

M Marinesco
S Sjogren
S Syndrome
NEWS

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Welcome to MSS News

Welcome to the first edition of the Marinesco-Sjogren Syndrome (MSS) Support Group Newsletter. We are excited about spreading the news about MSS and hope you find this newsletter helpful, whether you are a parent, relative, friend, or health-care professional. We plan to publish the newsletter two to three times a year. Suggestions for improving the newsletter and ideas for articles are always 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 will also be available at the website.

Medical Overview

By William Wilcox, M.D., Ph.D.
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Thank you to Mrs. Yinger for putting this support group, web site, and newsletter together. As you have no doubt discovered by now, MSS

is a rare condition and few physicians, even geneticists, are aware of it. At the present time, the diagnosis is made based only on the history and physical findings. Other disorders can look a lot like MSS and be confused with it. If you read the medical literature, you will see that some patients labeled with MSS do not have it.

What originally intrigued me about MSS was something I saw under the electron microscope about 9 years ago that had been seen by others. Patients with MSS have accumulation of fat inside cultured skin cells when we look at them under the electron microscope. While we have never seen this fat in any other condition, we do not know if you can have MSS and have normal skin cells. We are trying to get cultured skin cells and a medical history on anyone where the diagnosis of MSS is suspected.

MSS is an autosomal recessive condition, meaning that both parents carry the gene for it. Genes are a section of DNA that contains instructions to make a specific protein that carries out a specific process in the cell. Everyone

has two copies of the MSS gene- one we inherit from our mother, the other from our father. In the case of children with MSS, both copies of the gene are not functioning. One functioning copy of the gene is enough to make up for the non-working gene one so you are completely normal if you are a carrier. Everyone carries about 8 different recessive disorders amongst some 35,000 genes; you just don't usually know what you carry. MSS seems to occur around the world in all ethnic groups. I can only estimate the frequency at about 1:300,000 births.

There is no specific treatment for MSS, but physical and occupational therapy, orthotic devices and walkers, special education, and ophthalmology evaluations can greatly improve the ability of child with MSS to function at the highest level possible. Because of the visual and muscular problems, it is very easy to underestimate the intelligence of a person with MSS. Growth hormone, while it can be tried in MSS, has not been very successful in most genetic conditions with short stature, and involves daily injections of an expensive medication. As children with MSS reach the age of puberty, many of them will need treatments with hormones because their brains are not telling their bodies to make enough hormones to develop normally and to have the pubertal growth spurt.

Most of our research efforts on MSS have focused on trying to find the gene in a large family with MSS affecting many family members. We have so far narrowed our search down to approximately 4 million base pairs (a base pair is like one letter in a very long word) of DNA. While we have a candidate gene that we are testing, this is still a pretty large amount of DNA to look at. Soon we will see if MSS in other families could be due to the same gene or not. Sometimes even rare conditions can be caused by more than one gene. Every additional family that participates in the research will help us find the MSS gene(s). With the gene in hand, diagnostic testing will be easier, prenatal diagnosis would be possible, and we would be

much closer to understanding how the gene(s) can cause the condition. Knowing what causes the condition may allow us to devise treatments.

Family News

Here are short updates from a couple of our MSS families. You can read more about our children at the website under "Family Stories."

Misa is a 16-year-old girl who attends high school in Torrance, California. She is enrolled in a mixture of special day classes and regular-ed classes. This year she is trying Japanese and Algebra 2 in regular-ed, so far without any modifications, but some may be needed. Misa is active in the local AYSO (American Youth Soccer Organization) VIP division, is taking piano lessons, and is in a therapeutic horseback-riding program. She also participates in judo classes, as this is a family activity. Misa no longer has any kinds of active therapy - physical, occupational or speech. She does go to counseling to deal with some of the issues dealing with her limitations. Misa has been on growth hormone for many years, but is still very small – 65 lbs and 54 inches.

Kimberly, 10, is now a 5th grader in Simi Valley, California where she attends a special day class. Her favorite activities are playing with Barbie, riding her bicycle with training wheels at the beach, going to movies, and horseback riding. She has recently made excellent progress in using forearm crutches, although her red, four-wheel walker is still her most reliable means of getting around at school. Kimberly is making nice progress in reading, and she is becoming much more independent all-

around. Speech remains the most challenging area for her, although she can talk up a storm when playing with her pet hamster, Sammy. Kimberly is 46 inches tall and weighs 40 pounds.

NORD Conference

<http://www.rarediseases.org>

By Colleen Yinger,
Mother of Kimberly

I attended the Annual Conference of the National Organization for Rare Disorders (NORD) in Arlington, VA, October 10-13, 2002. About 200 people attended. It was a great opportunity to meet a wide variety of people affected by “rare” disorders (i.e. any disorder with fewer than 200,000 cases).

My previous experience with NORD over the last eight years was limited to the disease reports, networking program, and newsletters. But I soon realized that the organization is a group of highly dedicated, politically active individuals with many connections within the rare-disease community.

The conference confirmed my impression that there are rare disorders, very rare disorders, and then there’s MSS! Many of the disorders represented had support groups with thousands of people. Even the most rare ones had rapidly increased their membership as soon as they developed websites and raised awareness of their diseases among physicians. The MSS support group has not seen that growth. On the positive side, however, we are fortunate to have geneticists studying our disorder. For many other groups, the most challenging part has been convincing doctors to take an interest in their disorder.

The conference included sessions on the human genome project and its impact on both common and rare disorders, the role of clinical centers at the National Institutes of Health (NIH), expansion of the NIH Office of Rare Disorders, developing and bringing an orphan drug to market, planning for the special needs child, fund raising for non-profits, and many others.

The NORD conference was held in conjunction with the national meeting of Gaucher Disease, a lysosomal storage disorder affecting the liver, spleen, and bones, for which enzyme replacement therapy is now available. It was an interesting opportunity to observe in-person the impact of medical progress on their younger members, to gain some insight into the political and financial challenges they face, and to anticipate the issues (drug cost, insurance coverage, availability of home health care, and others) that a growing number of people with rare disorders may encounter over the next few decades.

The conference was exciting and motivating. I look forward to attending again – perhaps with one of you.

Ideas for Improving Walking

Several families provided suggestions for improving walking abilities of our MSS children. These ideas presented are not a substitute for medical and professional therapeutic advice. Check with your physician or physical therapist to see what activities are appropriate for your child.

- Increase the amount of time your child stands independently. Have

him stand while watching a favorite video or TV show or playing a handheld video game.

- Encourage cruising (moving without a walker) around the house and in the classroom.
- Provide reduced support when walking: just a finger to hold onto or a light grasp on her clothing.
- Transition to forearm crutches as soon as possible after good mobility with a walker is achieved.
- Do activities to improve quadricep strength: stair and hill climbing, tricycle or bicycle riding. Standard bikes and training wheels don't work for your child? Try <http://www.UcanBike2.com>
- Consider therapeutic horseback riding. It's great for trunk stability and lots of fun, too.
- Work on activities that improve trunk strength and stability such as balancing on a large therapy ball.

Have more ideas for helping our kids? Send them to us for inclusion in future newsletter issues or the website.

Financial Contributions

MSS research is moving slowly because of a lack of funds. Any amount can help buy needed research supplies. One way to contribute to MSS research is to make your check out to Cedars-Sinai Medical Center and send it to Dr. Wilcox with a letter attached **designating the funds exclusively for MSS research**. Cedars-Sinai is a non-profit research hospital and your donations are tax deductible.

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Website Updates

The website was revised over the summer to provide a better format for printing and easier navigation. The site now has four family stories. Do we have your story and picture?

Brochures Available

Brochures summarizing MSS symptoms, diagnosis, inheritance, prognosis, and research goals are available. The brochures also publicize the MSS support group and website. The brochures are appropriate for physicians to distribute to newly diagnosed families, or for families to share with friends, relatives, and professionals. To obtain copies of the brochures, email us with your name, address, and the number of brochures you would like.

MSS Included in New Rare Disorders Book

Marinesco-Sjogren Syndrome will be included in the book "The NORD Guide to Rare Disorders" scheduled to be released soon. The 1000-page text, published by Lippincott, Williams, & Wilkins, is written for physicians and other medical professionals to encourage earlier diagnosis of rare disorders. William Wilcox, MD, PhD. wrote the MSS article.

Volunteer Opportunities

Interested in helping out with the MSS support group? Here are some ways you can participate.

Website: Submit your family story if you haven't already done so, write new articles, suggest additions or improvements, and notify us if you detect any problems with the site (e.g. broken links). You can even be the webmaster.

Fundraising: Encourage friends, relatives, service organizations, or private foundations to make a donation to MSS research. Hold a fundraiser in your community.

Newsletter: Write an article for the next newsletter (e.g. a feature on your family, activities that have helped your child, or what you learned at a recent conference). Or become the newsletter editor.

Membership: Keep in touch with other support group members. Make a special point of greeting and helping out new members.

Legal: Help the MSS support group become a corporation and official 501(c)(3) nonprofit organization.

Librarian: Maintain the library of medical journal articles on MSS. Periodically search for and acquire new articles.

crutches and wears braces? Check out the book "Franklin's School Play", published by Scholastic Inc, to read about Badger as the stage manager at the school Christmas play.

Recommended Reading

"Karen" by Marie Killilea is a well written, inspiring true story about a young girl growing up in the 1940's with cerebral palsy. Although opportunities for the disabled have improved since then, many of the challenges remain the same. Killilea describes them in a touching but uplifting way.

Trivia

Did you know that the character "Badger" in the Franklin series of children's books uses forearm