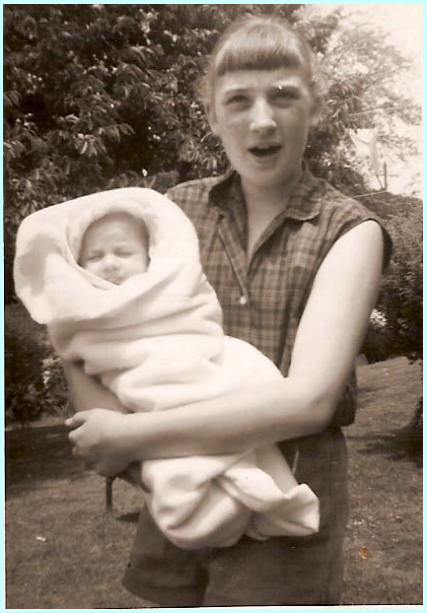
Clues Overlooked –Lives Lost

By Marianne S. Clancy

As I learned about the Surgeon General's Family History Initiative, I was struck by how different my own life might have been had any of the health professionals my family encountered over the years given family health history proper attention. How much loss and grief might have been prevented?



Angela with Baby Marianne: the sister Marianne never got to know.

For as long as I can remember, my mother and several aunts, uncles, and cousins had nosebleeds. Some were simply a nuisance, but others were profuse and lasted several hours. On more than one occasion, because of extensive blood loss, my mother's nosebleeds required transfusions. As a little girl, I vividly remember Saturday shopping trips with mom, which included stops at the ENT to have her nose cauterized. I also have had extensive nosebleeds since childhood and have become adept at disguising them while in business meetings or while giving lectures to university students.

We did not suspect there was more to it, however, until 1964, when my then 17 year old brother, Gary, had a routine chest x-ray, prior to entering nursing school, which revealed a "spot on his lung." He was immediately put in quarantine with suspected tuberculosis. After extensive testing at a major teaching hospital, it was determined that he had Osler-Weber-Rendu Syndrome, which HHT was called back then.

Shortly thereafter, my brother went through a several-hour operation to remove a section of lung containing a blood vessel malformation. As a child of seven, I distinctly recall my parents' hushed conversations and the extended time they spent at the hospital, two-and-a-half hours away, at my brother's side. Gary spent one month in the hospital and returned home 30 pounds lighter with a large scar on his chest. He had to drop out of college for a semester to recover, but came through well. At the time, he was misinformed that he would never have to worry about this again.

Gary's ordeal created much anxiety in our house because just five years earlier, my then 14 year

Gary and Angela

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old sister, Angela, died suddenly in my father's arms. Her death was attributed to "walking pneumonia" since she caught a cold after falling into a pond while ice skating. Since Angela died when I was a year old, I vividly recall my parents living with chronic grief. Understandably, my mother was barely able to function after my sister's death, and the possibility of losing another child was unbearable.

Then, just a few months later, a cousin who was in college suffered a pulmonary hemorrhage. At once, the calls came to our home and he was flown to Philadelphia and met by my brother's surgeon. He was diagnosed with HHT and went through the same surgery and long recuperation as my brother had. At that point, everyone in the family who suffered nosebleeds was sent to get chest x-rays. Seven large pulmonary arteriovenous malformations (AVMs) that could have led to another fatal situation were discovered in another cousin, who then underwent a partial lung removal at the same hospital and with the same surgeon as the other two. *Despite the emerging pattern, no one took an extensive look at family history, and there was no mention of follow-up therapy.*

It was a long time before anyone connected my sister's death possibly to this disorder, or the deaths of my mother's two sisters, age 16 and 18, who came home after school and just suddenly died. Nosebleeds were present in my two siblings and the cousins I described above, but were not severe. It was not until years later that we learned that the presence and severity of nosebleeds does not correlate with involvement of internal organs. Looking at an extensive family tree with 20/20 hindsight, it is clear that only two of my mother's ten siblings were unaffected.

Our family history is full of cases of suspected cerebral vascular hemorrhage, unexplained stroke, clubbed fingers, gastrointestinal bleeding –all clues that HHT was present, but the dots went unconnected for decades.

My mother suffered a stroke in 1969, when I was 12, and could not speak when I got home from school. Her blood count was very low as she suffered from chronic anemia. Doctors in a leading teaching hospital found a few lung AVMs, which they determined were small and did not need treatment. She was told by our local family doctor to take iron, B12 shots, and to "eat a lot of red meat" to keep up her iron stores. Unfortunately, those pulmonary AVMs contributed to my mother's many transient ischemic attacks (i.e. mini-strokes) through the years, and although her red-meat diet contributed to weight gain, she was too fatigued from blood loss to exercise. She died of a heart attack at the age of 64, when I was 21 years old.

Fast forward twenty years: After five years of marriage, my husband and I were thinking of starting a family and were referred to the chief of hematology at Harvard Medical School who, upon hearing my history, tested me for Von Willibrand's Disease. When I tested negative, he promptly told me I did not have it and not to worry.

I went through two successful pregnancies, but still had extensive nosebleeds lasting several hours. I again related our family health history, this time to a family physician in Virginia, and was told by a consulting hematologist that if I had HHT, there was nothing to be done about it, so why fret? That was in 1992. During the delivery of my third child, little did I know that I had untreated AVMs. I had had a little trouble breathing and much lightheadedness throughout the pregnancy. Fortunately, there weren't any complications.

Back in Maryland, my brother's 14 year old son was turning blue on the basketball court. My brother flashed back to my sister's collapse in my father's arms and quickly took action. It wasn't until then that we found a physician who understood HHT: Dr. Robert I. White, Jr. from Yale University. Dr. White had become fascinated with HHT while at Johns Hopkins University in the early 1970s. He created a non-surgical procedure called embolization to treat lung AVMs which has saved the lives of thousands of HHT patients.

My nephew was found to have low oxygen levels due to several large pulmonary AVMs and was treated successfully via embolization. Upon examination of my records, Dr. White recognized two previously missed AVMs in my lungs, and embolized these as well. Our family is now carefully followed every 3-5 years; we have come to understand more fully than ever the importance of follow-up and monitoring for individuals with HHT.

Dr. White continues to see countless people with HHT who have been misdiagnosed, and he is often quoted as saying to his colleagues that ***family history*** is one of the most important aspects of medical care. He's right.

Marianne with daughter Caroline at the Arlington HHT Foundation National Conference 2004

If a thorough family history had been taken at any number of points, there might have been a realization that nosebleeds, pulmonary hemorrhages, deaths in adolescence, gastrointestinal bleeding, strokes, brain abcesses, and migraines in multiple generations were more than just coincidental. That realization, furthermore, might have prevented death and disability in my family, and all the suffering and loss experienced by those impacted and those who loved them.

If only the dots had been connected earlier in my own family's case, I might have been able to experience life with the sister that I never got the chance to know. I might remember a mother who was not shattered by grief.

It is this knowledge of how often HHT is missed –and of how preventable so much of the suffering associated with it truly is—that has compelled me to serve the HHT Foundation over the last ten years, first on the Board of Directors, and now as Executive Director. I continue to feel driven to find a way to reach more families and to advocate for their needs so that no other child has to lose a sibling, and no parent –a child, when that loss could have been prevented.

There is nothing I can do about the past, but it gives me great satisfaction to spend my days working towards the Foundation's goals: hastening the diagnosis and improving the management of HHT so that countless children all over the world, including my own, can live long and productive lives free of needless death, disability, and loss.