

M Marinesco
S Sjogren
S Syndrome
NEWS

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About the Newsletter

This edition of the newsletter reviews MSS support group accomplishments over the past four years and our plan for the future, summarizes the status of MSS research, and includes numerous special needs news items and resources. Suggestions for improving the newsletter and ideas for articles are welcome. 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>

The MSS Support Group: Our First Four Years

The MSS website went live just over four years ago. Our accomplishments over that time include:

- The website is #1 for MSS on both the Google and Yahoo search engines.

- This edition of the newsletter is our 7th in just over two years. It is distributed by email and hardcopy to MSS families, selected physicians and other health care workers, and friends and relatives all over the world.
- We created a tri-fold brochure for families and a one-page medical summary for physicians. We respond to requests for MSS information from newly diagnosed families, physicians, care-givers, therapists, health-care workers, medical students, and even high school students preparing papers on rare disorders. We have had contact with more than 25 families from nine different countries in which MSS was diagnosed or was considered a possible diagnosis.
- We increased awareness about MSS by informing over 700 clinical geneticists in the US and Canada about our website and support group. We exhibited at the 2003 ASHG conference. Our effort at that meeting helped establish an international collaborative MSS research effort.

In that four-year time period, the following medical research progress on MSS has occurred:

- At least ten journal and conference papers were published by American, French, Finnish, Australian, and German physicians on MSS and clinically overlapping disorders.
- The genes for two similar disorders (CCFDN and CMRD) have been identified, helping to clarify MSS diagnosis.
- The published location of the MSS gene has been narrowed to about 1 million base pairs. (The complete human genome contains about 3 **billion** base pairs.) All of the known large family groups with classical MSS in the US, Norway, Turkey, and Finland map to chromosome 5q31.
- Exhibit at upcoming medical conferences, or contribute financially to offset conference fees.
- Provide legal or financial expertise in non-profit incorporation and 501(c)(3) formation.
- Help with publicity and fund-raising.
- Use your writing or computer skills to publish the newsletter or upgrade the website.

You can see how the MSS website (or any other website) has changed over the years by going to the following website and typing in the website of interest.

<http://web.archive.org/collections/web.html>

Where do we go from here? Our goals for the next few years are to:

- Expand our database of MSS families and improve communication with them. Establish an on-line email list to encourage communication between families.
- Increase awareness about MSS through our website, newsletter, and other publications, and by exhibiting at medical conferences (e.g. genetics and child neurology meetings).
- Support MSS research by establishing and maintaining contact with researchers and encouraging family participation in research activities.
- Incorporate and establish non-profit 501 (c)(3) status. Initiate fund-raising activities.
- Sponsor a NIH scientific workshop and/or family conference.
- Increase support group effectiveness through interaction with other lay advocacy groups.

We are eager to get family inputs on how we can better help you. Please email us to share your thoughts or to let us know if you can help in any of the following ways:

New MSS Technical Publications

J. Lee, B. Chodirker, A. E. Chudley, S. R. Sanders, C. Carriles, C. Powell, J. J. Mulvihill, S. Li, "No evidence of SARA2 gene mutation in our patients with Marinesco-Sjogren Syndrome (MSS)", ASHG Meeting, Toronto, Ontario, November 2004.

Harting, MD, A. Blaschek, MD, N. I. Wolf, MD, A. Seitz, MD, M. Haupt, MD, H. H. Goebel, MD, D. Rating, MD, K. Sartor, MD and F. Ebinger, MD, "T2-hyperintense cerebellar cortex in Marinesco-Sjögren syndrome", Neurology 2004; Vol 63: 2448-2449.

Anne Slavotinek, Jill Goldman, Kara Weisiger, Dana Kostiner, Mahin Golabi, Seymour Packman, William Wilcox, H. Eugene Hoyme, Elliott Sherr, "Marinesco-Sjögren syndrome in a male with mild dysmorphism," American Journal of Medical Genetics, 4 January 2005.

MSS in New NORD Brochure

MSS is included in the latest National Organization for Rare Disorders (NORD) brochure on Hereditary Ataxias published for physicians and other health care professionals. The brochure describes symptoms and signs of ataxia, types, and diagnosis. MSS is included in the list of autosomal recessive hereditary ataxias. The brochures are available at no cost and can be viewed at the NORD website: http://www.rarediseases.org/programs/hereditaryataxia_brochure.html

Upcoming Conferences

American College of Medical Genetics 2005 Annual Clinical Genetics Meetings, Dallas, Texas, March 17-20, 2005. www.acmg.net

European Society of Human Genetics (ESHG) Annual Meeting, Prague, May 7-10, 2005. www.eshg.org

Genetic Alliance, Bethesda North Marriott, Bethesda, Maryland, July 29-31, 2005, <http://www.geneticalliance.org/>

American Society of Human Genetics (ASHG) Annual Meeting, Salt Lake City, Utah, October 25-29, 2005. www.ashg.org

Child Neurology Society, Westin Century Plaza Hotel, Los Angeles, California, September 28-October 1, 2005. <http://www.childneurologysociety.org/>

US Individuals with Disabilities Education Act (IDEA) Update

On Friday, December 3, 2004, President Bush signed HR 1350 (IDEA 204) into law. The final text of the law passed (over 300 pages) is at:

<http://thomas.loc.gov/cgi-bin/bdquery/z?d108:HR01350:d108query.html>

A summary of the major changes by the National Association of Protection & Advocacy Services is available at http://www.oclb.info/pdf/NAPAS_IDEA2004_Summary.pdf

Genetic Alliance Interactive Guide

The Genetic Alliance interactive guide is a user-friendly source of information on starting and building advocacy groups. The guide provides information on defining goals, establishing the organization structure, recruiting, fundraising, publicity, supporting research, and more. The Genetic Alliance will continue to be a valuable source of information for us as we expand our support group efforts.

http://www.geneticalliance.org/ws_display.asp?filter=tools_interactive_guide

The Genetic Alliance also publishes a periodic newsletter. To subscribe to the newsletter, submit your email address at the following website.

http://www.galists.org/read/all_forums/subscribe?name=g_advocacy

US Tax Benefits for Parents of Children with Learning Disabilities

Tax benefits for children with disabilities may include deductions for medical expenses (therapy and tutoring under some conditions), disability related conferences, and child and dependent care credits. Contact your tax accountant or attorney for more details. A short introduction to tax-related issues is available at: <http://www.schwablearning.org/articles.asp?r=773>

NIH Roadmap

The National Institutes of Health (NIH) has established a roadmap to lead to new strategies for diagnosing, treating, and preventing disease, and new strategies for conducting research. You can read more about the new NIH initiatives at: <http://www.nihroadmap.nih.gov/>

Genetic Alliance BioBank™

Seven genetic advocacy organizations established the Genetic Alliance BioBank™, a repository owned and managed by advocacy groups for the standardized collection, storage, and distribution of biological samples and clinical data for research purposes. The goal of the repository is to focus and accelerate research, providing infrastructure for many advocacy groups.
<http://www.gabiobank.org/default.asp>

DrScore Physician Satisfaction

DrScore is a website designed to improve the quality of medical care by providing feedback on physician quality to patients and to health care providers. The website allows you to “score” your doctor or search for a doctor given geographic location and specialty. The site also provides information on medical school attended and residency information. DrScore is partnering with patient-advocacy groups.
www.drscore.com

Book Review:

“Straight Talk About Psychological Testing for Kids”

What types of testing are used to evaluate children? What are the advantages of school-based versus private testing? How can I use

testing to get help for my child? These are some of the questions addressed in the book “Straight Talk About Psychological Testing for Kids” by Ellen B. Braaten, Ph.D., and Gretchen Felopulos, Ph.D. Recommended by special needs organizations and available at: www.amazon.com.

Gene Patenting

Ever hear about “gene patenting” and wonder what it really means? If something is useful and required human intervention to produce, then the product or process may be patentable. Any patentable invention must be useful, novel and unobvious. One cannot get a blanket patent covering all the possible uses of a gene, or even a patent on the gene sequence itself. Some patents that might be allowed include a gene sequence that is indirectly patented, the use of the gene product, or possibly the method of purifying the gene product. In order to patent the use of a mutant gene for diagnostics, the filing party must demonstrate novelty and utility in diagnosing disease. Read more about gene patenting and other biotechnology issues at: <http://biotech.about.com/library/weekly/aa060900a.htm>