Also known as
- Babington disease
- Goldstein haematemesis
- Goldstein heredofamilial angiomatosis
- Goldstein syndrome
- Hereditary haemorrhagic telangiectasia
- Osler disease
- Osler syndrome
- Osler–Rendu–Weber syndrome
- Rendu–Osler syndrome

Osler–Rendu–Weber disease (hereditary haemorrhagic telangiectasia: HHT)
- occurs mainly in white people.
- is an uncommon autosomal dominant disorder characterized by multiple telangiectasia of the skin, and of the oral, nasal, conjunctival and gastrointestinal mucous membranes.
- These manifest from childhood and are liable to ulcerate and bleed, and so epistaxis and gastrointestinal haemorrhages are common.
- Arteriovenous fistula, especially of the lungs, liver and brain.
- Recurrent complications are severe anaemia, stroke and pulmonary hypertension.

The mechanism
- underlying the formation of the vascular malformations in HHT seems related to transforming growth factor beta 1 (TGFB-1) signaling defects adversely affecting matrix and connective tissue production.
  - TGF-beta signalling has a pivotal role in angiogenesis.

Several forms of HHT have been described:
- *Hereditary haemorrhagic telangiectasia 1*
  - Which predisposes to pulmonary and cerebral arteriovenous fistulae and early oral and nose bleeds, is related to mutation of the endoglin gene (ENG).
  - ENG is a receptor for transforming growth factor beta 1 (TGFB-1) and transforming growth factor beta 3 (TGFB-3).
- Hereditary haemorrhagic telangiectasia 2
  - Dermal lesions and hepatic vascular malformations are more frequent and appear earlier in life with later nose bleeds, it is related to mutation in the ALK1 gene.
Activin receptor-like kinase 1 (ALK-1 or ACVRL1) is a TGFB1 receptor.

- Hereditary haemorrhagic telangiectasia 3 has not yet been linked to a defective gene.
- Juvenile polyposis/HHT syndrome is caused by mutations in the SMAD4 gene, which modulates TGF.
- Hereditary haemorrhagic telangiectasia 4 has now been identified.

The diagnostic criteria for HHT include:
1. Spontaneous recurrent epistaxis
2. Multiple telangiectasis
3. Proven visceral arteriovenous fistulae
4. First-degree family member with HHT.
   - If three or four of these criteria are met, a patient has definite HHT, while two gives a possible diagnosis.

Treatment
- In mild cases of HHT, no treatment is necessary.
- Anaemia due to bleeding may necessitate blood transfusions.
- AVMs in critical organs may necessitate surgery or embolisation under radiographical control.
- In severe cases of HHT, recurrent epistaxis is treated surgically with nasal septum skin transplants by using skin taken from the lower trunk.
- Infra-red laser coagulation is well suited to the treatment of telangiectases in the skin and/or mucosal surfaces.

Prognosis
- Most patients with hereditary hemorrhagic telangiectasia (HHT) have a favorable prognosis.
- The prognosis depends on the degree of systemic involvement, especially involvement of the pulmonary, hepatic, and central nervous systems. Only 10% of patients die from complications of their disease.

Background
- First described by Sutton in 1864 and Babington in 1865 as a hereditary epistaxis disease.
- In 1896, Rendu described the disease as a pseudo hemophilia related to hereditary epistaxis.
- William Bart Osler in 1901 authored the first comprehensive description of the disease in three patients, and emphasized its familial nature.
- Weber (1907) recognized HHT as a clinical entity distinct from hereditary hemophilia, and Hanes (1909) named the syndrome hereditary hemorrhagic telangiectasia.

The main persons
- William Osler
  - He started to study Medicine at Toronto Medical School in 1868. He spent the longest period at University College, London, where Osler was the first to see platelets.
  - Osler returned to Canada to undertake general practice in Dundas, was appointed lecturer in the Institutes of Medicine at McGill and became Professor at a age of 26.
  - In 1888/1889, Osler accepted an invitation to be the first Professor of medicine at the Johns Hopkins University Medical School. Osler published his book 'Principles and Practice of Medicine' in 1892.
Osler wrote some of the early descriptions of platelets and classical papers on hereditary telangiectasia, lupus erythematosus

**Henri Jules Louis Marie Rendu**
- He started studying Medicine in Paris and became interne at the Hôpital Saint-Antoine.
- In 1877, Rendu received the degree of hospital physician
- He then returned to the Hôpital Necker as Head of the Department of Medicine and he received the ultimate accolade of election to membership of Academy of Medicine.

**Frederick Parkes Weber**
- was born on 8 May 1863, in London.
- Weber obtained his doctorate at Cambridge and worked at St.
- appointed honorary physician to the German Hospital, Queen Square, London.

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