

Hereditary Haemorrhagic Telangiectasia
(Oslo-Weber Rendu Syndrome)



WHAT IS HHT?

HHT is a genetic disease (which means you can be born with it).

HHT results in blood vessels that form incorrectly in some areas of the body.



These formations are called **telangiectasia** when they form part of the small blood vessels and arteriovenous malformations (**AVMs**) in the larger blood vessels.

These abnormal vessel can occur in the nose, mouth, gastrointestinal tract, the brain, lungs, liver and on the skin. They can rupture and bleed or lead to other complications.



HHT affects about 1 in every 5000 people worldwide. It is a chronic (lifelong) disease which cannot be cured but it can be safely managed.

You need to take control of your disease and work with your doctor to manage it properly. That way you can improve your quality of life.



The better the management the better the prognosis!

HOW IS HHT DIAGNOSED?

HHT is diagnosed by using the following criteria: (or by genetic testing)

- 1 Spontaneous and recurrent nosebleeds
- 2 Telangiectasia (small red spots) on the skin, lips or inside the nose and mouth.
- 3 A first degree relative (biological parent, child or sibling) who has HHT
- 4 The presence of AVMs in the lungs, liver, spine or brain (diagnosed on a screening scan)

CAUSE OF HHT



HHT is caused by a mutation (a change in the structure of the genetic material in the cell).

HHT is NOT caused by lifestyle habits or anything you have "done wrong", it is genetic. This means that HHT can be transferred to your children.



The chance of a child inheriting the HHT mutation from a parent with HHT is 50%. In other words, every child has a 1 in 2 chance of being born with HHT.

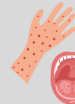


The parents, siblings and children of anybody living with HHT should be screened for HHT in order to diagnose them as early as possible. Early diagnosis, even if somebody does not have any of the symptoms, can prevent problems later in life.

COMMON SYMPTOMS



Spontaneous and recurrent nosebleeds



Small red spots on lips, tongue or skin (telangiectasia)



Fatigue (due to iron deficiency)



Gastro intestinal bleeding (e.g. dark stools)



Shortness of breath (due to lung AVM or anaemia)



Migraine headache (due to AVMs)



Three or more of the above-mentioned criteria indicates a definite diagnosis. Possible diagnosis for 2 and unlikely if only one criterion is met.

TREATMENT

The type of treatment you need will depend on how severe your HHT is and if/where you have AVMs. Nose bleeds will get worse as you get older.

Ideally, you should have different specialists treating your HHT and the complications:



HAEMATOLOGIST or SPECIALIST PHYSICIAN

- To give you intravenous iron (in a drip) to treat your Iron Deficiency Anaemia. It is important that you do not always need a blood transfusion!
- Also to do investigations to screen for AVMs



ENT SURGEON

To give you treatment to decrease your nose bleeds.

There are various options available that are specific to each individual patient.



GASTROENTEROLOGIST

They are often the first specialist to find a reason for your iron deficiency and play an important role in the diagnosis and management of telangiectasia in the stomach and gut.

IMPORTANT to REMEMBER



Always inform your doctor that you have HHT or if you suspect you might have HHT. Keep any records of your HHT with you and show it to every healthcare professional involved in your care.



Do not scuba dive or climb at high altitude, without consulting your doctor. These activities may put you at risk for severe complications or low levels of oxygen.



Any female with HHT who wish to fall or are already pregnant must discuss this with her doctor as such a pregnancy should be regarded as high risk (especially if she has lung AVMs). Females should avoid oral contraception as it poses a risk for blood clots. Alternative means of contraception such as intrauterine devices is recommended.



Genetic counselling is advised. Family planning and potential risks to mother and baby need to be discussed. If genetic testing is done you need to be informed about what the results entail.



Be careful with medication! Some medicines might cause you to bleed more easily (like certain pain medications e.g. aspirin and ibuprofen). Please discuss any new medications with your HHT healthcare provider.



Inform your dentist about your HHT diagnosis as you should use antibiotics before they do any form of invasive treatments (filling, root canal, etc).



When you are admitted for any procedure or you need to receive an "drip" (IV line) If you have a lung AVM you will need an IV filter as lung AVMs might cause micro-bubbles in your blood stream.



Please contact your doctor if you experience any new symptoms or a change in current symptoms. Seek urgent medical attention for any severe bleeding.



CureHHT website QR code:

www.bloodsa.org.za

Please note:

The information provided here is meant to be a general overview and should not be used as a substitute for professional medical advice. If you have any questions or concerns about your health, please consult with a healthcare professional.



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