

**M** Marinesco  
**S** Sjogren  
**S** Syndrome  
**NEWS**

Fall 2008  
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### About the Newsletter

In this edition of the MSS newsletter, we introduce a new family from Japan, highlight recent technical papers on MSS, and provide a variety of useful resources and activities for MSS families.

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](http://www.marinesco-sjogren.org/pubs.html)

The website and newsletter are not a substitute for medical or legal advice. Please see your physician or other professionals regarding diagnosis, therapy, devices, services, or activities discussed in the newsletter.

### Welcome Akina and Niina!



**Akina, Marina, and Niina**

Welcome to Akina, Niina, and their sister Marina. Akina and Niina live near Osaka, Japan and were recently diagnosed with MSS. Akina is twelve, attends junior high, and enjoys singing in the chorus. Niina is six years old and is learning to play the piano. The whole family enjoyed vacationing in Hawaii this past summer with their grandparents. The girls love the Tokyo Disney resort and all of the Disney characters.



### Look What I Can Do!

Akina, who is average height, uses crutches at school, but walks independently at home. Niina is small for her age and uses a walker. Akina and Niina had cataract surgeries at the ages of seven years and three years, respectively.

We are delighted to add these beautiful young ladies and their family to our support group.

### MSS Papers at 2008 ASHG Annual Meeting

Three poster papers on MSS will be presented at the upcoming American Society of Human Genetics (ASHG) Annual Meeting in Philadelphia, November 11-15, 2008. Abstracts can be found by going to the following link and searching on Marinesco-Sjogren or other text. <http://www.ashg.org/2008meeting/abstracts/fulltext/>

- (1) “Comparison of clinical features between *SIL1* mutation -positive and -negative patients in Marinesco-Sjögren syndrome”
- (2) “Identification of a novel mutation homozygous mutation in the *SIL1* gene in Marinesco-Sjögren syndrome (MSS)”
- (3) “Marinesco Sjögren syndrome (MSS): Novel *SIL1* mutations and description of a less severe phenotype”

### Recent MSS Technical Publications

Merlini, L., “Marinesco-Sjogren syndrome, Fanfare, and more”, *Neuromuscular Disorders*, February 2008.

<http://www.ncbi.nlm.nih.gov/pubmed/18207737>

Anttonen, A., et. al, “Novel *SIL1* mutations and exclusion of functional candidate genes in Marinesco-Sjögren syndrome”, *European Journal of Human Genetics*, February 2008.

<http://www.ncbi.nlm.nih.gov/pubmed/18285827>

Eriguchi, M, et. al., “Identification of a new homozygous frameshift insertion mutation in the *SIL1* gene in 3 Japanese patients with Marinesco-Sjögren syndrome”, *Journal of Neurol Science*, April 2008.

<http://www.ncbi.nlm.nih.gov/pubmed/18395226>

Sakai K, et. al, “Marinesco-Sjögren syndrome with atrophy of the brain stem tegmentum and dysplastic cytoarchitecture in the cerebral cortex.”, *Neuropathology*, April 2008.

<http://www.ncbi.nlm.nih.gov/pubmed/18410272>

Anttonen, Anna-Kaisa’s PhD Dissertation on MSS.

<https://oa.doria.fi/bitstream/handle/10024/35266/themolec.pdf>

## 25<sup>th</sup> Anniversary of NORD and Orphan Drug Act

The NORD (National Organization for Rare Disorders) 25<sup>th</sup> Anniversary Gala was held in Washington DC on May 20, 2008. The gala was held in conjunction with the International Conference on Rare Diseases and Orphan Drugs (ICORD) and celebrated the 25<sup>th</sup> anniversary of the Orphan Drug Act. Since the passage of the Orphan Drug Act, more than 1,100 treatments for rare diseases have entered the research pipeline and more than 300 have been approved by the U.S. Food and Drug Administration for marketing <http://www.rarediseases.org/>

## Upcoming Conferences

Child Neurology Society 37<sup>th</sup> Annual Meeting, Santa Clara, California, November 5-8, 2008. [http://www.childneurologysociety.org/annual\\_meeting/registration](http://www.childneurologysociety.org/annual_meeting/registration)

American Society of Human Genetics 58<sup>th</sup> Annual Meeting, Philadelphia, Pennsylvania, November 11-15, 2008. <http://www.ashg.org/2008meeting/>

## Genetic Nondiscrimination Act Signed into Law

The Genetic Information Nondiscrimination Act (GINA) was signed into law on May 21, 2008. GINA provides protections against genetic discrimination in both the health insurance and employment settings. The health insurance provisions of the law will take effect in 12 months and the employment protections will take effect in 18 months.

Read more about GINA at: <http://www.geneticfairness.org/index.html>

## “Patient Power” - Forbes Magazine

“Patient Power”, an on-line September 2008 article in Forbes Magazine, describes the power of patient advocacy groups and their impact over the past 20 years.

<http://www.forbes.com/healthcare/forbes/2008/0915/070.html>

## Pediatric Cataracts and Glaucoma Support Groups on the Web

Two on-line support groups are available to help children with pediatric cataracts or glaucoma.

The first is the Pediatric Glaucoma and Cataract Family Association:

<http://health.groups.yahoo.com/group/PGCFA-Support/>

The organization also maintains a database at:

<http://pgcfa.org/kb/>

The second, APHAKIC (Association of Parents Having A Kid In Contacts) discusses IOLs, glasses, contact lenses, and related issues.

<http://health.groups.yahoo.com/group/aphakic/>

## WikiGenetics

WikiGenetics is web-based encyclopedia on human genetics for the public. Topics include basic science, the human genome, genetic testing, and a genetic condition directory.

[http://www.wikigenetics.org/index.php/Main\\_Page](http://www.wikigenetics.org/index.php/Main_Page)

## US Presidential Candidates’ Health Care Proposals

A side-by-side comparison of the US Presidential Candidates’ Health Care Proposal is available at the following site:

[http://www.health08.org/sidebyside\\_results.cfm?c=5&c=16](http://www.health08.org/sidebyside_results.cfm?c=5&c=16)

## National Family Caregivers Month

November 2008 has been designated National Family Caregivers Month to honor the millions of Americans who provide care, support, and encouragement to family members with rare and chronic diseases.

[www.thefamilycaregiver.org](http://www.thefamilycaregiver.org)

## Ride-On Therapeutic Horsemanship on YouTube

An uplifting video about Ride-On Therapeutic Horseback Riding is available on YouTube. The video highlights the riders, ranging in age from young children to adults, who benefit from therapeutic horsemanship. There are a few short clips of Kimberly Yinger. She is wearing a blue Ride-On T-shirt. You can see her trotting, smiling as she climbs the stairs to ride, and high-fiving one of her “side-walkers”.

<http://www.youtube.com/watch?v=CkK29De1iGk>

## Next Chapter Book Club

The Next Chapter Book Club (NCBC) provides adolescents and adults with intellectual disabilities the opportunity to read and learn to read, talk about books, and make friends in a fun, community setting. Next Chapter Book Clubs across the country meet weekly in local bookstores and cafés to read and discuss books of their choosing. NCBC members range from those who read well to those who do not read at all. See the following website for locations and how to get involved as a participant or volunteer.

<http://nextchapterbookclub.org/index.asp>

## Enhancing Access to General Education

The Access Center strives to enhance access to the general education curriculum for students with disabilities. Their website resources focus on core content areas—language arts, math, and science—and on instructional and learning strategies to provide disabled students with access to rigorous academic content

<http://www.k8accesscenter.org/index.php>

## US Adaptive Recreation Center (USARC)

The United States Adaptive Recreation Center was founded in 1983 (as California Handicapped Skiers) to ensure that access to skiing is available to people with all types of disabilities. Registration for private ski lessons at Big Bear Mountain in Southern California starts in November. Call early for reservations, particularly for weekend lessons.

[www.usarc.org](http://www.usarc.org)

## Transition to Adult Healthcare

The Canadian Pediatric Society website discusses transition to adult care for youth with special health care needs. The article compares pediatric and adult-oriented care, discusses transition strategies, and provides transition recommendations and resources.

<http://www.cps.ca/english/statements/am/ah07-01.htm>

## Controlling Regional Center Costs – Report to the California Legislature

Regional Centers provide a variety of services to developmentally disabled children and adults in California. A December 2007 report to the California state legislature summarizes a variety

of cost-containment measures that the centers have used or are considering to reduce expenditures.

<http://www.cdcan.us/budget/2008-2009/ControllingRCCosts2007.pdf>

## Historical NIH Grant Data

The Computer Retrieval of Information on Scientific Projects (CRISP) Query Form allows the general public to access information about National Institutes of Health (NIH) grants from 1972 to the present. You can search by topic, principal investigator, institution, and other fields.

[http://crisp.cit.nih.gov/crisp/crisp\\_query\\_generate\\_screen](http://crisp.cit.nih.gov/crisp/crisp_query_generate_screen)

## All-Terrain Strollers and Wheelchairs

**Wheeleez**<sup>®</sup> wheels on trailers and mobility devices enables travel over sand, mud, turf, snow, and other terrain that is inaccessible to other wheels. [www.wheeleez.com](http://www.wheeleez.com)

The following site shows examples of how wheelchairs and beach strollers have been equipped for sand and other terrain.

<http://www.pierpontbay.com/projects.html>