Understanding HHT
More than a nosebleed

HHT ireland
What is HHT?
Hereditary Haemorrhagic Telangiectasia (HHT) is a rare genetic disorder of the blood vessels. It causes ArterioVenous Malformations (AVMs) in many vital organs.
What is an AVM?
**Arteries and Veins** are connected by capillaries which filter the blood, allow oxygen exchange and lead to normal blood flow.

**In HHT** patients, the capillary connection is often missing resulting in abnormal blood vessels (malformations). This gives rise to less filtering, faster blood circulation and less oxygen exchange, causing HHT symptoms.
Nosebleeds
90% of HHT patients suffer recurrent nosebleeds, which may result in anaemia and the need for iron infusions and blood transfusions.

Telangiectases
These are small AVMs and appear like tiny red dots on the face, skin, lips, mouth and fingertips.

*(Other symptoms may include shortness of breath, exercise intolerance, fatigue, migraine headaches, seizures, abdominal pain, intestinal bleeding).*

The manifestations of HHT are variable; patients can present with a varied combination of symptoms and signs.
HHT
The silent dangers
www.hhtireland.org
40% of patients have AVMs in the lungs and/or brain. These are the most dangerous manifestations of HHT because they may cause haemorrhage, strokes, or brain abscesses.

AVMs are often present in the gastrointestinal tract, They may bleed continuously, and are difficult to locate and treat.
HHT Timeline

Newborns:
Possible lung and brain AVMs at birth.

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When Pulmonary (lung) and Cerebral (brain) AVMs are not detected and treated they represent a serious danger for patients during their entire life.

**Age 12:**
Nosebleeds may begin. They can appear earlier, later or never at all.

**Age 30 to 60:**
Telangiectases become more evident. Increase in nosebleeds. Possible increase in gastrointestinal bleeding.

**Age 60+:**
Gastrointestinal bleeding may worsen. The dependence on iron infusions and blood transfusions may increase.
How is it transmitted?
HHT is an autosomal dominant hereditary disorder

It does not depend on the sex of the child or the parent.

A parent with HHT may transmit the gene; each child of an affected parent has a 50% chance of inheriting the disorder.

If you don’t have HHT you cannot pass it on.
A diagnosis is likely if 3 of these clinical criteria are present

1. Spontaneous & recurrent nosebleeds
2. Multiple telangiectases in typical locations (face, lips, oral cavity, fingers)
3. AVMs in other organs (lungs, liver, brain, gastrointestinal tract)
4. A 1st degree relative with HHT by these criteria

A cure does not exist but screening for manifestations in a HHT Specialist Centre can help prevent the most serious complications of HHT

1 person in every 5000 has HHT
90% of them don’t know it
HHT is a manageable disorder.
About us

HHT Ireland represents HHT patients and family members. Our primary aims are to promote awareness of the disorder, to provide support to affected families and to encourage early diagnosis in a Specialist Centre.

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