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## Genetics and Molecular Biology of Cerebral Cavernous Malformations and CCM Italia

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Summer 2012

# Angioma Alliance Newsletter

## Angioma Alliance is 10 Years Old

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Note: In this newsletter, the terms "cavernous angioma," "cavernous malformation," and "CCM" are used interchangeably.

In November, 2001, I learned that I had multiple cerebral cavernous malformations (CCMs) and that I needed brain surgery. I felt numb and scared. For all of you who have received this diagnosis, you know exactly how I felt when those words came out of my doctor's mouth. None of my friends had ever had any significant health problems. I told some of my best friends but they didn't know what to say, except that they were sorry. They had never had a friend experience a serious health condition before and they were unable to offer me any emotional support. In other words, they couldn't deal with it.

My upcoming brain surgery made them scared and nervous so they avoided me. I don't blame them. At that young age, I don't know what I would have done in their place. My family was scared and did the best they could to help me but I felt very alone. I turned to the Internet to learn about my condition. I was hoping to find somebody else out there going through what I was going through. I found a few scientific articles written by researchers. The articles I read explained CCMs in very clinical terms and often chronicled worse case scenarios. I was really scared now! I had never had surgery before and here I was facing brain surgery.

I started emailing anybody that mentioned anything on the Internet about having or knowing somebody with a CCM (this was before MySpace or Facebook). I emailed somebody in New York who told me to contact Connie Lee whose daughter had CCMs. I emailed Connie. Connie told me that her young daughter, Julia, had been through several brain surgeries. Connie asked where in my brain they would be operating and then told me that she thought I would be okay. This was the first time I had actually communicated with somebody who knew about CCMs and who had helped somebody undergo brain surgery. The first time somebody told me that I would be all right. I can't begin to explain how much that meant to me. The surgeon had told me that I would lose some of my vision in the surgery, as he would be going through the vision part of my brain to reach my CCM. But Connie had told me that I was going to be okay! I am still emotional now as I write this. That meant everything to me.

I did lose part of my vision in the surgery and it took me a long time to regain my strength and balance, and I do have permanent deficits. But thanks to Connie, I got through it. Just having that one person to talk to made all the difference.

In May, 2002, Connie Lee e-mailed me that she was starting a non-profit organization to help people with cerebral cavernous angiomas. She wanted to create an online community where people could offer support to one another, create awareness in the medical community, and help facilitate research. She asked if I would be on the Board of Directors to help with patient advocacy. It was an honor to be asked and, of course, I said "Yes!"

So this is how Angioma Alliance began. In August of 2002, Angioma Alliance's website was created. Angioma Alliance had no money and Connie's brother offered to create the website where accurate information about CCMs was posted. Soon after, the Community Forum was added to the website so people with CCMs could communicate with each other, ask questions, and receive support from people who knew exactly what they were going through.

Wonderful Board Members joined Angioma Alliance and, with their ideas and hard work, Angioma Alliance's first publication, a brochure for patients and doctors, was created. In December of 2002, Angioma Alliance's first newsletter was published and mailed to its first members. Angioma Alliance was off and running.

It should be noted that all this was accomplished solely with money donated from Angioma Alliance members, family, and friends (and Angioma Alliance still relies solely on money given and raised by its members and their loved ones).

Angioma Alliance held its first Family Conference in June, 2004, in Chicago. This was the first time that people diagnosed with CCMs and their families could meet each other in person. They could also attend seminars given by doctors and specialists to receive accurate information. Many other Family Conferences were to follow. Some attendees made lifelong friends and attended many conferences.

Soon, Angioma Alliance began to focus on scientific research. A treatment was needed that was not invasive like surgery. People with CCMs needed this treatment and they needed it now.

The first Scientific Workshop was held in November, 2005. The workshop was created so researchers from around the world could meet and

share their ideas and research on CCMs in the hope of finding a cure. Since 2005, the workshop has become an annual event with more and more researchers from all over the world attending. The 2011 Scientific Workshop was held in France with over 50 scientists from 20 laboratories in 9 countries. Collaborations from these workshops are a primary reason the science has moved from gene discovery to possible treatments in less than 10 years.

From these workshops, Angioma Alliance realized there was a need for a DNA and Tissue Bank so that researchers and scientists would have tissue (patient



*Participants in the first Angioma Alliance Scientific Workshop.*

tissue collected from brain surgeries) to use to further their research. In June, 2006, Angioma Alliance started the DNA/Tissue bank. The Angioma Alliance DNA/Tissue Bank quickly became the primary source of cavernous angioma tissue and patient DNA for use in research throughout the United States. We support more than 10 laboratories and have provided the tissue that has been used for several significant discoveries.

As our focus on science increased, the Board of Directors decided that Angioma Alliance should hire a full-time Chief Scientific Officer. In August, 2009, Dr. Amy Akers started in the position, and she has dedicated herself to driving research for a cure.

As the science advanced, researchers identified drugs that could possibly stop CCMs from bleeding. Drug trials were needed to test the effectiveness of these drugs. The drugs that have been identified as possible treatments have been in use for other conditions, but the scientists needed CCM patients to evaluate the drugs' effectiveness in treating CCMs. For

each drug trial, thousands of patients would be needed to test the drug. A CCM patient registry was needed (a list of people with CCMs who could be called upon to be in the drug trials if they wanted to be). With the hope of a cure or non-surgical treatment on the horizon, Angioma Alliance started a Patient Registry. The International Cavernous Angioma Patient Registry is an ongoing project and many more people are needed. So if you haven't signed up for Angioma Alliance's Patient Registry, please do so. Details for the registry are on the website and the Patient Registry is the only way we can find a treatment and cure for CCMs!

One of Angioma Alliance's original goals was to create a community of people affected by CCMs who could help and support each other. To further this goal, Angioma Alliance started a Peer Support Program in the fall of 2006 for Angioma Alliance members to help other members. This program would connect people who had CCMs and/or family members of people with CCMs with people who were going through what they were experiencing; people who were recently diagnosed, needed support with their symptoms, had an upcoming surgery, or had a family member with CCMs. The hope was that people affected by CCMs would not feel alone and would have the support they needed from other people affected by CCMs. Since its inception, this program has paired hundreds of Angioma Alliance members with newly diagnosed CCM sufferers.

Angioma Alliance took its crusade to Capitol Hill. Connie Lee, member Joyce Gonzales and Scientific

Advisor Dr. Leslie Morrison worked with Senator Tom Udall of New Mexico to help pass legislation calling for increased awareness of cavernous angiomas. Through much hard work and persistence this legislation was passed in the Senate in 2009.

The need for Angioma Alliance was clear and began to spread internationally. In 2005 Angioma Alliance UK was formed (AA UK). Last year Italy started CCM Italia and just this year Angioma Alliance Canada began (AA Canada).

Angioma Alliance has increased awareness in the medical community through exhibiting at medical conventions and starting a Neurology Residents Award. For the award, Neurology Residents in the United States are asked to write treatments for CCM patients using case scenarios. The best article receives a cash prize. The goal of the this award is to increase awareness of CCMs in the neurological medical community and awareness for our future neurologists!

It's hard to believe that just ten years ago, there was nowhere for a CCM patient to turn for information and support. Angioma Alliance is now an international community with websites, a Community Forum for people affected by CCMs to receive support and share information, a newsletter, family conferences, scientific workshops, programs to facilitate research, a peer support program, federal legislation, and programs to create awareness in the medical community.

I feel so lucky to have been a small part of Angioma Alliance's progress. To know that people can find Angioma Alliance's website, receive accurate information, and not feel alone is a wonderful feeling.

Many people have helped make Angioma Alliance what it is today including you, Angioma Alliance's members. Without its members and their generosity, there would be no funding to keep Angioma Alliance's programs and hopes for a cure alive. Angioma Alliance still has many goals to accomplish and with your help they can be reached.

*Kristen Dehn*



*A small group discussion at the 2011 Dallas conference.*

## News

### 2012 Patient and Family Conference

In June, Angioma Alliance and the University of New Mexico Department of Neurology jointly hosted the 7th Cavernous Angioma Patient and Family Conference. Held in Santa Fe, New Mexico, in the historic La Fonda Hotel, this one-day conference was attended by over 100 patients, friends and family members—all of whom have had their lives touched by cavernous angioma. This meeting provided attendees with the opportunity to network, develop relationships with one another, share experiences, and hear lectures from distinguished speakers.

The morning session opened with introductions from Beth Baca from the University of New Mexico as well as Angioma Alliance's Amy Akers and Christine Castellano. Karen Ball, President and CEO of the Sturge Weber Foundation, and Callyn Hall from the Rare Diseases Clinical Research Network, followed with short presentations. Callyn discussed the importance of participation in patient registries, while Karen spoke to the group about the role of the patient community and advocacy groups for rare disease research. These two talks remind us that participation in research studies and patient registries is essential to the progress of science and development of drug treatments.

To close out the morning session, leaders in the fields of neurosurgery, basic science and neurology presented three scientific talks.

Dr. Erich Marchand, a neurosurgeon at the University of New Mexico, described for the group the various treatment strategies for individuals with cavernous angioma. He talked about conservative management, traditional surgery by craniotomy and radio surgery.

The second presentation was from a genetic researcher, Doug Marchuk, PhD, of Duke University. Dr. Marchuk's lab is currently working on developing a drug therapy for cavernous angioma. His group uses mice and magnetic resonance imaging (MRI) as research tools and he discussed the recent scientific data using fasudil as a drug treatment in these mice.

Dr. Leslie Morrison, also from the University of New Mexico, is the neurologist who is currently running the Brain Vascular Malformations Consortium research project at UNM. This study is designed to investigate the genetic causes of variability of cavernous angioma—essentially addressing the question of why some people are seriously affected with lots of symptoms, while others, even in the same family, may have no symptoms. Dr. Morrison described some of her preliminary and unpublished data from the study.

To learn more about this study please see our last newsletter (Spring 2012) or visit the website: [rarediseasesnetwork.epi.usf.edu/BVMC/studies/6201-CCM.htm](http://rarediseasesnetwork.epi.usf.edu/BVMC/studies/6201-CCM.htm)

The afternoon session was divided into breakout discussion sessions and talks. The final presentation was from Dr. Davin Quinn, a psychiatrist from the University of New Mexico, who discussed some of the emotional factors involved in living with cavernous angiomas. Lastly, the meeting closed with a relaxing Qigong session led by Loretta Lopez-Shiver.

We would like to thank all those who attended and presented at this important meeting. A special thanks to Beth Baca who did a fantastic job organizing this wonderful event!

This wasn't my first Family Conference with the Angioma Alliance, as I had been to the 2007 Family Conference in Washington DC and have been on the Board of Directors since 2007.

My friend Mark Seymour has lived with me since his brainstem hemorrhage in 2006. As his power of attorney, I have dealt with hospital and rehab personnel: doctors, nurses, technicians, social services, social security employees, disability insurers, and, my favorite, the hospital billing staff!

Mark and I came to Santa Fe on behalf of the Angioma Alliance Board, thinking that we could also fit some vacation time in. What I didn't realize was how much this conference would change me.

In Santa Fe, I saw Physicians and Scientists who were so passionate about their work and excited about their new findings; they were open to questions given to them by the attendees and answered with such care and thoughtfulness, I was indeed impressed. As many of you know, this is not the side most of us see from our doctors and therapists on a regular basis.

Angioma Alliance has done so much for so many people. Please show your support and volunteer to have a fund raiser, join the peer support program, or financially support someone who is having a fund raiser.

*Christine Castellano*

## International Cavernous Angioma Patient Registry Reaches Milestone

The International Cavernous Angioma Patient Registry was started in Spring 2010 as a way to gather information about people with cavernous angiomas. Information in the registry provides a snapshot of the cavernous angioma community—when we were diagnosed, where our lesions are, what other medical problems we have, etc. It is also a way to gather a group of people who might be interested in speaking with researchers in the future when drug trials become a reality.

The Registry now has over 500 participants! This is an outstanding accomplishment. But, with your help, we can do even better. It is our goal to have 1,200 participants by the end of 2012. Please take 15 minutes today to sign up at [www.angioma.org/registry](http://www.angioma.org/registry). In particular, we would love to have more participants with the familial form of the disease. If all affected members of a family sign up, we can easily reach our goal. Take the time to sign up today. Remember, without you there can be no cure!

## CCM3 Action Update

Angioma Alliance's first action group, CCM3 Action, is off to a running start. CCM3 Action was formed to support those who are affected by CCM3, an ultra-rare mutation that appears to cause a more serious, possibly multi-systemic, form of cavernous angioma.

We now have a website, [www.ccm3.org](http://www.ccm3.org), which provides information specific to this mutation. The group has reached out to doctors throughout the nation who have patients diagnosed through genetic testing at either PreventionGenetics or GeneDX laboratories. As a result, several more CCM3 families have been identified.

The CCM3 Clinic at the University of Chicago is up and running! Already, two-thirds of United States CCM3 Action families have flown to Chicago to visit with Dr. Awad and his team. There, they obtained a very advanced 3 Tesla MRI with a CCM protocol that includes permeability analysis, which may be useful for future research. We hope to have the remaining families seen by the end of this year or early next year. These visits will help to document commonalities between affected individuals so doctors and researchers can gain a better understanding of what the illness looks like.

Our fundraising efforts have been quite successful. In addition to the Bakersfield MadoroM

Wine Auction, whose proceeds were restricted to CCM3 Action in February (see our Spring newsletter), a Long Island, New York, fundraiser was hosted by Eileen Benthall and her family to benefit CCM3 Action, raising over \$13,000. Eileen's 15-year-old daughter Johanna is severely affected by the CCM3 mutation. Johanna has had more than 70 surgeries including 15 cavernous angioma resections, a spinal fusion, a brain cancer resection, and numerous shunt revisions. Held at Martha Clara Vineyards, Eileen's event had over 150 attendees who danced, enjoyed a yummy dinner, and purchased chances to win any of 50 gift baskets. Attendees were brought to tears when Johanna sang Carrie Underwood's spiritual song "Temporary Home." We would like to offer a deep thank you to Eileen, her husband Steve, Johanna, and all of their helpers for their tireless work, even during times when Johanna was in hospital in medical crisis, to make this event a success.



*Eileen Benthall, Johanna, and her companion dog, Taffy*

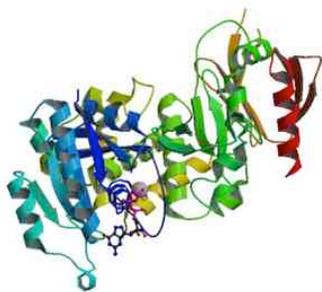
We are looking forward to the 2012 Angioma Alliance Scientific Meeting where we expect to hear exciting news from a number of labs that are already trying to find treatments. Our future plans as we move into 2013 and 2014 include funding CCM3 research directly and organizing a gathering of CCM3-affected families. We're grateful that we've been able to get such a strong start toward meeting our very ambitious goals.

*Connie Lee*

## CCM Research Update: Summary of Recent Publications

**CCM and pregnancy:** A group of Canadian researchers recently published findings from their investigation of how pregnancy affects the risk of CCM hemorrhage. To perform this study, the researchers interviewed nearly 200 women and reviewed their neurological and pregnancy histories. With this information, the researchers calculated the risk of hemorrhage for pregnant and non-pregnant women. The findings from this study conclude that, while some women did experience hemorrhage while they were pregnant, pregnancy itself does not change the risk of hemorrhage in women with CCM. (PMID: 22710379)

**Defining the structure of a protein and its interacting partners, CCM1:** CCM researchers at Yale University have recently used a highly sensitive technique called x-ray crystallography to describe the physical structure of the CCM1 protein molecule. This modeling defines the specific way that CCM1 protein interacts with its partner molecule, Rap1. The interaction with Rap1 is important for regulating the location and function of the CCM1 protein; therefore understanding the way in which these two molecules interact is an important step towards better understanding the biology of cavernous angioma disease. (PMID: 22577140)



*Above: Illustrated representation of CCM1 protein bound to Rap1, courtesy of the Protein Data Bank accession code 4DXA.*

**CCM protein research helps produce interesting results for cancer research:** In the brain, the CCM proteins are responsible from maintaining normal blood vessel structure. When the gene for one of these proteins is mutated and the protein loses function, CCM lesions form and can result in a variety of

symptoms. As far as we know, these CCM lesions are specific to the brain and central nervous system.

Interestingly, the CCM genes can be found throughout the body and in recent years, researchers have discovered that these genes may behave very differently when they interact with different groups of molecules in different cell types. In a collaborative project between research groups from Israel, Canada and Germany, the CCM2 protein was studied in the context of a rare pediatric form of nerve cancer. (This does not mean that people with CCM2 mutations are more likely to have cancer. The biological mechanisms of CCM2 affecting cavernous angioma are distinctly different from those affecting cancer.)

In these cancer cells, normal functioning CCM2 protein is essential for helping to stop tumors growing. In this recent study, while investigating the function of CCM2, researchers identified other molecules that interact with CCM2 in these tumor cells and those molecules are believed to be possible therapeutic targets for treating cancer. Future experiments may lead to better understanding of the role for CCM2 in cancer cells and/or whether these other newly identified partner molecules are involved in cavernous angioma disease. (PMID: 22782892)

*To read more about these studies, please use the [pubmed.gov](http://pubmed.gov) search engine and search for these papers by their PMID number listed above.*

## Scientific Update: Angioma Alliance Collaborations

**Angioma Alliance partners with the Maryland Brain Bank – brain donations for research:** Angioma Alliance has established a partnership with the Maryland Brain and Tissue bank. The Maryland group is a long-standing leader in the field of brain tissue preservation, storage and distribution for research. This collaboration will enable individuals to include brain donation for cavernous angioma research as part of end of life arrangements. To learn more about this program and how to enroll, please contact Amy Akers by email at [amy.akers@angioma.org](mailto:amy.akers@angioma.org).

*Amy Akers*

## Update on Planning the CCM Scientific Meeting

Each year Angioma Alliance hosts an annual conference for CCM researchers. The 2012 CCM Scientific Meeting will be the eighth in this series. Since the first workshop in 2005, this meeting has evolved into the premiere international conference for CCM investigators. Researchers travel from all over the world to be in attendance because no other scientific meeting brings together the same group of researchers to focus exclusively on the biology, treatment and care of cerebral cavernous malformations. This year, our goals for the meeting are as follows:

- To gather a diverse group of clinical and basic science investigators with research focused on CCM.
- To provide a training opportunity for young investigators studying CCM.
- To create an action plan for the design and implementation of clinical trials for CCM.

For this year, we plan to expand the program of the CCM Scientific Meeting to include a poster session and panel discussion. The poster session will focus on junior investigators and trainees to provide them with an opportunity to discuss their research projects and data with more senior investigators. The panel discussion will be a formal moderated discussion on clinical trials that will address topics including biomarkers, recruitment strategies as well as federal regulations and procedures. We are excited to announce that for the first time, we have confirmed speakers from both the National Institutes of Health (NIH) and the U.S. Food and Drug Administration (FDA) who will participate in this important discussion.

The 2012 CCM Scientific Meeting will be held in Washington, DC this November. A summary of the meeting will be published in our Winter Newsletter.

### Estimated cost of the CCM Scientific Meeting

The total cost of the 8th Annual CCM Scientific Meeting is estimated to be approximately \$30,000. Estimated costs for this two-day meeting are:

- \$6,000 Hotel: conference room & equipment rental
- \$15,000 Food: lunch, dinner & afternoon refreshments for all attendees
- \$1,000 Supplies: printing, binding & shipping of meeting programs
- \$6,000 Travel: travel scholarships to four students

*Amy Akers*

## Fundraising Q&A: Fun Run

Are you interested in starting a Fun Run Fundraiser? Here are some questions and answers that can help you get off the ground.

### How do I get started?

If you are interested in planning a Fun Run you can contact Rachel Hart at [rhart@angioma.org](mailto:rhart@angioma.org) or Savannah Hollis at [shollis@angioma.org](mailto:shollis@angioma.org). They will be happy to give you advice on how you can best organize your event.

### How many months ahead of time should I start planning?

You should allow at least 4-6 months to plan the event.

### What is my first step?

Look for a conveniently located, easily accessible walking trail or park. Tour the prospective locations and explain your criteria for the event to make sure the location fits your needs. Make sure the park director knows that you are with a nonprofit organization; discounts may be offered.

### How do I get sponsors?

Small businesses are more likely to sponsor your event to gain publicity. Visit prospective sponsors in person; it is harder to say no to a person than by e-mail. Let interested sponsors know that all donations are tax deductible, and that their logo will be placed on the event t-shirt (if you are printing t-shirts).

### How do I get volunteers?

Angioma Alliance members are eager and willing to help. Make your event known on social media websites and the Angioma Alliance website and forum. Recruit family and friends; they make great, reliable volunteers. In addition, Local Boy Scout and Girl Scouts troops often offer assistance for volunteer hours. Contact local high schools with the event information. Most high school students need volunteer hours to graduate.

*Rachel Hart*



# News from Angioma Alliance Canada

The Angioma Alliance Canada had an amazing summer season, with many positive developments. On June 2nd, 2012, the Angioma Alliance Canada hosted their first annual “Cavernous Angioma Family Conference”. The conference was well attended with more than 25 people from Canada and the US actively participating in the discussions following the presentations of the two main speakers. In the morning session, we heard first from the ambassador of the US Angioma Alliance, Dr. Connie Lee, who informed us about the latest research and facts about cavernous angioma. Our second speaker, Dr. Brent Derry from the Hospital of Sick Children in Toronto, explained the working of scientific research into the genetics behind cavernous angioma. His team works with the functions of specific genes in a worm model to explain the formations of lesions similar to those seen in humans. A brief explanation was given on Dr. Awad’s research into the workings of fasudil and how this possible treatment might work on Cavernous Angioma in humans. The afternoon was used to discuss the personal struggles that each of us faces and how we are coping with the deficits. The day ended with many of us having new understandings and a definite hope for a better future.

On July 1st 2012, also known as Canada Day, the Angioma Alliance Canada was honored, represented, and financial supported by a very dedicated and courageous mother Lea Labelle-McKinley who raises funds with her “Bounce to Bounce back” organization.

Her son Carter had his second brain surgery this past May to remove a lime sized Cavernous Angioma that was causing him pain and hardship.

Lea has been dedicated to raising funds for the Hospital of Sick Children in Toronto, specifically for the neurosurgery department which performed Carter’s lifesaving surgery not once, but twice. In the past two years she was able to raise over \$20,000 for their department, and this year even allocated \$1,000 for the Angioma Alliance Canada. In her own words written on her personal blog ([bouncetobounceback.wordpress.com](http://bouncetobounceback.wordpress.com)), “I am proud of our accomplishments over the past 2 summers and am inspired to continue raising money to support the amazing work being done at SickKids and Angioma Alliance Canada!”



Cavernous Angioma Family Conference at McMaster University in Hamilton, Ontario on June 2, 2012.

So we take a moment to reflect on the positive developments of this summer, and thank Dr. Connie Lee, Dr. Brent Derry, Lea Labelle-McKinley and all of our supporters for their amazing contributions to our organization’s success and growth. We look forward to continue our efforts this fall as we continue to “Work Towards a Cure!”

*Henk van der Wilt*



# Cavernoma Alliance UK Update

July 2012 saw Cavernoma Alliance UK (CA UK) pass an important milestone: the 500th member joined, and the charity continues to grow at an average rate of 2-3 individuals each week.

Whilst grants for charities are globally limited at this time, CA UK manages to cover costs, just. Thanks to the generosity and energy of many of its members, CA UK is able to maintain an office in Dorset with administrative help from Indispensable Iris one day a week. Surrounded by a wonderful board of trustees, I am constantly amazed by members who appear so grateful for the little we are able provide them. A case in point is Lee Smith whose son Zane has been diagnosed with the rare CCM3 mutation. Lee's tremendous fundraising efforts have provided CA UK with over £10,000. From packing groceries at his local store or jumping out of a plane, to organising a formal event in September, Lee's commitment is an example to us all. Lee's fundraising efforts have enabled CA UK to establish its first-ever welfare fund. Starting in the Autumn, this fund will be ring-fenced money allowing CA UK to assist UK based members who are financially challenged attend regional CaverHubs, CaverClinics or the London-based Forum.

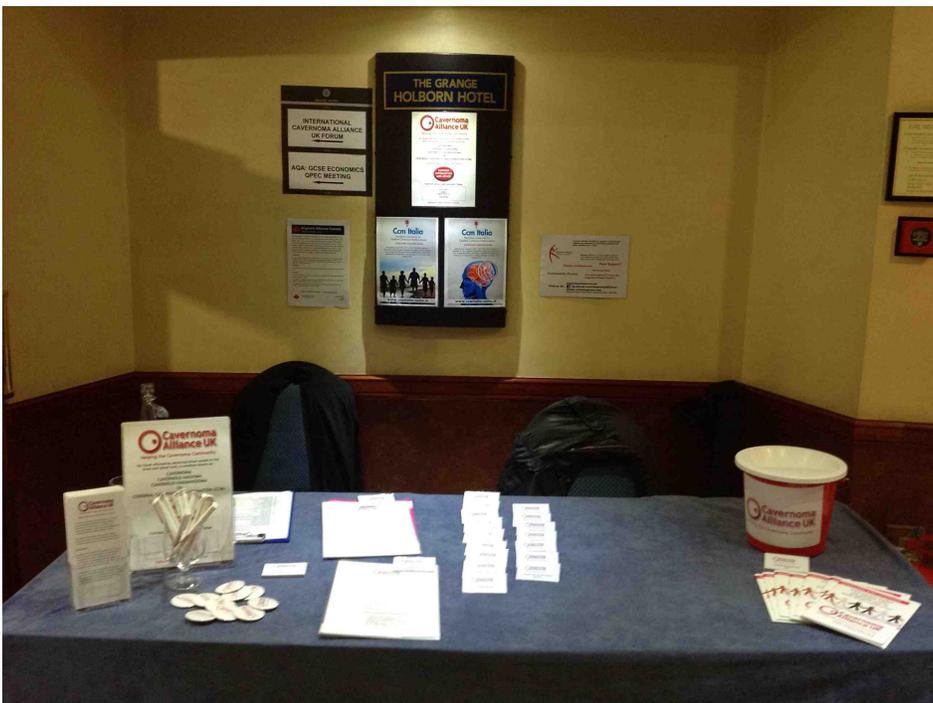
For its sixth International Cavernoma Alliance UK Forum, CA UK took over the Grange Holborn Hotel.

And thanks to Frank Gent, trustee, and Paul Oldham, webmaster, videos of the day can be viewed on our website: [www.cavernoma.org.uk](http://www.cavernoma.org.uk). Owen Sparrow, consultant neurosurgeon, University Hospital, Southampton, provided us with his cavernoma musings. Dr. Sofia Eriksson, consultant neurologist, National Hospital for Neurology and Neurosurgery, spoke on epilepsy and cavernoma, and Fran Brander, also based at Queen Square, lectured on physiotherapy following stroke or intracerebral haemorrhage. The workshops given by and for members were of great use covering aspects such as surgical intervention, non-intervention, genetics and a special workshop for carers. CA UK was also fortunate to host Professor Francesco Retta from the University of Turin who gave a short illustrated presentation on the genetics of CCM and the work of the patient-led CCM Italia.

Summer is the time to plan ahead. Coming up are three conferences: the British Society of Human Genetics, the 1st World AVM Congress in Edinburgh (featuring Dr. Robert Spetzler from the Barrow and a symposium on cavernoma) and the Society of British Neurological Surgeons in Yorkshire. Thanks to a grant from the Foyle Foundation, CA UK has also been able to restart its regional CaverHub programme. Thanks to the work of Simon Temple, trustee, and Leeds-based coordinator Joanne Smith, CA UK has a Yorkshire CaverHub in October; Godsall has arranged a Scottish CaverHub; and the programme is to commence on 6 October with Mr. Kitchen, consultant neurosurgeon and our senior medical adviser, in London.

A busy and productive time awaits CA UK this time during which I "celebrate" the 25th anniversary of my brainstem cavernoma.

*Ian Stuart*



*Cavernoma Alliance UK 2012 Reception (or the Tale of Two Chairs).*

## About Angioma Alliance

Angioma Alliance is a non-profit, international, patient-directed health organization created by people affected by cerebral cavernous angiomas (also known as cavernous malformations or CCM). Our mission is to inform, support, and empower individuals affected by cavernous angioma and drive research for a cure. We are monitored closely in our educational efforts by a Scientific Advisory Board comprised of leading cerebrovascular neurosurgeons, neurogeneticists, and neurologists.

Angioma Alliance  
520 W 21st Street, Suite G2-411  
Norfolk, Virginia 23517

Phone: 571-306-2873  
Fax: 757-623-0616  
info@angioma.org  
www.angioma.org

## Angioma Alliance Board of Directors

- Connie Lee, Founder / Ambassador
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- Eric Johnson, Ph.D., ACireGeneTics, LLC
- Douglas Marchuk, Ph.D., Duke University Medical Center, Dept. of Molecular Genetics and Microbiology
- Leslie Morrison, MD, University of New Mexico Medical School
- Rustam Al-Shahi Salman, MA, PhD, MRCP, Western General Hospital, Edinburgh, Scotland

## How You Can Help

Your contributions help fund conferences and forums, increase research, and enhance outreach and support efforts. To donate to Angioma Alliance, send a check or money order (using the enclosed envelope) or visit [www.angioma.org](http://www.angioma.org). You can also donate on line using a credit card with our Paypal connection.

### Consider a sponsorship

Sponsorships can maintain essential programs or help us expand the ways that we support the cavernous angioma community. Please contact us at [info@angioma.org](mailto:info@angioma.org) to discuss these or other sponsorship opportunities.

#### Travel to Scientific Meeting: \$1,500

Support the vital travel that allows Angioma Alliance to interface with governmental agencies and the scientific community on behalf of those with cavernous angiomas.

#### DNA/Tissue Bank Research Nurse: \$6,000/year

Support the research nurse who gathers and maintains information for the DNA/Tissue Bank which allows researchers to obtain material needed for projects that may one day lead to a cure for cavernous angioma.

#### Patient Registry: \$8,500

Support the ongoing costs of the International Cavernous Angioma Patient Registry which is an essential way to connect the patient and researcher communities in the shared goal of finding a cure.

#### Family Conference: \$15,000

Support this important gathering of people with cavernous angiomas and those that care about them for a weekend of networking and education.

#### Seed grant: \$35,000

Support a young researcher's pilot study that could be leveraged to obtain a \$100,000+ NIH grant.

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Editor: Kirk McElhearn, [kirk@mcelhearn.com](mailto:kirk@mcelhearn.com)