Ph.D. / Ulla Bengtsson, Ph.D.

Institution: 202 Sprague Hall
Biological Chemistry
University of California, Irvine
Irvine, CA 92697 USA

Project Title: "Restoration of normal myogenic pattern in FSHD: A nutritional approach."

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| \$30,000 | 3/1/2003 – 2/29/2004 | Year 1 |
| \$30,000 | 3/1/2004 – 2/28/2005 | Year 2 |

Goal: A clinically oriented project to study patterns of FSHD myogenesis in cell systems using compounds and nutritional agents that affect methylation, oxidative stress, chromatin structure and muscle cell differentiation. A major goal of this project is to an effective model system to assay target compounds effectively. The objective of this study is to identify therapeutic compounds to treat FSHD that can be taken as part of a nutritional regimen. Nutritional compounds are selected based on functional impact on myogenesis, availability as nutritional supplement and expediency for clinical trials.

FSH Society New York City Symphony and Song Benefit Concert
Grant: FSHS-NYSS-001

Researcher: Daniela M. Oliveira, Ph.D.

Institution: Ottawa Health Research Institute
501 Smyth Road
Ottawa, Ontario
Canada K1H 8L6

Project Title: "Identification of the mechanism regulating the Wnt-dependent activation of muscle progenitor cells."

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|----------|------------------------|--------|
| \$30,000 | 1/1/2005 – 12/31//2005 | Year 1 |
|----------|------------------------|--------|

Goal: The overall goal of the project is to identify genes regulated by the Wnt signaling pathway that are responsible for the myogenic differentiation and proliferation of CD45+/Sca-1+ muscle cells. In addition muscle satellite cells, another stem cell population within muscle (CD45+/Sca-1+ muscle cells) plays a physiological role in muscle regeneration. Identification of new therapeutic targets can be used to help stimulate the Wnt-target genes that might be used to enhance stem cell transplant in FSHD.

Grant: FSHS-NYSS-002

Researcher: York Marahrens, Ph.D. / Nieves Embade, Ph.D.

Institution: Department of HumAn Genetics
David Geffen School of Medicine
University of California, Los Angeles
Gonda Center, Room 4558
695 Charles E. Young Drive
Los Angeles, CA 90095 USA

Project Title: "Testing whether D4Z4 Perform Long Distance Gene Silencing via the Chromosome 4 Inactivation Network."

\$22,652

11/1/2004 – 10/31/2005

Year 1

Goal: A high risk and novel approach to test the hypothesis that long repetitive sequence on a chromosome, regardless of sequence, is tied into the network of long repeats responsible for chromosome inactivation and particular with FSHD the case of non-random mono-allelic autosomal inactivation. To test the hypothesis that the tract of D4Z4 repeats at 4q35 is tied into the chromosome 4 inactivation network and that D4Z4 deletions disturb chromosome 4 inactivation resulting in abnormal gene expression.

FSH Society Research & Education Fund

Grant: FSHS-FS-001

Researcher: Nieves Embade, Ph.D. / York Marahrens, Ph.D.

Institution: Department of Human Genetics

David Geffen School of Medicine

University of California, Los Angeles

Gonda Center, Room 4558

695 Charles E. Young Drive

Los Angeles, CA 90095 USA

Project Title: "Tethering Adenine (Dam) Methylase to the 3.3-kb FSHD Repeats to Identify Distant Genes that Physically Come in Contact with the Repeats."

\$30,000

3/1/2003 – 9/30/2004

Year 1

(interrupt/extend)

Goal: A high risk and novel approach to understanding chromosome interactions, epigenetics, gene expression in FSHD and with which other parts of the chromosome(s) the FSHD chromosome 4 D4Z4 repeats are coming into contact. To locate the FSHD gene(s) that interact with the D4Z4 repeats by tethering bacterial adenine methylase to sequences in or near the 3.3 kb repeats and then identifying adenine-methylation at distant sites on the same chromosome and/or other chromosomes.

FSH Society Lewis Family Research & Education Fund

Grant: FSHS-LEWI-001
Researcher: Graham J Kemp, M.D.
Institution: Faculty of Medicine
University of Liverpool
Liverpool L69 3GA, UK
Project Title: "Muscle damage by reactive oxygen species, muscle atrophy and effects of creatine supplementation in facioscapulohumeral muscular dystrophy."

\$13,650 1/1/2003 - 5/01/2005 Year 1.5
(interrupt/extend) (\$48,650 total see balance under Roberts)

Goal: This a pilot study designed to test the following hypotheses: (1 that muscle in FSHD shows evidence of damage by ROS in vivo;
2) that this is at least partly due to reduced anti-ROS protection; 3) that this is ameliorated by 6 months creatine treatment; and,
4) that this also partially alleviates muscle atrophy, even in the absence of training, and; 5) that this results in an increase in muscle strength and clinical indices. This is an open label pre-post protocol examining the effects of 6 months creatine supplementation in 10 patients with proven FSHD. ROS protection and damage will be studied in conchotome biopsies of deltoid. Muscle atrophy and its effect on body composition will be measured by whole-body quantitative magnetic resonance imaging (MRI). Muscle strength and effects on symptomatology will be be quantified. We will compare pre-creatine results with those of control subjects, and examine differences between post- and pre-creatine values.
This study has several possible benefits: it will contribute evidence of the therapeutic usefulness of creatine over a longer time span than earlier studies; it will throw light on mechanisms of muscle damage in FSHD; if ROS are indeed important then other compounds that reduce oxidative stress in muscle may be useful; lastly, the results will help in the design and interpretation of future placebo-control trials.

Grant: FSHS-LEWI-002

Researcher: Emma Ciafaloni, M.D

Institution: University of Rochester School of Medicine
 Department of Neurology
 601 Elmwood Avenue
 P.O. Box 673
 Rochester, New York 14642 USA

Project Title: Muscular Dystrophy.” “The Course and Outcome of Pregnancy and Delivery in Women with FSH

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|---------------------|----------|-----------------------|------------------------|
| see Delta Railroad) | \$0 | 1/1/2004 - 12/31/2004 | Year 1 (\$13,074 total |
| see Delta Railroad) | \$11,047 | 1/1/2005 - 12/31/2005 | Year 2 (\$12,973 total |
| | \$13,363 | 1/1/2006 - 12/31/2006 | Year 3 |

Goal: Very little is known about the course and outcome of pregnancy and delivery in women with muscular dystrophies. Our current ability to efficiently counsel women with muscular dystrophies when pregnant or planning a pregnancy is very limited due to the lack of studies addressing the issue of pregnancy and delivery outcome in this group. No specific attention has been paid to the possible interaction between gestation and progression of the myopathy. Objectives are: to increase our knowledge about the course and outcome of pregnancy and delivery in women with FSH muscular dystrophy; to assess the effect of pregnancy, delivery and post-partum on the progression of muscle weakness and muscle pain and on quality of life in women with FSH muscular dystrophy; and, to ultimately improve counseling, family planning and obstetric management of women with FSH muscular dystrophy

FSH Society, Inc. Small Grants

Grant: FSHS-SG-001

Researcher: Valery Kazakov, M.D., Ph.D.

Institution: Department of Neurology, Pavlov's Medical Institute, Lev Tolstoy str. 6/8,
197022, filial 1 St. Petersburg, Russia

Project Title: Travel Grant

\$952 4/1997

Goal: To assist with travel to the AAN/NIH/FSH Society International Research
Consortium Meeting

Grant: FSHS-SG-002

Researcher: Michio Hirano, M.D.

Institution: College of Physicians & Surgeons of Columbia University, P&S 4-443,
630 West 168th Street, 2nd Floor, Room 401, New

York, NY 10032 USA

Project Title: AAN Travel Fellowship Grant

\$1078.87 5/1997

Goal: To assist with travel to the AAN/NIH/FSH Society International Research
Consortium Meeting

Grant: FSHS-SG-003

Researcher: David Lacomis, M.D.

Institution: University of Pittsburg, Pittsburg, PA

Project Title: AAN Travel Fellowship Grant

\$1333.28 5/1997

Goal: To assist with travel to the AAN/NIH/FSH Society International Research
Consortium Meeting

Grant: FSHS-SG-004

Researcher: William Ted Brown, M.D., Ph.D.

Institution: New York Institute for Basic Research, Staten Island, NY USA

Project Title: Travel Grant

\$670.80 5/1997

Goal: To assist with travel to the Annual FSH Society International Research
Consortium Meeting. For opinion, estimate and consulting on establishing FSHD genetic testing in the
United States

Grant: FSHS-SG-005

Researcher: Rossella Tupler, M.D., Ph.D.

Institution: Howard Hughes Medical Institute, Program in Gene Function and Expression, University of Massachusetts Medical School, Lazare Research Building - 6th Floor - Room 660 A, 364 Plantation Street, Worcester, MA 01605 USA

Project Title: Travel Grant

\$233 10/1997

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-006

Researcher: Linda Surh, Ph.D.

Institution: Molecular Diagnostics Lab, Children's Hospital of Eastern Ontario, 401 Smyth Road, Room 3029, Ottawa, Ontario, Canada

K1H 8L1

Project Title: Equipment Grant

\$771.95 12/1997

Goal: To purchase needed equipment for pulse field gel electrophoresis required for accurate molecular genetics testing of FSHD.

Grant: FSHS-SG-007

Researcher: Meena Upadhyaya, Ph.D.

Institution: Institute of Medical Genetics, University of Wales, Heath Park, Cardiff, C214 4XN UK

Project Title: Travel Grant

\$1,346.14 7/1998

Goal: To assist with travel to the International Muscle Conference Australia

Grant: FSHS-SG-008

Researcher: Silvere van der Maarel, Ph.D.

Institution: Leiden University Medical Center, Dept. of Human Genetics,
Wassenaarseweg 72, PO Box 9503, 2300 RA Leiden, The Netherlands

Project Title: Travel Grant

\$845.24 11/1998

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-009

Researcher: Kevin Flanigan, M.D.

Institution: Eccles Institute of Genetics, Room 7290, University of Utah, 15 North 2030 East Street, Salt Lake City, Utah 84113 USA

Project Title: Small grant proposal for "QMA software/system and professional physical therapy resources to help with studies to answer definitively whether anticipation in disease severity and onset, gender effects, or parent-of origin effects exist in FSHD."

\$8,375 1/1/2000 – 9/30/2003 Year 1 (interrupt/extend)

Goal: To assist with data collection in clinical trials and patient acquisition at the University of Utah. To help University of Utah build a registry of special FSHD families (founder effect) in the region

Grant: FSHS-SG-010

Researcher: Yang (Ted) D. Teng, M.D., Ph.D.

Institution: Children's Hospital, 300 Longwood Avenue, Bader 3, Boston, MA 02115, USA

Project Title: Travel Grant

\$138.52 6/2000

Goal: To assist with travel to the NIH FSHD Symposium held at Bethesda, MD.

Grant: FSHS-SG-011

Researcher: Amy Csink, Ph.D.

Institution: Department of Biological Sciences, Carnegie Mellon University, 4400 Fifth Avenue, Pittsburgh, PA 15213 USA

Project Title: Travel Grant

\$840.70 10/2000

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG. Lecture on Apoptosis.

Grant: FSHS-SG-012

Researcher: Laura M. Palmucci, M.D., Ph.D.

Institution: Centro Malattie Neuromuscolari, Department of Neuroscience, University of Turin, via Cherasco 15, 10126, Turin, Italy

Project Title: Travel Grant

\$395.75 10/2000

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-013

Researcher: Peter Lunt, Ph.D.

Institution: Department Clinical Genetics, Institute of Child Health, Bristol Children's Hospital, St. Michael's Hill, Bristol, BS2 8FG England

Project Title: Travel Grant

\$776.93 12/2000

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-014

Researcher: Michel van Geel, Ph.D.

Institution: University Hospital Nijmegen, Department of Dermatology, Building M351, Room 26, PO-Box 9101, 6500 HB, (Rene Descartesdreef 1, 6525 GL), Nijmegen, The Netherlands

Project Title: Research Publication Grant "Support for doctoral thesis printing efforts."

\$2,500 7/2001

Goal: To facilitate printing and dissemination of research doctoral thesis book covering high level and detailed work in evolutionary aspects of FSHD and including the first notion of A/B allele in FSHD.

Grant: FSHS-SG-015

Researcher: Patrick Reed, Ph.D.

Institution: Department of Physiology, University of Maryland School of Medicine,
660 W. Redwood St., Baltimore, MD 21201 USA

Project Title: Travel Grant

\$700 7/2001

Goal: To facilitate travel to the 6th World Muscle Society Congress, Thursday,
6 September 2001, Snowbird, UT to allow fellow to
present recent data on FSHD. To facilitate thinking and research on
structural and patho-physiological models for FSHD.

Grant: FSHS-SG-016

Researcher: Rossella Tupler, M.D., Ph.D.

Institution: Howard Hughes Medical Institute, Program in Gene Function and
Expression, University of Massachusetts Medical School, Lazare Research Building - 6th Floor - Room
660 A, 364 Plantation Street, Worcester, MA 01605 USA

Project Title: Travel Grant

\$677.82 10/2001

Goal: To assist with travel to the Annual FSH Society International Research
Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-017

Researcher: Rune R. Frants, Ph.D.

Institution: Human Genetics, Leiden University Medical Center, PO Box 9503, 2300
RA Leiden, The Netherlands

Project Title: Travel Grant

\$534.23 10/2001

Goal: To assist with travel to the Annual FSH Society International Research
Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-018

Researcher: Richard Festenstein, Ph.D.

Institution: Gene Control Mechanisms and Disease, Room 5006, Clinical Research Building, MRC Clinical Sciences Centre, Department of Medicine, Hammersmith Hospital, Du Cane Road, London W12 ONN England

Project Title: Travel Grant

\$1,500 11/2001

Goal: To facilitate travel to the 6th World Muscle Society Congress, Thursday, 6 September 2001, Snowbird, UT to deliver Keynote

FSHD Lecture on: "Chromatin structure, gene expression, and disease."
To facilitate thinking and research on chromatin structure and models for FSHD.

Grant: FSHS-SG-019

Researcher: Sara T. Winokur, Ph.D.

Institution: 240 D, Medical Sciences I, Department of Biological Chemistry, University of California, Irvine, CA 92697 1700 USA

Project Title: Small Research Grant for "FSHD-Research ListServ."

\$7,000 3//2002 – open

Goal: To facilitate collaboration, sharing and biomaterials collection and distribution in the FSHD clinical and research communities.

Grant: FSHS-SG-020

Researcher: Davide Gabellini, Ph.D.

Institution: Howard Hughes Medical Institute, Program in Gene Function and Expression, University of Massachusetts Medical School,
Lazare Research Building - 6th Floor - Room 660 A, 364 Plantation Street, Worcester, MA 01605 USA

Project Title: Travel Grant

\$508.78 10/2002

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-021
Researcher: Rossella Tupler, M.D., Ph.D.
Institution: Howard Hughes Medical Institute, Program in Gene Function and Expression, University of Massachusetts Medical School, Lazare Research Building - 6th Floor - Room 660 A, 364 Plantation Street, Worcester, MA 01605 USA

Project Title: Travel Grant
\$678.69 10/2002

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-022
Researcher: Michael R. Green, M.D., Ph.D.
Institution: Howard Hughes Medical Institute, Program in Molecular Medicine, University of Massachusetts, 373 Plantation St., Suite 309, Worcester, MA 01605 USA

Project Title: Travel Grant
\$720.37 10/2002

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-023
Researcher: Yukiko K. Hayashi, M.D. and Kanako Goto, Ph.D.
Institution: Department of Neuromuscular Research, National Institute of Neuroscience, NCNP, 4-1-1 Ogawa-Higashi, Kodaira, Tokyo 187-8502, Japan
Project Title: FSH Society Kiichi Arahata, M.D. Memorial (KAM) International Travel Fellowship Grant
\$2,761.84 10/2002

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG. To facilitate continuity of Japanese FSHD research

Grant: FSHS-SG-024
Researcher: James Marshall, Ph.D. and Kylie DeBoer, Ph.D.
Institution: Sydney IVF, Level 11, O'Connell Street, Sydney NSW 2000 Australia
Project Title: FSH Society Kiichi Arahata, M.D. Memorial (KAM) International Travel Fellowship Grant
\$2,000 10/2003
Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG. To present initial data on in vitro fertilization techniques for FSHD to the FSHD research community.

Grant: FSHS-SG-025
Researcher: Valery Kazakov, M.D., Ph.D.
Institution: Department of Neurology, Pavlov's Medical Institute, Lev Tolstoy str. 6/8, 197022, filial 1 St. Petersburg, Russia
Project Title: Research Grant
\$650 11/2003
Goal: To assist with cost of DNA genotyping study of Russian FSHD samples via Dr. Upadhyaya in Cardiff

Grant: FSHS-SG-026
Researcher: Kevin Flanigan, M.D.
Institution: Eccles Institute of Genetics, Room 4420, University of Utah, 15 North 2030 East Street, Salt Lake City, Utah 84112 USA
Project Title: Travel Grant
\$854.04 11/2003
Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-027

Researcher: Silvere van der Maarel, Ph.D.

Institution: Leiden University Medical Center, Dept. of Human Genetics,
Wassenaarseweg 72, PO Box 9503, 2300 RA Leiden, The Netherlands

Project Title: Travel Grant

\$956.14 11/2003

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-028

Researcher: H. Lee Sweeney

Institution: University of Pennsylvania, School of Medicine, B400 Richards Building,
3700 Hamilton Walk, Philadelphia, PA 19104-6085

Project Title: Conference Grant

\$500 12/2003

Goal: Conference Support Grant for 1st "New Directions in Biology and Disease of Skeletal Muscle," San Diego, CA USA January 25-27, 2004.

Grant: FSHS-SG-029

Researcher: Alberto Luis Rosa, M.D., Ph.D.

Institution: Washington State University – Spokane, WSU Spokane Health Science,
310 North Riverpoint Blvd., PO Box 1495, Spokane,
WA 99210-1495 USA

Project Title: FSH Society Kiichi Arahata, M.D. Memorial (KAM) International Travel Fellowship Grant

\$1,506.02 7/2004

Goal: To assist with travel to the 7th Annual Summer School in Myology to lecture on FSHD with J. Adoni Urtizberea, M.D. and for
travel to Mons-Hainut, Belgium via rail to meet with Alexandra Belayew, Ph.D. on DUX4 and FSHD.

Grant: FSHS-SG-030

Researcher: Silvere van der Maarel, Ph.D.

Institution: Leiden University Medical Center, Dept. of Human Genetics,
Wassenaarseweg 72, PO Box 9503, 2300 RA Leiden,
The Netherlands

Project Title: Travel Grant
\$1,184.97 11/2004

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-031

Researcher: George W.A.M. Padberg, M.D., Ph.D.

Institution: c/o Anjali Kali, Department of Neurology, 326, UMC St Radboud, PO
Box 9101, 6500 HB Nijmegen, The Netherlands

Project Title: Travel Grant
\$1,656.19 12/2004

Goal: To assist with travel to the Annual FSH Society International Research Consortium Meeting as satellite to the ASHG.

Grant: FSHS-SG-032

Researcher: Richard Lemmers, Ph.D.

Institution: Leiden University Medical Center, Dept. of Human Genetics,
Wassenaarseweg 72, PO Box 9503, 2300 RA Leiden, The Netherlands

Project Title: Research Publication Grant
\$851.19 03/2005

Goal: To assist with publication, production and distribution of doctoral thesis on FSHD.

Grant: FSHS-SG-033
Researcher: Petra van Overveld, Ph.D.
Institution: Leiden University Medisch Centrum, Dept. Urology/Endocrinology,
Stafcentrum Endocrinologie C4-R, Albinusdreef 2, 2333
ZA, Leiden, The Netherlands
Project Title: Research Publication Grant
\$851.19 03/2005
Goal: To assist with publication, production and distribution of doctoral thesis
on FSHD.

Grant: FSHS-SG-034
Researcher: Silvana van Koningsbruggen, Ph.D.
Institution: Leiden University Medical Center, Dept. of Human Genetics,
Wassenaarseweg 72, PO Box 9503, 2300 RA Leiden, The Netherlands
Project Title: Research Publication Grant
\$851.19 03/2005
Goal: To assist with publication, production and distribution of doctoral thesis
on FSHD.

Grant: FSHS-SG-035
Researcher: Kristen Bastress and Marcy Speer, Ph.D.
Institution: Duke University Medical Center, Box 3445, Durham, NC 27710 USA
Project Title: Research Project Grant
\$3,800 04/2005
Goal: To assist with travel to Leiden to research and collaborate on non
chromosome 4 linked FSHD samples with distal and proximal deletions in May 2005.

Total: \$1,115,212.84