



ANGIOMA ALLIANCE NEWSLETTER

because brains shouldn't bleed

Comings, Goings, and How We're Different

On Thanksgiving morning, while most Americans were preparing for the big feast, I had the pleasure of addressing a group of dedicated, compassionate medical professionals and researchers at Federal University of Rio de Janeiro's public hospital. I shared the story of Angioma Alliance, our successes and our challenges, to offer the Brazilian research and medical community alternative ideas to advance treatments during their current fiscal crisis. After my presentation, the feedback from the audience impressed on me how different the Angioma Alliance model is from traditional approaches, and how this has helped us achieve so much so quickly over the last ten years.

What makes us different? Angioma Alliance is, at heart, an organization that facilitates collaboration and the optimal shared use of resources. In a world where competition, secrecy, and top-down directives predominate, we have always broken the mold. Collaboration occurs between and among patients, researchers, doctors, government, and industry. We create spaces online, in meetings, and in multi-disciplinary collaborative projects for exchanges in which everyone's voice is valued, from the most senior surgeon to the newly diagnosed patient.

For example, in 2014, Angioma Alliance reached out to BioAxone BioSciences, who had not previously heard of cavernous angioma, and introduced them to our academic researchers at our Scientific Meeting. This week, BioAxone announced they had received over \$4 million in NIH funding to develop the first ever medication specifically for cavernous angioma, advancing the work of the consortium we helped to build. And, Angioma Alliance identifies and manages

critical resources—DNA, cavernous angioma tissue, medical information, genetic testing, our funding—to make the utmost use of what a patient advocacy group can offer to our wide, international network.

We take risks to drive research and to create community, but we do so strategically and creatively, preferring lower-cost, pinpoint initiatives that have the potential to yield high returns. You will read about such a new initiative in this issue on page 8 with the introduction of our small grant program, but this is just the latest example of a central tenet. Because of our approach, we provide a model for rare disease organizations around the world.

In February, I will participate in a three-day intensive cavernous angioma workshop in New Mexico that will bring together health professionals, researchers, and patients. As you may know, New Mexico has the largest population of people with the illness in the world. The goal of this workshop is to determine which cavernous angioma research questions are important to the patient community. Participants will begin new research projects based on these questions by creating working groups and action plans. We are honored that cavernous angioma was chosen by the national organizers, the Patient-Centered Outcomes Research Initiative and the Alan Alda Center for Scientific Communication, as a disease community of interest. Our patient voices count in New Mexico, in the greater United States, and around the world.

The coming months will be critical. Our Scientific Advisory Board will be completing clinical care guidelines that we can use as we begin to certify Angioma Alliance Clinical Centers in university hospitals around the United States. The first clinical drug trials should start recruiting late this year. And we have a major public event planned at a baseball game in Cincinnati in May, and others later in the year. Please check our website for updates.

Finally, we need you to educate your legislators about the CCM-CARE bill so we can continue to support our researchers as they seek government grant funding and as we advocate for better healthcare. There is nothing you can do that is more important to our mission of driving research. Please read more about the legislation on page 2. Without you, there can be no cure.

Connie Lee

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The terms cavernous angioma, cavernoma, cavernous malformation, and CCM are used interchangeably in this issue.

News

Take Action!

Senator Tom Udall and the New Mexico Congressional delegation introduced legislation called the CCM-CARE Act. In the Senate, it's known as S. 1391. In the House, it is known as H.R.2480.

The CCM-CARE Act calls for:

- The development of three Clinical Coordinating Centers, each of which would be capable of coordinating multi-center cavernous angioma clinical drug trials.
- Cavernous angioma clinical training in Centers that have the possibility of participating as secondary sites in a clinical drug trial.
- CDC to begin to surveil the incidence of cavernous angioma. There has never been an attempt to do this.
- FDA to pay special attention to Investigational New Drug Applications submitted for cavernous angioma clinical drug trials because we currently have no treatments available.
- Encourage NIH to focus more funding on cavernous angioma research.

Talking Points

Please call your legislators to let them know how important this legislation is to you. Ask your legislator to co-sponsor this bill and, especially, to contact the National Institutes of Health, the FDA, and the CDC to ask what is being done currently for cavernous angioma research.

Please see our website for a script for telephone calls. If you are able to schedule a face-to-face meeting or phone conversation with a member of the Congressional staff, please write us at info@angioma.org so that we can prepare you.

Free Genetic Testing Program Continues

Angioma Alliance's free genetic testing program is continuing through 2016. We will provide free genetic testing for individuals with multiple cavernous angiomas in the United States and Canada who are not able to get coverage for testing through their insurance company. To express your interest, please register with the Angioma Alliance Susan Sukalich International Patient Registry at www.angioma.org/registry.

Angioma Alliance Night at the Cincinnati Reds

Please join us on Monday, May 2, 2016 at the Great American Ballpark, when the Cincinnati Reds host the San Francisco Giants and celebrate Angioma Alliance Night. We will be raising awareness in the stadium and in homes around the US among fans who are watching or listening to the game. We are also hoping to arrange a small patient conference with expert speakers the afternoon of May 2. Please check our website or Facebook page for more details.

Job Posting: Director of Resource Development

Would you like to work for Angioma Alliance, or do you know someone who might be a good fit? We are looking for a Director of Resource Development.

The position requires an experienced development professional who will assist us in securing the resources to further the mission of the organization during a very exciting time. Reporting directly to the President, the Director is responsible for strategic development planning, donor prospecting, donor relations, and event development and support.

This new, full-time telecommuting position is an opportunity for a self-directed individual to make a difference. To learn more, visit angioma.org/pages.aspx?content=468

Ryan Challenge Complete

We would like to express our gratitude to our anonymous philanthropists who have so generously donated \$100,000 to Angioma Alliance as a matching grant, and to those of you who rose to the Ryan Challenge to allow us to meet their match. These funds help us to continue all of our current programs including free genetic testing, to offer small research grants, and to begin the process of certifying Angioma Alliance Clinical Centers. These steps are speeding our progress toward better treatments and a cure.



New Jersey Devils Host Angioma Alliance

The New Jersey Devils game on January 2 was the venue for an Angioma Alliance get-together, fundraiser, and awareness event. Before the game, we enjoyed meeting up at our reception at a local hotel, sharing our stories and catching up with old friends.

Once in Prudential Center, our public awareness efforts began. Anne DeMichiel, who has multiple affected family members, including Angioma Alliance Board Member Julie DeMichiel, did a fantastic job singing the National Anthem to the sold-out arena. Between periods, 12-year-old Trent Clayton, just two days before his brain surgery, rode the Zamboni and won the hearts of the crowd. After the game, all our members in attendance were able to go on the ice for a photo and a shot at the goal from the blue line. This was a special evening for our attendees and introduced hockey fans in New Jersey to cavernous angioma.

Organizing Angioma Alliance Professional Sporting Events

As you've been reading, Angioma Alliance has joined with several professional sports teams to create successful awareness and fundraising events. Perhaps you're wondering how a Cavernous Angioma Awareness Night can happen with your local team. We can provide guidance and support to help you and your nearby Angioma Alliance members bring that dream into reality. Here's some insight to get you started:

- Sporting events can provide a unique opportunity to raise awareness. While we may receive donations or sponsorships as a result of an event, this is not our main purpose. To that end, our first

request of a team is for a designated "Cavernous Angioma Awareness Night" or "Angioma Alliance Night."

- We are much more likely to receive honorary treatment from a team if we have a connection to a current or retired player. If you know a player who would be willing to be our Ambassador during a game, tell us!
- A sporting event can include several opportunities for fundraising. T-shirt sponsors, ticket sales, grants and game experience auctions are just a few of the ways a sporting event can both educate and provide resources we need to continue our work.
- Large sporting events are a great opportunity to bring Angioma Alliance members together. We always include a gathering before the game and, in some cases, may be able to organize a patient education event.

The first step in planning is to contact us. Let us work with you to plan, budget, and find local members and key personnel with the team. Learn more at angioma.org/pages.aspx?content=469.

Peer Caring Committee Established

We have established a United States peer caring committee to expand our peer support program. We currently have more than 30 members who will revise our one-on-one peer support program and initiate new support efforts to connect our community. We are growing, but our personal relationships with other members are our heart and soul. No one with cavernous angioma should feel alone. If you'd like to join the committee, please email Connie Lee at clee@angioma.org.

Interview: Christy Osborne

Connie Lee: Christy Osborne is going to be producing a podcast series for Angioma Alliance. I wanted to ask her a few questions about her own experiences.

Christy Osborne: It's fairly new for me. I'm actually, right now, 17 weeks past surgery. In the beginning, in 2014, we had gone on a cross-country trip. When we returned, I felt dizzy and light-headed. I had a headache. After that, I went to take a bath and, apparently, I passed out. After I passed out, I was pushed to go to the doctor – for days – before I actually went. When I finally went to the doctor, they were worried I had had a stroke. So, I went to a CT scan. When I went through the CT scan, they felt they needed to do an MRI. In the meantime, I went to the neurologist and I failed the test, miserably. After the MRI, they saw I had a lesion and so we went from there. I had no idea what a cavernoma was or anything. I just wanted to research it as best as I could.

After the diagnosis, we continued from there and tried to figure out, what do you do? I was lost. So, I researched on the internet and found Angioma Alliance and that changed my whole view of what was going on. We went to the doctor, the neurosurgeon, and he said, "You know, you're inoperable. It's really deep and I don't want to operate. You should just go live your life." I wasn't going to accept that. I felt that, after reading through the forums and all the information on Angioma Alliance, there was more that could be done. I could tell from the discussions he was having with me that there was more to this than he possibly knew. So, I reached out to all the

members and talked to them on the forums, and I actually was in contact with [Angioma Alliance Chief Scientific Officer] Amy Akers.

In the between time, my neurosurgeon told me he wanted to try to do an angiogram on me because he thought I might have an attached AVM. Knowing what I know now, I probably shouldn't have had that done. But, we did it and I had a stroke. They couldn't stop the bleeding and it went to my brain. Some of the bleeding went to the cavernoma and irritated it, and it bled again.

After that, I had many deficits. I was blind for 7-8 days. When my vision started returning, on one side, colors would cover my vision. It was very strange. I couldn't lift things, couldn't cook, couldn't go to the bathroom by myself. I was dizzy all the time. It took a while to try and cope with it. The main thing we were worried about was if I was blind. What happened? How do I get my sight back?

I reached out again and talked to Amy Akers. She

said, "You have to get a second opinion." After getting the names of doctors, I looked them all up and I found Dr. Spetzler, sent in my scans under their second opinion program, and they said they would take me. He said I'd had bleeds. He said, "I may not be able to fix everything from the stroke, but let's give it a try. Let's get it out." He said, "I can do it."

So that changed our world. We spent the whole summer doing things we wanted to do, preparing for the surgery. We went in September last year and had the surgery. We were really worried about deficits. When I came out of the surgery, I had trouble



with my speech. I had trouble reading. I had trouble writing. I could not write – it was a complete change for me. But, when I came out of surgery, my vision was clearer. I had some ringing in the ear before surgery, and it was gone. So, he can't explain why those things happened, but I'm not going to ask questions. I happy how it ended up.

With therapy, with video games, with trying to write some more, and with reading to my son, I am starting to improve my speech. I can write – I'm writing in my journal. It just takes time. It's slowly coming around.

CL: Can you tell me a little bit about the podcast – your plans for the podcast and what you hope to achieve with it?

CO: Yes, my husband and I, we play video games together because we had read a story about video games helping someone who had brain surgery. He said, "You know, you should try this." So, I started playing them. I saw people streaming their games, and I saw some of them were streaming their games for charity. We had donated to Angioma Alliance, we bought the t-shirts, and we went to the Red Sox game which was fantastic. I said, "I want to do more. Now that I'm out of surgery I want to help more. What can we do?" For Christmas, my husband got me a [home] broadcasting studio. And he said, "Here you go. What can you do?" I started seeing that those same streamers were making podcasts. And I thought, "This is great

for the millennials. They put in their podcasts, they listen to them, they select them, they subscribe to them, and they can listen to them on their way to work. This reaches a whole new audience."

I think that the human story, when people hear each other's stories about feeling the same way, "what did you go through?" it really touches people. It's good to have the forums and that form of communication, but to actually hear someone's voice and know that you can empathize with them - it's very powerful. I'm trying to create that powerful story.

CL: I'm looking forward to hearing them and learning from them.

CO: I want to talk to other people and see what their experience is. I think that there's so much hope and so much work to be done that the more areas that we get to, whether it is podcasts or streaming or the Google groups, that it reaches more and more people. You'll create more than an audience. You'll create a community of support.

CL: Is there anything else you'd like to share or tell us?

CO: Just that I'm excited to start the podcasts. I've got it set up. I'm ready to start interviewing people. I want to say, I will be trying to find words. I will make mistakes when I'm trying to make the podcast, but it's the authenticity of what the situation is.

CL: Thank you very much. Thank you for talking with us.



Research Update

The following is a summary of several recent publications, each of which have a connection to Angioma Alliance through our Scientific Meeting, Biobank and/or a research collaboration.

Italian research team identifies a new potential target for drug development

Autophagy is a normal process by which cells in our body manage waste to get rid of old or damaged cellular components. Improper regulation of autophagy plays a role in several human diseases. In a recent study from the University of Torino, Italy, this process was also shown to have a connection to CCM. When the CCM1 gene is mutated and not functioning correctly, this team showed that there is also a decrease in autophagy. This discovery identifies a potential new target for drug development.

The lead author of this study was one of the trainee travel award recipients at the 2015 CCM Scientific meeting.

Defective autophagy is a key feature of cerebral cavernous malformation: <http://www.ncbi.nlm.nih.gov/pubmed/26417067>

Angioma Alliance DNA/Tissue Bank samples are used to help identify previously unknown function of CCM3

Normally, the blood vessels of the brain have specialized connective junctions that create very tight bonds between cells to ensure that blood cannot leak into the brain. These junctions are disrupted in the vessels of a CCM lesion, a hallmark feature of the illness. In a recent study from the University of Michigan, researchers used tissue from the Angioma Alliance's DNA/Tissue Bank (www.angioma.org/DNA) to investigate the role of the CCM3 protein in regulating these specialized junctions and maintaining blood vessels that are non-leaky.

PDCD10 (CCM3) regulates brain endothelial barrier integrity in cerebral cavernous malformation type 3: role of CCM3-ERK1/2-cortactin cross-talk: <http://www.ncbi.nlm.nih.gov/pubmed/26385474>

The drug Sulindac is effective in decreasing lesion size and number in CCM3 mutant mice

In a previous study, a research team from Milan and Paris identified an important role for a signaling network, termed the TGF β pathway, in CCM disease.

In the disease state, the pathway signals inappropriately and causes blood vessel cells to change and grow uncontrollably. Drugs that block TGF β decrease both the size and number of CCM lesions in mouse models. In a recent study, the drug Sulindac, blocks a molecule related to the TGF β pathway, and also reduces number and size of CCM lesions in CCM3 mice. Sulindac is used clinically for other indications including colon cancer.

Sulindac metabolites decrease cerebrovascular malformations in CCM3-knockout mice: <http://www.ncbi.nlm.nih.gov/pubmed/26109568>

The BMVC Study team identifies genetic risk factors that may help predict long term disease outcome

The Brain Vascular Malformations Consortium (BVMC) is a research team that studies three blood vessel diseases: Sturge Weber Syndrome (SWS), Hereditary Hemorrhagic Telangiectasia (HHT) and Cerebral Cavernous Malformations (CCM). The CCM study is aimed at investigating the genetic factors that contribute to variable clinical severity that is observed between CCM patients, even those within the same family. In a recent collaborative study, the investigation team identified several genetic markers related to oxidative stress, a known biological factor contributing to CCM disease, which correlate with clinical severity of CCM and may serve as predictors of disease outcome.

Angioma Alliance is a BVMC research partner and we are currently seeking study participants with CCM1 gene mutations to participate in a new line of investigation. For more information, please visit: <http://www.rarediseasesnetwork.org/cms/bvmc/Get-Involved/Studies/6201-CCM>, or contact amy.akers@angioma.org.

Cytochrome P450 and matrix metalloproteinase genetic modifiers of disease severity in Cerebral Cavernous Malformations type 1: <http://www.ncbi.nlm.nih.gov/pubmed/26795600>

Amy Akers

Highlights from the 2015 CCM Scientific Meeting

In November, 2015, Angioma Alliance hosted the 11th annual CCM Scientific Meeting. With nearly 80 attendees and representatives from a vast majority of the world's CCM research labs, this was our largest meeting yet. Scientists traveled from across the United States, Canada, Brazil, Italy, France, Germany, and Australia to attend. No other meeting, workshop, or conference brings together such a diverse group of researchers who study CCM disease from every angle.

We are committed to hosting this annual series to foster the development of novel collaborations and to speed the pace of research for a cure. As the meeting continues to grow each year, we aim to maintain an informal workshop-like atmosphere in which speakers are encouraged to share unpublished data for open discussion among the group. Oral presentations about current research studies and open discussion time make up a majority of the meeting agenda. Because the focus of the meeting is on unpublished data, we will not share specific results in this summary; instead please see the Research Update article in this newsletter for a summary of the latest published research. Below is a general summary of some of the highlights from the meeting.



Connie Lee opening the 2015 Scientific Meeting

An important component of the meeting is that it provides a training opportunity; graduate students, postdoctoral trainees and medical residents are encouraged to attend, share their research, and

participate in discussions. This year, Angioma Alliance was able to provide Trainee Travel awards to eight outstanding young scientists:

- **Maria Sole Cigoli** is a graduate student in the Medical Genetics Unit at Niguarda Ca' granda Hospital, in Milan, Italy. She presented her research on using deletion testing as a genetic testing diagnostic tool for CCM patients.
- **Bart-Jan de Kreuk, PhD**, is a postdoctoral fellow at the University of California in San Diego, where his studies focus on how the CCM1 protein is involved in maintenance of blood vessel integrity.
- **Maged Fam, MD**, is a postdoctoral scholar at the University of Chicago. His talk at the CCM Meeting focused on the planning of a clinical trial for atorvastatin therapy in CCM disease. See below for details.
- **Romuald Girard, PhD**, is carrying out postdoctoral research at the University of Chicago, focused on identifying markers in the blood that can be measured to reflect the severity of CCM disease.
- **Ben Lant, PhD**, is a postdoc at The Hospital for Sick Children in Toronto, Canada, where he uses a tiny worm (*C. elegans*) to study the function of the CCM3 protein.
- **Noemi Rudini, PhD**, is a postdoctoral scholar at the FIRC Institute of Molecular Biology in Milan, Italy. She presented a talk describing her current studies aimed at understanding the molecular signals that control how CCM lesions develop.
- **Eliana Trapani** is a graduate student at the University of Torino, Italy. She presented a talk describing her unique studies to investigate how defects in the normal cellular process of autophagy (this is the cell's own waste and recycling program) impact CCM disease.
- **Hussein Zeineddine, MD**, is a postdoctoral scholar at the University of Chicago. He presented his research on using advanced MRI techniques as tools to measure the effects of an experimental drug in future clinical trials.

During this two-day meeting, we heard presentations on a variety of topics including protein biology, molecular signaling, blood vessel and lesion biology, human genetics, and drug development. This is the first year that we were able to dedicate an entire afternoon session to drug development and clinical trials. The session included a talk from the lead scientist at BioAxone BioSciences, who is interested in developing a new molecule for the treatment of recurrent bleeds in CCM. This project is in the early development phase, and is an example of a collaborative project that began because a group of scientists met one another at the CCM Meeting.

Other presentations in this session included discussions on repurposing currently approved drugs for other illnesses to become possible CCM treatments. As an update, the small simvastatin trial out of the University of New Mexico has completed recruitment and is analyzing that data to determine if there is an effect of the drug treatment on lesion permeability, as measured by permeability MRI. Furthermore, we are excited to announce that the University of Chicago has planned a Proof of Concept Trial with Atorvastatin. The goal of the study is to determine if treatment with this drug reduces iron deposition (a marker of bleeding that can be measured by MRI) in patients with CCM. To participate in this trial, participants must fit a number of criteria, including having had a symptomatic CCM bleed within the last 2 years. This study is in the planning phase; we anticipate beginning to recruit for this trial late in 2016. To learn more about the study, please visit: www.Clinicaltrials.gov identifier: NCT02603328.

Additionally, for updates and to be notified when this study begins recruitment, be sure to register for the Susan Sukalich Angioma Alliance International Patient Registry: www.angioma.org/registry.



Issam Awad and Brent Derry.

Angioma Alliance Offers Research Grants

Angioma Alliance is offering small grants of \$5,000 - \$15,000 for pilot projects and bridge funding. A grant may be used to provide funds to researchers to test new hypotheses and generate preliminary data. Having pilot data in hand increases the chances a traditional larger grant application will be funded. Alternatively, researchers may use funds to bridge a gap between grants received from other entities, such as NIH.

In addition to providing funding, Angioma Alliance is able to support research with DNA and tissue samples and with access to de-identified clinical information. We can assist study recruitment through our international patient registry and our extensive patient network. We can be helpful with study design and in identifying collaborative partners.

Grants will be offered on a rolling basis throughout the year. We wish to thank our many generous donors for making this program possible. Complete application instructions can be found at angioma.org/pages.aspx?content=467.



International News



Cavernoma Alliance UK

Cavernoma Alliance UK (CAUK) is committed to ensuring that people gain free access to information about the condition as well as being introduced to the support available in their area. So we would like to use this section of the Angioma Alliance newsletter to update you on how we do this through some aspects of our recent work.

From their first contact with CAUK, an individual receives an information pack containing our three new information booklets about cavernoma, a personal information booklet (which encourages the recipient to record times and dates of appointments with medical professionals, as well as allowing them the opportunity to chronicle their own personal journey), and a DVD of clinicians and members of CAUK discussing their experiences of cavernoma.

The information folder that we send is based on the members' need. For example, CAUK has recently been working with a surgeon to produce a specialist booklet on spinal cavernoma. CAUK also has folders for ages 5 to 11, 12 to 16, adults, and clinicians. For children, we have a cartoon booklet and accompanying DVD to explain the condition. We have also developed CaverFamilies to provide children, their parents, and their siblings, with an opportunity to meet others in a

similar position at a residential weekend. Ollie Holt, a member of CaverFamilies, was recently given a WellChild Charity Award by their patron, His Royal Highness Prince Harry, (pictured below) for Ollie's bravery and cheerfulness in dealing with cavernoma.

CaverHubs are usually one-off events with a professional local speaker discussing cavernoma. (For example, CAUK are to run CaverHubs in Glasgow, Newcastle, Sheffield, London, Southampton and



Bristol during International Brain Awareness Week in March 2016.) However, a fixed CaverHub, which has become an annual event, occurred at the National Hospital for Neurology and Neurosurgery in October 2015. The Autumn London CaverHub was given by Dr Kirsty Harkness, a consultant neurologist from the University of Sheffield. Dr Harkness spoke about the Medical Management of Cavernomas arguing that, at the moment, and until a clinical trial had been adopted, it remained impossible to know whether a further bleed from a cavernoma following, for example, surgery of a deep lesion was due to intervention, or whether this was the cavernoma's natural history

A CaverCentre is a fixed discussion/support group run by volunteers, such as the operational CaverCentre in Liverpool, which is about to have its fourth meeting. An experimental CaverCentre in Leeds was trialled in November. The participants at this first Leeds meeting brought in two speakers: Dr Jenny Thomson, clinical geneticist, who spoke on cavernoma, prenatal testing and genetics; and Mr Kenan Deniz, consultant neurosurgeon, Leeds General Infirmary, who gave a talk on cavernoma management. This successful day will be replicated by a second meeting in Leeds during March.

CaverBuddies (a voluntary role providing confidential emotional and practical support to other members who may be dealing with the direct and indirect effects of cavernoma) is based on the idea that the person best placed to understand the affected person is someone who has been through similar experiences themselves. So in November, eight CAUK members attended the first CaverBuddy training day run by three trustees and Angela Yeomans, community worker. A second training day with nine more participants is scheduled for early February. A voluntary CaverBuddy Co-ordinator has been selected and CAUK hopes that this successful programme will soon be rolled out across the country.

In addition to colleagues from affiliated organisations in North America, Canada, Brazil, Italy, France, Germany, and the Philippines, all CAUK members are invited to reserve now to attend our free Tenth International Forum at the Grange Holborn Hotel, London, 11 June 2016, with talks by a

neurosurgeon, neurologist, neuropsychologist, and a selection of five member workshops.

Finally, as part of our 10th anniversary celebrations, CAUK produced CaverNews, containing articles from members and clinicians about living with cavernoma and researching the condition. An electronic copy can be accessed by going to the information slider of the website, www.cavernoma.org.uk.

Ian Stuart and Angela Yeomans

Angioma Alliance Canada

Angioma Alliance Canada (AAC) has been busy planning the 5th Annual Canadian Cavernous Malformation conference, which will be held on Saturday, June 11, in Toronto, Ontario, Canada. This event will be held once again at The Sick Kids Hospital at The Peter Gilgan Center for Research & Learning.

We have a dynamic line-up of speakers. At this time, we are awaiting one more confirmation regarding the neurosurgeon who will speak. On the agenda so far is Connie Lee, the founding President of Angioma Alliance. She is a clinical psychologist and the mother of a daughter with multiple cavernous malformations. She will share with us her personal story and news from our US partner Angioma Alliance. Next on the agenda is Dr. Brent Derry, the Senior Scientist at the Hospital for Sick Kids, and a supporter of Angioma Alliance Canada. Dr. Derry will discuss the latest scientific news, and what his lab is doing to work toward a cure for CCM. Shawn Mulvihill, President of the Angioma Alliance Canada, will discuss his personal story as well as other news.

Full details regarding the conference will be added to the AAC website www.angioma.ca as well as our Facebook page in the near future.

If you are a Canadian and would like to volunteer to be a board member for AAC, please contact us at angiomaalliancecanada@gmail.com for details.

We look forward to seeing you at the conference in June!

Shawn Mulvihill

CCM Italia

CCM Italia and AIAC are continuing to promote research networking at the national and international level, and raise awareness of CCM disease among the Italian population and health care institutions.

In particular, a multidisciplinary cooperation among clinical and research groups of the CCM Italia network has recently provided novel insights into the understanding of CCM disease pathogenesis and treatment (Marchi et al., 2015 EMBO Mol Med; www.ncbi.nlm.nih.gov/pubmed/26417067).

Moreover, a joint international cooperation involving research groups of the University of Torino in Italy and the Universities of California, San Francisco, and New Mexico in the United States has recently contributed to the identification of genetic susceptibility factors of potential clinical value that may provide information about current status or future risk of CCM disease severity and progression, as well as novel therapeutic options (Choquet et al., 2016, Free Radic Biol Med; www.ncbi.nlm.nih.gov/pubmed/26795600).

In this light, it is noteworthy that a novel Italian clinical and research unit from Cagliari, the major city of the Sardinia region, has recently joined the CCM Italia research network. This unit has documented expertise in clinical and genetic studies related to CCM disease, and, during the last decade it has collected and analyzed a large cohort of CCM patients in Sardinia, the second largest island in the Mediterranean Sea, identifying and characterizing distinct mutations of the CCM genes, including the unique founder CCM1 C329X “Common Sardinian Mutation” (CCM1-CSM) (Cau et al., 2009; www.ncbi.nlm.nih.gov/pubmed/19454328). The Cagliari Unit will contribute to verify whether the genetic modifiers of CCM disease severity identified in the homogeneous cohort of Hispanic CCM patients carrying the founder CCM1-CHM mutation (Choquet et al., 2016) are replicated across different ethnic groups, including Italian cohorts of CCM patients carrying distinct CCM gene mutations.

On December 18th and 20th, 2015, Massimo Chiesa, the President of the Associazione Italiana Angiomi Cavernosi (AIAC), and Prof. Francesco Retta, helped raise awareness of CCM disease by participating on regional and national radio and TV programs, including the Telethon-RAI TV telethon, a major annual Italian television event focused on genetic diseases with a big live audience (see related posts on the AIAC Facebook page: www.facebook.com/AIAC-Associazione-Italiana-Angiomi-Cavernosi-212620812109830).

This year, the slogan of the Telethon was “Non mi arrendo” (“I don’t give up”), which fully expresses the strong courage and willingness of CCM patients to strive for better quality of life, as well as the strong commitment and hope of researchers to develop novel safe and effective therapeutic strategies for treating CCM disease.

For more information, please visit the CCM Italia (www.ccmitalia.unito.it) and the Associazione Italiana Angiomi Cavernosi (AIAC) (www.ccmitalia.unito.it/aiac) websites.

Francesco Retta



Massimo Chiesa, President of AIAC, during a tandem skydiving experience.

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 treatments and a cure. We are monitored closely in our educational efforts by a Scientific Advisory Board comprised of leading cerebrovascular neurosurgeons, neurogeneticists, and neurologists.

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How You Can Help

Your contributions help fund our research initiatives toward a cure and our patient support programs. To donate, please send a check or money order in the enclosed envelope or visit our website at www.angioma.org to donate with a credit card.

Sponsorships can maintain essential programs or help us expand our support for the patient and research community. Sponsors are acknowledged with logo placement, naming opportunities, or appropriate other recognition.

Sponsorships are available for the following:

Newsletter

This newsletter reaches thousands of patients and donors both in print and online. It is the only patient-directed source of information for the cavernous angioma community. If you would like to reach this community and support our efforts, please contact us.

Website

Our website has a global reach, and is always in the top three search results for cavernous angioma. It is the first place newly diagnosed patients look for

information and support. In addition to being a patient resource, the website provides information to medical support staff, researchers and the general public.

Events

Angioma Alliance members host multiple events throughout the year, from Cavernous Angioma Awareness Night at major league sporting events to smaller Fun Runs and tournaments. Sponsorship opportunities are always available with varying levels of public exposure depending on the event.

Scientific Meeting

Our scientific meeting offers a variety of opportunities to support and reach the research community, including travel awards and sponsored speakers, breaks, and meals.

DNA and Tissue Bank and Genetic Testing

The DNA and Tissue Bank is the major source of cavernous angioma biological samples for labs around the world, and we have provided the raw materials for several major published studies.

Contact us at info@angioma.org to discuss any of these sponsorship opportunities.