

Genetic Study for Chiari with or without Syringomyelia

The Duke Molecular Physiology Institute (DMPI), formerly the Duke Center for Human Genetics, is actively recruiting families who have two or more family members with Chiari type 1 malformation, with or without syringomyelia. These family members must be related to each other by blood, and BOTH must be willing to participate.

Study participation involves these steps:

- Contact the study coordinator.
- Provide blood samples from family members both with and without Chiari malformation.
- Answer questions about family and medical history.
- Complete a medical questionnaire.
- Review of medical records and MRIs to confirm the diagnosis of Chiari malformation.
- Potentially ask other family members (parents, siblings, children) to participate in the study.

If your family meets these criteria and is willing to participate in the study, please contact the study coordinator.

CM1/S Study Coordinator:

Heidi Cope, MS, CGC
E-mail: chg_chiari@dm.duke.edu
Phone: (919) 684-0655

There must be those among us whom we can sit down and weep and still be counted as warriors.

-Adrienne Rich

Providing Research, Education and Support since 1988



Registration Is Open For The ASAP Chiari & Syringomyelia Conference

Long Island, NY

July 19-23, 2017

Conference Speakers

The following speakers have agreed to participate in the ASAP annual conference:

Richard Anderson (Columbia)
Allison Ashley-Koch (Duke)
Marat Avshalumov (IOM at NSPC)
Ulrich Batzdorf (UCLA)
Ghassan Bejjani (UPMC)
Paolo Bolognese (Chiari NSC)
Andrew Brodbelt (Liverpool, UK)
Marco Castori (Rome, Italy)
Palma Ciaramitaro (Turin, Italy)
Brian Dlouhy (Univ. Iowa)
Robert Duarte (NYS Pain Society)
Neil Feldstein (Columbia)
Clair Francomano (Harvey Inst.)
David Frim (Univ. Chicago)
Lorenzo Genitori (Florence, Italy)
Timothy George (Dells' Children)
Atul Goel (Mumbai, India)
David Goldstein (NIH)
Rodney Grahame (London, UK)
Gerald Grant (Stanford)
Dan Heffez (Chiari Institute, WI)
Ian Heger (MCG)
John Heiss (NIH)
Fraser Henderson (Metro NSG Group)
Bermans Iskandar (Univ. Wisconsin)
Robert Keating (Washington Children)
J Klekamp (Quakenbruck, Germany)
Petra Klinge (Brown)
Ilzumi Koyanagi (Hokkaido, Japan)
Roger Kula (Chiari NSC)
Lance LaCerte (Aurora, CO)
Jorge Lazareff (UCLA)
David Limbrick (Wash Univ. St Louis)

Yung Liu (Beijing, China)
Kenneth Liu (UVA)
Yongli Lou (Zhengzhou, China)
Tina Loven (Mercy Clinic, MO)
Cormac Maher (Univ. Michigan)
Anne Maitland (Mt. Sinai)
D Marino (Canine Chiari Institute)
Bryn Martin (Univ. Akron)
Arnold Menezes (Univ. Iowa)
John Mitakides (Beavercreek, OH)
Alon Mogilner (NYU)
Kevin Muldowney (Rhode Island)
Misao Nishikawa (Osaka, Japan)
Ed Oldfield (UVA)
Fabrice Parker (Paris, France)
Harold Rekate (Chiari Institute, NY)
Brandon Rocque (UAB)
Peter Rowe (Johns Hopkins)
Juan Sahuquillo (Barcelona, Spain)
Francesco Sala (Verona, Italy)
Wouter Schievink (Cedars Sinai)
Victor Shakhnovic (Moscow)
Abe Shulman (SUNY Brooklyn)
Konstantin Slavin (Univ. Illinois)
Mark Souweidane (Cornell)
Marcus Stoodley (Sidney, Australia)
Charles Tator (Toronto, Canada)
Laura Valentini (Milan, Italy)
Monica Wehby (Portland, OR)
Nicholas Wetjen (Mayo Clinic)
Jeffrey Wisoff (NYU)
Shokei Yamada (Loma Linda, CA)



A Word From Our President

This year is flying by! Spring is around the corner and we have lots of excitement on the horizon for ASAP and the CM/SM community. We are not slowing down anytime soon. The spring fundraising season is upon us, with multiple events taking place between now and the summer. In addition to this, we are gearing up for the national conference. We are

busy writing letters for sponsorship, auction items, raffle items, welcome bag goodies. The work seems endless but it is so worth it. ASAP does all this with two full-time employees and a handful of volunteers. If anyone is interested in helping us obtain auction or raffle items or sponsors, please let us know.

I honestly cannot believe we are having our 29th conference in July. This year the conference will host the highest concentration of experts on the topics of Chiari, syringomyelia and related disorders ever assembled. At this time, there are over 60 speakers from around the world. We are truly excited and so appreciative to the doctors organizing this meeting. It is truly a world-class event! We are also deeply indebted to the doctors attending and presenting at the conference. Many of them have been presenting for years, mostly at their own expense, in order to serve our community. They are the best of the best and we owe them our deepest thanks. Lastly, I need to thank our conference committee in advance for all their hard work. If you have ever thought about attending our conference before, now is the time to do it. As I stated earlier, this will be the highest concentration of medical professionals in this field EVER assembled in one place. If you are looking for answers, our conference is the place to be!

In our quest to do more for the people in our community, we developed the L.E.A.R.N program. It makes the conference available to anyone interested in attending. ASAP members who raise 5500 points receive a

complimentary package to the ASAP national conference. This package includes tuition for the event, hotel room (one per family), and travel allowance (airfare up to \$350 or mileage credit of .40/mile up to \$250). Rules and regulations are available on our website or by contacting the ASAP office.

ASAP continues to support people in the community by taking calls and answering endless emails. We also provide a listserv and message board. Our chapter leaders are busy in the community hosting support groups in their area.

ASAP's Research Committee is a team of doctors that look over the many incoming grant requests for the best research. After reviewing the applications, they make recommendations to the Board of Directors. The prospective research grants are vetted for the tangible benefits to the CM/SM community and then voted upon. Only those that will result in direct benefit to our community are accepted and funded.

In closing, I would like to thank everyone who responded to our last mailing. The money raised through this appeal will continue to fund our various programs, allowing us to continue to serve our community. You may still participate even if you have not received a mailer, simply call our office or go to ASAP.org to make a donation. Every bit of support, no matter how small, is deeply appreciated.

I look forward to seeing you at the conference this summer. It is a chance of a lifetime you will not want to miss.

Gentle Hugs always,
Patrice
Patrice_Schaublin@ASAP.org

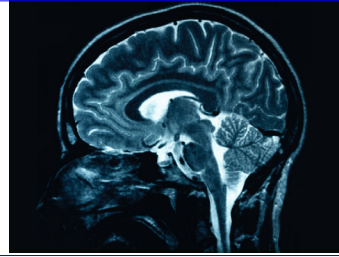
Your life will become better by making other lives better.
Will Smith

Please Note: Articles in this newsletter are not intended as a substitute for medical advice and do not necessarily represent the viewpoints of the editor, Medical Advisory Board or Board of Directors. Please contact your doctor before engaging in any new therapy or medication.

www.ASAP.org

Exercises for Your Brain

1. Test your recall. Make a list (grocery items, things to do, anything else that comes to mind) and memorize it. An hour or so later, see how many items you can recall. Make items on the list as challenging as possible for the greatest mental stimulation.
2. Let the music play. Learn to play a musical instrument or join a choir. Studies show that learning something new and complex over a longer period of time is ideal for the stressed mind.
3. Do math in your head. Figure out problems without the aid of pencil, paper, or computer; you can make this more difficult and athletic by walking at the same time.
4. Take a cooking class. Learn how to cook a new cuisine. Cooking uses a number of senses: smell, touch, sight, and taste, which all involve different parts of the brain.
5. Learn a foreign language. The listening and hearing involved stimulates the brain. What's more, a rich vocabulary has been linked to a reduced risk for cognitive decline.
6. Create word pictures. Visualize the spelling of a word in your head, then try and think of any other words that begin (or end) with the same two letters.
7. Draw a map from memory. After returning home from visiting a new place, try to draw a map of the area; repeat this exercise each time you visit a new location.
8. Challenge your taste buds. When eating, try to identify individual ingredients in your meal, including subtle herbs and spices.
9. Refine your hand-eye abilities. Take up a new hobby that involves fine-motor skills, such as knitting, drawing, painting, assembling a puzzle, etc.
10. Learn a new sport. Start doing an athletic exercise that utilizes both mind and body, such as yoga, golf, or tennis.



Symptoms Associated with Chiari and Syringomyelia

Brain Fog

“Brain fog” isn’t a medical condition. It’s a term used for certain symptoms that can affect your ability to think. You may feel confused or disorganized or find it hard to focus or put your thoughts into words.

Neurological conditions can change the way your brain talks to the rest of your body. Other factors that might contribute are poor sleep habits, medication, depression, chronic fatigue and pain.

If you notice you are having a problem remembering things, recalling words or focusing, you might want to talk to your doctor. He can help you to pinpoint the cause and offer solutions to help. In some cases memory exercises and learning can help with brain fog.

Tinnitus

Tinnitus (TIN-ih-tus) is the perception of noise or ringing in the ears. A common problem, tinnitus affects about 1 in 5 people. Tinnitus isn’t a condition itself — it’s a symptom of an underlying condition, such as age-related hearing loss, ear injury or a circulatory system disorder.

Cape Effect

Many patients report severe pain in the neck, upper back, and shoulders. Doctors refer to this as the “cape effect” of syringomyelia — meaning pain in the area where a cape is draped over the shoulders.

Nystagmus

Nystagmus is an involuntary, rapid and repetitive movement of the eyes. Usually the movement is side-to-side (horizontal nystagmus), but it can also be up and down (vertical nystagmus) or circular (rotary nystagmus). The movement can vary between slow and fast, and it usually involves both eyes.

People with nystagmus may not be aware of their eye movements because what they see usually does not appear shaky to them. People with nystagmus may tilt or turn their head in order to see more clearly. This helps to dampen or slow down the eye movements.

Acquired nystagmus occurs later in life and has a variety of causes, including an association with serious medical conditions. Unlike children with nystagmus, adults who acquire nystagmus may see images as shaky.

**MAY IS SYRINGOMYELIA
AWARENESS MONTH**

Chronic Pain

Depression

Pain and the problems it causes can wear you down over time and affect your mood. Chronic pain causes a number of problems that can lead to depression, such as trouble sleeping and stress. Disabling pain can cause low self-esteem due to work, legal or financial issues.

The Immune System

The link between the nervous system and the immune system is strong. Thinking in terms of the “fight or flight” system, the body generally has one basic overriding instinct: to survive. When chased by a lion or burned by a stove, the reactions and responses are short-term and quick. In order to divert energy to muscles for running or moving, the body can shut down more long-term, high-energy processes like digestion, sexual and immunity functions. Actually, this relationship is more complex than a simple shut down procedure because during the initial stress-response the nervous system fine-tunes and enhances parts of immunity. But as the stressor continues, the nervous system triggers the shutdown of immunity and actually starts to disassemble it.

Research

In an article published by the Department of Anesthesiology, School of Medicine, Pain Research Center, Salt Lake City, Utah, the author reviews the potential physical and psychological consequences of chronic pain and the importance of implementing effective therapeutic strategies to mitigate the harms associated with inadequate treatment.

Results

A review of recent literature examining the neurobiology and pathophysiology of chronic pain reveals that this highly prevalent condition negatively impacts multiple aspects of patient health, including sleep, cognitive processes and brain function, mood/mental health, cardiovascular health, sexual function, and overall quality of life. Furthermore, chronic pain has the capacity to become increasingly complex in its pathophysiology, and thus potentially more difficult to treat over time. The various health complications related to chronic pain can also incur significant economic consequences for patients.

Conclusion

Like other chronic conditions, it is important that chronic pain is managed with the objective of minimizing or avoiding its associated long-term effects. Early and effective treatment strategies, including analgesic therapy that controls pain intensity, are essential to improving outcomes and returning patients to normal levels of function.

Mindfulness Meditation and Chronic Pain

Learning to relax your body and mind takes practice, especially if you are experiencing pain. But recent studies have shown that committing even a minimal amount of time to learning meditation techniques can in turn have a positive impact on your pain. In fact the perception of pain can be significantly reduced after as little as three 20 minute sessions of meditation spread out over three days. Meditation is not likely to be a cure for pain-related issues but it should bring you meaningful relief. There is no right or wrong way to meditate. Typically all you need is a quiet, dark room. You can start with as little as 5-10 minutes a day. The important thing is not to put it off. Start right away and over the next few days you may find significant relief from your chronic pain.

Syringomyelia Awareness Month

Do something special during the month of May to promote awareness of syringomyelia! Take photos of your event or project and email to the ASAP office (info@ASAP.org). We will publish your story in the next edition of *ASAP Connections* or post on our website.

Suggestions for promoting awareness:

- Have your governor or mayor declare May as Syringomyelia Awareness Month with a proclamation. This is a newsworthy event. Get media coverage of you receiving the document.
- Local newspapers are always looking for personal stories for their Lifestyle or Health section. Contact your local paper to publish a story about syringomyelia.
- Plan a walk or awareness event in your town.
- Start a social media campaign to spread awareness.
- Give a presentation about syringomyelia at your local schools or social clubs (Lions, Women’s Clubs, etc.).
- Help your local scout troop learn about syringomyelia.
- Share information with classmates, co-workers, neighbors, church family and friends.
- Make a YouTube video and tag ASAP.
- Tweet #maysyringomyeliaawarenessmonth.
- Learn more about syringomyelia. Knowledge is power!
- Honor someone with syringomyelia through a donation to ASAP.
- Ask your employer to create a workday giving program.

Let the world know syringomyelia exists!

Beef-N-Brew Bowling Night

When: Saturday, April 22, 2017, 7:00 - 10:00 PM

Where: Kingpin Lanes, Egg Harbor Township, NJ

\$25.00 - Beef-n-Brew ticket includes roast beef, baked ziti, salad, dessert, soft drink and coupon for game at a later date.

\$30.00 - Beef-n-Brew with one game of bowling (includes shoe rental)

Raffle and silent auction, an autographed guitar from Vince Gill will be auctioned in addition to other great prizes.

<http://ASAP.kintera.org/Beef-n-Brew> for more information



Yogi's 2nd Community Fun Day and Ziti Luncheon

When: Saturday, May 20, 2017, 10:00 AM

Where: Seneca Falls Elk Lodge, Seneca Falls, NY

Free Admission

\$10.00 - Adult lunch ticket

\$8.00 - Child lunch ticket (12 and under)

Lots of fun activities for the entire family
Tickets 50 cents each

Raffle and silent auction

For more information visit:
<http://ASAP.kintera.org/Yogi2017>

Jessica's 2nd Annual Walk & Roll For A Cure

When: Saturday, August 13, 2017, 10:00 AM

Where: Gouverneur Fairgrounds, Gouverneur, NY

Free Admission

\$30.00 - Pre-registered walk participants will receive an event t-shirt

Raffle and silent auction, Elvis impersonator, face painting, lots of fun activities for the whole family

<http://ASAP.kintera.org/Jessica2017> to register, make a donation, volunteer or for more information,

Molly's 3rd Annual Chiari & Syringomyelia Walk and Roll

When: Saturday, September 16, 2017, 10:00 AM

Where: Bretton Woods Recreation Center, Germantown, MD

Free Admission

\$5.00 - Dove release, purchase a balloon dove in honor of or in memory of a loved one

\$30.00 - Walk participant to receive an event t-shirt

Everyone is welcome to participate. Register online
<http://ASAP.kintera.org/MollysWalkNRoll>



Remember ASAP...

When It's Time to Remember Loved Ones

Our appreciation to everyone who made a recent donation to ASAP on behalf of their friends and loved ones.

We will send an acknowledgement card to individuals or families when you make a \$5 (or more) donation to the organization. Please indicate whether the gift is 'in honor of' or 'in memory of' and provide name and mailing address of the recipient.

In Honor of Person

Donor

Jacqueline Babitts

Steven & Rosa Babitts

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Nancy Earon

Camile Malensek

Cindy, Becky Sprunger &
Family

Bill Squires

Jim, Becky, Sara & Molly
Sprunger

Jeanie O'Sullivan

Jim, Becky, Sara & Molly
Sprunger



ASAP History

As we began preparing for the next conference it seems only fitting that we look back twenty-nine years to our first meeting.

Colorado Springs 1988

ASAP's first conference was a two-day affair in the foothills of the beautiful Rocky Mountains. Fifty-two people attended, 20 with syringomyelia. No one quite knew what to expect since it was ASAP's first get-together but it set the tone for all future conferences. It was a weekend of instant friendships that lasted for many years, a time of hope and learning and a time of pride in what ASAP had accomplished in just one short year. It was also a weekend of personal struggles for some. For Barbara White it was her acceptance of the necessity for a wheelchair.

One attendee, Diana E. wrote, "To me, Conference 1989 was like a family reunion even though I had never before met anyone there. I was really nervous about going, but five minutes after walking in, I knew I was with friends."

Inez S. wrote "Many times since the conference in June when I have had a down time, I will recall the good and fond memories, the tears and the laughter we shared and look forward to the next time we meet." Her husband added, "We would like to thank you for all the hope, support and encouragement you gave us. You enriched our lives so much with your wise and understanding words. We felt your love and your pain and we will never forget you."

Contact Information

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Associated Disorders of Chiari Malformation

Perhaps a contributing factor in the misdiagnosis or unsuccessful treatment of Chiari is the fact that it may not be the only disorder or disease responsible for our symptoms. Many know about tethered cord syndrome and Ehlers Danlos syndrome (EDS) but are you familiar with these?

Mitochondrial Disorders

According to the United Mitochondrial Disorders Foundation, mitochondrial diseases result from failures of the mitochondria, specialized compartments present in every cell of the body (except red blood cells).

Mitochondria are responsible for creating more than 90% of the energy needed by the body to sustain life and support organ function. When they fail, less and less energy is generated within the cell. Cell injury and even cell death follow. If this process is repeated throughout the body, whole organ systems begin to fail.

The parts of the body, such as the heart, brain, muscles and lungs, requiring the greatest amounts of energy, are the most affected. Mitochondrial disease is difficult to diagnose because it affects each individual differently. Symptoms can include seizures, strokes, severe developmental delays, inability to walk, talk, see, and digest food, combined with a host of other complications. If three or more organ systems are involved, mitochondrial disease should be suspected.

Although mitochondrial disease primarily affects children, adult onset is becoming more common.

Empty Sella Syndrome

Empty sella syndrome is a rare disorder characterized by enlargement or malformation of a structure in the head known as the sella turcica. The sella turcica is a saddle-shaped depression located in the bone at the base of skull (sphenoid bone) in which resides the pituitary gland. In empty sella syndrome, the malformed sella turcica is often either partially or completely filled with cerebrospinal fluid. As a result, the pituitary gland is often compressed and flattened so that the sella turcica appears empty. Most individuals with empty sella syndrome do not have any associated symptoms. Occasionally, headaches or pituitary dysfunction may occur. Empty sella syndrome may occur as a primary disorder, for which the cause is unknown (idiopathic), or as a secondary disorder, in which it occurs due to an underlying condition or disorder such as a pituitary tumor or trauma in the pituitary region.

The symptoms of empty sella syndrome may vary from one person to another. In most cases, especially in individuals with primary empty sella syndrome, there are no associated symptoms (asymptomatic). Often, empty sella syndrome is discovered incidentally on x-ray examination when individuals are being evaluated for other reasons.

The most common symptom potentially associated with empty sella syndrome is chronic headaches. However, it is unknown whether headaches develop because of empty sella syndrome or are a coincidental finding. Many individuals with empty sella syndrome have high blood pressure (hypertension).

In rare cases, individuals with empty sella syndrome have developed increased pressure within the skull (benign intracranial pressure), leakage of cerebrospinal fluid from the nose (cerebrospinal rhinorrhea), swelling of the optic disc due to increased cranial pressure (papilledema), and abnormalities affecting vision such as loss of clarity of vision (visual acuity).

The pituitary gland is usually not affected. The pituitary gland is a small gland located near the base of the skull that stores several hormones and releases them into the bloodstream as needed by the body. These hormones regulate many different bodily functions. Although a rare occurrence, some abnormal or decreased pituitary function can occur (hypopituitarism). A specific finding in some affected individuals, including children, has been isolated growth hormone deficiency.

Individuals with secondary empty sella syndrome are more likely to develop abnormalities affecting vision and decreased function of the pituitary.

Mast Cell Activation Syndrome

Mast cell activation syndrome (MCAS), also commonly referred to as mast cell activation disorder (MCAD), is an immunological condition in which mast cells inappropriately and excessively release chemical mediators, resulting in a range of chronic symptoms, sometimes including anaphylaxis or near-anaphylaxis attacks. Primary symptoms include cardiovascular, dermatological, gastrointestinal, neurological and respiratory problems.

Unlike mastocytosis, where patients have an abnormally increased number of mast cells, patients with MCAS have a normal number of mast cells that do not function properly and are defined as hyper-responsive. MCAS is still a poorly understood condition.

MCAS is often found in patients with Ehlers–Danlos syndrome (EDS) and postural orthostatic tachycardia syndrome (POTS). There are no known causes, but the condition appears to be inherited. Symptoms of MCAS are caused by excessive chemical mediators inappropriately released by mast cells. Mediators include leukotrienes and histamines. The condition may be mild until exacerbated by stressful life events, or symptoms may develop and slowly trend worse with time.

ASAP's Mission: to improve the lives of persons affected by syringomyelia, Chiari malformation and related disorders while we find the cure.

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ASAP Connections

The American Syringomyelia & Chiari Alliance Project (ASAP) is a tax-exempt 501(c)(3) organization. Our goals include providing a clearinghouse for information on syringomyelia (SM), Chiari malformation (CM), and related conditions.

We offer a supportive network of programs and services and fund research to find better therapies and cures. ASAP is supported by tax deductible donations.

ASAP Connections is published quarterly for ASAP members. Your contributions of articles, letters, and photos are encouraged. The editor reserves the right to edit any article in order to accommodate space.

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ASAP Connections Editor: Patricia Maxwell

ASAP's Chiari & Syringomyelia Conference

July 19-23, 2017

Attendee Registration Form

Pre-conference registration closes July 10

Attendees may register at conference for a slightly higher fee.

Contact Information

Name: _____

Address: _____

Phone: _____ Email: _____

Additional Attendees

Please add contact information if different from above. List each child's name and age..

Name: _____

Name: _____

List additional attendees and contact information if needed on back.

Fees

Number

Sub-total

Adult registration by July 10 *

_____ x \$175.00 _____

Children's registration: ages 5 - 15 *:

_____ x \$ 95.00 _____

Additional banquet tickets: **

_____ x \$ 75.00 _____

Family pack: 2 adults, 2 children (ages 5-15) *

_____ x \$490.00 _____

*Includes Wednesday reception, Saturday banquet & breakfast (Thursday, Friday, Saturday and Sunday)

** For those who will only attend the banquet on Saturday evening

Special diet request: _____ Number of vegetarian entrees banquet: _____

Note: By submitting this form and/or attending the conference, you agree to allow ASAP to use photographs and/or videos taken of you in ASAP's promotional materials. You understand that ASAP will not publish or record any personally identifiable information without your express written consent.

Payment Information

I want to make a donation. Scholarship Fund \$ _____ Conference Sponsor \$ _____

Payment Type: ☐ Check ☐ Credit Card Total Amount Enclosed (see sub-total above): \$ _____

Credit Card Type: ☐ Visa ☐ MasterCard ☐ Discover ☐ American Express

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Credit Card Number: _____ Expiration Date: _____

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ASAP Conference Registration
PO Box 1586
Longview TX 75606