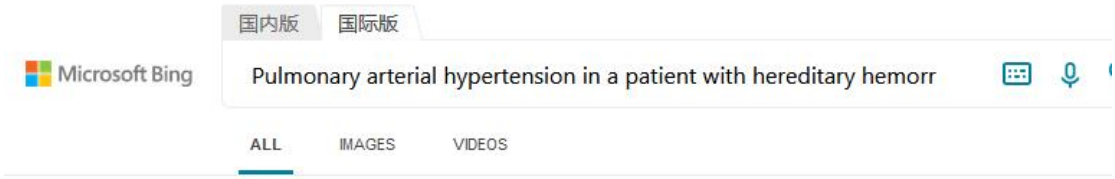

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Mar 01, 2013 · Introduction. **Hereditary hemorrhagic telangiectasia** (HHT), or Rendu–Osler–Weber syndrome, is an autosomal dominant disorder with variable penetrance that is characterized by the presence of epistaxis, telangiectasias and vascular malformations in the **pulmonary**, gastrointestinal, hepatic and cerebral regions. 1 It is a rare disease, with an estimated prevalence of 1:5000 to 1:10 000 .

Cited by: 6 **Author:** Alejandro Raimondi, Isabel Blanco, Xavier...
Publish Year: 2013

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Pulmonary Arterial Hypertension in a Patient With Hereditary Hemorrhagic Telangiectasia ... With the reported **family** history, and given the fact that HHT is **hereditary** in nature, a genetic study was done. BMPR2 gene sequencing was normal, and complete sequencing of the ACVRL1

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A subset of **patients** with hereditary hemorrhagic telangiectasia (HHT) develops pulmonary hypertension (PH) by mechanisms including pulmonary arterial hypertension, high flow, and elevated pulmonary arterial wedge pressure (PAWP). We aimed to describe echocardiographic and hemodynamic characteristics of patients with coexisting HHT and PH.

Cited by: 18 **Author:** Melissa A. Lyle, Eric R. Fenstad, Michae...
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Pulmonary arterial hypertension (PAH) and hereditary hemorrhagic telangiectasia (HHT) are distinct **clinical entities caused by germline mutations in genes encoding members of the TGFβ/BMP superfamily: BMPR2 in PAH and ACVRL1, ENG, or SMAD4 in HHT. When PAH and HHT occasionally co-exist within the same family, ACVRL1 mutations predominate**. We report a ...

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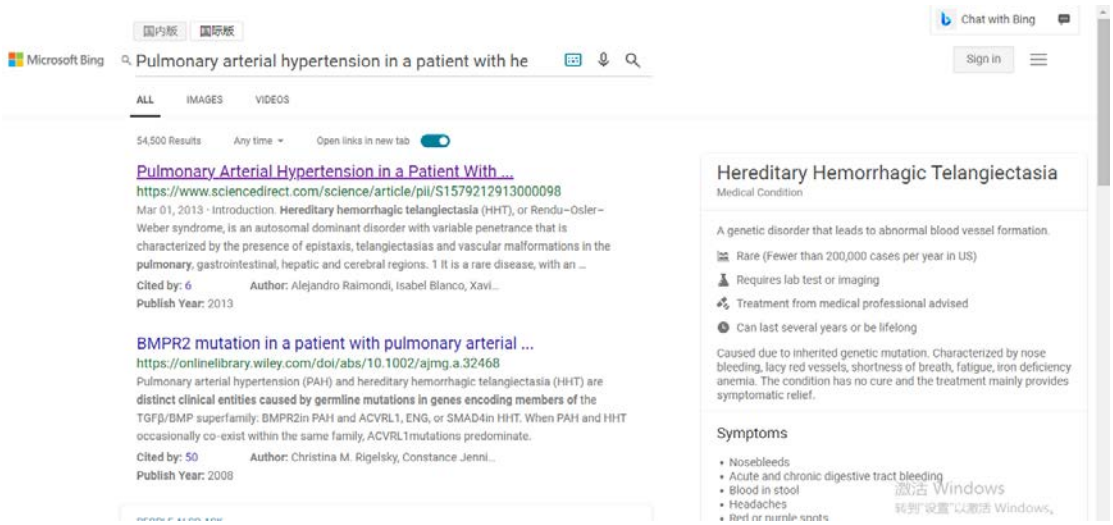
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Cited by: 50
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Hereditary Hemorrhagic Telangiectasia
Medical Condition
A genetic disorder that leads to abnormal blood vessel formation.
Rare (Fewer than 200,000 cases per year in US)
Requires lab test or imaging
Treatment from medical professional advised
Can last several years or be lifelong
Caused due to inherited genetic mutation. Characterized by nose bleeding, lacy red vessels, shortness of breath, fatigue, iron deficiency anemia. The condition has no cure and the treatment mainly provides symptomatic relief.
Symptoms
Nosebleeds
Acute and chronic digestive tract bleeding
Blood in stool
Headaches
Red or purple spots
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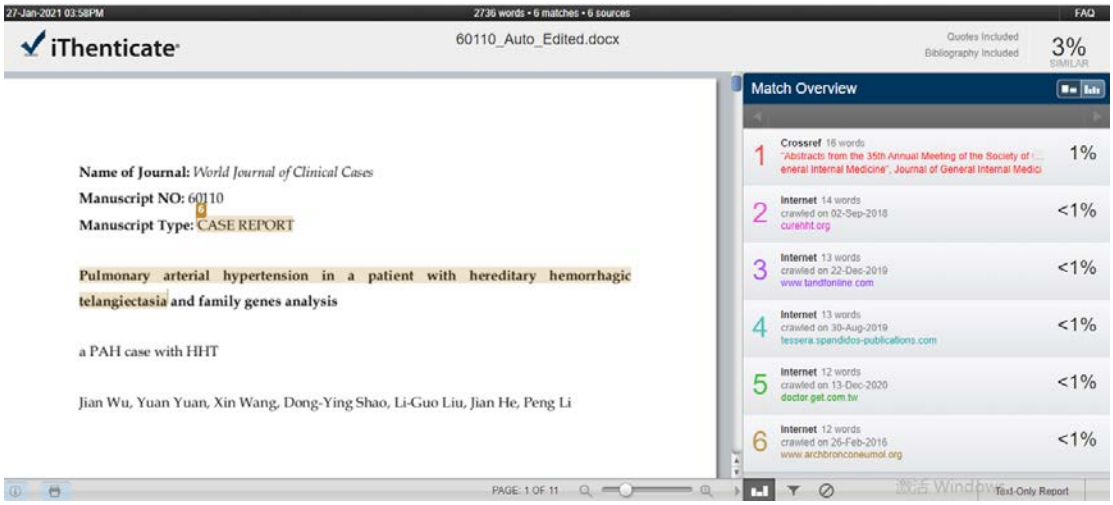
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Mar 01, 2013 · Hereditary hemorrhagic telangiectasia (HHT) is **an autosomal dominant disorder characterized by the triad of epistaxis, telangiectasia and vascular malformations**. Pulmonary vascular complications associated with this disease include pulmonary arteriovenous malformations (AVM) and, less frequently, pulmonary hypertension (PH).

Cited by: 6 **Author:** Alejandro Raimondi, Isabel Blanco, Xavier...
Publish Year: 2013

Pulmonary hypertension in a patient with hereditary ...

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Feb 01, 2013 · A young male **patient** reported for evaluation of progressive easy fatigability, accompanied by a recent history of recurrent haemoptysis. His clinical examination was unremarkable except for evidence of **pulmonary arterial hypertension** (PAH). Routine investigations (haemogram, coagulogram, serological tests for connective tissue disorders and a sputum Ziehl Neelsen stain for ...

Cited by: 7 **Author:** Davinder Chadha, Ajay Handa, Abhishek ...
Publish Year: 2013

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Publish Year: 2016

Case report: Pulmonary hypertension in a patient with ...

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The intersection of genes and environment: development of **pulmonary arterial hypertension in a patient with hereditary hemorrhagic telangiectasia** and stimulant exposure. Chest 2012; 141:1598–600. [PMC free article]

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