Objectives and posttest can be found on page 169.

Hereditary Hemorrhagic Telangiectasia: Children Need Screening Too

Lynne A. Sekarski, Lori A. Spangenberg

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant blood vessel disorder characterized by the presence of arteriovenous malformations (AVMs), epistaxis, and mucocutaneous telangiectases. AVMs are present in lungs, brain, liver, and spine. Children and adults share the same manifestations, with epistaxis and skin telangiectases being the most common. Parents often seek medical attention for their children after an adult in the family is diagnosed. There is debate whether manifestations of HHT are present at birth or develop after puberty, thus making recommendations for evaluation or screening of children in families with HHT uncertain. In the authors' pediatric HHT center, potentially life-threatening manifestations of HHT have been identified in asymptomatic children under 12 years of age. Treatments for HHT include embolization and surgery, laser, and hormone therapy. It is imperative for nurses and other health professionals to recognize this disease and become familiar with evaluation and treatment options.

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu syndrome, is an autosomal dominant disorder that affects blood vessels (HHT Foundation International, 2010b). Many organs and multiple body systems are affected by this blood vessel dysplasia (Mei-Zahav et al., 2006). HHT is characterized by the presence of epistaxis, mucocutaneous telangiectases, and arteriovenous malformations (AVMs) in solid organs. In the United States, about 1 in 5000 individuals are thought to have HHT (HHT Foundation International, 2010b).

The age of clinical presentation of HHT is highly variable among individuals and within families with HHT. Some individuals present with epistaxis prior to their teen years, and others are diagnosed after a life-threatening event, such as a stroke, seizure, severe anemia, or hypoxemic incident. Because young children rarely present with epistaxis or skin findings, nurses and other health care clinicians must be aware of HHT and consider it when a family history is suspicious for symptoms. Some symptoms of HHT are characteristic of other medical diagnoses, including heart and pulmonary disease or a bleeding disorder, making the diagnosis and recognition of the disease difficult. The HHT Foundation International (2010b) notes most patients with HHT are still undiagnosed because of the overlap of symptoms with other conditions. The purpose of this article is to present signs and symptoms of HHT so the practicing clinician can identify the potential diagnosis and have information for treatment and referral.

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Clinical Manifestations

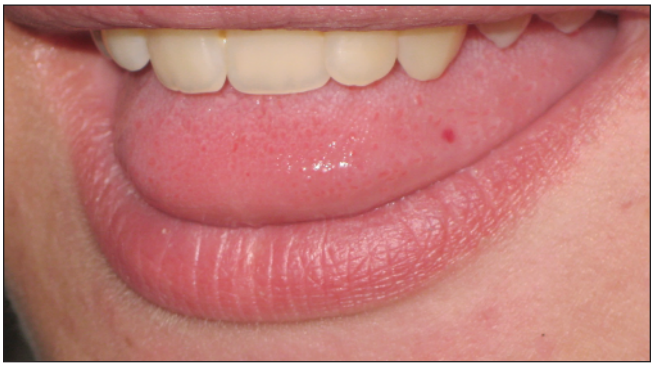
Epistaxis

Epistaxis is the most common symptom of HHT and is present in over 90% of patients. (Faughnan et al., 2009). Although epistaxis is not usually the presenting symptom in children, it will develop in most patients by adolescence, with a median age of onset at 12 years (Faughnan et al., 2009). Other patients do not develop epistaxis until they reach their 40s, at which time almost 90% of HHT patients have nosebleeds.

Epistaxis varies in frequency and severity among individuals and family members. For some patients, it can be an occasional annoyance, and for others, a daily disruption. In the authors' pediatric population, some children do not have nosebleeds, some have two to three a week or month, and a small group has three to five per day. Because pediatric patients with HHT under 10 years of age initially have a lower incidence of nosebleeds, the disease or need for screening may go unrecognized by parents and health care providers (Mei-Zahav et al., 2006).

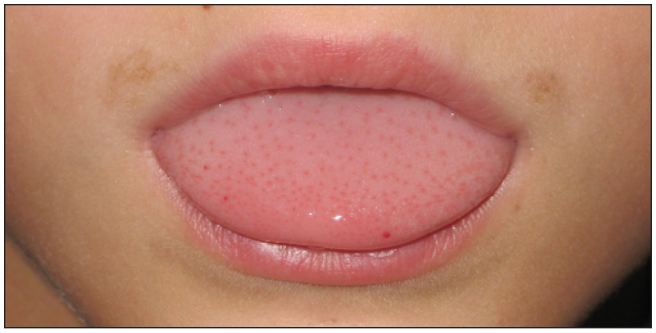
Children without nosebleeds or other symptoms of HHT can have arteriovenous malformations in their lungs or brain that require intervention (Mei-Zahav et al., 2006). It can be difficult for nurses and health care providers to distinguish abnormal nosebleeds from typical nosebleeds because many people without HHT have occasional or bothersome nosebleeds. Epistaxis is the most bothersome symptom for patients with HHT and can be severe enough to lead to anemia, requiring transfusions, iron therapy, or laser surgery (Shovlin, 2009). Investigators are exploring hormone therapy and other medications as possible treatment modalities for recurrent nosebleeds that negatively affect the quality of life of patients with HHT.

Figure 1.
Telangiectasia on the Tongue of a Young Boy



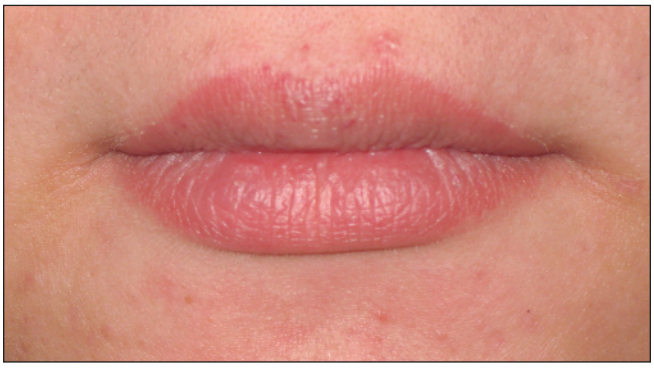
Source: Photo courtesy of Dr. Andrew White.

Figure 2.
Three Telangiectases on the Tongue of a Boy with HHT



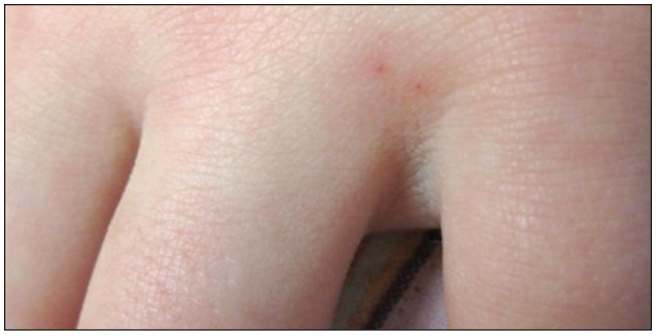
Source: Photo courtesy of Dr. Andrew White.

Figure 3.
Lip Telangiectases on Young Boy with HHT



Source: Photo courtesy of Dr. Andrew White.

Figure 4.
Telangiectasia on the Hand of a Young Patient with HHT



Source: Photo courtesy of Dr. Andrew White.

Telangiectases

Telangiectases are dilated blood vessels found on the face, hands, lips, tongue, and gastrointestinal (GI) tract (Faughnan et al., 2009). These skin lesions are most commonly referred to as "red spots" (see Figures 1-4). Because many people have red spots on their skin and do not have HHT, it is important for the health care professional to be aware that red spots are characteristic manifestations of HHT and require differentiation from benign red spots. Children, however, do not characteristically have telangiectases; many patients with HHT will not develop these mucocutaneous findings until 30 or 40 years of age. About 25% of HHT patients will develop gastrointestinal (GI) bleeding from these dilated vessels in the GI tract. Some patients with GI bleeding and associated anemia may require frequent surveillance with endoscopy and/or blood or iron transfusions (HHT Foundation International, 2010b).

Arteriovenous Malformations

Arteriovenous malformations (AVMs) that characterize HHT are found in the lungs, brain, spinal cord, and liver. AVMs are abnormal connections between arteries and veins. AVMs in HHT result in arteries that are directly connected to veins, causing these fragile vessels to potentially rupture and bleed (HHT Foundation International, 2010b). These abnormal blood vessels must be carefully monitored and treated.

One-third of patients will have a pulmonary arteriovenous malformation (PAVM) in their lifetime (Shovlin, 2009). Others will have two to three PAVMs, and some will have multiple PAVMs that require continued monitoring and treatment. Patients with a PAVM may present with no objective symptoms, or they may have shortness of breath with activity, difficulty lying flat to sleep, clubbing of their fingers, brain abscess, or hypoxemia with an oxygen saturation less than 95%.

PAVMs with a feeding vessel 3 mm or larger are amenable to embolization (HHT Foundation International, 2010b). Embolization of the vessel is a procedure in which a coil, plug, or glue-like substance is placed in the abnormal vessel. By stopping the blood flow to the abnormal vessel, more oxygenated blood is delivered throughout the circulatory system, increasing oxygenation and decreasing symptoms of hypoxemia. Embolization of AVMs should be performed by experienced interventional radiologists or cardiologists and in conjunction with pulmonary angiography. Pulmonary angiography is considered to be the gold standard test for PAVM determination (Tabori & Love, 2008).

Cerebral Arteriovenous Malformations

Cerebral arteriovenous malformations (CAVMs) are present in 20% of patients with HHT, and the rare spinal AVMs are found in 1% to 2% of patients (HHT Foundation International, 2010b). Infants as young as

3 weeks have been diagnosed with a CAVM in the authors' center. Patients with CAVMs may be asymptomatic or may present with stroke, seizures, neurological changes, or brain abscess. Treatment should be considered if an AVM is equal to or greater than one centimeter in size because removing the AVM decreases the risk for future brain hemorrhage. Effective treatment may include surgery, embolization, stereotactic radiation (gamma knife), or a combination of these treatments (HHT Foundation International, 2010b). Decisions regarding treatment of patients with a CAVM should be made by a multidisciplinary team on an individual basis. Variables that should be considered when deciding on treatment include the location of the AVM, depth of the AVM, the ability to access it, and all risks involved in surgery and long-term affects (HHT Foundation International, 2010b).

Diagnostic Genetic Testing

The diagnosis of HHT is made by satisfying the Curacao criteria or by having a positive genetic test (Faughnan et al., 2009). A guide to making the diagnosis based on clinical criteria (the Curacao criteria) was established in 1999 by the Scientific Advisory Board of the HHT Foundation International, Inc., and remains unchanged (Shovlin et al., 2000). These criteria include:

- A first-degree family member (parent, sibling, or child) with HHT.
- Epistaxis.
- Telangiectases.
- Arteriovenous malformation in a solid organ.

Patients are diagnosed with definite HHT if they have met three out of the four criteria (Shovlin et al., 2000). If an individual has at least two criteria, they have possible HHT. In the United States, health care professionals believe over 90% of individuals with HHT have yet to be diagnosed (HHT Foundation International, 2010b).

In 2003, commercial genetic testing for HHT became available in the form of a blood test (HHT Foundation International, 2010b). The two main genes that have been identified as contributing to HHT are the endoglin (ENG) mutation on chromosome 9q33-34 and the activin-receptor-like kinase 1 (ACVRL1 or ALK1) on chromosome 12q (HHT Foundation International, 2010b). These disease subtypes are known as HHT1 and HHT2, respectively. Another type of HHT has been found in an overlapping group of patients with juvenile polyposis syndrome (JPS) and is identified as SMAD4 (Haidle & Howe, 2008).

Juvenile polyposis is a condition in which individuals are predisposed to polyps forming in their gastrointestinal tract (Haidle & Howe, 2008). The number of polyps varies from a few to over a hundred in one's lifetime. The polyps specifically found in the stomach, small intestine, colon, and rectum are termed juvenile because of their type and not in relation to when they present. Most juvenile polyps are benign but can cause bleeding and anemia. The combination of HHT (ENG) and SMAD4 occurs in about 20% of individuals with SMAD4 mutation (Haidle & Howe, 2008).

In addition to these types, several hundred HHT-causing mutations have been described (Gedge et al., 2007). Genetic testing for HHT will provide an answer only 75% of the time; therefore, many families are reluctant to have the genetic test performed due to cost, odds of not finding the mutation, or future potential problems with insurance coverage (Gedge et al., 2007). Controversy regarding future insurance coverage for a child sometimes clouds the decision to carry out this test. The most affected and senior member of the family living with HHT (the proband) should have the genetic testing first.

After the family's particular genetic mutation is initially identified, other members can be tested (Shovlin, 2009). The average cost for the first genetic test in a family is about $1500 (Cohen et al., 2005). Each additional affected family member is usually charged $250. Some insurance plans will cover the cost of the genetic test, but families are often responsible for the cost. Genetic testing can be useful if a genetic mutation is identified. Knowing the family mutation and having a member with a positive genetic test allows for making the diagnosis in other members of the same family because the positive test satisfies one of the Curacao criteria. This can be especially helpful in diagnosing children because they do not often meet clinical diagnostic criteria (Mei-Zahav et al., 2006). About 30% of patients who meet the clinical criteria for a diagnosis of HHT will have a negative or indeterminate genetic test (Faughnan et al., 2009). If a child has not had a genetic test or if the genetic test failed to identify a gene, it is necessary for the child to have periodic screening and evaluation for the disease every 3 to 5 years.

Children present for screening or evaluation of HHT after an adult family member or sibling has been diagnosed or is suspected of having HHT. In the past, it was thought screening of children in families was not necessary until adolescence because of the age at which the abnormal blood vessels usually develop and the disease presents itself. Although most people with HHT are diagnosed in adulthood by 40 years of age, some pediatric patients have life-threatening manifestations that need to be diagnosed and treated well before reaching an adult age (Faughnan et al., 2009). In this disease, manifestations or symptoms vary within family members. For some, HHT is a nuisance, while for many others, it is life-threatening and requires vigilant medical care and intervention. Children are diagnosed with HHT using the same criteria as adults. Increased awareness and recognition of HHT symptoms in adults will hopefully open up avenues for increased screening and diagnosis in children.

Evaluation and Screening Process

Patients from birth to 21 years of age are referred to the authors' HHT center by their primary care providers, medical specialists, and the HHT Foundation Inc., or self-referral. The nurses and physicians review information obtained during the initial contact to obtain necessary records and facilitate an evaluation appointment for one or more family members. This evaluation includes a comprehensive history and physical and diagnostic testing. Every child with suspected HHT should be screened by physicians familiar with this disease. Physicians with expertise in HHT can be found in one of the 33 HHT Centers of Excellence throughout the world (HHT Foundation International, 2010b).

As nurses at the center, the authors are one of the first contacts for the families. It is imperative that necessary clinical and demographic information is obtained to plan the evaluation. In collaboration with the expert HHT pediatrician, a plan of care and evaluation are formulated, and a visit is planned. Most patients travel long distances to get to the center. There are only 12 HHT Centers of Excellence within the United States, and some do not provide pediatric

care. The HHT Foundation International provides a comprehensive list of HHT Centers of Excellence.

An initial screening appointment consists of a thorough history, a physical examination, pulse oximetry, a contrast (or bubble) echocardiogram (CE), and brain magnetic resonance imaging (MRI) with and without contrast. The family history is crucial in determining the symptoms, as well as the extent of HHT within the family. The authors' team meets with the family and patient simultaneously during the interview and asks questions related to specific problems or symptoms of HHT. These appointments are arranged so the clinic visit and testing are completed in one to two days. Infants and young children are unable to tolerate the testing without sedation because any movement interferes with some tests. Sedation is usually well tolerated in children when managed by experienced nurses and physicians in the pediatric care setting. Health care professionals in the HHT center must weigh the risk of sedation versus the knowledge gained from having the tests. At the authors' center, a child life specialist is available to assist pediatric patients in preparing and completing the necessary radiology tests. These screening tests can be physically demanding and can provoke anxiety regardless of the age or developmental level of the patient.

Contrast echocardiography (CE) is the first-line test used to evaluate the presence of intrapulmonary shunting seen with AVMs and has been determined to be a very sensitive screening tool (Gossage, 2003). Patients are injected with agitated saline during transthoracic echocardiography. The test is indicative of a pulmonary AVM if bubbles are seen in the left heart after three to five heart beats (Gossage, 2003). Bubbles identified immediately are usually diagnostic of patent foramen ovale (PFO). It is important that the cardiologist be familiar with distinguishing these findings in the pediatric patient. If the CE is positive, the patient will be scheduled for a chest computed tomography scan with angiography (CTA) because a positive CE alone cannot diagnose a PAVM or describe the size and location of the shunting vessel (Oxhoj, Kjeldson, & Neilsen, 2000).

Brain MRI with and without contrast (MRI/MRA) is done to evaluate for the presence of cranial arteriovenous malformations (CAVMs). A baseline MRI with and without contrast is completed whether or not the patient has evidence of abnormal neurological symptoms. CAVMs occur in 5% to 20% of all HHT patients and have been diagnosed in patients despite a normal neurological examination (HHT Foundation International, 2010b). Interpretation of brain MRIs and CTAs should be done by neuroradiologists with HHT experience to ensure a correct diagnosis.

At the end of the evaluation and screening appointment, the patient and family will know whether they have any symptoms of HHT and if they can be diagnosed with possible or definite HHT. The physician and nurse team at the authors' center provide education about living with HHT. In this pediatric population, education is focused on nasal hygiene and supportive care for nosebleeds, sub-acute bacterial endocarditis (SBE), changes in activity and exercise restrictions, avoidance of aspirin and non-steroidal anti-inflammatory drugs (NSAIDS), and the need for a filter with intravenous infusions (Peiffer, 2009). Patients who do not know whether they have a PAVM or have a known or treated PAVM should follow the American Heart Association (AHA) guidelines for prophylactic antibiotics (HHT Foundation International, 2010a).

NSAIDS should be avoided in patients with suspected HHT because of anticoagulant properties that could contribute to prolonged bleeding (HHT Foundation International, 2010b). Filters are recommended for use with any intravenous infusion to decrease the chance of air embolism (Peiffer, 2009). Families are encouraged to continue regular medical care with their primary care physician and maintain communication with the HHT center as needed. Families or health care providers should call their local HHT center with symptoms of decreased exercise tolerance, decreased oxygen saturation, increased frequency and severity of nosebleeds, hemoptysis, hematochezia, and change in neurological status or behavior in their child.

It is believed that AVMs grow slowly, and patients can wait three to five years between evaluations; however, some children experience an increase or change in symptoms sooner and should be evaluated. Nurses are crucial in communicating changes in a child's symptoms to the HHT physician. Parents call with questions regarding many aspects of their child's health and whether or not their concern is related to HHT. Triaging these phone calls requires expertise of the disease and its presentation in children; even then, it can be difficult to decide if the symptoms are related to HHT. The following case studies are being shared to demonstrate the variability of symptoms that exists in children with HHT.

Pediatric Case Examples Of HHT

Tim

Tim was born to a family in which his father has HHT manifested by telangiectases, epistaxis, and PAVMs. Tim's father was diagnosed as a young adult based on clinical findings. A few years prior to Tim's visit to the authors' pediatric HHT center, Tim's father had genetic testing that revealed the endoglin mutation or HHT1. He has two older sisters, each with positive family history and epistaxis. Using the Curacao criteria, one sister has definite HHT (she has skin telangiectases, positive family history, and epistaxis). His other sister has possible HHT, having only two criteria, a positive family history and epistaxis. At four weeks of age, Tim presented to a local emergency unit with episodes of stiffening after feeding, suspicious behavior for seizure activity. He was anemic and posturing, and was quickly admitted to the pediatric critical care unit. His evaluation revealed a tangle of blood vessels measuring 2x3 cm in the left frontal region of his brain that was promptly resected. Given his CAVM and positive family history, he meets criteria for possible HHT. At discharge, he had mild right hemi-paresis.

At 3 years of age, he returned to the center and had progressed in his neurodevelopmental milestones; he was talking, walking, and feeding himself. He had right-sided weakness and seizures that were treated with daily antiepileptic medicine. He had nosebleeds two times per week and no skin telangiectases. At this visit, Tim satisfies the criteria for definite HHT because he has nosebleeds, a CAVM, and positive family history. His CE at this visit revealed intrapulmonary shunting, and his brain MRI did not reveal any new AVMs.

Given his positive CE, a computed tomography of the chest with angiography (CTA) will be completed at his visit in one year to determine if he has a PAVM. Dependent on these findings, he will continue to require interval brain MRI, chest CTAs, a review of

his interval history, and physical examinations to guide future therapy.

Andrew

Andrew presented to the authors' HHT center as a 21-month-old whose umbilical cord blood was tested at birth and revealed the ENG mutation. At his first visit, he accompanied his sisters to an appointment for an interval evaluation for them. Andrew's father has HHT manifested with epistaxis, skin telangiectases, and PAVMs. His 17-year-old brother has HHT and has epistaxis, skin telangiectases, and intrapulmonary shunting. Andrew did not have nosebleeds or skin telangiectases.

At this visit, recommendations were made to have CE in one to two years and to use prophylaxis for dental work. He returned to the clinic at 5 years of age for follow up and had developed a few telangiectases on his face and lower lip, but no nosebleeds or other symptoms. When Andrew visited the center at 9 years of age, CE showed intrapulmonary shunting, and his brain MRI revealed a 2 cm right parietal AVM and three tiny AVMs in the left parietal region. He reported nosebleeds once per month, another telangiectasia on his right arm, and an oxygen saturation of 96%. He did not have shortness of breath with exercise or any other complaints. He had a craniotomy with successful removal of his large CAVM.

Andrew returned in the fall for CTA to further define his intrapulmonary shunting, and this revealed multiple PAVMs, of which three were embolized. He will have repeat CTA and brain MRI in two to three years to guide further treatment. Andrew is attending school and participating in all his activities.

Ann

At 6 years of age, Ann was noted to have oxygen saturations of 89% to 94% during anesthetic induction for a tonsillectomy at an outside hospital. As a follow up to the lower-than-normal oxygen saturations, Ann was evaluated by medical specialists at a larger hospital system in her home state and given a definitive diagnosis of HHT. Ann met all four Curacao criteria used to make a diagnosis of HHT because her evaluation revealed positive family history of HHT on the father's side, multiple skin telangiectases in the characteristic locations, occasional epistaxis, and a 3x3x2 cm PAVM in the right lower lobe discovered after undergoing a cardiac catheterization procedure.

Successful closure of the PAVM through embolization helped increase Ann's oxygen saturations from a baseline of 80% to about 93% at rest without oxygen supplementation. Ann enjoyed more childhood activities after the procedure due to less exercise intolerance and fatigue. She experienced minimal problems related to HHT over the next two years. At 9 years of age, Ann had a follow-up visit with testing that revealed multiple small PAVMs too small to embolize due to the tiny vessels feeding these PAVMs.

Ann presented to the authors' pediatric HHT center at 11 years of age with complaints of intermittent chest pain, nosebleeds, occasional hemoptysis, exercise intolerance, and hypoxemia with oxygen saturations of 80% on room air. Ann had a pulmonary angiogram that identified innumerable AVMs in both lungs. There were five PAVMs with feeding arteries large enough to accept coils for successful embolization. Ann returned to the center at 12 years of age and had a CTA that revealed more diffuse bilateral PAVMs with no pattern to their distribution. At this point in Ann's life, she could not keep up with her friends while at play. Collaboration among several well-known HHT specialists offered difficult and unattractive treatment options that included obliterating entire segments of the lungs more heavily involved with AVMs or continuation of monitoring her health with support measures as needed. The family chose continued monitoring of her health with supportive measures.

Ann returned to the center at 13 years of age with more frequent complaints of chest pain and increasing need for supplemental oxygen. Her shortness of breath led her to sleep in an upright position. The family noticed occasional periorbital, perioral, and peripheral cyanosis. During this visit, her CTA revealed a new finding of right ventricular hypertrophy with increasing numbers of subcentimeter AVMs scattered bilaterally throughout the lungs that were again too small to embolize.

At this visit, new findings of two CAVMs were noted on a head MRI/MRA. Her neurological examination was normal. Neuroradiologists offered options for possible treatment of the CAVMs, such as brain surgery, gamma knife, or possible embolization. The patient and family declined these treatment options and returned home. They returned 6 months later for re-evaluation with her oxygen saturations between 70% to 80% at rest on room air. She admitted to using her oxygen much more frequently at this point. Repeat pulmonary angiograms showed innumerable sub-centimeter AVMs in both lungs with more recruitment of collateral vessels from the chest wall, right hemi diaphragm, and celiac artery.

Because the preponderance of the AVMs is in a single segment, segmental obliteration by embolization is not an option because it would not improve the patient's oxygen saturations or poor prognosis. One final consultation was made with the lung transplant team. The pulmonologists and cardiothoracic surgeons reviewed her care and thought that transplant would be risky because of potential for hemorrhage, given the presence of cranial AVMs. The patient and family decided to return home with hospice care and comfort measures. Ann died at age 16 after a stroke.

Ann's 17-year-old brother, Nick, also has HHT. His case is not as complicated as his sister's was in her teen years. He has skin telangiectasias, nosebleeds, and several small PAVMs that are too small to treat with embolization. He continues to return to the center every 2 to 3 years for evaluation.

Charlie

Charlie, almost 5 years of age, was referred to the authors' pediatric HHT center for routine screening of possible HHT due to positive family history. The paternal grandfather is known to have definite HHT, and Charlie's mother has possible HHT. Charlie has epistaxis and cutaneous telangiectases on his face and hands. He denied chest discomfort, shortness of breath, headaches, and exercise intolerance, but oxygen saturations in the examination room registered only 91% to 92%. Charlie's CE was positive for intrapulmonary shunting, and his brain imaging study was normal. Further testing included a CTA and subsequent pulmonary angiography. The CTA revealed multiple PAVMs, with the largest in the right lower lobe, and multiple small peripheral PAVMs scattered throughout bilaterally.

One month after this CTA, the patient had pulmonary angiography with embolization of a complex right lower lobe AVM that was found to have two feeding arteries and two draining veins ranging in size from 3 to 6 mm. This unusual AVM pattern was noted to be like a tree with branches.

Charlie was discharged home on the same day as the outpatient procedure. He is to have repeat CTA in 1 to 2 years. Charlie attends school and participates in physical activity with much more energy. He could have suffered complicated, life-altering health problems due to the PAVM if it had not been treated.

Discussion

The above cases illustrate the variability of presentation and subsequent morbidity of this disease. The youngest child seen at the authors' center with a CAVM is 4-week-old Tim. He is the only patient with symptoms associated with his CAVM, and it was reported as suspected seizure activity. Andrew and Ann did not have any symptoms of a CAVM, yet CAVMs were identified through brain imaging. Andrew's case is especially representative of the center's pediatric population in that he had only "red spots" and no other clinical symptoms.

Knowing Andrew had the gene for HHT, it was difficult for the health care team to decide whether or not to recommend sedating him for the screening tests needed to identify AVMs. The family wished to postpone the screening until he was 8 years of age, and the center honored their request. All were surprised that pulmonary and cranial AVMs were found in this asymptomatic patient.

Charlie came to an evaluation at the center without any noticeable symptoms. The first indication that he may have HHT was the oxygen saturation of 91% in an otherwise presumably healthy school-aged child. It is important to note that he came to the center because of a family history of HHT, which is the most frequent reason children are referred for an evaluation. Ann had low oxygen saturation as her presenting symptom. Her case was one of the most complicated because of her diffuse PAVMs and subsequent CAVMs. Each of these children presented to the authors' center with varying symptomotology, which is why it is so important to educate nurses and health care professionals to be alert to the signs and symptoms of HHT.

Recommendation

Children have a 50% chance of having HHT if one of their parents has the diagnosis (HHT Foundation International, 2010b). Screening children of these affected adults is necessary to detect life-threatening medical problems. If children are not screened because they do not have nosebleeds or skin telangiectases, life-threatening manifestations can be missed, as described in these cases. PAVMs and CAVMs requiring treatment have been detected in patients with and without recognizable symptoms. Regular HHT evaluations by experienced HHT specialists can help detect treatable problems that may cause significant morbidity and/or mortality if left unidentified. It is important for nurses and other health care professionals to consider HHT as a possibility when they encounter a child with epistaxis, cutaneous telangiectases or "red spots," complaints of symptoms suspicious for solid organ AVMs, or family history suspicious for HHT.

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