Telangiectasia Self Help Group

NEWSLETTER NO.17 – 2010

Congratulations 25th

Dear Member,

I am delighted to welcome you to this edition of the Telangiectasia Self Help Group's Newsletter in which we celebrate the Silver Anniversary of our formation. I can hardly believe that it was 25 years ago when I decided to compile a register of sufferers of the medical condition known as Hereditary Haemorrhagic Telangiectasia [also known as Osler-Weber-Rendu disease] in order to put affected families in touch with one another. Little did I realise that "like Topsy" it would grow to the present membership of nearly 700 and reach across the world to contact other HHT Patient Support Groups and Medical Specialists and their Students. Much of this achievement would not have been possible without the support of yourselves and the Medical Professionals. I would like to pay tribute to all the Medical Specialists who are so dedicated to the treatment and care of sufferers with this condition and who unfailingly support our Self Help Group. My special thanks go to Matthew Fletcher - our WebMaster - whose Technical expertise is invaluable in creating and managing our Website.

Included in this edition are reports from our Medical Specialists, our 2nd HHT U.K. Patients Conference and the 8th HHT International Conference. Also I have included items of congratulations from our friends in the HHT Foundation International Inc. Some of our members have also kindly contributed articles - for which many thanks - which deals with their experiences of HHT and the medications and treatments they have found helpful. Please do get in touch if you would like to communicate feedback concerning any item in this Newsletter.

Finally I would like to send my sincere thanks to all of you who have sent donations which have enabled the production of this Newsletter and of the Information Packs which are sent to all who request them.

With best wishes

Diana Lawson

Congratulations 25th

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Mrs Diana Lawson *Co-ordinator/Organiser*

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NEWSLETTER NO.17 – 2010

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Welcome to the web section of our newsletter.

I would like to thank both Diana and Roger for their sterling efforts with the Telangiectasia Self Help Group, with Diana's perseverance and organisation skills, the TSHG wouldn't be here today in its current form and celebrating its Silver (25th) Anniversary.

Since our last newsletter, our website has continued to grow in hits. The number of hits are in excess of 36,000 for 2009 which reinforces the fact that there is a continued demand for information on HHT.

Our busiest month was January, followed by February and then March. It's suspected that this is due to the 2nd UK HHT Family Meeting that took place in February.

Having noted sources of the hits by country, the majority of our hits are from the UK closely followed by North America and Europe. Our website has seen many hits from all over the globe.

It's also been refreshing to receive communication from the Medical profession seeking advice regarding treatments and typical symptoms of their patients.

Our plans for 2010 are to continue to support members and to provide continued updates regarding HHT diagnosis, management of symptoms and to reduce continued expenditure by distributing our newsletter and other communication via electronic mail.

If there is anything else that you our members would like to see on our website, please do not hesitate to contact either Diana or myself

Matt

Webmaster
TSHG

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HHT Foundation International, Inc.

(Osler-Weber-Rendu Syndrome)

Congratulations Telangiectasia Self Help Support Group
25 Years of HHT Advocacy!

On behalf of the HHT Foundation International, it is an honor to wish you a Happy 25th Anniversary! The TSHSG was a ***pioneer*** in 1985 through the establishment of a registry for all HHT affected families. You were able to recognize the critical need for HHT families to link to one another.

Through our combined advocacy efforts and those throughout the world, we have been on a journey seeking advanced screening and treatment options for our families affected by this uncommon disorder.

The first patient meeting of HHT patient advocacy organizations from Europe, South America and the United States held in Santander, Spain in 2009, allowed all of our groups to discuss common issues that affect us all. We are excited about collaborations that will help our worldwide community!

HHT families from throughout Great Britain are now much more educated about HHT and have received treatment through your two outstanding Centers of Excellence. Additionally, there have been great research strides through the collaboration of HHT patients and the HHT Treatment Centers. This is all possible through your ongoing commitment to the HHT community in the United Kingdom.

Best Wishes for continued success and, together, we will find a cure for HHT in our lifetimes!

Warmest Regards,

Marianne S. Clancy

Marianne S. Clancy
Executive Director

Happy Silver Anniversary TSHSG!

My goodness, the Telangiectasia Self Help Support Group is having its Silver Anniversary in 2010! That certainly is longer than the HHT Foundation has been in existence. It was only starting the process of forming in 1989.

Twenty years ago this past summer, I found a name and telephone number for an HHT Registry in a library book on genetic disorders. I called the number. And suddenly, my family was no longer the only people in the world with HHT. But I never imagined that, in 2010, there would be a well-known international foundation and many, many HHT treatment centers all over the whole world. Or that I would be a part of it.

I must be getting old, though, because when Diana Lawson asked me to write about our history together (the HHTFII and the TSHSG), I honestly could not remember when we were first associated. So I searched some of my older records and finally found mention of Diana as Coordinator in 1992 in our 4th newsletter. It included information on how to get in touch with her if had HHT and you were located in the UK.

Here it is exactly as it was printed in that newsletter.....

> For those of you with family in Great Britain, you might like to know that there is a TELANGIECTASIA SELF HELP SUPPORT GROUP there which operates entirely on voluntary donations. To receive their Information Sheets or Newsletters, write to:
>
> **Mrs. Diana Lawson**
> **39 Sunny Croft, Downley**
> **High Wycombe, Bucks, HP13 5UQ**
> **or call: 0494 528047**

Since that time (wow, it's 18 years!) I have referred many, many hundreds of people to the TSHSG, and to the British HHT treatment centers.

But it was not until 2001 that we eventually met Diana, and Roger too, in Denmark at the HHT Scientific and Medical Conference. And we met again at the next Scientific and Medical Conferences in Bonaire in the Dutch Antilles and the Canary Islands in Spain. And finally in 2005, Roger and Diana paid a visit to Vancouver, near where my husband and I live and together we all had a very pleasant visit seeing the sights together.

So much has happened in the world of HHT in the last 18 to 20 years. So much has been accomplished. So I'd like to take you on a little trip down Memory Lane, of the things that stand out in my mind as the most impressive.

- The first scientific conference was held in Edinburgh in I think 1992 or 1993 (you see, my memory is going!). It was attended by somewhere between 25 and 30 HHT scientists and physicians, most of whom are still coming to these conference, which are held every other year. They have been held in France, Italy, Curacao and each time there are more and more attendees. At the recent conference in Spain this year there were approximately 200 HHT scientists and physicians. It's so encouraging to know that these people really care this much about HHT and about making life better for the people who have it.

- In 1995, after begging the Lay Board to please set up a website (they were a bit nervous about the internet at that time), we finally did go online at www.hht.org, and since then I have been <hhtinfo@hht.org> , answer every single email that arrives. And may I say that there are hundreds. Almost all inquiries about HHT come from the internet nowadays, and I am charged with supplying them all with the needed information and referring them to the nearest HHT treatment center. There is an average of about 350 new contacts every year and another 350 questions from people who have already contacted us, but need further information and support. And the best of all, many of these inquiries are from family physicians in many different countries who want information to be able to help their patients.

- And in keeping with my part as information provider on HHT, over the years I have accumulated stacks of information from the Global Research and Medical Advisory

Board, innumerable medical articles and personal and anecdotal stories from people with HHT. So much information that I am able to send people information on exactly what they need. If I cannot answer it, I go to one of the people on the GRMAB for assistance.

- There is an online Discussion Forum where people with HHT can go to compare notes on the HHT website and another one in the UK. There is a Facebook page where people with HHT can post messages as well.
- Where once there was only the Yale HHT Treatment center in New Haven CT., there are now over 30 HHT treatment centers around the world, all with very experienced teams of specialists. Isn't that wonderful!!
- Where once there was only the TSHSG in Britain, there are now support groups like the TSHSG all over the world, in Israel, Spain, Germany, Denmark, Central America, the United States, and other countries. New groups are forming all the time. People helping people.
- There are now many patient information conferences held by these support groups that provide information and support to the people with HHT who live within their boundaries. The TSHSGwas the first to do so and has already provided two such helpful conferences.
- And one other note, something that stands out in my mind because it affects me so personally. The HHT logo with the earth and the family in a circle was designed in 1993 by my mother, who of course, also had HHT. It has become the symbol for HHT all around the world. I know she would be very proud.

One last important point to make … just like the TSHSG, the HHT Foundation relies on the generosity of its membership for funding. We have no other funding. We hope someday that we will all be able to have enough funding to find a cure for HHT.

And so, I wish for the members of the Telangiectasia Self Help Support Group another 25 years of success and a very bright future.

Trish Linke
Patient Education Liaison
HHT Foundation International Inc.
hhtinfo@hht.org
www.hht.org

25 years 1985–2010

Letters from our members including their personal experiences connected with HHT and their queries.

Dear Diana,

I and my two eldest children and their spouses were very pleased to attend the 2nd UK HHT Patients Meeting in February 2009 in London at the Hammersmith Hospital. After registration, continental breakfast and the Conference welcome we had a series of very informative talks covering the various aspects of HHT by very Specialist Doctors. After a coffee break we had more talks and a general discussion by all the speakers and the audience when our questions could be asked. We had a very nice lunch followed by workshops in the different suites allocated to the Specialists. In these workshops we were able to meet the Doctors more informally and ask about the aspects of HHT we were personally interested in - in my case PAVMs and nosebleeds. I had a PAVM embolised in Leeds General Infirmary over seventeen years ago and at that time I did not realise that I had HHT and that it was an inherited condition. I was so pleased to find out that because of a previous PAVM I should have a CT scan at least every five years. Back in the conference room we were shown the mouse models and an update on the current clinical research and ways forward were indicated. After which a final discussion with all the Doctors and Specialists was held and then the day concluded with an informal reception.

I would like to add my thanks to Dr. Claire Shovlin and to Mrs. Diana Lawson for all the hard work they must have put in to organising such an event (at which all tickets I believe were taken). As a direct result of going to this meeting, I got a referral to the Hull HHT Centre where I met Dr. Graham Robinson the Centre's Director. Following a CT scan I have just had a Pulmonary Embolisation performed by Dr. Robinson in Hull Royal Infirmary. Everything went very well, so very grateful thanks to the team at Hull Royal Infirmary. I am also now under the care of Professor N. Stafford the ENT Consultant for HHT at Castle Hill Hospital for my nosebleeds and this treatment is still ongoing.

Myself and my family thought the Conference was a resounding success and we look forward to the next UK Patients Meeting.

Yours sincerely,

Kathleen Dowker.

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Dear Diana

Just a few lines to update my personal story. Since I last wrote of my experience with grafts and laser treatments at the Royal National Throat Nose and Ear Hospital, my life has greatly changed.

Back in 2008 my nose bleeding increased to the point of needing yet more laser treatment, so, at the age of 70, in January 2008 Professor Lund surgically closed my right nostril.

This worked well, but I continued to bleed from the left side. After a very heavy bleed and emergency treatment, I finally decided enough was enough and requested a second closure in January 2009.

Although I now have no sense of smell and eating can be quite difficult because I need to take a breath whilst chewing, my daily activities – walks to and from school with my granddaughter, gardening, shopping, general housework, bending down and lifting – can now go ahead without any problems. I no longer need to take tissues, cotton wool etc. when I leave the house, and bloodied tissues in pockets and handbags are a thing of the past.

Having tried various "treatments" such as No-Bleed, tranexamic acid etc., nothing really worked for me. Now I can fly away on holiday, sit in a restaurant without checking where the toilet is in case of a nosebleed, and have glass of wine or two without worrying.

So to sum up, after so many years of embarrassing moments I can plan ahead and look forward to a better quality of life.

My thanks to the clever doctors and nurses at the Royal National Throat Nose and Ear Hospital who have changed my life for the better.

Ann Price, January 2010

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Dear Mrs. Lawson,

Telangiectasia Self Help Group

Thank you for newsletter No.16 that I have recently received. I have found the contents of great interest and I feel that my own experiences may be of help to other members.
I have had major bleeding problems in my stomach and from my nose and give below details of the problems and subsequent treatment.

Stomach

I had regular stomach bleeds which resulted in me bringing up, at times, as much as 1 to 2 litres of congealed blood. I was referred to Sheffield Hallamshire Hospital were I received Argon Laser treatment (via endoscopy) This has been a great success and has given me relief for, at best, up to 24 months before further treatment is needed.

Nose

I was having bad nose bleeds a number of times each day, which was necessitating me having blood transfusions at roughly 2-week intervals. At this time I got very weak. I was referred to Sheffield Hallamshire Hospital, under Mr. Woolford the ENT consultant. Mr Woolford had invented a plug made out of soft touch plastic and he asked me to give one a try. An impression is taken of both nostrils with a link piece joining the pieces together. The impression is done in a similar manner to the one used for hearing aids with quick set epoxy. The plug is called an obdurator. The attached photograph shows the obdurator. Please contact the TSHG if you would like a copy.
This plug has transformed my life, before it was not safe for me to leave the house. I have now been on holidays, including cruises. The obdurator is absolutely magic. I wear it at all times except when having a meal or when I am in bed.
I take Cyklokapron (Tranexamic acid) 3 times a day to help clotting.

Please spread the word to any similar sufferers.

Pulmonary Embolism

I had a blood clot in my lung and was not allowed to be treated with Heparin or Warfarin as my bleeding at that time was so severe. I was fitted with a Vena Cava filter to prevent any further clot material reaching vital organs.I have scanned a page of the descriptive leaflet of the filter. Please contact the TSHG if you would like a copy.

I hope this information can be of use and give the relief that I have gained

Kind regards & best wishes

M Brown

Marjorie Brown

Mrs Diana Lawson Coordinator/Organiser
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Emily's Story

When first being diagnosed with HHT I was at the age of 15. This was at first, very scary news as I didn't really know what this meant to me. My mother had a liver transplant due to HHT when I was around 5 years old. My feelings at first were very anxious and sad as you can understand. For me this meant having pulmonary angiography and embolization of my left basal PAVM, which is a small stent being placed into my lung to prevent problems, such as a stroke. The procedure performed by Mr Jackson went well and I was back home in Morecambe after an overnight stay in St Marys. I couldn't really believe the news as I was the youngest out of my siblings only to be diagnosed, being anxious I emailed the team at London asking questions about the procedure, which looking back now I can't thank them enough for! From the day I was diagnosed to the present day the support from the team is world class which gives me great piece of mind. Throughout the whole process of having the operation the team were so understanding and supportive. Thankfully everything went well; after all they are the world's specialists! Living with HHT hasn't changed my day to day life; I am fit, well and enjoy a full active life style. I am proud to be part of research and to potentially help thousands of others. The commitment in London and personal history has inspired not only me, but family and friends to fundraise for my Nana's charity specially dedicated to HHT,(The Margaret Straker Memorial Fund). With the help of my family and friends we are planning more fundraising events later this year.

Emily Morrow

If you have had any experience of using the following products could you please let me know so that I can pass on this information to other members of the TSHG:-

Sterimar - Colostrum - Veinwave - Homeopathic Phosphorous 30 - Merocal Standard Nasal Dressing - Rapid Rhino.

Many thanks - Diana Lawson.

Mrs Diana Lawson Co-ordinator/Organiser

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2nd UK HHT PATIENT'S AND FAMILY CONFERENCE.

The 2nd UK HHT Patient's and Family Conference was held at the Hammersmith Conference Centre in Hammersmith Hospital on Saturday 28th February, 2009. This event was well attended by 130 delegates all of whom had come to listen to and learn from the Medical Specialists and Prestigious Speakers who are committed in their efforts to treat and cure the condition known as Hereditary Haemorrhagic Telangiectasia. The Conference Centre looked after us brilliantly providing delicious food, comfortable seats, relaxing areas, an excellent lecture room [with all technical modern equipment] and ample car parking.

The morning programme entailed presentations by the invited Specialists, after each presentation there was an opportunity for an answer and question session. During the lunch break delegates were able to meet each other socially and also approach the Specialists with more individual questions. The afternoon featured four workshops which concentrated on certain aspects of HHT. Delegates were able to choose which workshop to attend. Each workshop was timed to last about 20 minutes and were operated on a rolling programme, so that everyone could attend the workshop of their choice. The meeting concluded with a general discussion forum back in the lecture theatre. Before delegates left for their homeward journey an informal reception rounded off the day.

This event was such a thrill for all participants - I'm not sure who was more enthusiastic - the delegates or the Specialists! Morale was high after such an exhilarating day. My grateful thanks go to all those who were involved in the organising and planning of the day, to all the contributors and especially to Dr. Claire Shovlin. For those of you who were unable to attend I have attached a copy of the Conference Programme.

Diana Lawson.

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CONFERENCE PROGRAMME
Saturday 28th February 2009

MORNING

09.00-9.30	**REGISTRATION** *and continental breakfast*	
09.30 -9.35	**CONFERENCE WELCOME:**	
09.35-11.00	**CLINICAL HHT 1: OVERVIEW & BLEEDING:**	
09.35-09.55	Overview of HHT	Dr Claire Shovlin
09.55-10.20	Nosebleeds in HHT	Professor Valerie Lund
10.20-10.40	Gastrointestinal and hepatic HHT	Dr Andrew Thillainayagam
10.40-10.50	Diet and HHT	RD James le Couteur
10.50-11.00	Iron treatments	Kandarp Thakkar MRPharmS
11.00-11.30	***Coffee Break***	
11.30-12.30	**CLINICAL HHT 2:**	
11.30-11.50	Pulmonary AVMs	Dr James Jackson
11.50-12.10	Shunts, diving, migraine & stroke	Dr Peter Wilmshurst
12.10-12.30	Pregnancy and HHT	Mr Andrew McCarthy
12.30-12.50	Children and HHT:	Dr Nicky Coote
12.50-13.00	The patients' perspective	CN Robert Hickey
13.00-13.15	*Clinical discussion*	*All morning speakers*
13.15-14.15	***Hot Buffet Lunch (Ratatouille or Beef Strogonoff)***	

AFTERNOON

14.15-15.30	**HHT WORKSHOPS**	

Conference attendees will be invited to rotate around workshops every 15-20 minutes. These will provide an opportunity to ask more informal questions in small groups

Workshop 1: *Oak Suite:*	PAVMs and GI Dr Jackson, Dr Wilmshurst, Dr Thillainayagam
Workshop 2 *Maple Suite*	Nose bleeds and diet Professor Lund, Mr James le Couteur
Workshop 3 *Ash suite*	Children, and general pharmacy issues Dr Coote, Mr Kandarp Thakkar
Workshop 4 *Hawthorne*	Genetics (Basic or Advanced) Dr Claire Shovlin

15.30-15.45	***Tea Break***	
15.45- 17.00	**SCIENCE AND FUTURES:**	
15.45-16.10	Mouse models of HHT	Dr Helen Arthur
16.10-16.35	Clinical research update	Dr Claire Shovlin
16.35-17.00	Ways Forward: EuroHHT	EuroHHT members
17.00-17.30	**FINAL DISCUSSION**	All faculty
17.30-18.00	***Informal reception.***	
18.00	***Conference Closes***	

If you have any questions that you do not want to ask in person, write them down on a piece of paper and give to one of the conference faculty.
We will then discuss your questions in the final discussion of the day.

The road to genetic testing in HHT – a personal story

Professor Mary Porteous, Service Lead SE Scotland Genetic Service

In February 1989 I moved to Newcastle to take up my first genetic job as the Catherine Cookson Research Fellow. As most of you know, Catherine suffered from HHT and was keen to push research forward. She had a list of people that had contacted her when she wrote of her own experiences of HHT in her autobiography, "Our Kate" and suggested that I might contact these people to see if they would help with research to identify the genetic cause of HHT. I was fortunate to be welcomed into many of your homes and was touched by the generosity of the HHT "community". Nowadays such a project would be impossible; by the time consent had been obtained from the national research ethics committee and the individual research and development departments my two years of funding would be at an end.

Armed with family trees, clinical details and DNA from blood samples I worked in the lab with my technician Livvy Kirk to try and identify the location of the HHT gene. Progress was slow. I could tell where the gene wasn't but not where it was. In the autumn of 1989 I took a poster to the American Society of Human Genetics meeting in Baltimore Maryland where I met up with a fellow clinical geneticist Alan Guttmacher from Vermont who told me about a very large Vermont family with HHT. DNA from this family had been passed to Francis Collins, the hero of the 1989 meeting after cloning the gene responsible for another serious genetic disease, cystic fibrosis. I made it my mission to hunt Francis Collins down and cornered him over a donut two mornings later. Francis was charming and passed me swiftly to another member of his team who was about to set up his own research group, Doug Marchuk. Doug and I hit it off straight away and agreed to share data of the HHT "mapping project". In 1992 I visited Doug, then working at Ann Arbor, Michigan to use a new technology called microsatellite marker to try and find the elusive HHT gene. Again, no joy. However 3 months later, Doug's team finally identified the location of the first HHT gene, endoglin. Over subsequent years it has been a source of great joy to me to be associated with the identification of the second gene, ACVRL1 and to contribute to the understanding of how changes in these genes can cause specific clinical features.

Over recent years my research direction has changed and I have been more focussed on colorectal cancer but I have always been interested to follow developments in the field of HHT. The molecular diagnostic laboratory in Edinburgh hosts the mutation database on behalf of the HHT Foundation recording genetic changes found in Endoglin and ACVRL1 associated with the clinical pattern of HHT. We also offer a genetic testing service for HHT. This testing is generally accessed through local Genetic Services. HHT remains a clinical diagnosis and the recently published guidelines have confirmed the "Curacao criteria" are still robust in making the diagnosis. Genetic testing may be of value in a family where the diagnosis is uncertain in a family member. Access

to genetic testing across the UK is patchy – the budget each Department has available for such tests is limited and local criteria are applied. The most costly part of testing is finding the genetic change in the first affected member of the family to be tested. If and when the family change is demonstrated, other family members can be tested more easily.

Any families that took part in my original research should mention this to their local genetic service as we will waive the costs of identifying the family genetic change in the first affected member tested.

Although I am not actively involved in HHT research I am still very interested in the work of others. It has been a privilege to be part of the HHT community and I am delighted to be part of the 25th anniversary edition of the TSHG newsletter. The last 25 years have seen a quantum leap in our scientific understanding of the basis of HHT and I am optimistic that the next 25 years will see the same sort of advances in treatment.

Prof Mary Porteous
SE Scotland Genetic Service
Western General Hospital
Crewe Road
Edinburgh
EH4 2XU

Gums, teeth and antibiotics: The dental issues in HHT

Article written by Claire L. Shovlin, PhD FRCP, Lead Clinician HHTIC London

For many years, people with HHT and pulmonary AVMs (PAVMs) have been advised to use antibiotics before having a dental procedure. This is called 'antibiotic prophylaxis'. However, there has been great confusion in HHT circles following new guidance to dentists issued in 2007 and 2008 [1, 2]. Although those recommendations referred to heart patients, they have led to dentists being reluctant to give out antibiotics to people with HHT and PAVMs.

If you are confused about the advice you are receiving from your doctors and dentists, or unsure why you need to think about your teeth, then this article may be helpful. It is based on the advice we give to people attending the HHTIC London centre in the UK, and stems from several years of discussions between HHT/PAVM specialists Dr Claire Shovlin and Dr James Jackson, microbiologist Dr Kathy Bamford, and very importantly, the Chairman of the British NICE Dental Committee, Professor David Wray. The summary for dentists is published in the British Dental Journal [3] and can be accessed in full at the journal website.

Background:

Bacteria (bugs) from around the teeth and gums can leak into the bloodstream, and if they are not cleared properly, can set up infections at distant sites away from the mouth. For people with pulmonary AVMs and HHT, our biggest concern is that such bacteria can lead to the development of a brain abscess. Ever since this was first recognised, antibiotic prophylaxis has been recommended for people with PAVMs, based on the advice given to people at risk of endocarditis due to heart disease.

At HHTIC London, we have been particularly concerned about this issue for a number of years. In 2000, we approached the British Society for Antimicrobial Chemotherapy and Dental Formulary Sub-Committee who approved a PAVM-specific card for dentists, making it easier for people with PAVMs to explain why they needed antibiotics. From 1999, the team were also gathering 6 years worth of evidence to work out which people with PAVMs were more likely to have a brain abscess. When these data were finally available, they strengthened the link between dental microbes and brain abscess [4]: Most of the bugs grown from PAVM-associated brain abscesses were bugs that had come from the gums, and lots of the PAVM-brain abscess patients described recent dental procedures that would lead to having more bugs in the blood stream [4].

Yet at the same time, we were receiving several calls a month from dentists and PAVM patients, saying that because of the advice from the American Heart Association and British National Institute of Clinical Excellence (NICE) [1,2], our card was invalid and antibiotics should no longer be given.

Who was right?

Why do heart patients not need antibiotics any more?

The AHA and NICE withdrew recommendations for antibiotics for people with heart disease because of 1) the low risks of endocarditis without antibiotics; 2) the lack of evidence that antibiotics prevented complications; 3) the knowledge that for some people, antibiotics could cause harm; and 4) Recognition that bugs get into the blood stream from the gums following everyday events such as tooth brushing.

Their main conclusion was to recommend that preventative action was taken long before the trip to the dentist, not by taking antibiotics, but by making sure the gums and teeth were in as healthy a condition as possible.

What about people with PAVMs?

Unfortunately, the risk of brain abscess for someone with HHT and PAVMs is a lot higher than the risk of endocarditis for a heart patient. There are several potential reasons for this, which we discuss in our article [3]. But importantly, it means we can't use the same arguments as for heart patients. By highlighting that HHT/PAVM risk of brain abscess is substantially higher than endocarditis risks from heart disease, we have said that for now, antibiotic prophylaxis should still be given to HHT/PAVM patients.

> ***BOX : WHAT SHOULD I DO?***
>
> ***1: Everyone in HHT families should look after their teeth, and tell all the children in the family to look after their teeth too. It will probably be good for you in other ways too, and will definitely do you no harm.***
>
> ***2: When you go to the dentist, if you have PAVMs, let them know that you are not like a heart patient and that at the moment, you still need "antibiotic prophylaxis". This is important for any procedure including a 'scale and polish'***

And in the future?

We still can't be sure we are right. We need further research in the area, and have therefore designed a small trial, for which the HHT Foundation is trying to obtain funding.

References

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2) Wray D, Ruiz F, Richey R, Stokes T, Guideline Development Group. Prophylaxis against infective endocarditis for dental procedures-summary of the NICE guideline. Br Dent J. 2008;204:555-7.

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Need for Antibiotics before Dental Treatment

Recently published NICE guidance on antibiotic prophylaxis against infective endocarditis has been widely circulated to doctors and dentists. This recommended that patients at risk of this heart infection no longer need to take antibiotics before dental treatment. As a result fewer people are now offered antibiotics before dental treatment.

NICE, however, did not consider patients with pulmonary arteriovenous malformations (PAVMs) and hereditary haemorrhagic telangiectasia (HHT) and so the NICE guidelines don't apply to them.

Patients with pulmonary arteriovenous malformations (PAVMs) and hereditary haemorrhagic telangiectasia (HHT) are also at risk of infections from dental bacteraemias caused by dental treatment. There is a particularly strong link between dental bacteria and brain abscess.

In HHT/PAVM circles, antibiotic prophylaxis has been recommended before dental treatment and in contrast to cardiac patients with infective endocarditis, a high proportion of the PAVM-brain abscess group had experienced identifiable events known to be associated with bacteraemia in the weeks preceding their abscess

Antibiotic prophylaxis, therefore, should still be given to PAVM/HHT patients so you need to ensure your dentist is aware that such patients are different from the patients considered by NICE.

Also, all HHT/PAVM patients and their families should ensure that they pay particular attention to maintaining good oral hygiene since this is known to reduce the risk of bacteria entering the bloodstream.

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NEWSLETTER NO.17 – 2010

8th HHT INTERNATIONAL SCIENTIFIC CONFERENCE.

The 8th HHT International Scientific Conference was held in Santander, Spain from May 27 - 31st 2009. The International HHT Scientific Conference takes place every 2 years and it provides an opportunity for Scientists, Physicians and Clinicians with an interest in HHT to meet and discuss their work. This 8th International Scientific Conference was hosted by the Spanish HHT Community and organised by the HHT International Committee. It was held in the Palacio De La Magdalena situated in a picturesque park on the Peninsula De La Magdalena. There were 213 delegates in attendance, many of whom had travelled great distances across the world to be present at this Conference. These conferences continue to consolidate human and scientific relationships between the HHT Community and inspire new research into this disease resulting in the expansion and flourishing of HHT Treatment Centres around the world. It was particularly heartening to realise that many of the original 49 delegates from the very 1st International Scientific Conference held in 1996 in Edinburgh, Scotland were also present in Santander.

After an informal social gathering during registration on the evening prior to the conference a full programme took place with continuous lectures and workshops running alongside each other in Scientific, Clinical and Basic Research. There were opportunities to socialise and look at the posters displaying the work in progress during the meal breaks. All the presentations were of a very high standard and some reports of exceptional research into HHT instigated animated debate and discussion. Dr. Claire Shovlin presented the results of the work funded by our own TSHG families and her findings were well received. Once again it appears that the variation experienced in different countries in the funding or non-funding of research and treatment of rare diseases greatly affects the outcome for the patient.

At this conference the involvement and treatment of nosebleeds continued to be focused on and the specialists are still looking for answers in the research results so that a common criteria can be achieved. The Curacao Criteria is still a guideline and valid for HHT1 and HHT2, meanwhile new tools are being developed for diagnosis in some areas. The practice of screening for PAVMs, CAVMs and AVMs varies across the medical world. PAVM sufferers particularly should be aware of the need for antibiotics when they have dental issues - see reports in this Newsletter. In the future it is necessary to pool data for successful treatment of PAVMs, Stroke complications and the standardisation of the Echo method used in diagnosis.

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8th HHT INTERNATIONAL SCIENTIFIC CONFERENCE [continued]

The very first meeting of the representatives of the Patient Support Groups was held during the afternoon of the 28th May 2009. This event had been longed for by all involved for some years, so it was particularly exciting to hold the inaugural meeting in the splendid dining room of the Magdalena Palace. Seated around the table were delegates from the Patient Support Groups in the following countries:- Israel, Italy, Germany, Spain, Norway, Guatamala, Argentina, Ireland, France and myself, representing the UK. We were all invited to give presentations outlining the history of the formation of our group and explaining how we were able to inform our members and enquirers about HHT. We also were asked about our present membership numbers and our plans for the future. Fortunately there was a multi-language interpretation system in place so that we could understand each other! I was amazed to discover that the TSHG was the most senior group - and indeed was the very first to be formed. I found myself giving advice to other newly formed groups and introducing Medical Specialists to the relevant Patient Support Group representatives. As a group we decided to join together to apply to the EU for grants which are available to further the work and treatment of Rare Diseases. United we can request help with a louder voice! There are many stages to such applications so I guess that it will take some time before we see any results. We also decided to look into the possibilities of setting up an International Website to which we could all contribute so that sufferers throughout the world could find information about HHT in their own language.

At the end of the Conference prizes were awarded and thanks and praise was given to all those who had contributed to the organising and running of such a successful event. The future for HHT research is exciting and the HHT patient community worldwide is awaiting the results of the expanding volume of HHT research. We look forward to the future of science leading the way to eliminate HHT and to "wrest from Nature, the secrets which have perplexed philosophers in all ages, to track their sources, the causes of disease....." [quoted by Sir William Osler at the beginning of 1900s].

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Epistaxis in HHT

Although often referred to as Osler-Rendu-Weber disease, HHT was probably first described by Sutton nearly 40 years earlier in 1864 when he referred to '*Curious cases of recurrent epistaxis associated with internal hemorrhages and telangiectases of the skin*'.

Nosebleeds are by far the commonest symptom in HHT, being the presenting symptom in 90% of patients and a serious problem in 80% by the age of 30. The fundamental problem is that the red spots or telangiectasia have fragile walls, which lack muscle that would normally allow the vessel to shut down when damaged so any minor trauma starts the bleeding and this carries on until the blood clots. Spots in the nose are therefore very vulnerable simply due to the constant airflow, whereas they are protected in other parts of the body. Fortunately, not all nosebleeds are serious and there is a wide range of severity from infrequent mild spotting to frequent, severe bleeds which require hospital admission and regular blood transfusions. Furthermore there does not appear to be any natural progression from mild to severe – some people just seem to be more fortunate than others. Consequently there is also a long menu of treatments, largely dependant on severity and whilst none are truly curative, the frequency and severity of nose bleeds can often be reduced for varying periods of time. Many of these treatments aim to protect the telangiectasia in various ways.

From a practical point of view, my treatment menu based on treating nearly 300 patients remains as shown (Figure 1), with coagulating lasers such as the KTP, neodymium YAG and argon helping in the milder cases together with the Hammersmith triple ointment regime if it can be used without making the nose bleed when the ointments are applied. In the severely affected, I still use closure of the nose which by stopping airflow, stops the bleeding but also of course means that one has to breathe through the mouth. This seems a reasonable price to pay however, if one's nose is permanently blocked anyway with clot and the bleeding is very disruptive and even life-threatening. From a quality of life perspective, this procedure has the greatest positive impact. In between these two ends of the spectrum are a number of other treatments such as grafting the nose, hormone treatments and medicines which alter clotting, all of which can be helpful in some people.

Another seemingly logical approach might be to block off the main blood vessels supplying the nose, particularly as this can now be done either using interventional radiology to perform embolisation or nasal surgery using endoscopes to access the blood vessels. Because the nose has an excellent (too excellent, one might think) blood supply, it is not possible to completely shut down the blood supply at the level of the telangiectasia themselves, only diminish it. Thus whilst these treatments can be extremely helpful in an acute situation to help with a massive nosebleed, they do not help so much in the longer term for more chronic bleeding. However, they can be used very successfully in the lungs and other organs.

Generally in medicine there is an increasing interest in outcomes of treatment, not just in terms of reduction of the specific symptoms but also the effect that this has on quality of life - and this has been looked at in HHT. To qualify improvement, there has been some work done on grading the severity of the bleeding. My own rule of thumb has depended simply on whether or not the nose bleeds have resulted in the

need for regular blood transfusions and not surprisingly sufferers know themselves whether their nosebleeds are mild, moderate or severe. Measuring the haemoglobin in the blood is a rather less accurate indicator, probably because it is affected by other factors. A recent paper tried to put together the various grading systems (Table) and this may be helpful for future research projects.

I and my co-workers are very interested in looking at what factors trigger the nose bleeds and also how the condition and its treatment affects you and your family. I hope that with the help of the HHT society that we will be able to explore these aspects with you in the future.

Reading List

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