Marinesco Sjogren Syndrome NEWS

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About the Newsletter

The gene responsible for MSS has been identified! This edition of the newsletter focuses on this exciting discovery and its possible impacts on MSS patients and families. Congratulations to the researchers on their work and recent publications.

Feel free to distribute the newsletter by email or to print copies for interested individuals. Email us if you wish to be added to or removed from the newsletter mailing list. Current and back issues of the newsletter are available at the website on the publications page. http://www.marinesco-sjogren.org/pubs.html

MSS Gene Identified

The gene for MSS, Sil1, was recently discovered. Several labs around the world, including my own, are sequencing the gene in all the cases we have. We plan to share information on the problems the patients have so that we can once and for all define what constitutes MSS. Problems in other genes can cause MSS-like disorders. It is also possible that there is more than one MSS gene, although all large families I know of show linkage to the same region on chromosome 5. My laboratory has so far identified several mutations in the gene. After we verify the mutations, we will be notifying the families. So far, all the mutations are predicted to eliminate the function of the protein made by the gene Sil1.

The MSS protein is involved in helping other newly made proteins fold correctly. If proteins cannot fold correctly, the cell destroys them and the cell is stressed. How this causes MSS is not clear, but stressed cells do not act normally. Fortunately, there is a mouse model of MSS that is born normal but develops ataxia. That mouse model will help us understand MSS better and will allow us to test therapies.

In human MSS, it is unlikely that a specific therapy would improve the ataxia, but it might improve muscle strength, growth, and mental function.

Once a research laboratory has identified the mutations in your family, it would be possible to perform prenatal diagnosis. Dr. Shibo Li in Oklahoma directs a clinical molecular genetics laboratory and has worked with me on MSS for

years. His laboratory can verify the mutations and perform the prenatal testing. Research laboratories in the United States are not allowed to do such testing.

Studying the mouse and testing therapies will take substantial amounts of money. At this juncture, no pharmaceutical company will be interested. If you have the resources, please make a tax deductible donation to one of the research laboratories working on MSS.

If you are interested in looking for a Sil1 mutation in your family, please contact me at (310) 423-6673 or by email: william.wilcox@cshs.org.

Bill Wilcox, MD, PhD
Medical Genetics Institute
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MSS Publications

L. Zhao, C. Longo-Guess, et. al., "Protein accumulation and neurodegeneration in the woozy mutant mouse is caused by disruption of SIL1, a cochaperone of BiP", Nature Genetics, August 14, 2005.

I. Mahjneh, A.-K. Anttonen, et. al., "Myopathy is a prominent feature in Marinesco-Sjögren syndrome; A muscle computed tomography study", Journal of Neurology, September 15, 2005.

J. Senderek, Michael Krieger, et. al., "Mutations in SIL1 cause Marinesco-Sjogren syndrome, a cerebellar ataxia with cataract and myopathy", Nature Genetics, November 13, 2005.

A. Anttonen, I. Mahjneh, et. al., "The gene disrupted in Marinesco-Sjogren syndrome encodes SIL1, an HSPA5 cochaperone", Nature Genetics, November 13, 2005.

H. Zoghbi, "SILencing misbehaving proteins", Nature Genetics, November 13, 2005.

Congratulations, Graduate!

Congratulations to Misa, 19, on her graduation from high school. Misa is now attending her local community college. Awesome job!

Misa (left) and her sister, Cammi, celebrate on their graduation day.

National Patient Travel Helpline

The National Patient Travel Helpline provides information about all forms of charitable, long-distance medical air transportation. Their objective is "to ensure that no financially-needy patient is denied access to distant specialized medical evaluation, diagnosis, and treatment for lack of a means of medical air transportation.

Services include airline ticket assistance and private and corporate aviation resources.
Phone: 1-800-296-1217
Website: www.PatientTravel.org

NIH Exploring Biospecimen Availability for Disease Research

The issue of availability of high-grade biospecimen and clinical data for research constitutes a barrier to rare disease research. The Genetic Alliance has moved to establish a "biobank" for which it charges patient advocacy groups appropriate fees to participate and to maintain the biospecimens. However, many rare diseases groups do not have the funds for such an endeavor, and many more rare diseases do not have an advocacy group. The Trans-NIH (National Institutes of Health) Rare Diseases Research Working Group has formed a planning committee to determine the issues that need to be explored and to identify the significant players of biospecimen collection, storage, and dissemination. The planning committee will provide recommendations to the NIH for a conference that will lead to a clear understanding of the needs and a plan for implementation.

Genetic Alliance Conference Presentations

Presentations from the July 2005 Genetic Alliance Annual Meeting in Washington DC are available at the Genetic Alliance website. Topics include strategic planning, communication, fund raising, and organization for advocacy groups, research, drug development, public policy, and lobbying.
http://www.geneticalliance.org/ws_display.asp?filter=conference05

Emergency Preparedness for Special Needs Children

The American Academy of Pediatrics (AAP) and the American College of Emergency Physicians have created a tool (Emergency Information Form) to assure prompt and appropriate care for Children with Special Health Care Needs. The form is designed to ensure that a child's complicated medical history is concisely summarized and available when it is needed most - when the child has an acute health problem at a time when neither parent nor pediatrician is immediately available. The information can be retrieved from a central repository through MedicAlert®. Additional information is available at the AAP website:
http://www.aap.org/advocacy/emergprep.htm

Genomics Updates

A weekly on-online newsletter from the Center for Disease Control (CDC) provides information about the impact of human genetic research on disease prevention and public health.
http://www.cdc.gov/genomics/update/current.htm

The National Human Genomics Research Institute (NHGRI) website has a wide range of articles on the status of human genome sequencing, genome comparisons between humans and chimps, genetics education, ethics issues, and more.
http://www.genome.gov/12513430

Exceptional Parent Magazine

Exceptional Parent Magazine has been providing information, support, ideas, encouragement & outreach for parents and

families of children with disabilities, and the professionals who work with them for 34 years. On-line resource: http://www.eparent.com

California Regional Center Service Standards

The Lanterman Act (Welfare & Institutions Code) is the backbone of the Department of Developmental Services/Regional Center system in California. The Lanterman act governs services in the state and sets the standards by which regional centers and service providers must comply to ensure families are treated fairly. Copies of the act can be requested at: http://www.dds.ca.gov/Statutes/RequestForm.cfm

Dolphin Assisted Therapy

Island Dolphin Care is a 501(c)3 not for profit organization that provides dolphin therapy to children with critical illnesses, disabilities and special needs from all over the world. The facility, located in Key Largo, Florida, provides 1-day and 5-day dolphin-assisted therapy programs. http://www.islanddolphincare.org/

Family Friends

Family Friends is an intergenerational volunteer program that matches men and women 55 years and older with families who have children with disabilities and chronic illnesses. The program was started by the National Council on the Aging in 1984. There are currently 35 programs throughout the country, striving to join the skills, knowledge, and compassion of older adults with the needs of families. http://www.family-friends.org/index.htm