Curaçao's diagnostic criteria for Hereditary Hemorrhagic Telangiectasia (HHT, Rendu-Osler-Weber syndrome)

(Shovlin C.L. et al., . Am. J. Med. Genet. 91:66-67, 2000)

1. Epistaxis: spontaneous, recurrent nose bleeds

2. Telangiectases: multiple, at characteristic sites (lips, oral cavity, fingers, nose)

3. Visceral lesions such as gastrointestinal telangiectasia (with or without bleeding), pulmonary arteriovenous malformation (AVM), hepatic AVM, cerebral AVMs, spinal AVM

4. Family history: a first degree relative with HHT according to these criteria

Diagnosis of HHT
Definite: 3 criteria are present
Possible or suspected: 2 criteria are present
Unlikely: <2 criteria are present

Note:
All offspring of an individual with HHT are at risk of having the disease since HHT may not manifest until late in life. If there is any concern regarding the presence of physical signs, an experienced physician should be consulted. Coagulation disorders should be excluded. The presence of visceral abnormalities in children should prompt a particularly careful check of other family members. These criteria are likely to be further refined as molecular diagnostic tests become available in the next few years.