Angioma Alliance: The First Five Years

While there was no party or fireworks celebration, Angioma Alliance marked an important milestone in May. We have now been serving the community of people affected by cavernous angiommas for five years.

The past five years have seen many changes. We have moved from the kitchen table to the halls of state and federal governments. We have reached across the oceans to touch affected individuals, families, physicians and researchers wherever they are.

We have come face-to-face in three national family conferences in the United States and, in June, 2007, one conference in the United Kingdom. We have brought researchers together twice for scientific workshops and have helped to grow this dedicated group who are working to find a cure.

We are participating more and more in research both as facilitators, via our DNA/Tissue Bank, and as investigators, as you will see in our request for research participants in this issue (see page 3).

We have encouraged physicians, both new and seasoned, to learn more about our illness so they can provide the highest quality care. We have worked toward educating the public and are recognized as the voice of those who are affected by this illness.

No one who has a cavernous angioma has to feel uninformed anymore. You can find accurate information on our website and in our literature, written specifically for patients or loved ones, that can help you to make better decisions about your treatment.

No one who has a cavernous angioma has to feel isolated anymore. Via our toll-free number, peer support program, and info@AngiomaAlliance.org email address, if you have access to a computer or phone, you can connect with someone who will
provide support and shared experience.

While much has changed, one thing has stayed the same. As you will read in an article on page 5 of this issue, our Community Forum remains the heart of our online support. It is a virtual venue that is often the first place newly diagnosed individuals or those who are simply new to us come into contact with others who have the illness. The Community Forum was an original part of our website and has continued to serve as a major force in breaking down the walls of isolation.

While there is much more to do, our first five years have been phenomenal. We want to thank every volunteer who has served, every affected individual who has reached out, and every donor who has given to make this organization what it is and will become.

Angioma Alliance Donors - First Quarter 2007

Beacons $5000+
HEDCO Foundation

Leaders $1000-4999
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In honor of Dr. Dan, Monica, and Scout Brooks
Claire and Richard Brooks

In honor of Heather Kurpiewski
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Hugh and Nancy Bonnar
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Toni Garubo
Thomas and Rosemary Kurpiewski
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In memory of Jenae Gallegos
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In honor of Jake and Sam Neuman
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In honor of Allison Ruggles
Anne Antonealis
Margaret and Kenneth Dickinson
Brian and Kathy Duddy
Leroy and Wilma Jolly
Richard and Patricia Kowalczyk
Barry McAloney
Irene and Ronald Ruggles
Brian and Edie Tonis
Mary and Wallace Varonko

In honor of the Ryan family
Howard Bonde

In honor of Mark Seymour
William Calloway

We’ve included photos of some of the significant get-togethers of our past. We hope you enjoy them and continue to work with us to see where we go over the next five years.

Connie Lee
Letter from the President

If you’ve been reading the Angioma Alliance newsletter for the last few years, I am sure you are impressed by the ways this organization has grown and expanded its reach. We are now five years old and there is no way I could have guessed at the outset that we would have come this far so soon. We have moved beyond our website roots and have become active participants in research, in physician education, in public awareness, and in federal and state advocacy. We continue to expand the ways in which we inform and support those affected by cavernous angiomas throughout the world.

The dramatic growth and importance of our work comes with increased costs, and we count on your support to meet these. We also would like to do more, particularly in the area of research. If you received this newsletter in the mail, it came with an envelope that encourages you to mail in a donation. If you are reading this online, we have a link www.AngiomaAlliance.org/donate.html that allows you to use Paypal to donate, or you can mail a check to the address listed on the last page of this newsletter. If you have a connection with a grant-making organization or if you would like to host a fundraiser, please contact us so that we can work together to explore these possibilities. As you’ve seen, your past generosity has borne much fruit. If you have not given recently, please take a few moments to begin or renew your support. Angioma Alliance is the only voice for those with cavernous angiomas—please allow us to be heard loud and strong.

Connie Lee

Volunteers Needed

We need individuals to serve on two committees and as peer support volunteers. For information on any of these opportunities, please call us at 1-866-432-5226 or contact us by e-mail at info@AngiomaAlliance.org.

Fundraising Strategy Committee. We are looking for folks who have some experience organizing fundraisers who can work together to brainstorm and develop a “signature” event for Angioma Alliance, much like Race for the Cure and cancer research. We’d like to create an event that could be replicated easily anywhere.

Family Conference Committee. We are looking for volunteers to help plan and organize this year’s scientific workshop and family conference. There are many jobs to be done, both big and small, and you don’t necessarily have to be attending to help.

Peer Support Volunteers - We are looking for peer support volunteers who are either parents of a child under 12 who has a cavernous angioma or adults who have been living with a seizure disorder and are currently on medication. To be a peer support volunteer, we ask that you be at least 2 years from your or your child’s diagnosis and have 1-2 hours a week to give either for email or phone support. Additional information about our peer support program and a volunteer application can be found at http://www.angiomaalliance.org/support.html.

DNA/Tissue Bank and Patient Registry

The BioBank and Patient Registry project is quickly evolving into a major research effort. We have hired a part-time registry nurse, Angie Prolog, to manage enrollment of new participants and maintenance of the registry. We now have more than 60 participants. Since our last newsletter, we have begun acquiring frozen tissue from cavernous angiomas removed during surgery, we have completed genetic testing of those members currently enrolled in the bank, we have created the structure of a clinical database, and we’ve developed two brochures to spread the word to physicians and potential participants. If you have already signed up for the BioBank, please expect a call from us very soon to set up an interview time. For those who are interested in participating (particularly individuals who have surgery scheduled anywhere in the US and those who have had a surgery at St. Joseph's in Phoenix in the last 5 years), please call us at 866-432-5226 or e-mail us at Biobank@AngiomaAlliance.org.
Q&A: Ian Stuart

If you walked past Ian Stuart in the streets of Dorchester, in Dorset, England, you probably wouldn’t suspect that he is ill. He looks like a fit, independent 44-year-old, with a broad smile to boot. A button pinned to his lapel is the only distress signal, which reads—should your eyes come to rest on the words—"I have a condition similar to Parkinson's Disease." In fact, Stuart, who founded Angioma Alliance UK in 2005, is challenged by a host of difficulties—double vision, a Holmes Tremor on the right side of his body, and his right arm in particular; trouble with balance and movement, and slurred speech—due to a brainstem cavernous angioma that was discovered 20 years ago while he was working toward his Ph.D. at the University of California at Santa Barbara.

Less than three weeks before the first international forum organized by Angioma Alliance UK (set for June 16, 2007, in London), I called Ian to ask him about his history with the illness and get some insight into his daily life.

“A friend of mine said to me, ‘Don’t be a show off,’” Ian says, cracking a joke before telling me how his difficulty getting around in public prompted him to don the button initially. “I walk up the stairs just using my left arm to hold the handrail—I can’t use the right—so it was borne by the fact that people were saying, ‘Wow. How rude this person is!’ And I wanted to make a statement, and make them appreciate that there are people with hidden diseases. It’s the white stick syndrome. It’s to educate and inform, most importantly.”

It’s easy to see how a candid sign like this one could inspire curiosity or, depending on the audience, a mini-interrogation. In Ian’s experience, the response is decidedly cold. “It’s the cultural difference,” he explains. “In the U.S., they say, ‘Oh hey, what’s up?’ In the UK, they look away. And it’s like a bad smell: they don’t want to be caught breathing your bad air.”

“I feel with Angioma Alliance I have a mission; that I’m on a quest. I have got a purpose in my life.”

In a sense, the button is Ian’s scarlet letter, although he bears his mark in the name of education. “Shakespeare said something once, that you wear your heart on your sleeve,” he muses. “I wear my heart on my lapel. It’s not a burden, it’s for people to read. And to top it off I wear my Angioma Alliance badge just above it so they can connect the two. And they may not know what an angioma is, but it might encourage them to check it out.”

Networking augments Ian’s role as an educator (he insists he’s no good at it). Recently, he represented Angioma Alliance UK at the opening of the Walton Centre for Neurology and Neurosurgery in Liverpool and at a member forum of the Long-Term Conditions Alliance. (See Out and About, page 10.)

“I guess I’m a good person to represent Angioma Alliance because on the outside, people say ‘Oh well, there's nothing wrong with him, he's fine. He looks fit,’ and things like that. But underneath, or inside, there is this problem and really they just have to be with me for a minute to know there's something wrong.”

Ultimately, Ian’s role as a representative for cavernous angiomas isn’t negotiable. “I live and breathe in my condition,” he says. “One can’t do otherwise. So at least I can do it for some value, which is
demonstrating this situation to others; allowing others to understand.”

These days, while he’s not assembling PowerPoint presentations about cavernous angiomas, serving as coordinator for Angioma Alliance UK, writing newsletter articles and countless other activities, Ian is gearing up for this month’s international forum. I asked him what part of the event he’s most looking forward to.

"If there's something I'm most nervous about, I have a whole list," he jokes. "I'm excited to meet all the members who make up Angioma Alliance UK. That's going to be exciting for me. The surgeons... I've met them in the past. I'm excited to hear what they're going to talk about. They sound very interesting and captivating. But first and foremost are the members and the fact that, for example, I'll meet Sacha Bonsor." [She is the author of “Dipped into Oblivion” and Health Editor at the London Times].

It’s this spirit of community and shared experience that seems to inspire Ian. “I feel with Angioma Alliance UK I have a mission,” he says, “that I’m on a quest. I have got a purpose in my life.”

“I guess I’m lucky … I feel pretty lucky and fortunate to be able to see my life in a different way now; in a way that appreciates beauty and harmony where politicians only see distraction. I see the goodness behind what people are and who they are; and I think very strongly that it is a condition which allows me to embrace the good things in this world and to an extent, other people... But I do feel fortunate, in a perverse sort of way. You could call this article ‘Ian’s persion’.”

Ian has a good sense of humor. He laughs when I tell him he seems to have a very balanced view of his life and the world at large. In physical terms, maintaining his balance is a daily frustration. But he also maintains an addictive optimism that can't be interrupted by a tedious cross-continental phone line, sounds of Chicago traffic drifting into my apartment, or his slurred speech.

At this point in time, surgery is not a viable option for Ian. "I'm an interesting case in that doctors really don't know what to do with me,” he says. "[The angiomia] is probably best left alone and, you know, quality of life is the important thing. At the moment I have my mobility and my independence. Everyone deals with the uncertainty of their lives,” he adds. My uncertainty is more certain.”

Rebecca Palmore

Visit the Angioma Alliance UK web site for more information about cavernous angiomas in the United Kingdom.
http://angiomaalliance.org.uk

The Community Forum: A Virtual Support Group

On the Angioma Alliance website, there is a section called the Community Forum that allows registered members to share their questions and experiences, and allows visitors to get a glimpse into what living with cavernous angioma is like. As of this writing, the Community Forum has over 760 registered members from all over the world, from all walks of life. On a typical day, one can read posts on topics such as “Another seizure last night”, “What is in the pons (and why don’t I know this yet)?” “Vision Therapy” and “I am feeling better”. Members announce upcoming surgeries, new hemorrhages or hospitalizations, describe challenges conquered, and offer a voice of experience to those newly diagnosed. And new members ask questions—often those questions that arise when first confronted with cavernous angioma—and receive thorough, compassionate answers from those who have been there before.

Although the number of registered members is large, the Community Forum continues to be true to its name. It is a community of support and encouragement, of shared setbacks and achievements. Individuals come to know each other intimately and many form relationships outside of the forum. Having a cavernous angioma can be a very isolating experience—one doesn’t necessarily look ill, few affected people know others with the illness, and, for those who use the forum most often, disability may keep them from traveling any distance from home. On the Community Forum, all are welcome—please come visit us there sometime.
Another Day in the Frontal Lobe
Katrina S. Firlik
Random House, 2006
288 pages - $24.95 (hc) $14.95 (pb)

Dr. Katrina Firlik, who describes herself as "part scientist, part mechanic", is in a minority: with 4,500 neurosurgeons in the US, or about one for every 65,000 people, only 5 percent of them are women. Recounting how she came to be a neurosurgeon in a hospital environment, Dr. Firlik gives a stark and realistic overview of the work involved in this profession, and how she got there.

"Surgery is usually a last resort," Dr. Firlik writes. "Neurosurgeons complain that neurologists wait too long and try too many medications before sending patients over. Neurologists complain that neurosurgeons are too aggressive in recommending surgery." Yet for those with cavernous angiomas, surgery may be the best, or even the only solution to their problem.

Contrary to popular belief, neurosurgeons are not really brain surgeons; at least not exclusively. They spend much more time operating on spines and spinal injuries than brains. The brain is quite a solid box; the spine is much weaker. When it's car vs. car, the spine loses out much more often. Nevertheless, Dr. Firlik only mentions spine surgery in passing; the focus of this book is on the brain, perhaps because it is more impressive to lay people.

She then briefly explains how she came to choose neurosurgery during medical school, the rigors of the training required to specialize as a neurosurgeon after four years of medical school, and the quirks of her internship and residency, the period during which, "residents evolve from lowly interns to fully fledged neurosurgeons."

Dr. Firlik points out how some people may choose which surgeon they want for their operation. Discussing how some neurosurgeons spend most of their time in the operating room, and others try to get as many hours in the lab as possible, she points out that, "a lay person might assume that the surgeon with his name on the greatest number of papers . . . is the guy to go to for a certain type of surgery. While this may be the case, . . . the reality could also be that this is the guy who spends far more time in the lab than in the OR. A great mind for science and great hands do not necessarily go together."

Firlik discusses the different types of neurosurgeons. There are those who specialize in such things as aneurysms, who are usually called in at night or on holidays to operate in emergency conditions. Then there are the spinal specialists, who perform well-paid spine fusions working nine-to-five hours. Then there are the radiosurgeons, the kind who work with radiation such as the gamma knife; they, says Firlik, are "often the smartest ones around, because they have time to read."

Interestingly, the hospital where Dr. Firlik did her residency was the first in the US to have a gamma knife, and she briefly discusses this form of treatment when talking about her term in the stereotactic radiosurgery department. Firlik tells one story about a patient with an AVM (arterio-venous malformation) who died after surgery, and another who chose to live with an especially large AVM and its uncertainty. She then asked the questions that many of this review's readers will ask themselves: "It's clear that the brain can accommodate quite nicely to the over-bearing presence of a malformation, but can the mind be trained to accommodate just as well? When inaction is the best action, how do you prevent fear..."
itself from becoming an illness? Does the fear simply wear out, or does it have to be forced out?" She then says, "I have found that handling a patient's anxiety can be more complicated, and sometimes even more time-consuming, than the surgery itself."

Dr. Firlik is an esthete; she likes the way the brain looks, and she occasionally tosses in statements about how much she marvels at the way the human body works, how the brain leads to the mind, or waxes philosophical about everyday life. She explains, at one point, how she broke down and cried when telling a patient that his brain tumor was fatal, and manages to express a deep understanding of the human condition.

This book can be gruesome at times, as Dr. Firlik describes some of the more bizarre cases she has dealt with, and may not be for the squeamish. Some of the stories have sad endings, and others end happily. In the end, they probably all add up to the complex life of a neurosurgeon: dealing with one of the most eloquent parts of the human body is fraught with both great risk and great possibilities to heal.

*Kirk McElhearn

Visit Dr. Katrina Firlik’s web site at [http://www.katrinafirlik.com](http://www.katrinafirlik.com), to find out more about her book, learn about the brain, and see her intriguing drawings of parts of the brain and some of the tools of her trade.

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**Calendar**

**Angioma Alliance Family Conference 2007**

**November 9-11, 2007**

DoubleTree Hotel  
1515 Rhode Island Ave  
Washington, DC 20005

The 2007 Family Conference will be held at the DoubleTree Washington Hotel in Washington DC. It will begin with a reception at 8 p.m. on Friday, November 9 and end at 1 p.m. Sunday, November 11 with a wrap-up session. This is Veteran's Day weekend in the United States and we hope this will allow more out-of-town folks to attend without missing much work time for travel.

The conference will feature expert presentations, opportunities for socializing, and structured conversation groups. The list of speakers will be announced on our website within the next month.

Accommodations will be available at the DoubleTree Washington hotel at a rate of $149 per night for a single or double room. We will provide more reservation information in the coming months. We do not plan to offer organized child care at this conference, but we can help you make arrangements if you would like to bring your kids.

This year's conference will be held back-to-back with our annual Scientific Workshop. The Friday evening reception will be an opportunity for family conference attendees to mingle with many of the researchers and physicians who are dedicating their work to discovering better treatments for cavernous angioma.

Please mark your calendars and plan to attend. Our conferences are always educational and inspirational, and you won’t want to miss this one.
News

Caring Bears

Brianna Hnath and her Girl Scout troop have been busy making stuffed bears to send to Angioma Alliance members who are in the hospital for a surgery or a bleed. They are decorating the bears with a red bow, a Get Well Soon tag, and an ice pack (on the head, of course). We’d like to thank members of Girl Scout troop #34 in Reading, PA for their caring support.

Advocacy

NIH - Angioma Alliance has been working with Eugene Golanov, who is the new Program Director at NIH responsible for administering cavernous angioma research grants, and with our scientific advisors to increase focus on cavernous angioma research. We have had several productive conference calls and look forward to continuing our relationship. Dr. Golanov is enthusiastic about expanding cavernous angioma research not just as a benefit for those who have the illness, but also for its applicability to other diseases that involve blood vessel growth (angiogenesis).

GINA - On April 25, the Genetic Information Nondiscrimination Act (H.R.493) passed the House of Representatives by a 420-3 vote. Reps. Flake (AZ-6), Paul (TX-14), and Royce (CA-40) were the only nay votes on the bill. This bill makes it illegal for employers and insurance companies to discriminate based solely on a genetic testing result. The bill must now pass in the Senate where it is facing the challenge of being held in committee rather than going to the full Senate floor for a vote. If it is successfully passed in the Senate, it must also be signed by the President. We extend our thanks to all who contacted their congressional representatives to express support for the bill.

Physician Education

Dr. Mike Berg, an epilepsy specialist at the University of Rochester and a strong supporter of Angioma Alliance, has applied to the American Academy of Neurology to offer a half-day continuing medical education course on the diagnosis and treatment of cavernous angioma to the neurologists attending AAN’s annual convention. The convention attracts more than 10,000 neurologists annually, and while not all would be attending the course, if the course is accepted, we hope to attract a sizable number. Dr. Berg will be joined by Dr. Issam Awad, Dr. Jose Biller, Dr. Leslie Morrison, and Dr. Doug Marchuk in offering presentations.

Neurology Residents Award

The Neurology Residents Awards will be announced on June 20th, just past the deadline of this newsletter. Look for award winners in our next issue.

Igive.com

Did you know that Angioma Alliance members have raised over $1000 by doing their online shopping through Igive.com and designating us as their charity? If you go through the Igive.com portal to reach a retail site, any purchase you make results in a donation to Angioma Alliance at absolutely no cost to you. Simply visit www.igive.com, register, and bookmark the site for future shopping.
Wine-Tasting Fundraiser

Kandance Weems-Norris hosted a wine-tasting fundraiser on May 2nd that raised more than $6000. Attendees were treated to a selection of wines donated by sommelier Steve Meyn and to presentations about cavernous angioma and the work of Angioma Alliance given by Michelle Hnath and Christine Castellano. Thank you to Kandance, Steve, Michelle, and Christine.

Participants Needed for Research

Angioma Alliance now has several research projects underway in collaboration with other institutions. We are looking for several types of individuals in the US or Canada to participate:

1. Individuals who have had genetic testing and have been told they have a CCM2 exon 2-10 deletion as the cause of their hereditary cavernous angiomas.
2. Individuals who have had surgery to remove a cavernous angioma at St. Joseph’s Hospital in Phoenix at any time in the last 5 years.
3. Individuals who have cavernous malformations (cavernous angiomas, cavernous hemangiomas) on the skin in addition to cavernous malformations in the brain or spine. We are also enrolling affected family members of these individuals.
4. Individuals who have an upcoming surgery scheduled at any hospital.

If you fit any of these criteria, please email us or call us at 1-866-432-5226. Participating in research is one way to take an active role in finding a cure for cavernous angioma. It is both empowering and much needed.

New Angioma Alliance Treasurer

Karen Asbury has joined the Angioma Alliance board of directors as Treasurer. Karen has had a long career in accounting; currently she is a Manager in Information Technology supporting centralized financial and human resource systems. Karen and her husband Mitch were married in San Diego in 1980. Mitch experienced his first seizure in 1978 while in boot camp; this prompted his discharge from the Air Force. He was told his epilepsy was probably caused by a childhood fall and was not genetic.

Mitch was later diagnosed with multiple cavernous angiomas. Mitch has undergone two surgeries; a left temporal lobectomy in 1994, and a craniotomy in the right occipital junction in 2004. The family migrated to Chesapeake, Virginia after Mitch’s first surgery to be closer to Karen’s large family. They have three children and a two-year-old granddaughter.

Their middle child, Callie (age 22), has since been diagnosed with multiple angiomas through MRI. Callie and Mitch have received confirmation that they are affected by a change in the CCM3 gene. Mitch’s 77-year-old father has been recently diagnosed with multiple angiomas; thankfully he remains non-symptomatic.

Angioma Alliance in the Media

The Discovery Health Channel’s program Mystery Diagnosis during the week of May 14th featured our own Joyce Gonzales and her search for a diagnosis of her spinal cavernous angioma. The segment was well received, resulting in numerous calls and emails for information as well as doubling the traffic to our website. Way to go, Joyce!

Also, the New York Times will run an article about the common Hispanic mutation in New Mexico in one of its forthcoming Tuesday issues. You can check the print version of the newspaper, or you can look on the New York Times website, in the Health section: http://www.nytimes.com.
International Update

Out and About

The Board of Angioma Alliance UK voted unanimously that a representative attend the open day at the Walton Centre for Neurology and Neurosurgery in Liverpool. Due to health and other factors, it was decided that I travel to Liverpool. Also, because of the distance involved from Dorset, the Board recommended that I stay overnight.

The Walton Centre is unique to the National Health Service. It is the only specialist trust in the UK that provides neurology, neurosurgery and pain management services. Based in Liverpool, the Centre serves over 3.5 million people from Merseyside Cheshire, parts of Lancashire, Greater Manchester, the Isle of Man and North Wales. However, with an international reputation in some areas of expertise, referrals are accepted on the basis of need from other parts of Britain and Europe.

I was curious how the event organizer was to carry out the tour, as it was only to last three hours, including lunch! The Chairman, Ken Hoskisson, introduced the session and the tour guides, which included Jane Findlay, Tony Murphy, Julie Elwill, and Sara Vinas, Corporate Services Manager, to guide us through the labyrinthine hospital. We were introduced to Graham Mitchell, Modern Matron, Carol Scott, Practice Educator; Donna White, Neuro X-ray, and Caroline Finnegan, Neurophysiology; Paul Cresswell, Consultant, Neurophysiology; was in attendance too. We had little opportunity to ask questions before we were whisked on to the next part of the hospital.

The finale was conducted by the delightful Miguel Aguirrereburuade, Pathology: laboratory manager and Professor Gus Baker, Clinical Neuropsychologist. They gave us a lecture accompanied by a PowerPoint presentation. The Walton Centre is an excellent facility. I just wish we had had more time!

I recently had the honor of representing Angioma Alliance UK at a member forum of the Long-term Conditions Alliance (LTCA - www.ltca.org.uk), at the Montecalm Hotel, London. My expenses were met by the kindness of Pfizer through the LTCA. Earlier this year, the Board of Angioma Alliance UK decided that we should become members of the LTCA. The group were kind enough to waive their £40 ($75 approx.) membership fee for small charities for the first year.

This was a worthwhile day. In addition to networking with small groups such as the Gorlin Syndrome Group and the Pernicious Anaemia Society, I met individuals from larger charities such as the Parkinson’s Disease Society and Diabetes UK.

LTCA recently had an AGM, which was a useful day. I managed to meet and talk with Melissa Winter from the Genetic Interest Group, which will be very important as we learn more about the CCM gene.

Meanwhile, preparation is underway for the First International Angioma Alliance UK Forum, to be held in London, England, on Saturday 16th June 2007. We are lucky to have Connie Lee, president, Angioma Alliance, assuming her role as “honorary coordinator” of Angioma Alliance UK watching over us and the board wrestle with technical matters (will it be the “right” computer? What happens if the bulb on the projector breaks down?!) But all will be well, I hope.

The final line-up will be Dr. Rustam Al-Shahi Salman, MRC clinician scientist and honorary consultant neurologist, Western General Hospital, Edinburgh, Scotland. His presentation will be: “Epidemiology, symptoms and prognosis.” Mr. Neil Kitchen, Consultant Neurosurgeon and Associate Clinical Director, National Hospital, for Neurology and Neurosurgery, Queen Square, London. Mr. Kitchen’s presentation will be "Surgery for Cavernoma." Then we have Mr. Eric W. Johnson, Director Molecular Diagnostics and BioBanking, Wisconsin, USA. Dr. Johnson’s paper is called "Basic Genetics: An Overview of CCM Genetics and Gene Testing." After lunch we have two speakers: Mr. Ian Sabin, Consultant Neurosurgeon, Royal London Hospital, Whitechapel. The title of Mr. Sabin’s presentation is "Star Wars: The Gamma Knife Strikes Back." Then Dr. Jonathan Berg, Senior Lecturer and Honorary Consultant in Clinical Genetics, University of Dundee, will speak on “Cerebral Cavernous Malformations - if I have one, what does it mean for my family?”

(If you are wondering about surgeons’ titles being “Mr.”, this comes from a long-standing tradition that the cutting of patients’ is performed by “mere” surgeons, the real skill lies with the non-operating doctors.)

Ian Stuart

Editor’s note: The forum was held on June 16th and attracted more than 80 attendees. The event was a smashing success with informative presentations and an opportunity for many questions and discussion. The forum was taped and may be available online via streaming video in the near future.
Research

Seizures

Researchers at the Hospital for Sick Children in Toronto examined the records of 126 children who underwent temporal lobe surgery for intractable epilepsy caused by a wide variety of abnormalities including cavernous angiomas. They found that for those who had temporal lobe surgery for lesions like tumors or cavernous angiomas, 83% experienced permanent improvement in the severity of their seizure disorder. The most common complications of surgery were visual problems, usually in the form of “blind spots” in the visual field. It was not known whether these resolved with time.


A multi-center retrospective study was conducted that involved 168 patients with a single cavernous angioma who were experiencing epilepsy. All of these patients underwent surgery to remove the cavernous angioma and were followed for three years after surgery. More than two thirds of the patients were seizure free throughout the three year post-operative period. The best outcomes were seen for patients who were more than 30 years old at the time of surgery, whose cavernous angioma was in the mesiotemporal lobe, whose cavernous angioma was less than 1.5 cm, and who did not have generalized seizures. These authors recommend that surgery be considered for all patients with a single cavernous angioma who are experiencing seizure.


Genetics

Gault et al have performed a prospective study in which they conclude that on average, those with the CCM1 mutation appear to have a less severe course of the illness than those with a CCM2 or CCM3 mutation. While this study does not tease out CCM2 vs CCM3 severity, it does give further evidence (along with the article published by Denier, et al, that describes a higher risk of hemorrhage for those with the CCM3 mutation) that it is important to know which mutation an individual has before making treatment decisions. It may be that individuals with CCM2 and CCM3 mutations will need a more aggressive treatment approach.


In January, 2007, Liquori, et. al. reported that CCM2 deletions are a common cause of the familial form of CCM. This has led researchers to begin screening their DNA samples for these deletions. Ortiz, et. al. tested Spanish and Portuguese samples and found that samples they held from 11 different families had the same deletion (a 14 bp in exon 5). No familial relationship between these families could be found.


Spinal Cavernous Angiomas

By analyzing the medical records of the 14 patients treated for cavernous angioma in the spine between 1989 and 2002, researchers at Johns Hopkins examined the symptomatic course of the 10 patients who were managed conservatively (without surgery). Remarkably, in this group of conservatively managed patients, no patients were found to have additional acute hemorrhages and their symptoms remained the same or better over the period they were studied.


Who We Are...

Angioma Alliance is a non-profit, international, patient-directed health organization created by people affected by cerebral cavernous malformations (CCM). Our mission is to improve the quality of life for those affected by CCM through education, support, and promotion of research. 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 will help fund conferences and forums, increase research, and enhance outreach and support efforts. Our pins, car decals and wristbands support the mission and growth of Angioma Alliance. Share these meaningful gifts with your friends and family.

Each donation of $10 or more will come with a CCM lapel pin thank you gift. Our “little red guy” pin is a wonderful way to increase awareness of cerebral cavernous malformation (CCM), our little known illness. Increasing public awareness can go a long way toward increasing research funding and improving quality of life for those with cavernous angioma. Each pin comes with cavernous angioma business-size information cards that can be handed to anyone who might have questions.

Angioma Alliance has its own magnetic car ribbon! These unique ribbons are the larger 3 1/2” x 8” size and are available for $5 each, including shipping. Educate while you travel!

To donate or order Angioma Alliance merchandise, send a check or money order (using the enclosed envelope) or visit www_angiomaalliance.org and purchase all of your Angioma Alliance gear in one fast, easy transaction! You can donate and purchase using a credit card with our Paypal connection.

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