

HHT Foundation International, Inc.

Where We've Come From

By Trish Linke, Patient Education Liason; and Dr. Robert I. White, Jr., Interventional Radiologist and Director of the Yale HHT Center of Excellence

Have you ever wondered how a rare disorder was discovered, how a Foundation is established, and how a small group of people can have such a tremendous impact on people around the world? Well, here's the story of Osler-Weber-Rendu Syndrome, also known as Hereditary Hemorrhagic Telangiectasia (HHT).



Dr. Osler



Dr. Weber



Dr. Rendu

In 1864, there was a report about recurrent epistaxis due to degeneration of the vascular system and impaired nutrition. In 1896, Dr. Rendu of France recognized the combination of hereditary telangiectasias and epistaxis as a specific entity, followed by reports of Dr. Osler of Johns Hopkins Hospital, USA and Dr. Weber of Germany. The disorder took on the name of the three key researchers / clinicians, Osler-Weber-Rendu Syndrome as it's known in the United States or Rendu-Osler-Weber Syndrome as it's known throughout Europe. In 1909, Dr. Hanes introduced the term "Hereditary Hemorrhagic Telangiectasia" since it better described the disorder.

H Hereditary, genetic, autosomal dominant vascular disease

H Hemorrhagic, blood

T Telangiectasia, vascular abnormalities vary from small (telangiectasias) to large (arteriovenous malformations – AVM)

Now, jump forward 80 years to the 1970's at Johns Hopkins Hospital in Baltimore, Maryland (USA). This was a very fertile time in radiology in which the subspecialty of cardiovascular and interventional radiology was developed. One of the areas of interest was embolotherapy (occlusion) of arteries and veins using image guided catheters by radiologists with special expertise in this area. Dr. Robert I. White, Jr., Interventional Radiologist, and his associates conducted a number of investigations of different materials that could be placed in arteries to control bleeding. By 1978, they had developed, in conjunction with Becton-Dickinson Corporation, a detachable balloon for use in blocking large arteries and veins to control hemorrhage from trauma or from benign and malignant tumors. Serendipity often occurs in medicine, and a patient with multiple pulmonary arteriovenous malformations (PAVMs) and HHT was admitted to the internal medicine service under Dr. Peter Terry, now Vice president of the HHT Global Research and Medical Advisory Board. Dr. Terry and Dr. White spent some time with

this patient and offered her an alternative to surgery, namely, the occlusion of her PAVMs using detachable balloons. After several catheterizations and occlusions, this first patient responded favorably with a rise in her oxygen, which had been sustained for over 15 years. They first reported a similar patient in the New England Journal of Medicine in 1980. Subsequently, they found a report in 1978 of a patient treated in Indiana using stainless steel coils and a 1977 report from East Germany (now United Germany) using homemade stainless steel coils. Their patient was the third patient in the world to be treated by embolotherapy and the first patient to be treated with detachable balloons.

Between 1978 and 1982, Dr. Terry and Dr. White treated 17 additional patients with good results. During 1983, Ms. Sharon Victor, a patient from Palo Alto, CA was treated successfully at Johns Hopkins Hospital. Dr. White mentioned to her that this therapy seemed unique and it would be wonderful if someday a foundation could be formed to better inform patients and healthcare professionals about the various options for patients with HHT and PAVMs.

In 1985, Dr. White learned that Dr. Bruce Jacobson, a biochemist at the University of Massachusetts, formed the first HHT patient registry. They began a series of telephone calls and conversations and Dr. Jacobson asked Dr. White to be on the Medical Advisory Board. Over the course of the next 3-5 years, they corresponded. During this time, Dr. White moved to Yale School of Medicine as Chair of the Department of Radiology.

In 1990, Ms. Sharon Victor, Dr. Bruce Jacobson, Mr. Dennis Donahue, a lawyer in West Haven, CT, and Abby Meyers of the National Organization of Rare Diseases (NORD), met in Dr. White's office at Yale to discuss the formation of the new HHT Foundation. The foundation later became an International organization, followed up by obtaining non-profit status, 501(c)(3).

By May 1991, fifteen patients and/or their families met in New Haven, CT for the first organizational meeting of the HHT Foundation International, Inc. Over a period of 48 hours, the foundation was born and over the course of the first year, the foundation experienced some tumultuous events as business was conducted by long distance calls between various board members. By May 1992, the foundation seemed to be on the right track and the first patient meeting was held in Columbus, Ohio on July 25-26, 1992.

From 1992 to 2000, the Foundation evolved. In 2001, we hired an Executive Director, Marianne Clancy, who still holds that position and the Foundation began to raise money and awareness around the world. We now have 23 HHT Centers of Excellence across the United States and throughout the world, we've held 13 national patient and family conferences, we've hosted 7 International HHT Scientific Conferences, partnered with NIH and CDC in the United States and grown to over 8,000 members, donors, contacts, and professionals.