Gastrointestinal Bleeding in HHT

Introduction:
About 25% of those with HHT will develop clinically significant bleeding in the gastrointestinal (GI) tract. It can range from mild to severe. Telangiectases can be found anywhere in the gastrointestinal (GI) system, including the esophagus (swallowing tube), the stomach, the small intestines, and the colon (large intestines). Most commonly, however, the stomach and the beginning of the small intestines are involved. Telangiectases in the GI tract look similar to telangiectasia on the skin. Telangiectases in the GI tract do not cause pain or discomfort. Symptoms of GI bleeding are black or bloody stools and/or anemia. Anemia (low blood count) in turn can cause fatigue, shortness of breath, chest pain or lightheaded feelings.

Bleeding from the stomach or intestines is generally treated only if it is causes anemia (low blood count). Iron replacement therapy is the first line of defense. If iron therapy does not control the anemia, transfusion and endoscopic treatments using a heater probe, bicap or laser are options. Hormonal treatment has also been helpful in some people.

Detailed description:

Severity: broad range- very mild disease requiring little or no therapy to severe, persistent bleeding requiring repeated transfusions.

Age of Onset: Unusual before age of 40; typically presents in those age > 50. As a patient ages, the problems caused by HHT in the GI tract may become increasingly severe.

Risk factors: No particular foods, activities or medications have been identified as contributing to GI bleeding in HHT - though as most with HHT know, anticoagulants (blood thinners) and anti-inflammatory agents such as ibuprofen and aspirin can worsen bleeding. It is not known why 25% of those with HHT develop significant GI bleeding and 75% do not.

Appearance: Telangiectases can be found anywhere in the gastrointestinal system including the esophagus (or swallowing tube), the stomach, the small intestines, and the colon (or large intestine), though most commonly involved is the stomach and the beginning of the small intestines. Telangiectasia in the GI tract look quite similar to telangiectasia on the skin, appearing as small red spots or distinct areas of delicate, lacey red vessels lining the inner surface of the stomach or intestines. These telangiectasia are fragile tangles of abnormal veins and arteries which can frequently bleed. The bleeding is usually slow but is often persistent.

Symptoms: Telangiectases in the GI tract do not cause stomach pains or cramps, but they can cause a broad range of other symptoms that may be mistakenly attributed to other problems. General symptoms may be those associated with anemia or a low blood count, the result of chronic blood loss which is greater than the body's capacity to make new blood cells to compensate for losses due to bleeding. These symptoms, which are not specific to GI bleeding, may include fatigue, shortness of breath, chest pain or lightheaded feelings. More specific signs of GI bleeding are related to the effects of blood in the GI tract, which can cause stools to become black, tarry, and have a distinctive foul odor. Iron supplements taken by mouth, Pepito-Bismol, some foods and blood swallowed from nosebleeds can also turn stools black and it may be difficult to distinguish between these different causes. Small losses of blood in the GI tract (common in HHT) are not sufficient to change the color of the stool. Nevertheless, slow bleeding can lead to a low blood count and related medical problems.

Diagnosis: In addition to obtaining a detailed account of symptoms and performing a physical examination, a physician will seek the initial verification of chronic and significant blood loss by obtaining a blood count, (often referred to as a CBC or Complete Blood Count). In people with HHT, special attention must be given to a low blood count for two reasons: (1) It is of vital importance not to dismiss anemia as being due solely to nosebleeds because nosebleeds may account for only some of the anemia, particularly when the extent of anemia seems disproportionate to the amount of nose bleeding. (2) People with HHT may develop medical problems unrelated to
HHT (such as ulcers or colon cancer) which can cause similar symptoms and signs of GI blood loss. Anemia found in someone with HHT should not be dismissed as being caused by HHT without good reason or further evaluation at some point. Although these points may sound obvious, they are emphasized as fundamental steps that may be overlooked. These errors may cause GI bleeding to go undiagnosed and untreated or to be mistakenly attributed to HHT while another cause of GI bleeding is being overlooked.

Further work-up of gastrointestinal blood loss in a patient with HHT should, at some point, involve inspection of the stomach and colon by endoscopy and colonoscopy which allow the physician to see the inner lining of some of the GI tract. These tests are helpful in identifying other causes of GI bleeding such as ulcers, cancer or colitis and also permit us to determine if telangiectases are present in the areas lining the stomach or intestines. However, the presence of telangiectases does not tell us if telangiectases are the principal culprits contributing to the blood loss.

Treatment:
Gastrointestinal (GI) bleeding can be one of the most difficult manifestations of Hereditary Hemorrhagic Telangiectasia (HHT) to treat. There is currently no treatment available that completely and permanently stops all GI bleeding in HHT patients.

Direct local therapy is sometimes effective in treating GI bleeding caused by HHT. For telangiectases accessible for endoscopic therapy, treatment can be attempted by laser, injection, cautery or heater probe. These treatments may be useful in patients who have fewer telangiectases (usually younger patients) or in whom one or a few sites can be identified as responsible for much of the bleeding.

Often systemic therapy such as oral medication is necessary. The first line of medication is iron. Iron, a necessary building block of red blood cells, is depleted by chronic blood loss. Low levels can limit the body's capacity to make new red blood cells and compensate for blood loss. However, oral iron may irritate the stomach or cause constipation for some people. Some formulations may be better tolerated than others, particularly slow release formulations such as SloFe. Oral iron requires some acid in the GI tract to help with absorption. Often people with GI bleeding and HHT are placed on acid lowering medication such as cimetidine (Tagemet), ranitidine (Zantac) or omeprazole (Prilosec) in an attempt to help the stomach heal. While there is no evidence to guide or support the use of these medications in HHT, they may reduce the absorption of iron. If your physician does not wish to discontinue one of those medications, small doses of vitamin C (i.e. 200 mg. with each dose of iron) can enhance iron absorption. Intravenous (IV) iron may be a useful alternative as well, particularly if the bleeding is more severe and the body's need for iron is greater than can be fulfilled by oral iron alone.

If repeated blood transfusions are necessary, other therapies should be attempted. Most experience has been obtained with hormonal treatment. Estrogen and progesterone have been useful in as many as 60 - 70% of patients with chronic GI bleeding due to HHT. The dose is particularly important. The therapy has been considered unsuccessful for some people who have been prescribed a low or inadequate dose. If someone is likely to benefit from estrogen/progesterone, an effect is usually noticed within 2 - 4 weeks. NOTE: You must be screened and treated for PAVMs before starting hormonal replacement. While it may be tremendously helpful for some, estrogen/progesterone is either ineffective, poorly tolerated, contraindicated or rejected by others. Relatively minor, but unpleasant, side effects can occur, such as retaining fluid, sleep disturbances and gastrointestinal upset. The increase in clotting factors can cause serious complications in people with significant heart disease and many men may be uncomfortable with or refuse estrogen/progesterone because of the feminizing effects. In addition, estrogen/progesterone at the high doses most commonly used in HHT may not be advisable in women with a history of breast cancer. Danazol is a medication that acts as a weak male hormone that has been used for a long time for other medical disorders. It has benefited half of the small number of patients who have taken it for GI bleeding and has been well tolerated. Danazol is a possible therapy for GI bleeding in HHT, particularly for men. Other therapies are being explored.
Summary:

Diagnosis:
1. anemia should not be dismissed as due to nosebleeds alone
2. anemia can be due to other causes not related to HHT: colon cancer, ulcers

Evaluation:
1. a blood count (a CBC or Complete Blood Count)
2. endoscopy, colonoscopy

Treatments:
1. Iron, oral or intravenous
2. blood transfusions
3. endoscopic therapy
4. Hormonal therapy: estrogen/progesterone (dose is very important)
5. Danazol
6. Amicar
7. Octreotide

References: