NHS
Hereditary Haemorrhagic telangiectasia
Rare Disease
Collaborative Network

Imperial College
London

2022 Information Sheet for Hereditary Haemorrhagic Telangiectasia (HHT)

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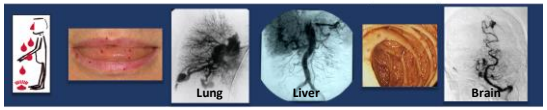
WHAT IS HHT?

- HHT is caused by a single change in one of 4 genes, and usually runs in families.
- HHT affects more than 10,000 people in the UK- though most are not diagnosed.
- HHT usually (but not always) causes nosebleeds that if regular, lead to anaemia.
- People with HHT may have obvious blood spots- but usually they are difficult to see.
- HHT usually causes one or more abnormal blood vessels in the lungs and/or liver.
- Less commonly, HHT causes abnormal blood vessels in other parts of the body.



HOW DO YOU KNOW IF SOMEONE HAS HHT- AND WHAT DO YOU DO?

Anderson, Sharma , Alsafi , Shovlin. Thorax. 2022
***Pulmonary arteriovenous malformations may be the only clinical criterion present in genetically confirmed hereditary haemorrhagic telangiectasia*.**

This important paper emphasises it can be very difficult to tell who has HHT. Please bring this to the attention of anyone in the family who thinks they cannot have HHT because they do not have nosebleeds, or blood spots. HHT can only be ruled out if the HHT gene change is known for the family AND a family member is shown not to have that gene change. Most people with HHT are well, working, and exercising. But screening and "Wise HHT Public Health" is important to limit complications in later life:

- **Anaemia** is common if nosebleeds occur several times a week, as a normal diet does not replace the lost iron. We arrange expert ENT care for regular nosebleeds. We also guide people to what their iron requirements are likely to be, confirm with blood tests, and try to find a low-dose tablet that they can take without side effects from the gut, or bleeds.
- **Pulmonary AVMs** affect ~1 in 2 with HHT. ~1 in 2 of these have preventable complications. PAVM screening once in adult life, and pre-symptomatic treatments are needed for all.
- **Hepatic AVMs** affect ~1 in 2 with HHT. ~ 1 in 20 of these have complications, but these do not occur "out of the blue". We investigate people if we suspect liver AVMs are present, and otherwise let doctors know AVMs are likely to be found if liver scans are performed.
- **Cerebral AVMs** affect ~1 in 20, and ~ 1 in 20 of these have complications which can be 'out of the blue'. On the other hand, treatment risks are "not negligible". Following European practice, we make sure everyone has a discussion, understands the evidence, and makes the right decision for them about a scan if they are well. Some choose scans, others do not.
- **Other AVMs** (eg pancreatic) or internal vascular abnormalities are found in ~1 in 5 people if scans are performed for other reasons, but these rarely if ever require treatment. Early discussion with experts is recommended to prevent unnecessary anxiety or interventions.
- **With appropriate medical care, life expectancy is normal for the HHT population,** likely reflecting protection from certain cancers and heart attacks. You can find more detail and references at https://www.imperial.ac.uk/people/c.shovlin/page/hht-and-pavms.html

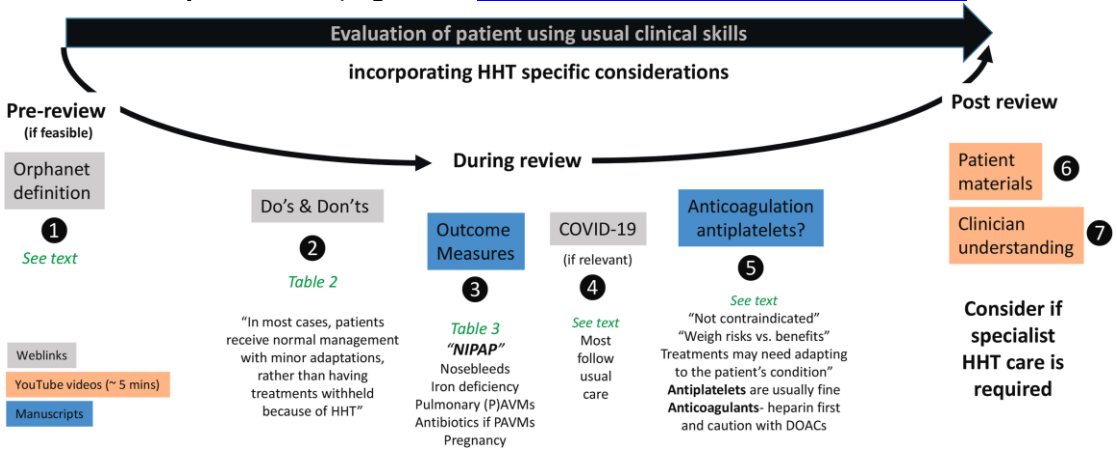
HOW CAN MY LOCAL DOCTORS KNOW WHAT TO DO IF I SEE THEM?

Please ask them to look at the 2022 European Reference Network's Frameworks for HHT:

Shovlin, Buscarini, Sabbà, Mager, Kjeldsen, Pagella, Sure, Ugolini, Torring, Suppressa, Rennie, Post, Patel, Nielsen, Manfredi, Lenato, Lefroy, Kariholu, Jones, Fialla, Eker, Dupuis, Droege, Coote, Boccardi, Alsafi, Alicante, Dupuis-Girod. Eur J Med Genet. 2022
***The European Rare Disease Network for HHT Frameworks for management of hereditary haemorrhagic telangiectasia in general and speciality care*.**

These Frameworks were developed using combined experience of looking after more than 15,000 people with HHT across 6 countries for decades, and more than 40 monthly meetings, led by the relevant specialists. The Frameworks distinguish expert HHT care from non-expert practice, with 100% agreement between the European HHT clinicians:

- There are detailed sections for hospital specialist clinicians.
- **Importantly, to guide any healthcare professional, there is a simple flow chart for any consultation, *Figure 1*** at https://pubmed.ncbi.nlm.nih.gov/34737116/:



WHERE CAN I GET MORE INFORMATION?

Our Trust website below provides extra links to VASCERN materials, including to:

6 HHT from VASCERN HHT: https://www.youtube.com/watch?v=0YjWf7Agn40
6 An Overview of HHT: https://www.youtube.com/watch?v=z2gALD8xSNE

AND COMING SOON:

The **NHS Rare Disease Collaborative Network (RDCN)** for Hereditary Haemorrhagic Telangiectasia is operating behind the scenes to address unanswered questions in HHT, particularly treatments for bleeding and liver AVMs. We will update as soon as we can.