原文題目(出處)：	Marathon of eponyms: 15 Osler–Rendu–Weber disease (Hereditary haemorrhagic telangiectasia) Oral Diseases 2011;17:125-27
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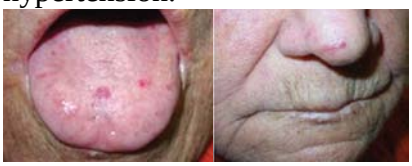
內文：

- 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-1or 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.

題號	題目
1	Hereditary haemorrhagic telangiectasia 好發於? (A) white people, children (B) White people, old people (C) Asian people, children (D) Asian people, old people
答案(A)	出處：Marathon of eponyms: 15 Osler–Rendu–Weber disease 遺傳性出血性血管擴張症之介紹及治療
題號	題目
2	Hereditary haemorrhagic telangiectasia 的病因?
	(A) 血液凝固問題 (B) 凝血因子缺乏 (C) 基因缺陷所導致的血管畸形病變 (D) 環境汙染
答案(C)	出處：Marathon of eponyms: 15 Osler–Rendu–Weber disease 遺傳性出血性血管擴張症之介紹及治療 Osler-Weber-Rendu Syndrome --Dental Implications