Marinesco Sjogren Syndrome NEWS

Summer 2003
Colleen Yinger, Editor

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About the Newsletter

Highlights of this newsletter include an update from Sylvain's family, Misa's experiences with full inclusion, ideas for adapting sports for the physically disabled, and a medical update on MSS and MSS-like conditions.

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 also available at the website on the publications page. http://www.marinesco-sjogren.org/pubs.html

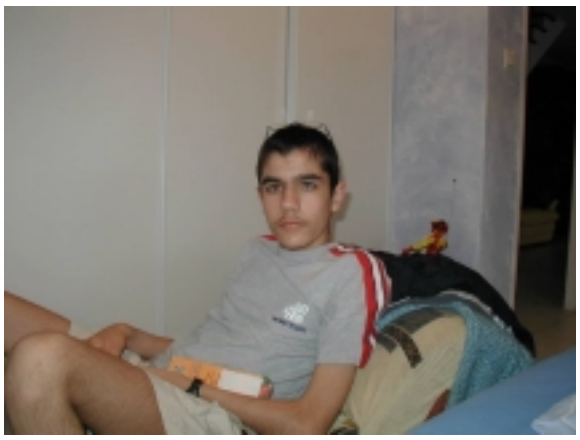
MSS Exhibit at ASHG Meeting

The MSS Support Group will be exhibiting at the American Society of Human Geneticists (ASHG) meeting at the Los Angeles Convention Center November 5-7, 2003. It will provide us an opportunity to raise awareness about MSS in the genetics community and to network with other support groups.

We encourage geneticists to visit our booth (#139 in the main exhibit hall). Families, please start thinking about pictures you would like to include in the exhibit.

ASHG meeting website: http://www.ashg.org/genetics/ashg/menu-annmeet.shtml

Family News: Update on Sylvain
by his parents



Sylvain, 17 years old, at his home in France

Sylvain is doing well. He is 17 years old, 1.57 m (62 inches) tall, 37 kg (81 lbs), and continues on growth hormone treatment.

He is working rather well at school even though he remains slow. He can walk by himself indoors, but needs help outside and when the ground is uneven. He wears and removes his contact lenses independently.

Sylvain loves video games, photography, reading, and swimming. He was third at the last French handicap national championship in his classification. He is a very happy young boy. His voice is changing into a young man (no hypogonadism). He has no idea of what he will want to do in the future, just like many teenagers of his age.

Read more about Sylvain and his younger years at the MSS website.
http://www.marinesco-sjogren.org/sylvain.html

Education: What About Full Inclusion?
by
Misa's mother

My daughter, Misa, is now 17 years old and has MSS. She is very small in stature, has low muscle tone, walks with forearm crutches, and has some speech and processing delays. She started off her education in the Los Angeles County program for the Orthopedically Handicapped (OH) at age 3. She remained in that program until the middle of first grade. At that time we were dissatisfied with the "mainstreaming" experience and the large age range in her OH class (6-11 year olds). We opted to have her fully included at our neighborhood elementary school.

We knew that the academics would be a struggle for Misa, but with her persistence and our help, we thought that she could be successful. We also thought that the social experience would be most beneficial for Misa. Misa's experience in elementary school was a very positive one. She was supported well by the teachers, resource staff and her peers. In hindsight, I realize that the friendship of one very special girl made this experience as close to normal as we could have hoped.

Middle school proved to be much more challenging and we opted to place Misa in the Special Day class for some of her academics. She continued to flourish academically, but without her special friend, Misa's social life was not happy. More people knew her name than anyone else on campus, but no one was truly her friend. Her size, maturity level and limited ability to participate in activities seemed to be barriers to connecting with her peers. We tried many outside activities to get Misa more involved and counseling to help her deal with some of her issues about being different. None of them seemed to make her happier. My feeling was that she needed a friend. Many avenues were explored at school, including a game club at lunch time, circle of friends and esteem building experiences like being a teacher's assistant and office assistant.

High school started out as more of the same. She knew lots of the kids but she didn't quite fit in. Being at a high school with the OH program on campus, we tried having her take one class with the OH kids. Misa didn't feel like she was one of them and opted out of that class when given the choice. Interestingly, this year, as a sophomore, she tried adapted PE with the OH class. She has befriended some of the kids in the OH class and has initiated some social outings and communications online and on the phone. It has become apparent to us that our choice to fully include Misa has somewhat excluded her from her natural peer group.

For academic reasons, I know that we made the right choice for Misa to be fully included. For social reasons, I question our decision. Misa is different from most of the other kids in many ways – size, physical ability, maturity level and many life experiences. Like most kids, she has found a niche where she feels most comfortable – with kids that are most like her.

I would like for other students to benefit from Misa's experience. I urge parents of children that are considering full inclusion or who already have fully included children to consider some of the negative social aspects of full inclusion. You can develop all the "circles of friends," buddy groups and social clubs that you want but you can't make another child be your child's friend. Those children may eat lunch with your child or play a game with them. That makes them a temporary companion, not a friend. Friends feel a connection to one another. Please make sure that your child's full inclusion experience is not fully excluding them from their natural peer group.

Kids on the Move: Sports and Recreation

Ready to try out a new sport? Several of our MSS kids have participated in some really fun activities. Here are ways in which some sports are adapted for the physically disabled. If you have a picture of your child participating in a sport, please send it to us so we can create a new "Kids on the Move" page on the MSS website. Always remember to use good safety sense and wear your helmet!

Cycling: Toddler seats for the smallest riders, bikes with sturdy training wheels (available on 16-inch and some 18-inch models), trailer bikes with handle bars and pedals (optional gears, too) that attach behind an adult bike, extra large tricycles, standard tandem bikes for near full-size riders, and hand bikes for wheelchair users.

Sidewalk scooter or "California chariot": These heavier scooters provide a more stable alternative to the typical lightweight scooter. *Editor's note: Has anyone tried a Segway?*

Snow skiing: Options include skis with outrigger poles or a sit-down style of ski. At least two members of our support group have been skiing and enjoyed it immensely.

Ice-skating: Some rinks have ice-skating "walkers" that children can use. Some facilities even allow the child to wear tennis shoes rather than skates on the ice.

Horseback riding: Therapeutic horseback riding lessons as well as hippotherapy (therapy on horseback) are popular across the country. Some programs offer "cart riding" for those that cannot ride on a horse.

Bowling: Many bowling alleys have ramps to launch the ball. Bumpers to prevent "gutter balls" are always helpful (even for some of us adults!)

Special Olympics and Therapeutic Recreation: Park and recreations districts offer a wide variety of sports programs for the disabled. Soccer, swimming, tennis, and softball are popular. Some areas offer VIP AYSO soccer and Challenger Little League Baseball programs.

Technical Publications

Recent technical publications on MSS and similar disorders include:

"Orthopaedic manifestations of Marinesco-Sjogren syndrome," Reinker, K., et. al.,

Journal of Pediatric Orthopaedics, May-June 2002.

"Linkage to 18qter differentiates two clinically overlapping syndromes: congenital cataracts-facial dysmorphism-neuropathy (CCFDN) syndrome and Marinesco-Sjogren Syndrome," C. Lagier-Tourenne, et. al., Journal of Medical Genetics, Nov 2002.

"MR Imaging Features in Marinesco-Sjogren Syndrome: Severe Cerebellar Atrophy Is Not an Obligatory Finding," Reinhold, A., et. al., American Journal of Neuroradiology, May 2003.

"Mutations in a Sar1 GTPase of COPII vesicles are associated with lipid absorption disorders," B. Jones, et. al., Nature Genetics, May 2003.

Medical update

by

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It is becoming clear that there is more than one type of MSS. Congenital cataracts-facial dysmorphism-neuropathy (CCFDN) has only been reported in gypsies in Europe, but could account for some of the other cases in the medical literature. My impression of these cases is that they have abnormal nerve function and tend to show deterioration in function with age. The gene maps to chromosome 18, but is not yet known.

MSS with chylomicronemia has abnormalities of the fats in the blood and vitamin E levels. People with this type do not gain weight well and may have diarrhea. The gene (Sar1) maps to chromosome 5. So far, all the MSS cases I know of have normal blood lipids and vitamin E levels. We have almost completed our examination of the Sar1 gene in every MSS case that we have samples on and have found no mutations. We are also looking for mutations in another candidate gene.

In addition to all these MSS-like conditions, I am aware that some patients with mitochondrial disorders can look like MSS. They have abnormal urine biochemical studies and generally elevated blood lactic acid.

Needless to say, the situation is very confusing because there is no gold standard test for MSS. It remains a clinical diagnosis. As soon as the genes for MSS and CCFDN are found we can begin to figure out much better what problems are associated with mutations in each gene and begin to think about specific therapies.

Books about Genetics

April 2003 marked the 50th anniversary of Watson and Crick's publication of the double helix structure of DNA and the celebration of the essential completion of the mapping of the human genome. Here is a sampling of recent books about genetics written for the lay reader.

"Cracking the Genome: Inside the Race to Unlock Human DNA" by Kevin Davies

"Genome: The Autobiography of a Species in 23 Chapters" by Matt Ridley

"The Human Genome, A User's Guide" by R. Scott Hawley and Catherine A. Mori.

Conferences

Genetic Alliance Conference, August 1-3, 2003, Arlington, VA. The Genetic Alliance

is a great source of information for lay advocacy groups (that means us!).
http://www.geneticalliance.org/members/conference.html

The National Organization for Rare Disorders (NORD) will not have a conference this year. Watch for the next one in 2004.
http://www.rarediseases.org

Medical Research Advocacy

Research!America is a national, not-for-profit, membership-supported public education and advocacy alliance working to make medical and health research—including research to prevent disease, disability and injury and to promote health—a higher national priority. Their website has updates on congressional funding for the National Institutes of Health (NIH). The site also provides suggestions for advocating for greater funding of medical research.
http://www.researchamerica.org/

Toll-Free Hotline For Air Travelers With Disabilities

The U.S. Department of Transportation (DOT) has established a toll-free hotline to assist travelers with disabilities. The hotline will provide general information to consumers about the rights of air travelers with disabilities, respond to requests for printed consumer information, and assist air travelers with time-sensitive disability-related issues that need to be addressed in "real time." The line is staffed from 7 a.m. to 11 p.m. Eastern time, seven days a week. Air travelers who experience disability-related air travel service problems may call the hotline at 1-800-778-4838 (voice) or 1-800-455-9880 (TTY) to obtain assistance.