

M Marinesco
S Sjogren
S Syndrome
NEWS

Summer 2007
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About the Newsletter

In this edition of the MSS newsletter, we welcome a new family from Australia, provide an update on our group, and announce plans to exhibit at the ASHG meeting in October.

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 on the website at:

www.marinesco-sjogren.org/pubs.html

Welcome to Dominic and Daniel!

We welcome Dominic and Daniel, two brothers from Australia, to our group. Dominic is 12 and excited about being a teenager soon. Daniel is 11, delightful and full of energy. The Yinger family was happy to meet them when Dominic, Daniel, and their parents recently came to the United States to visit relatives, Disneyland, Sea World, and other highlights of California.

The boys use walkers for short distances and wheelchairs for longer distances. They had cataract surgeries at the ages of 3-4 years. They are small in size, but large in spirit!



Dominic, Kimberly, and Daniel have fun together!

The MSS Support Group - Who are We?

Our support group now consists of over 25 families diagnosed with (or possibly having) MSS. About 40% of the families live in the US and 60% live in other countries. Our international families are from all over the world including: France, United Kingdom, Austria, Pakistan, Canada, Brazil, Australia, Germany, Mexico, Turkey, and Spain.

Here is the latest on a few of our families:

Misa (21) continues to attend Community College. She was planning to get a job or do volunteer work during the summer.

Tammy and Isaac Lau recently participated in a disability art fair near their home in Hawaii. Tammy sold the greeting cards she paints, and Isaac did a weaving demonstration. Isaac also participated in a karate exhibition.

Kimberly (15) just started 10th grade. She attended summer school, took swimming and horseback riding lessons, and enjoyed family vacation this summer. She recently started special needs gymnastics, which is very fun and great exercise, too. More info at: <http://www.bigfungymnastics.com/>.

MSS Exhibit at Upcoming ASHG Meeting

The MSS support group will be exhibiting at the American Society of Human Genetics (ASHG) Annual Meeting, October 23-27, 2007 in San Diego, California. Look for us at booth #337. See the ASHG website for more information about the meeting.

<http://www.ashg.org/genetics/ashg/menu-annmeet.shtml>

We encourage families to send us a picture (by email or postal mail) that will help add a personal touch to our exhibit. We would like to receive pictures from many countries and from MSS individuals young and old. If you have sent us a picture in the past and would like us to use that one, please send us a brief email giving us permission to do so. Most of the meeting attendees have never seen a patient with MSS, so this is a great opportunity to showcase our families and demonstrate the diversity of people affected by MSS.

Yinger Email Update

The Yinger family has a new email:

Old: csyinger@adelphia.net

New: csyinger@roadrunner.com

The MSS email address is unchanged, and is the best way to contact us since it is associated with the website and is unlikely to change: mss@marinesco-sjogren.org

Long-term MSS Study in Eight Japanese Families

A 1995 MSS paper from the Japanese Journal of Rehabilitation Medicine is available online. The paper (in Japanese) provides a long-term follow-up study of 14 patients from 8 families, including motor impairment and ambulatory capacity. Go to: <http://ci.nii.ac.jp/naid/110001862441/en/> and click on pdf at the top-right corner to see the full paper. Thank you to one of our families for the following translation of the legend in Figure 2, page 37 of the paper showing changing ambulatory capacity with age for each of the patients.

- Able to walk without support (white)
- Can cruise or walk with stick (medium)

- Unable to walk, can maneuver wheelchair (dark)
- Dependent on others for movement, upper & lower body affected (black)

Genetic Information Nondiscrimination Act Passes House

The Genetic Information Nondiscrimination Act (GINA) passed 420-3 in the United States House of Representatives on April 25, 2007. The Act would prevent misuse of genetic discrimination in insurance and employment. Fear of misuse of genetic information causes many people to avoid genetic testing or to opt out of participation in medical research.

http://www.geneticalliance.org/ws_display.asp?filter=policy.leg.nondiscrim

House Approves Taxpayer-Funded Research Access

The U.S. House of Representatives approved a measure directing the National Institutes of Health (NIH) to provide free public online access to agency-funded research findings within 12 months of their publication in a peer-reviewed journal. A similar measure has been approved by the Senate Appropriations Committee and will be considered by the full Senate later this summer. More info at:

www.taxpayeraccess.org.

Upcoming Conferences

NORD Annual Conference, September 28-30, 2007, Hilton Washington DC/Rockville Executive Meeting Center in Rockville, Maryland. www.rarediseases.org

American Society of Human Genetics (ASHG) Annual Meeting, October 23-27, 2007, San Diego, CA.

<http://www.ashg.org/genetics/ashg/menu-annmeet.shtml>

Technical Papers on MSS and Related Topics

Ni M, Lee AS, "ER chaperones in mammalian development and human diseases", FEBS Lett, April 25, 2007. (Contains a paragraph on the role of the co-chaperone SIL1 in MSS and an extensive discussion on the role of BiP in protein folding diseases).

Nicula C, Stanila L, Cristea A, "Marinescu Sjogren syndromme--case report", Oftamologia, 2007;51(1):35-9.

"Structural approach by X-ray crystallography to understand the mechanism of molecular chaperone action", Bracher, A. (website)

http://www.biochem.mpg.de/en/research/rd/hartl/andreas_bracher/index.html

Okada, M, et. al., "Clinicopathological features of 14 patients with Marinesco-Sjogren syndrome due to SIL1 mutation", 49th Annual Meeting of the Japanese Society of Child Neurology, July 5-7, 2007, O-117.

http://child-neuro-jp.org/english/images/49thJSCN_e.pdf

Schulz, S, et. al., "Congenital cataract, ataxia, external ophthalmoplegia and Dysphagia in two siblings. A Marinesco-Sjögren-like syndrome",

http://www.ncbi.nlm.nih.gov/sites/entrez?Db=pubmed&Cmd=ShowDetailView&TermToSearch=17712737&ordinalpos=1&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_RVDocSum

Join the MSS Listserv

Our MSS listserv is approaching 20 subscribers. To join the listserv, send us an email requesting to be added to the listserv, or register directly by going to

http://www.galists.org/read/all_forums/subscribe?name=mss and providing the requested information (email address, optional name, and password). After you register, you can submit email to the listserv by posting email to: mss@listserv.galists.org.

You can access the archives (read all previous postings) by going to <http://www.galists.org/read/?forum=mss> and logging in with email address and password.

The listserv is a low-volume list that enables families to communicate with each other. We also use the listserv as a means for sharing information about MSS or other relevant topics in a timelier manner than the newsletter. When you sign up for the listserv, your name and email remain private (except to the list administrator) unless you choose to post to the listserv. Please join and introduce yourself!

Websites for Special Needs Families

Here are several websites providing useful information about health care, family matching, public policy, and more for special needs families.

www.familyvoices.org : national (US) grassroots organization that provides information and education about ways to ensure and improve family-centered health care for children and youth with disabilities and chronic conditions.

www.p2pusa.org : national (US) non-profit organization committed to assuring access and quality in Parent-to-Parent support across the country.

<http://www.edcm.org.uk> : highlights the needs of families with disabled children in the United Kingdom.

Benefits of Respite Services

A publication by the National Information Center for Children and Youth (NICHCY) with Disabilities describes the benefits of respite services, resources, and issues to consider when selecting respite care.

<http://www.nichcy.org/pubs/outprint/nd12txt.htm>

Genetic Alliance Resource Repository

The newly updated Genetic Alliance Resource Repository contains best practices, tools, and tips contributed by members of the Genetic Alliance Network to help the advocacy community succeed. Although the information is largely geared towards advocates, there is a lot of interesting information for families including genetic resources, public policy information, and presentations from Genetic Alliance conferences.

<http://www.resourcerepository.org/>

Adaptive Equipment Resource

Here is a good on-line resource for all kinds of adaptive equipment – walkers, bicycles, therapy supplies, and more.

<http://www.adaptivemall.com/>

College Scholarships

The ChairScholars Foundation offers college scholarships, up to \$20,000 over four years, for the severely physically disabled.

<http://www.chairscholars.org/>