

The Curaçao Criteria

Consensus Diagnostic Criteria for HHT

by Claire Shovlin, Ph.D., MA, MRCP, Senior Lecturer/Hon Consultant in Respiratory Medicine
Imperial College School of Medicine (NHLI), Hammersmith Hospital, England

At the last HHT Scientific Advisory Board Meeting in Curaçao, one of the issues discussed was the lack of consensus criteria to diagnose individuals with HHT. The diagnostic criteria proposed ten years ago have been subtly modified in publications from each group of HHT clinicians and researchers.

As a result, we were encouraged to develop criteria which would be applicable to all groups around the world. These criteria will shortly be published in the American Journal of Medical Genetics, and a summary is provided here. Making a clinical diagnosis of HHT in individuals displaying the classical triad of nosebleeds, telangiectasia, and a suitable family history is relatively simple, but this accounts for only a proportion of cases. We were concerned that many physicians were overlooking the significance of subtle disease manifestations, particularly involvement of internal organs. We were also concerned that in HHT families, nosebleeds alone were sometimes used to establish the diagnosis yet nosebleeds are common in the general population too.

THE CURAÇAO CRITERIA

1. NOSEBLEEDS, spontaneous and recurrent.
2. TELANGIECTASES, multiple, at characteristic sites including lips, oral cavity, fingers and nose.
3. INTERNAL LESIONS such as
 - * Gastrointestinal telangiectasia (with or w/o bleeding)
 - * Pulmonary AVM
 - * Hepatic AVM
 - * Cerebral AVMs
 - * Spinal AVM
4. FAMILY HISTORY- a first degree relative with HHT according to these criteria.

The HHT Diagnosis is Definite if 3 criteria are present, Possible or suspected if 2 criteria are present, and Unlikely if fewer than 2 criteria are present.

Three features of these new diagnostic criteria should be noted. First, they spell out in detail exactly what is required for each manifestation of HHT. For example, as there is no agreement amongst doctors as to the precise number of episodes or severity of nosebleeds needed to satisfy a diagnosis of HHT, the criteria merely highlight that nosebleeds should occur spontaneously on more than one occasion, with night-time bleeds being particularly suspicious. Secondly, and perhaps more importantly, these criteria allow us to clearly distinguish between individuals who definitely have HHT, and those in whom it is suspected, and allow each label to be added to medical records to ensure appropriate follow-up and medical management.

Finally, we emphasize that all offspring of an individual with HHT are at risk of having the disease since HHT may not manifest until late in life. We recommend that if there is any concern regarding the presence of physical signs, an experienced physician should be consulted. These criteria are likely to be further refined as molecular diagnostic tests become available in the next few years.

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REFERENCES

Plauchu H, de Chadarévian J-P, Bideau A and Robert J-M. 1989. Age-related profile of hereditary hemorrhagic telangiectasia in an epidemiologically recruited population. American Journal of Medical Genetics, 32:291-7.
Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJJ, Kjeldsen AD, and Plauchu H. 1999. Diagnostic Criteria For Hereditary Hemorrhagic Telangiectasia (Rendu-Osler-Weber Syndrome). American Journal of Medical Genetics In Press.