

M Marinesco
S Sjogren
S Syndrome
NEWS

Fall 2004
Colleen Yinger, Editor

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About the Newsletter

This edition of the newsletter provides a family update, back to school educational ideas, and several resources for future planning for children with special needs. 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 available at the website on the publications page.

<http://www.marinesco-sjogren.org/pubs.html>

New Email Address

The MSS support group has a new email address. The website is unchanged. Please update your email addresses to contact us at: mss@marinesco-sjogren.org

Family Update: Kimberly

Kimberly is now 12 years old and just started 7th grade. She is taking a break from her therapeutic horseback riding lessons to play

soccer in the fall league for disabled kids (AYSO VIP). She can now play soccer comfortably with her forearm crutches. This is a big step from last year when she used her walker. We keep reminding her that in soccer you can't use your arms, and that includes crutches!



Kimberly, 12, plays soccer

We are excited about the gross motor progress Kimberly has made in just the past couple of years. Two years ago, it was very difficult for her to walk with crutches. Today, she walks confidently with them, and can walk rather long distances unassisted when adequately motivated. Chocolate cake anyone?

Kimberly's growth has been significantly faster the past few years (about 2 to 2 ½ inches each of the last three years). She is now 50 inches and weighs about 55 lbs. She has had both of her cataract surgeries and is doing well with IOLs and glasses (bifocals) to correct some remaining nearsightedness. She looks much more robust than she did as a little girl. Her hair, which used to be extremely thin, is now full. Her teeth continue to be a problem (no enamel) and will eventually require extensive dental work (not characteristic of MSS). Otherwise, she is very healthy and progressing in all areas.

New MSS Technical Publications

A.-K. Anitonen, I. Mahjneh, B. Udd, H. Somer, M. Somer, A.-E. Lehesjoki, "Refining the Marinesco-Sjogren syndrome locus on 5q31", ASHG meeting poster paper, October 2004. <http://www.ashg.org/genetics/ashg04s/index.shtml>

Hurricane Damage to Southeast US

The southeast region of the United States, particularly Florida and Alabama, suffered extensive wind and flood damage in the hurricanes in September. We send our thoughts to any of our MSS families and their friends and relatives who may have been affected by the severe storms in those parts of the country.

International MSS Research

An international collaborative group is closing in on the gene for MSS. More families with MSS are needed. If you want to help with the effort, please contact:

William Wilcox, MD, PhD
(310) 423-6673
William.Wilcox@cshs.org

Back to School: Reading, Writing, and Keyboard Hints

For many of us, the new school year recently started. Some of our MSS children have difficulties with reading, writing, or keyboarding. Here are a few suggestions that might help.

Reading:

- (1) Turn on captions on the TV screen so they see the written words along with the voice.
- (2) Try books on tape. If the reading is too fast, consider taping your own versions of books, preferably on a variable-speed tape recorder so you can adjust the speed as their skills improve.
- (3) Tape record your child's reading so they can listen to themselves read and hear their progress.
- (4) Try using a "slot card", a piece of lightweight cardboard with a slot cut just the right size to include one line of text. Use it to uncover one line of the book at a time to help focus and tracking.
- (6) Look in educational catalogs or teachers' stores for classic books written at easier reading levels. (www.acadcom.com, www.pcieducation.com)
- (7) Visit the library frequently.
- (8) Flashcards can be fun. (Editor's note: A friend of mine reported that her daughter likes to teach Barbie to read using flashcards. Then one day, we found Kimberly trying to teach her pet hamster. Hamster still isn't progressing too well.)

Transition to Adult Health Care

Writing:

- (1) Work with clay or squeeze squishy balls to improve hand strength.
- (2) Put colored salt or baking powder in a shallow pan and practice letter and word writing.
- (3) Practice large letters on a white board.
- (4) Try using stencils to control hand motion.
- (5) Write in clay to add resistance.
- (6) Have fun with “rainbow writing” – writing a name or word in many colors on top of each other.
- (7) Use larger pencils for better control.

Keyboarding:

- (1) Investigate keyboards with larger size keys, available in both “ABC” and QWERTY” formats. Infogrip (www.infogrip.com) is a good source for assistive technology.
- (2) Consider a key guard to reduce the likelihood of multiple key hits. Since they are custom made for a particular keyboard, they tend to be expensive (close to \$100).
- (3) Adjust the character repeat rate on your computer (Windows: my computer, control panel, keyboard) so holding a key down longer than normal does not generate multiple letters.
- (4) Many programs for improving keyboarding skills and word processing are available. Don Johnston’s “Write:OutLoud” (available at www.infogrip.com) has been recommended as an excellent word processing program. The following is a list of some typing programs that I have heard about, but I have no specific experience with any of them.

Type for Fun

Type to Learn, Type to Learn Jr.

Jumpstart Typing

Mavis Beacon Teaches Typing

AVKO Systems Individualized Keyboarding

Pumba and Timon

Mario Brothers

A free supplement, “Improving Transition for Adolescents with Special Health Care Needs from Pediatric to Adult-Centered Health Care,” is available from the American Academy of Pediatrics. The publication contains articles addressing health care transition for young adults with complex chronic health conditions and disabilities. Complementary copies can be obtained by completing an electronic form at the website:

<http://aappolicy.aappublications.org/cgi/content/full/pediatrics;110/6/S1/1304>

Family Handbook on Future Planning

A Family Handbook on Future Planning is a guide to help families develop a future plan for their sons or daughters with cognitive, intellectual or developmental disabilities that provides personal, financial and legal protections for these individuals after the parents either die or can no longer provide care or support. The handbook contains chapters on financial planning, trusts, guardianship, wills, and government benefits, and is available at the website:

<http://www.thearc.org/publications/futureplanninghandbook.doc>

Financial Planning for Individuals with Special Needs

“Financial Firms Focus on Parents of Disabled” appeared in the Wall Street Journal on August 26, 2004. It discusses financial planning for individuals with special needs and provides resources for families. It also discusses the Qualified Disability Savings Plan, to be presented to the President within the next two months, which would allow money to be saved

in tax-advantaged accounts for disabled children.

<http://webreprints.djreprints.com/1060361049008.html>

Open Access to Medical Research

The National Institutes of Health is developing policy guidance that would require that final peer-reviewed manuscripts of NIH published research be placed in PubMed Central, the digital library maintained by the National Library of Medicine, within six months after publication in a scientific journal. Language promoting this direction is included in a report accompanying a House Appropriations Committee bill.

Open access supporters say it's only right that taxpayers be able to read research articles that their dollars help fund without paying \$20 to \$30 to read an individual article or the pricey subscriptions to the scientific journals. The Association of American Publishers says that the legislation would threaten the continued survival of many scientific, scholarly and medical publications and professional societies.

Genetic Alliance Conference

The annual Genetic Alliance conference was held July 23-25, 2004, at the Key Bridge Marriott in Arlington, VA. Over 20 workshops were offered, as well as a plenary presentation by author Steve Olson - "Stories from the Genome", a discussion about outreach to underrepresented communities, and stories of how advocates have been effective in overcoming obstacles with innovative efforts. The next conference will be held July 29-31, 2005 in Rockville, MD.

Upcoming Conferences

NORD (National Organization for Rare Disorders) Conference, October 15-17, 2004, Bloomington, MN.

<http://www.raredisorders.org>

ASHG (American Society of Human Genetics) Annual Meeting, October 26-30, 2004, Toronto, Ontario, Canada

<http://www.ashg.org>

Family History Tools

ASHG is collaborating with the National Society of Genetic Counselors and the Genetic Alliance to develop a national awareness campaign on the importance of family history information and to encourage all Americans to gather their family history information, including health history. The website describes how to collect and record a family history.

<http://www.ashg.org/genetics/ashg/educ/007.shtml>

Two Molecular Biology Pioneers Die

Francis Crick, co-discoverer of DNA, died in July. He was 88. Crick's discovery with James Watson in 1953 laid the basis for the science of molecular biology.

Maurice Wilkins, whose X-ray images of the structure of molecules were instrumental in the discovery, died in October, also at the age of 88.

Watson, Crick, and Wilkins won the Nobel Prize for Medicine in 1962 for describing the structure of DNA.