Marinesco Sjogren Syndrome NEWS

Winter 2004
Colleen Yinger, Editor

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About the Newsletter

This edition of the newsletter focuses on our experiences at the American Society of Human Geneticists (ASHG) meeting, plus a variety of other topics relevant to the disabled community. 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

ASHG Meeting

The MSS support group exhibited at the American Society of Human Geneticists (ASHG) Meeting at the Los Angeles Convention Center, November 5-7, 2003. Our poster and handouts (see pages 4,5) helped raise awareness about MSS, and we identified some geneticists that might have patients with MSS. Some very positive steps were taken towards international collaboration on MSS research.

We'd like to thank the Lau family for their generous donation towards the exhibit expenses. A special thanks to Misa's mother, Chrissy, for helping to staff the booth. Thank you to Kimberly's Uncle Art for the wonderful poster artwork.

Several participants at the meeting pointed out that the colors of our MSS logo (red, yellow, and blue) are the colors of the Romanian flag. It turns out that our selection of logo colors is a fitting tribute to Dr. Marinesco, the highly respected Romanian neurologist after whom Marinesco-Sjogren Syndrome is named.

Colleen Yinger and Chrissy Kaichi staff the MSS exhibit at the 2003 ASHG Meeting in Los Angeles.

New Technical Publications

The following papers about MSS and related disorders have been published since our last newsletter.

Varon, R., Gooding, R., et. al., "Partial deficiency of the C-terminal-domain phosphatase of RNA polymerase II is associated with congenital cataracts facial dysmorphism neuropathy syndrome," Nature Genetics, Vol. 35, No. 2, October 2003.

Support Group Contact Information

Do we have your most current address, phone, and email address? If not, please drop us a line with your latest information. We do not give your personal information to anyone without your explicit permission. But maintaining your contact information allows us to provide you with the latest MSS news and research and inform you about new families that would like to correspond.

You can contact us by email (marinesco-sjogren@pacbell.net), through the website (www.marinesco-sjogren.org), or by mail:

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Genetics: Policy and Ethics

The United States Senate approved S.1053, the Genetic Information Nondiscrimination Act on October 14, 2003 by a vote of 95-0. The bill provides protection against discrimination on the basis of genetic information in the issuance of insurance and the setting of premiums, and prevents misuse of genetic information in the workplace. It is now up to the House to pass the bill and the President to sign it.

The National Human Genome Research Institute (NHGRI) website provides a good summary of policy and ethics issues related to genetic research.
http://www.genome.gov/PolicyEthics/

For a fun website about genetics, genetic disorders, gene therapy, and more, visit
http://gslc.genetics.utah.edu/

The Public Broadcasting System (PBS) in the United States recently aired a five-part television special on DNA. Their website contains summaries of the five episodes as well as a good DNA resource list.
http://www.pbs.org/dna

Upcoming Meetings

The next Annual Genetic Alliance Conference will be held July 23–25, 2004 at the Key Bridge Marriott in Arlington, VA (close to National Airport). The Genetic Alliance is an international coalition comprised of millions of individuals with genetic conditions and more than 600 advocacy, research and health care organizations that represent their interests. The Genetic Alliance also maintains a listserv that discusses many issues important to lay advocacy organizations like ours.
http://www.geneticalliance.org

The 2004 Annual Meeting of the Child Neurology Society (CNS) will be held October 13-16 in Ottawa, Canada.
http://www.childneurologysociety.org/

The 2004 Annual Meeting of the American Society of Human Geneticists (ASHG) will take place October 26-30 in Toronto, Canada.
http://www.ashg.org

US Presidential Candidates 2004

The National Organization on Disability website provides links to United States presidential candidate online statements about disability-specific issues.
http://www.nod.org/election2004.html

California State Long-Term Care Tax Credit

California allows a $500 long-term care credit (per applicable individual) against net income tax that can be used by eligible caregivers. Long-term care needs must be certified by a physician, and are generally defined as requiring substantial assistance to perform activities of daily living. No credit is allowed for any caregiver whose adjusted gross income exceeds $100,000. Some other restrictions may apply, so ask your accountant or tax attorney for advice. The credit is obtained by submitting California Form 3504.
http://www.ftb.ca.gov/

National Park Service Golden Access Passport

The United States National Park Service provides a free golden access passport for citizens or permanent residents of the United States who are blind or permanently disabled. The Golden Access Passport is a lifetime entrance pass to national parks, monuments, historic sites, recreation areas, and national wildlife refuges that charge an entrance fee. The Golden Access Passport admits the pass signee and any accompanying passengers in a private vehicle if a park has a per vehicle entrance fee. Where a per-person entrance fee is charged, the Golden Access Passport admits the pass signee, spouse, and children. The Golden Access Passport also provides a 50% discount on federal use fees charged for facilities and services such as camping, swimming, parking, boat launching, and tours.
http://www.nps.gov/fees_passes.htm#goldenaccess

The following poster was displayed at our exhibit at the November 2003 ASHG Annual Meeting.

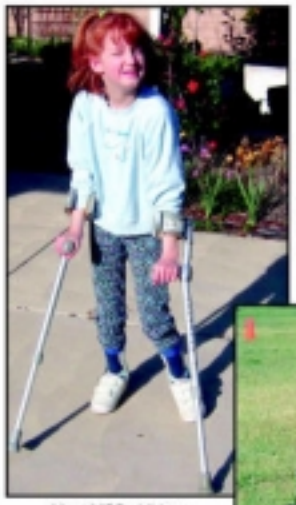
Marinesco Sjogren Syndrome

Clinical Diagnosis:

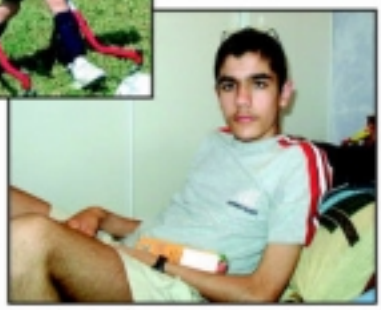
- Cerebellar ataxia
- Cataracts in early childhood
- Muscle weakness
- Very short stature (post natal)
- Mental retardation: moderate to near normal

Other common features:

- Hypergonadotropic hypogonadism
- Cerebellar atrophy
- Skeletal abnormalities
- Progressive weakness in adulthood



Most MSS children use walkers or forearm crutches.



Cataracts usually appear in early childhood, often suddenly.

Four MSS cases from three families: ages 22, 11, 17, and 23.

Two siblings exhibit very short stature typical of MSS.

Inheritance and incidence:

- Autosomal recessive
- Gene maps to 5q31
- Pan-ethnic, but very rare
- Over 100 cases reported
- Life expectancy near normal

The following summary was distributed at our exhibit at the November 2003 ASHG Annual Meeting and is also on our website.

Marinesco-Sjögren Syndrome

Marinesco-Sjögren Syndrome (MSS, OMIM 248800) is a rare, autosomal recessive disorder featuring cataracts, cerebellar ataxia, mental retardation, muscle weakness, short stature, and frequently hypergonadotropic hypogonadism.

MSS is usually evident at birth because of hypotonia. The cataracts are often not present at birth but may appear rapidly during childhood. Motor milestones are significantly delayed with ataxia becoming noticeable by the time the child can sit. Most patients are eventually able to ambulate with a walker. Linear growth is poor and pubertal development may not occur because of hypergonadotropic hypogonadism. Mental retardation is generally mild to moderate in severity if it is present at all. Dysarthria is common. Neurologic deterioration is slow to absent and prolonged survival is possible, but the muscle weakness may be progressive in adulthood. Less commonly reported features include optic atrophy, brachydactyly, and cone epiphyses.

MSS is inherited as an autosomal recessive trait with complete penetrance in both sexes. The genetic defect is currently unknown, but the gene has been mapped to chromosome 5q31. Over 100 cases have been reported. It is panethnic, but very rare except in genetic isolates, such as one in rural Alabama.

The diagnosis should be suspected based on the clinical symptomatology. An ophthalmologic exam (cataracts) and magnetic resonance imaging of the brain (cerebellar atrophy particularly involving the vermis) can be helpful. Muscle biopsy findings are generally non-specific, although ragged red fibers and abnormal mitochondria have been reported. Multilamellar inclusions can be present in muscle and conjunctival biopsies as well as in cultured fibroblasts. Metabolic testing is normal.

Treatment is supportive and based on symptomatology. Removal of the cataracts and placement of an artificial lens implant is often required to preserve vision. Physical and occupational therapy, special education, and computers are essential given their visual and motor problems as well as dysarthria. Hormonal replacement therapy is needed if hypogonadism is present.

The differential diagnosis includes: MSS with chylomicronemia (607692), congenital cataracts, facial dysmorphism, and neuropathy (604168), mitochondrial disorders, and the carbohydrate deficient glycoprotein syndromes.

For more information contact the Marinesco-Sjögren Syndrome Support Group (http://www.marinesco-sjogren.org) or Dr. William Wilcox, Cedars-Sinai Medical Center, 8700 Beverly Blvd., SSB-3, Los Angeles, CA 90048, telephone (310) 423-6673, email william.wilcox@cshs.org.