Marinesco Sjogren Syndrome

NEWS

Spring 2007
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About the Newsletter

In this edition of the MSS newsletter, we welcome 5-year old Bronnie and her family to our group. We provide an update on genetic nondiscrimination legislation currently being considered by Congress and provide resources related to income tax deductions, travel for the disabled, sibling support, and more.

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 Bronnie and her family!

We welcome Bronwen (Bronnie) and her family to our group. Bronnie lives in Wales, United Kingdom (UK) near the capital city of Cardiff with her parents Lynn and Anthony. Her grandfather, Alun, first contacted us about three years ago when Bronnie was just two years old and MSS was suspected.

Bronnie at her 5th birthday party with dad Anthony

A lot has happened since that time. Bronnie developed cataracts in both eyes at three years of age, underwent two cataract surgeries, and currently sees well with IOLs and glasses. She is now five years old and attends a mainstream primary school with the help of a classroom assistant. Bronnie is very happy, has a wonderful imagination, knows her letters, and loves books and telling stories. She is below average height for her age.

Lynn and Anthony's current priorities are potty training and improving mobility. Bronnie has a walker and wears leg splints at school and for exercises at home. However, she is not highly motivated to be mobile. Her physiotherapy and speech/language services have been sporadic – a pitfall of the UK National Health Service. The family would appreciate some good ideas for getting Bronnie moving!

Genetics Update

by

Dr. William Wilcox

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My genetics fellow, Dr. Fatih Ezgu, and I have been busy testing all the samples that have been sent to us for mutations in Sil1. In addition to looking for mutations in the DNA, when we have cultured cell lines, we are also examining Sil1 at the messenger RNA and protein level.

In people with typical MSS, we are finding Sil1 mutations about 2/3 of the time. The remainder that we have been able to study all have normal levels of Sil1 protein. There is clearly another gene for MSS.

All the people who have many features in addition to those of typical MSS do not so far have Sil1 mutations.

We are continuing to accept specimens for Sil1 testing and plan to examine other candidate genes for the cases without Sil1 mutations. We need more cases to really understand what features are part of Sil1 and non-Sil1 MSS. Whenever a treatment is tested, it will be important to know if there are Sil1 mutations or not.

Genetic Nondiscrimination Legislation

The Genetic Information Nondiscrimination Act (GINA) was recently introduced into the United States House of Representatives and Senate in January 2007. The proposed law prohibits discrimination in both health insurance and employment. Fear of misuse of genetic information often prevents people from having genetic tests or makes them unwilling to participate in clinical trials or other research. If you are concerned about these issues, please contact our representatives to support GINA using the simple directions (including suggested language) at the Coalition for Genetic Fairness website:
http://www.geneticfairness.org/contact_gina.html

Exhibit at ASHG conference

We are tentatively planning an MSS exhibit at the 2007 American Society of Human Genetics (ASHG) Annual Meeting Oct 23-27 in San Diego, California.

If you are an MSS family, consider helping us in one of two ways. First, we need help to staff the exhibit. No specialized knowledge is required, and all materials (displays, fact sheets, and brochures) will be provided. You also get the opportunity to attend some of the conference technical sessions and visit the other exhibits and poster papers. Second, we would also appreciate any financial contributions you can make to help offset the cost of the exhibit (a

total of about $1000, including booth fees, furniture rental, and printing expenses).

If you are a physician or researcher planning to attend the conference, please visit us at our booth. If you let us know in advance that you will be attending, we may be able to arrange an informal get-together.

Recent Publications on MSS and Related Topics

Macario, A., et. al., "Molecular Chaperones: Multiple Functions, Pathologies, and Potential Applications", Frontier in Bioscience 12, 2588-2600, January 1, 2007.
http://www.bioscience.org/inpress/2257.pdf

Annesi, G., SIL1 and SARA2 mutations in Marinesco-Sjogren and chylomicron retention diseases, Clin Genet. 2007 Mar;71(3):288-9.

Upcoming Conferences

Annual Clinical Genetics Meeting, March 21-25, 2007, Nashville, Tennessee.
http://www.acmgmeeting.net/

Family Voices Conference (for children with special health care needs), May 23-26, 2007, Washington, DC.
http://www.familyvoices.org/info/nc/index.php

Genetic Alliance Annual Conference: The Year of the Advocate, July 27-29, 2007, Bethesda North Marriott, Bethesda, MD.
http://www.geneticalliance.org/ws_display.asp?filter=2007.july

American Society of Human Genetics Annual Meeting, October 23-27, 2007, San Diego, CA.
http://www.ashg.org/genetics/ashg/menu-annmeet.shtml

Dr. Jan Senderek Wins Award

Congratulations to Dr. Jan Senderek on his 10,000 Euro award from the Maximillian May Stiftung, a foundation that fosters German researchers who work on neurodegenerative disorders in children. Dr. Senderek and his team discovered the Sil1 gene that causes MSS over a year ago. Read more on page 9 (in German) at:
http://www.mh-hannover.de/fileadmin/mhh/download/ueberblick_service/Info_06.12/06-15_Aktuelles.pdf

Sibling Support

The Sibling Support Project of the Arc of the United States provides SibNet and SibKids, listservs for and about brothers and sisters of people with special health, developmental, and emotional needs.
www.siblingsupport.org

The Sibling Center provides support to well siblings of children with a medical condition.
http://www.cpmc.org/services/sibcnt.html

In-Home Support Services

In-Home Support Services (California) helps pay for services to allow the elderly or disabled to live safely at home. Authorized services may include housecleaning, meal preparation, laundry, grocery shopping, personal care services, accompaniment to medical appointments, and protective supervision. More information about eligibility and how to apply is available at:
http://www.cdss.ca.gov/cdssweb/In-HomeSup_173.htm

Tax Benefits for Parents of Children with Disabilities

Several on-line resources are available to learn about US income tax benefits that may be applicable to families with disabled children. Deductions **may** include: medical expenses, diagnostic evaluations, therapy, tutoring (some restrictions), and transportation expenses. See your tax accountant or attorney for advice.

http://www.irs.gov/pub/irs-pdf/p3966.pdf

http://www.irs.gov/pub/irs-pdf/p907.pdf

Guided Tours for the Disabled

Guided Tour, Inc. has provided travel experiences for persons with developmental and physical challenges, ages 17 and above, since 1972. Destinations include the US, Canada, Mexico, and Europe. The company operates out of Philadelphia, PA. The trips include transportation, lodging, and most meals. More information is available at: http://guidedtour.com/

The MSS Support Group provides this resource for information only. Families are solely responsible for researching and selecting appropriate programs for their children or other family members.

"Special Needs Mama" On-line Magazine

Literary Mama, a monthly, on-line literary magazine for the "maternally inclined" recently introduced a new feature "Special Needs Mama". The website publishes high-quality columns about the daily experiences of special needs motherhood. The articles are available at: http://www.literarymama.com/columns/specialneedsmama/

Join the MSS Listserv

Our MSS listserv continues to grow. 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.

After you register, 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!

On the Bookshelf

Kushner, H.S., "When Bad Things Happen to Good People". Rabbi Kushner's practical perspective on how people can better deal with severe problems that enters their lives. His son suffered from a very rare genetic disorder, Progeria, which causes premature aging.