HHT,

Osler-Weber-Rendu Disease.

ACVRL1-Related …

ENG-Related ….

Hereditary Hemorrhagic Telangiectasia

Autosomal Dominant Inheritance.

*Thorax 1999;**54**:714–729. Claire L Shovlin, Michelle Letarte*

www.geneclinics.org

Nosebleeds: 95%

Spontaneous and recurrent

Mucocutaneous telangiectases: 95%

Lips, oral cavity, fingers, and nose

Visceral AVMs:

Pulmonary	33%
Cerebral / Spinal	11%
Gastrointestinal / Hepatic	25%

Family history:

First degree relative with HHT according to these criteria.

Clinical Diagnosis.

Definite three or more findings are present

Possible or suspected
two findings are present

Unlikely
fewer than two findings are present

Nosebleeds:
Mucocutaneous telangiectases:
Visceral AVMs:
Family history:

GeneClinics HHT Recommendations 2004

Clinical: **Annual Review**

Cerebral: **MRI / MRA as early as possible.**
No repeat if neg (?)

Pulmonary: **Contrast Echo (Ie.Pulmon Shunt Study !!) & CXR**
100% Sensitivity
Chest CT if either +ve.
Pulmonary Angio if CT +ve
Review acc. to age, sympt, FH or every 2-3 yrs
? Role for shunt study and DLCO

GI: **FBE, Faecal occult blood +/- endocsocpy**
Hepatic US – elderly, Female, symptomatic.

Avoid – Aspirin, NSAIDs, antiplatelet drugs and anticoagulants.