Marinesco Sjogren Syndrome

NEWS

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About the Newsletter

Highlights of this newsletter include an ophthalmologist's overview of cataracts in MSS, a report on our first MSS family meeting, and an introduction to a new family from Brazil.

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 will be exhibiting at the American Society of Human Geneticists (ASHG) Meeting in Los Angeles, November 5-7, 2003. Look for us at booth #139 in the exhibit hall.

Cataracts in Marinesco-Sjögren Syndrome

Kevin M. Miller, M.D.
Professor of Clinical Ophthalmology
Jules Stein Eye Institute
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Cataract is a clouding of the normally clear crystalline lens of the eye. The root word for cataract actually translates "waterfall." In advanced cases of cataract development, the lens can turn completely white and look like rushing water.

Cataracts are typically associated with the aging process. Almost everyone develops visually significant cataracts that necessitate surgery at some point in life.

When children develop cataracts, it is often because of genetic disorders. Autosomal dominant cataracts are fairly common. Autosomal recessive cataracts also occur. The specific gene defect for most inherited congenital cataracts is unknown, although several genes have been worked out. The

specific defect in Marinesco-Sjögren syndrome is unknown.

Cataracts have no medical treatment. If they are visually significant, surgery is indicated. When are they considered visually significant? This is not always easy to answer. Watching a child's behavior and seeing whether they are visually attentive is one way. If a child seems to lose visual ability as years pass by, cataracts should be suspected. Ultimately, it takes an ophthalmologic examination to determine the extent of cataract development. If an ophthalmologist has a difficult time seeing the back of the eye or if visual acuity is inappropriately reduced, cataract surgery may be indicated.

Cataract surgery can be performed at any age, including shortly after birth. The decision to perform cataract surgery is generally easy to make. The decision to implant an intraocular lens, however, can be complicated. Generally speaking, ophthalmologists are more prone to implant a lens after the age of two or three. By this time the eye has reached nearly adult proportions. Before then, however, the eye grows rapidly and the lens implant power calculation is difficult. If a lens is selected that has an appropriate power for the eye at age two, the eye will continue to grow and the lens power will be too high by age ten. Therefore, the child will become very nearsighted. Before age two or three, most ophthalmologists prefer to leave the eye aphakic, that is, without an implant, and correct the eye with a contact lens or glasses. A lens implant can be put in once the child is older.

In most instances, cataracts develop in both eyes at about the same time, although one cataract might be slightly worse than the other. When only one eye has a visually significant cataract, it is important to remove the cataract early so that the eye does not develop amblyopia. This is a condition in which the optic nerve fails to establish a good connection with the brain.

In the case of the patient with whom I had experience, the cataracts were of the posterior polar variety. This is a particularly difficult type of cataract to remove because the lens capsule on the back side of the cataract is congenitally absent. This means that, as soon as the cataract is removed, the posterior capsule can tear wide-open leading to vitreous loss and difficulty implanting an intraocular lens. An ophthalmologist who performs the surgery on patients with posterior polar cataracts should be comfortable handling the potential complications.

Once a cataract has been removed, it is important for the ophthalmologist to follow the child on a regular basis to optimize glasses and/or contact lens prescriptions and to treat amblyopia, if present. If a child is left aphakic at a young age, they may be considered for secondary lens implant surgery when they are six or seven years of age or older.

There are no significant long-term problems associated with cataract surgery other than the possibility of posterior capsule opacification, and this is not an issue if the capsule is congenitally absent. If the capsule remains intact at the time of surgery and becomes cloudy in the subsequent months or years, it can be opened using a neodynium:YAG laser.

Technical outcomes of cataract surgery usually are excellent with current techniques and lens implants. What makes for a good visual outcome, however, is early detection and early intervention. Parents or caregivers have to be alert to the possibility of cataract, and an ophthalmologist should be involved at

an early stage to assess the impact of cataract on visual development.

First MSS Family Meeting

The first meeting of the MSS family support group was held on August 25 in the Los Angeles area. Three families with four MSS children attended. Other families participated by telephone.

Family members took advantage of the opportunity to discuss medical issues, mobility, education, job opportunities for the disabled, and financial planning.

William Wilcox, MD, PhD, gave an overview of the status of MSS research at Cedars-Sinai Medical Center in Los Angeles. He reported that the location of the MSS gene is known to within four to six million base pairs. Blood samples from more families are needed to help map the gene more precisely. Identifying the gene is important for doing accurate diagnosis, allowing prenatal diagnosis, and understanding the physiology of the disorder (perhaps eventually leading to treatment).

Isaac (22), Kimberly (11), Misa (17), and Tammy (23) at the MSS family meeting

Diagnosis is currently based only on symptoms (a clinical diagnosis). Some other disorders reported in the literature may look similar to MSS. These include CCFDN (congenital cataracts, facial dysmorphism, neuropathy syndrome), MSS/CMRD (chylomicron retention disease) and some mitochondrial disorders. Cataracts tend to appear later (not as infants) in MSS. Children with MSS generally improve physically and academically throughout childhood. Progressive weakness may occur in adulthood. Life expectancy is close to normal. Dr. Wilcox described cultured skin cells from several families suggesting that MSS may be a lysosomal storage disorder.

Family News: Introducing our Newest Family

Raphaela (13) and Roberto (23) with their mother

We welcome Raphaela (13), Roberto (23), and their family to our MSS support group. Raphaela and Roberto live with their parents, Regina and Renato, and older brother, Rodrigo, in Brazil.

Roberto developed cataracts at the age of three years. As a child he had low tone, balance problems, and used a walker. In his teenage years he required surgery for scoliosis (curvature of the spine) and foot problems. Roberto now walks a little around the house and uses a wheelchair for traveling long distances. He receives physical therapy three times weekly. He is 1.57 meters (62 inches) tall. He completed high school, currently takes a computer science course, and is trying to decide what to do with his future.

Raphaela's cataracts appeared at four years of age. She used a walker when she was young, but now walks almost independently, holding hands for longer distances. She is about 1.30 meters (51 inches tall). Raphaela receives physical therapy three times weekly for strength, flexibility, and motor coordination, hoping to prevent the scoliosis that affected her brother. She is making progress physically and academically. Neither Raphaela nor Roberto has hypogonadism.

Visit the family stories page of the MSS website to see more pictures of the family. http://www.marinesco-sjogren.org/family.html

MSS Support Group: Who Are We?

Since the creation of the MSS support group and website three years ago, a total of 20 families have contacted us indicating their child has been diagnosed with or is suspected to have MSS. Nine of the families have more than one affected child. Eleven families reside in the United States. Nine families live in

seven other countries: United Kingdom, France, Italy, Austria, Pakistan, Canada, and Brazil. We are quite an international group! Ages range from three years to over thirty.

Since initially contacting us, a few families have received revised diagnoses. Some of the other children do not have all of the symptoms that are considered characteristic of typical MSS: cerebellar ataxia, childhood cataracts, very short stature, and some degree of mental retardation. We look forward to the day when the identification of the MSS gene will allow timely, definitive diagnosis of MSS.

New MSS Publications

Chudley A., "Genetic landmarks through philately: Georges Marinesco (1863-1938)", Clinical Genetics, 2003 Oct; 64(4): 297-299.

Lagier-Tourenne, C, et.al., "Homozygosity Mapping of Marinesco-Sjogren Syndrome to 5q31", European Journal of Human Genetics, 2003 Oct;11(10):770-778.

Regional Workshop for Rare Disease Organizations

The second regional workshop sponsored by the Office of Rare Diseases (ORD) of the National Institutes of Health (NIH), entitled "Gaining Access to Research Resources", will take place on November 7-9, 2003 at the Argonaut Hotel, 495 Jefferson St., in San Francisco. This workshop is for leaders of national patient support organizations focusing on a rare disease.

The organizations should be located along the West Coast or in a northwestern or southwestern state. Similar meetings will be held in other parts of the country in 2004.

Partial travel support can be provided. For information, call Jean Campbell, NORD's Vice President for Development, at (203) 744-0100, ext. 211, or Dr. Stephen Groft, PharmD, Director of the Office of Rare Diseases at the NIH at (301) 435-6041.

Moving from Activity to Connections

Some problems that many of our MSS children face are establishing close friendships and finding a role in life once the school years are completed. The following website presents some interesting ideas about how to move from individual activities designed to keep people occupied towards deepening connections, meaningful relationships, and community contributions.
http://www.communityworks.info/

Insurance Problems?

Consumers in the United States can request the assistance of their State Insurance Commission online in resolving insurance issues. The National Association of Insurance Commissioners has a web site that can assist consumers in various ways. One section allows consumers to click on their state and directs them to the complaint form for the Insurance Commission in their state. Go to http://www.naic.org/cis/fileComplaintMap.do and follow the links to your state.

Looking Ahead: Genetic Alliance Conference

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). Read more about the role of the Genetic Alliance at: www.geneticalliance.org.