

M Marinesco
S Sjogren
S Syndrome
NEWS

Fall 2007
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About the Newsletter

In this edition of the MSS newsletter, we highlight our successful exhibit at the recent genetics conference in San Diego, including the posters we displayed. Phil Conway shares information about his new venture for helping disabled children in the United Kingdom.

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

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.

ASHG Annual Meeting

Colleen Yinger hosted an MSS exhibit at the American Society of Human Genetics (ASHG) meeting on October 24-26, 2007 in San Diego, California.

The booth provided MSS posters, brochures, and medical summaries to clinical geneticists all over the world – US, Mexico, South America, Europe, Asia, and the Middle East. The attendees enjoyed seeing MSS pictures from many ethnic backgrounds, including the opportunity to compare parents and siblings with their MSS family members.

Many researchers were interested in contrasting symptoms of MSS with other diseases, learning about the Sil1 gene, and sharing information about mouse models. We interacted with several representatives from the National Institutes of Health (NIH) Office of Rare Disorders. The conference also allowed us to network with a wide variety of disease support groups, including groups representing hereditary ataxias, muscular dystrophy, and developmental disabilities.

Thank you to the nine families who provided pictures for the display (see last two pages of the newsletter). The pictures included MSS families from six countries and were very well received. Other disease support groups were enthusiastic about how we captured the energy of the children in their sports and other activities. Also, we say thank you to one of our families for their generous financial support for the conference exhibit.



Colleen Yinger Staffs the MSS Booth

We would appreciate feedback from physicians and researchers on how we can use conferences more effectively. Is exhibiting at ASHG or ACMG meetings more productive? What conferences are you most likely to attend? Would you consider participating in a one-day MSS research conference if it were held just prior to an ASHG or ACMG (or some other) meeting? Would an exhibit at a pediatric neurology conference promote improved/earlier MSS diagnosis? What European conference is best suited to an MSS exhibit? Please email us with your suggestions.

Glaucoma monitoring

Children who have cataract surgery, particularly when they are very young, are at increased risk for developing glaucoma. Glaucoma can lead to blindness by damaging the optic nerve and

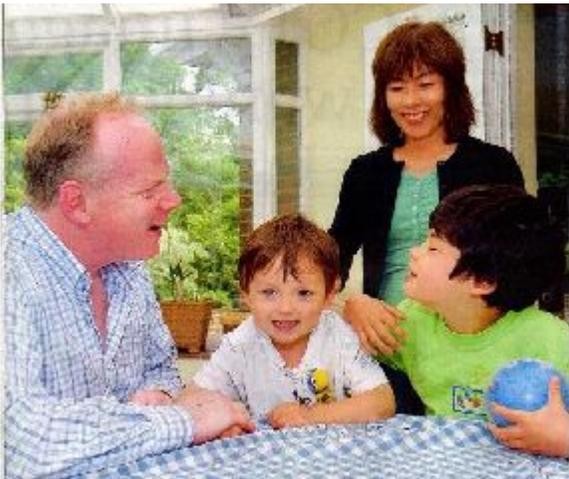
usually involves elevated eye pressures. Measuring pressures can be difficult in young or non-compliant patients. Some of our families report that measuring pressures is easier when the ophthalmologist uses the following tonometer by iCare. See your eye care professional for additional information. http://www.tiolat.fi/overview_icare.htm

MSS and Related Technical Publications

- Online Mendelian Inheritance in Man (OMIM), Description of the Sil1 gene and all reported variants that cause MSS. <http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=608005>
- Weitzmann, A., et al., "The heat shock protein 70 molecular chaperone network in the pancreatic endoplasmic reticulum - a quantitative approach", FEBS Journal, http://www.ncbi.nlm.nih.gov/sites/entrez?Db=pubmed&Cmd=ShowDetailView&TermToSearch=17850331&ordinalpos=1&itool=EntrezSystem2.PEntrez.Pubmed.Pubmed_ResultsPanel.Pubmed_RVDocSum
- Morano, Kevin A., "New tricks for an old dog: The evolving world of Hsp70", full article on-line. http://www.stress07.com/binx/morano_1b.pdf
- Shaner, L, and Morano, K., "All in the family: atypical Hsp70 chaperones are conserved modulators of Hsp70 activity", March 2007, full article on-line. <http://www.pubmedcentral.nih.gov/articlerender.fcgi?artid=1852889>
- Okada, M, et al., "Rimmed vacuoles in children: Highly specific indication for SIL1 mutation in Marinesco-Sjogren syndrome", World Muscle Society, Neuromuscular Disorders Journal, October 2007. <http://journals.elsevierhealth.com/periodicals/NMD>

Cool2Care

Phil Conway, father of Shaun (as included in the family section of the MSS website), is starting a new venture in the United Kingdom to help families with disabled children. "Cool2Care" will help families by recruiting, training and placing care workers into families with disabled children. The lack of skilled help is a major problem in the UK. Phil & Keiko searched for 2 years before finding Helena, their current care-worker from Poland.



Phil, Adam, Keiko, and Shaun

"It was a very difficult time for us" says Phil. "We knew we needed help with Shaun to get on with our lives, but we couldn't find anyone. Now that we have regular and structured help 5 days a week, funded by our local government authority, life is much easier".

Cool2Care is a non-profit Community Interest Company based in UK. It has just started hiring staff to find and train care workers, and market the service to families. It is aiming to raise £100,000 in its first year and has launched an appeal to help raise funds. If anyone is interested in donating to this worthy cause, please visit the website www.cool2care.co.uk where there is further information and a secure way to make a donation online.

Ski Season is Here

The ski season is here – at least for those of us in the northern hemisphere! Search the internet for “adaptive skiing” or visit the website <http://www.sitski.com/pg3.htm> for adaptive ski and sport programs in the US, Canada, and Europe. Here are two organizations that provide adaptive skiing lessons in southern and northern California.

- www.usarc.org - Lessons available at Big Bear Mountain, north of Los Angeles.

- www.disabledsportseasternsierra.org - Lessons available at the Mammoth Mountain ski area in northern California.

Special Needs Alliance

The Special Needs Alliance is a national, non-profit organization helping individuals with disabilities, their families, and the professionals who represent them. The alliance includes disability law attorneys, many who are parents and siblings of children with disabilities. Their website includes useful information about special needs trusts, financial planning, conservatorship, and other aspects of special needs legal planning.

<http://www.specialneedsalliance.com/>

Tammy's Blog

Tammy Lau's blog is available at: <http://welcometotammylaublog.blogspot.com/> Check it out to see pictures of her 28th birthday celebration and her recent vacation to the mainland.

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Clinical Diagnosis

- Cerebellar ataxia
- Cataracts in early childhood
- Muscle weakness
- Mental retardation

(moderate to near-normal)



Common Features

- Short stature (post-natal)
- Hypergonadotropic hypogonadism
- Cerebellar atrophy
- Skeletal abnormalities



Prognosis

- Progressive weakness in adulthood
- Life expectancy near normal



M S S Marinesco Sjogren Syndrome



Inheritance and Incidence

- Autosomal recessive
- Pan-ethnic, but very rare
- Over 200 cases reported

Pathogenesis

- Sil1 mutations in about 50% of cases
- Co-chaperone of BiP
- Accumulation of misfolded proteins



Animal model

- Woozy mutant mouse
- Ataxia and degeneration of Purkinje cells