Key Role for Otolaryngology in a Hereditary Hemorrhagic Telangiectasia Center of Excellence

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ABSTRACT

OBJECTIVES: Review the initial experiences of an HHT Center of Excellence and uderstand which HHT patients require otolaryngologic evaluation and interventions

METHODS: Retrospective review of all patients referred to our HHT Center of Excellence from its inception in May 2010 to June 2012. Clinical presentation, radiographical imaging, genetic testing, otolaryngologic treatments, and other operative interventions were analyzed.

RESULTS: In our cohort of 144 patients, 98 have HHT based on Curacao clinical criteria, and 16 have possible HHT or are undergoing further workup. Of the 98 with HHT, 20 have sought otolaryngologic evaluation at UCLA for relentless epistaxis or oral bleeding. Eleven (55%) have required OR intervention, primarily KTP laser ablation of symptomatic nasal and oral telangiectasias. Additionally, 47 patients have required embolization, neurosurgical clipping, or radiation for enlarging or symptomatic arteriovenous malformations in the lung, brain, and abdomen.

CONCLUSIONS: An HHT Center of Excellence is important in providing comprehensive care for patients with this rare disease with significant clinical sequelae. Otolaryngologists are critical members of this multidisciplinary team, performing interventions in the clinic and operating room to improve quality of life for these patients.

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Background

Hereditary hemorrhagic telangiectasia (HHT), also referred to as "Osler-Weber-Rendu Syndrome," is a multisystem disorder that affects approximately 1 in 5000 people.[1] HHT is autosomal dominant, and mutations within the signaling pathway in vascular endothelial cells lead to vascular dysplasia. The disease is genetically heterogenous, with multiple subtypes, and hundreds of identified mutations which lead to abnormal vessel formation.[2]

Telangiectasias of the skin, GI tract, nasal mucosa, lips, and oral cavity frequently cause recurrent bleeding and hemorrhage. Arteriovenous malformations (AVMs) commonly occur in the lungs, brain, and liver leading to complications of thrombosis, embolus, and the shunting of blood.[1]

These vascular malformations continue to grow with time and require lifelong surveillance.[1] Because of the multisystemic nature of this disease, a multidisciplinary team approach consisting of an otolaryngologist, interventional radiologist, pulmonologist, neurosurgeon, cardiologist, gastroenterologist, hepatologist, and hematologist is critical. In the United States, this need for multidisciplinary care and lifelong surveillance led to the development of HHT Centers of Excellence. UCLA was established as an HHT Center of Excellence in 2010 and is one of 12 centers in the country. Our objectives were to review the initial experience of an HHT Center of Excellence and analyze which patients require otolaryngology evaluation and treatment.

METHODS

We performed a retrospective review of all patients from the Southwestern United States referred to our HHT Center of Excellence from its inception in May 2010 through June 2012. Clinical presentation, radiographical imaging, genetic testing, otolaryngologic treatments, and other operative interventions were analyzed. Patients were diagnosed as having definite, possible, or unlikely HHT based on the Curacao clinical criteria **(Table 1)**.[3]

RESULTS

Based on the Curacao clinical criteria of having at least 3 criteria **(Table 1)**, 98 of the 144 patients referred to and evaluated at our HHT Center of Excellence have HHT. Sixteen patients met 2 of the 4 criteria, or are undergoing further work-up, and possibly have the disorder. 30 patients met less than one of the four criteria and do not have HHT.

Table 1: Patients with HHT based on Curacao Criteria

HHT Diagnosis	# of Patients
Definitie (3-4 criteria met)	98
Possible (2 criteria met)	16
Unlikely (<2 criteria met)	30

Criteria: 1. Epistaxis; 2. Telangiectasias; 3. Visceral telangectasias or arteriovenous malformations; 4. Family history of HHT in 1st degree relative

Of the 98 patients with confirmed HHT, 20 (20%) have been referred to otolaryngology for further evaluation for recurrent epistaxis or persistent oral bleeding. Of these 20 patients, 11 (55%) have need intervention in the operating room. Ten of the eleven patients have had KTP laser treatments for symptomatic oral, oropharyngeal, and nasal mucosal telangiectasias. Two of these nine patients have had multiple treatments over the last two years. The eleventh patient did not have KTP treatments, but had excision of an enlarged telangiectasia from the hard palate for repeated acute hemorrhages requiring transfusion; she has had significant improvement since surgery.

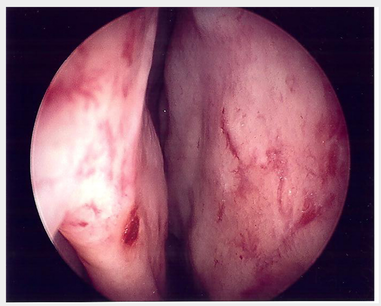

Figure 1: Nasal Telangiectasias

RESULTS

Additionally, 47 patients have been treated for symptomatic AVMs in other locations or are being followed closely with radiologic imaging to rule out enlargement of previously treated AVMs. Several of these patients have had interventions for multiple AVMs at different locations in the body for a total of 53 interventions (**Table 2**). These interventions have included embolization, neurosurgical clipping, or radiation for AVMs in the lung, brain, abdomen, and pelvis.

Table 2: Non-otolaryngology interventions

Intervention	Number of Patients
IR embolization of	
Pulmonary AVMs	**36**
Cerebral AVMs	**4**
Liver AVMs	**1**
Nasal AVMs	**1**
Pelvic AVM	**1**
Resection of pulm AVM	**2**
Neurosurgical clipping of cerebral aneurysm	**2**
Brain radiation	**3**
Partial colectomy	**1**
Bronchoscopy and laser coagulation	**1**
Upper endoscopy and laser coagulation	**1**

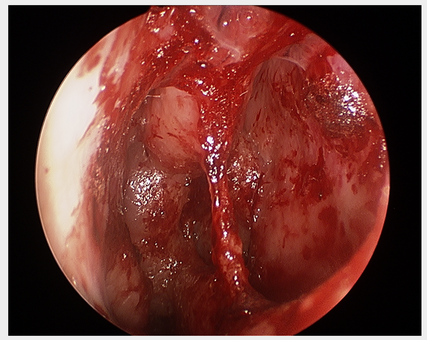

Figure 2: Nasal telangiectasias with large septal perforation.

DISCUSSION

HHT Centers of Excellence allow early diagnosis, screening and treatment. Severe complications of HHT, such as stroke, lung and brain hemorrhages can be prevented. In addition, early intervention and close monitoring can improve HHT patients' quality of life and minimize the morbidity associated with severe epistaxis and GI bleeds.

Otolaryngology plays a key role in managing these patients, especially considering that most common symptom of HHT is epistaxis which occurs in >90% of patients[1]. In just 16 months, 20% of the patients with HHT at our center have been referred to otolaryngology, including 11 patients who have needed interventions in the OR. In addition to these 20 patients, 10 additional patients were seen by outside otolaryngologists before the opening of UCLA HHT Center.

CONCLUSIONS

An HHT Center of Excellence is important in providing comprehensive care for patients with this rare disease with significant clinical sequelae. Otolaryngologists are critical members of this multidisciplinary team performing interventions in the clinic and operating room to improve quality of life for these patients.

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