BIOGRAPHICAL SKETCH

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NAME	POSITION TITLE
Rabi Tawil, MD	Professor of Neurology
eRA COMMONS USER NAME (credential, e.g., agency login) RTAWIL	

EDUCATION/TRAINING *(Begin with baccalaureate or other initial professional education, such as nursing, include postdoctoral training and residency training if applicable.)*

INSTITUTION AND LOCATION	DEGREE *(if applicable)*	MM/YY	FIELD OF STUDY
Emory University, Atlanta, Georgia	BS	6/1979	Biology
American University of Beirut, Lebanon	MS	6/1981	Physiology
Faculty of Medicine, Amer. U. of Beirut, Lebanon	MD	6/1985	Medicine

A. Personal Statement

I have been committed to clinical and translational research in FSHD since embarking on my neuromuscular fellowship in 1991. Our group has the most extensive experience in the development of clinical trial tools for FSHD as well as the conduct of clinical trials in FSHD. Additionally we have been involved in number of translational studies both independently and in collaboration with other institutions. We are one of the few centers with expertise in performance of needle muscle biopsies which has allowed us to develop the largest FSHD biorepository. The present application is a natural extension of my present collaboration with Dr. Stephen Tapscott. The development of new biomarkers for FSHD is of vital importance at this juncture as targeted treatments are likely to emerge in the next several years.

B. Positions and Honors

Positions and Employment

7/85 - 6/86 American University of Beirut Medical Center: Internship in Medicine
7/86 - 7/88 Good Samaritan Hospital, Baltimore, MD: Resident in Internal Medicine
7/88 - 6/91 Univ. of Rochester Medical Center, Rochester, NY: Resident in Neurology
7/91 - 7/93 Univ. of Rochester Medical Center, Rochester, NY: Fellow in Neuromuscular Disease
7/91 - 6/93 Instructor in Neurology, Univ. of Rochester School of Medicine and Dentistry, Rochester, NY.
11/93 -11/98 Asst. Prof. of Neurology, U of Rochester School of Medicine and Dentistry, Rochester, NY
11/96 -11/98 Asst. Prof. of Pathology and Laboratory Medicine (Neuropathology), Univ. of Rochester School of Medicine and Dentistry, Rochester, NY.
11/98- 11/05 Associate Professor of Neurology, Pathology and Laboratory Medicine (Neuropathology), Univ. of Rochester School of Medicine and Dentistry, Rochester, NY.
11/05-present Professor of Neurology, Pathology and Laboratory Medicine (Neuropathology), Univ. of Rochester School of Medicine and Dentistry, Rochester, NY

Honors

1979 Associate Member: Sigma Xi, Emory University Chapter
1991-1993 Muscular Dystrophy Association Fellowship: "Muscle protein synthesis in FSH muscular dystrophy"
1994-1995 Buswell Fellowship Award; University of Rochester.

Professional Societies

1992-Present Member, American Academy of Neurology
2007-Present Fellow, American Academy of Neurology
2001-Present Member, American Neurological Association

C. Selected Peer-Reviewed Publications

Tawil R, Forrester J, Griggs RC, Mendell JR, Kissel J, McDermott M, King W, Weiffenbach B, Figlewicz D, and the FSH-DY Group. Evidence for anticipation and association of deletion size with severity in facioscapulohumeral muscular dystrophy. Ann Neurol 1996; 39:744-748.

The FSH-DY Group. A prospective, quantitative study of the natural history of facioscapulohumeral muscular dystrophy (FSHD): Implications for therapeutic trials. Neurology 1997; 48:38-46.

Orrell RW, Tawil R, Forrester J, Kissell J, Mendell J, Figlewicz DA. Definitive molecular diagnosis of facioscapulohumeral dystrophy. Neurology 1999; 52, 1822-1826.

Forbes GB, Griggs RC, Moxley RT, Thornton CT, Tawil R. K-40 and Dual-Energy X-ray Absorptiometry Estimates of Lean Weight Compared: Normals and Patients with Neuromuscular Disease. NY Acad Sci 2000; 904:111-114.

Kissel JT, McDermott MP, Mendell JR, Pandya S, King WM, Griggs RC, Tawil R, and the FSH-DY Group. Double-Blind, Placebo-Controlled Trial of Albuterol in Facioscapulohumeral Muscular Dystrophy. Neurology 2001; 57: 1434-1440.

Osborne RJ, Welle S, Venance SL, Thornton CA, Tawil R. Expression profile of FSHD supports a link between retinal vasculopathy and muscular dystrophy. Neurology 2007;68:569-577.

Klooster R, Straasheijm K, Shah B, Sowden J, Frants R, Thornton C, Tawil R, van der Maarel S. Comprehensive expression analysis of FSHD candidate genes at the mRNA and protein level. Eur J Hum Genet. 2009. 17:1615-1624.

Tawil R, van der Maarel S, Padberg GW, van Engelen BG. 171st ENMC international workshop: Standards of care and management of facioscapulohumeral muscular dystrophy. Neuromuscul Disord. 2010 Jul;20(7):471-5.

de Greef JC, Lemmers RJ, Camaño P, Day JW, Sacconi S, Dunand M, van Engelen BG, Kiuru-Enari S, Padberg GW, Rosa AL, Desnuelle C, Spuler S, Tarnopolsky M, Venance SL, Frants RR, van der Maarel SM, Tawil R. Clinical features of facioscapulohumeral muscular dystrophy 2. Neurology. 2010; 751548-54.

Lemmers RJ, van der Vliet PJ, Klooster R, Sacconi S, Camaño P, Dauwerse JG, Snider L, Straasheijm KR, Jan van Ommen G, Padberg GW, Miller DG, Tapscott SJ, Tawil R, Frants RR, van der Maarel SM. A Unifying Genetic Model for Facioscapulohumeral Muscular Dystrophy. Science. 2010 Aug 19.

Geng LN, Yao Z, Snider L, Fong AP, Cech JN, Young JM, van der Maarel SM, Ruzzo WL, Gentleman RC, Tawil R, Tapscott SJ.DUX4 activates germline genes, retroelements, and immune mediators: implications for facioscapulohumeral dystrophy. Dev Cell. 2012; 22(1):38-51.

Lemmers RJ, Tawil R, Petek LM, Balog J, Block GJ, Santen GW, Amell AM, van der Vliet PJ, Almomani R, Straasheijm KR, Krom YD, Klooster R, Sun Y, den Dunnen JT, Helmer Q, Donlin-Smith CM, Padberg GW, van Engelen BG, de Greef JC, Aartsma-Rus AM, Frants RR, de Visser M, Desnuelle C, Sacconi S, Filippova GN, Bakker B, Bamshad MJ, Tapscott SJ, Miller DG, van der Maarel SM. Digenic inheritance of an SMCHD1 mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2.Nat Genet. 2012 Dec;44(12):1370-4. doi: 10.1038/ng.2454.

Statland JM, McDermott MP, Heatwole C, Martens WB, Pandya S, van der Kooi EL, Kissel JT, Wagner KR, Tawil R. Reevaluating measures of disease progression in facioscapulohumeral muscular dystrophy. Neuromuscul Disord. 2013 Apr;23(4):306-12. doi: 10.1016/j.nmd.2013.01.008

Statland JM, Tawil R. Risk of functional impairment in facioscapulohumeral muscular dystrophy. Muscle Nerve. 2013 Jul 19. doi: 10.1002/mus.23949.

Tawil R, Shaw DW, van der Maarel SJ, Tapscott SJ. Clinical trial preparedness in facioscapulohumeral muscular dystrophy: Outcome measures and patient access. Neuromuscular disorders 2013. Doi: http://dx.doi.org/10.1016/j.nmd.2013.07.009

D. Research Support

Ongoing Research Support

MDA (Tawil) 1/1/2013-1/1/2014
Exploratory Biomarker Study in FSHD

Support for study of potential serum and tissue biomarkers in FSHD.

R01 NS061795-01A2 (McDermott/Tawil) 1/1/10-1/1/15
NIH
Biostatistics and Data Management for a Trial of Corticosteroid Regimens in DMD

Support is for the statistical and data management component of a study (FOR-DMD; Griggs, R., PI) comparing three different corticosteroid (CS) regimens for the treatment of Duchenne muscular dystrophy (DMD).

1P01 NS069539-01 (Tapscott) 04/15/10-03/31/15
Fred Hutchinson Cancer Center (NIH)
The Pathogenesis of Facioscapulohumeral Muscular Dystrophy

The pathophysiology of FSHD is currently unknown and there are no validated targets or mechanisms that can provide the basis for therapeutic development. The mail goal of this study is to identify and validate specific mechanism for the pathophysiology of FSHD.

7R01FD003710 (Benatar) 11/22/10-6/30/15
University of Miami (FDA)
The Effectiveness of Prednisone for the Treatment of Ocular Myasthenia (EPITOME)

The primary goal of this study is to evaluate the efficacy and tolerability of prednisone in patients with newly diagnosed ocular myasthenia whose symptoms have failed to remit in response to a trial of cholinesterase inhibitor monotherapy (pyridostrigmine).

Completed Research Support

R01 AR054366 (Welle) 08/09/07-06/30/11
NIH
Muscle protein synthesis and gene expression in myostatin-deficient mice

The goals are to determine the effects of postdevelopmental loss of myostatin activity on muscle mass and function, and to examine potential molecular mechanisms for these effects.

HHSN26420052274C (Moxley) 9/30/05-9/29/10
NIAMS
National Registry for Myotonic Dystrophy and Facioscapulohumeral Muscular Dystrophy Patients and Family Members

The major goal of this study is to compile a registry of the above mentioned diseases within the USA.

5 U54 RR019482-02 (Griggs) 9/28/03-9/27/08
NIH
Nervous System Channelopathies Pathogenesis and Treatments

The goals of this project are: 1. begin characterization of the phenotype/natural history of each disease, 2. devise outcome measures for treatment trials, and 3. access quality of life. All goals are in preparation for pilot clinical trials of novel treatments.

1R01 NS049639-01A2 (Amato) 9/15/05-5/31/08
NIH/NINDS
A Pilot Study of Etanercept in Dermatomyositis

The goal of this study is to assess the safety and tolerability of etanerept in patients with Dermatomyositis.

Wyeth Pharmaceuticals(Tawil) 3/1/05-5/27/2008
A Double-blind, Placebo Controlled, Randomized, Multiple Ascending Dose, Safety Study of MYO-029 Administered to Adult Patients with Becker, FSH and Limb Girdle Muscular Dystrophy

The goal of this drug company initiated clinical trial is to perform a trial of MYO-029 in patients with Becker, FSH and Limb Girdle Muscular Dystrophy.

2 U54NS048843-08(Moxley) 9/30/08-8/31/13
NIH
Muscular Dystrophy Cooperative Research Center

The goal of this program project is to establish optimal methods to assess treatment efficacy in DM1 patients and to identify potential new therapies.