Stroke…It Can Happen to Anyone

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Objectives:

- Define Stroke
- Describe signs and symptoms of stroke
- Discuss a case study involving a stoke because of HHT

Stroke (CVA)

- A condition where a blood clot or ruptured artery or blood vessel interrupts blood flow to an area of the brain. A lack of oxygen and glucose (sugar) flowing to the brain leads to the death of brain cells and brain damage, often resulting in an impairment in speech, movement, and memory.

Elderly??

Young??

Babies/Children??

Stroke in the U.S.

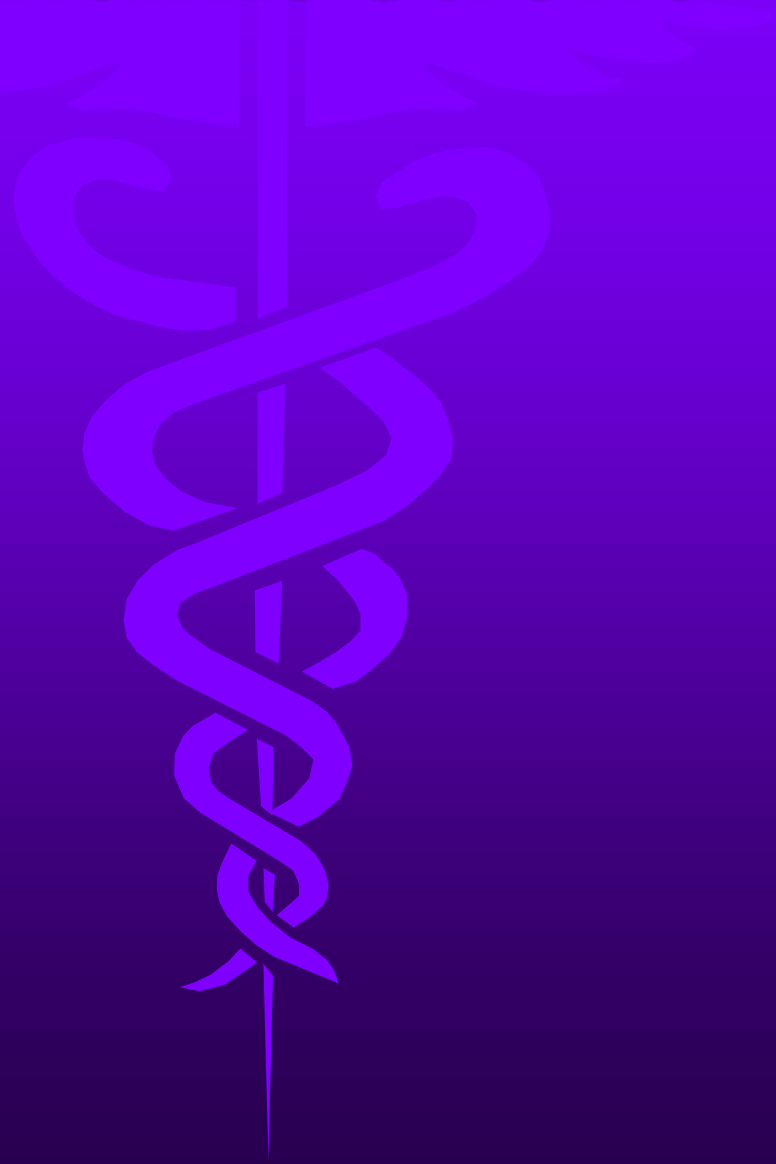
- 730,000 new or recurrent strokes each year
- 3rd leading cause of death in the United States: second leading cause worldwide.
- Leading cause of serious, long-term disability.

Types of Stroke

- Ischemic Stroke
- Hemorrhagic Stroke

Ischemic Stroke

- 75%
- Blood Clot



Hemorrhagic Stroke

- Blood Vessel
- Artery

Signs and Symptoms

- Face — Sudden weakness or droopiness of the face, or problems with vision
- Arm — Sudden weakness or numbness of one or both arms

Signs and Symptoms

- Speech — Difficulty speaking, slurred speech, or garbled speech

- Time — Time is very important in stroke treatment. The sooner treatment begins, the better the chances are for recovery.

Initial Testing

- Emergent brain imaging with CT or MRI is mandatory in all patients with sudden neurologic deterioration or acute stroke.

Initial Testing

- Electrocardiogram
- Complete blood count including platelets
- Cardiac enzymes and troponin
- Electrolytes, urea nitrogen, creatinine
- Serum glucose
- Prothrombin time and international normalized ratio (INR)
- Partial thromboplastin time
- Oxygen saturation
- Lipid profile

Testing

- Liver function tests
- Toxicology screen
- Blood alcohol level
- Pregnancy test in women of child-bearing potential
- Arterial blood gas if hypoxia is suspected
- Lumbar puncture if subarachnoid hemorrhage is suspected and head CT scan is negative for blood
- Electroencephalogram if seizures are suspected

Other Test???

Fever

Treatment and prevention

- Blood Pressure
- Anticoagulant therapy
- Smoking
- Diabetes

Hereditary Hemorrhagic Telangiectasia (HHT)

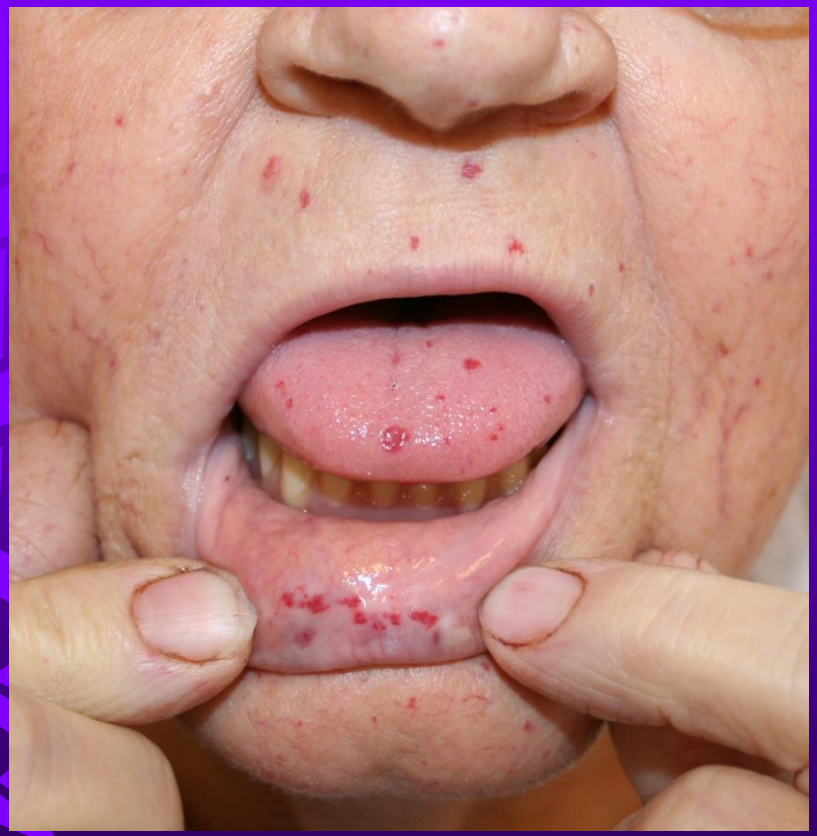
- Osler-Weber-Rendu
- Genetic disorder of the blood vessels
- Affects approximately 1 in 5000

HHT

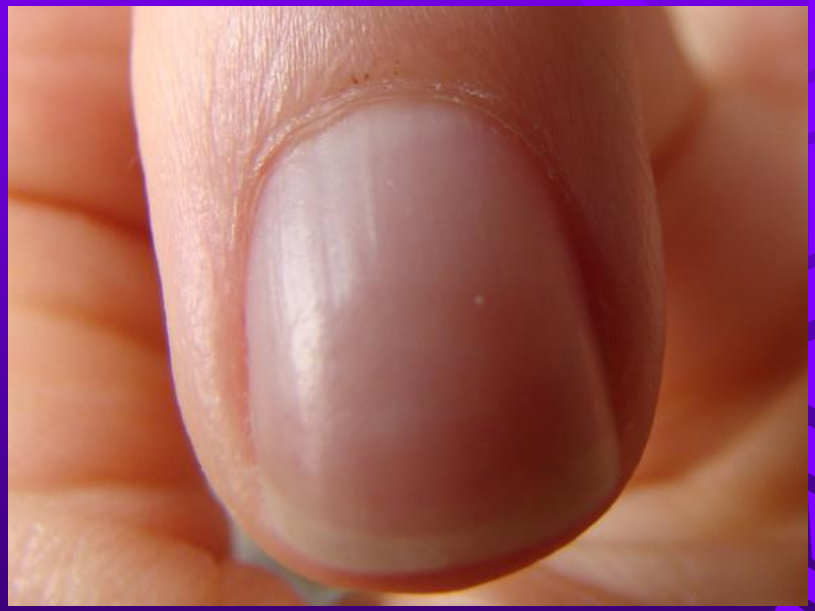
- Disorder involving abnormalities of the blood vessels, not a clotting disorder

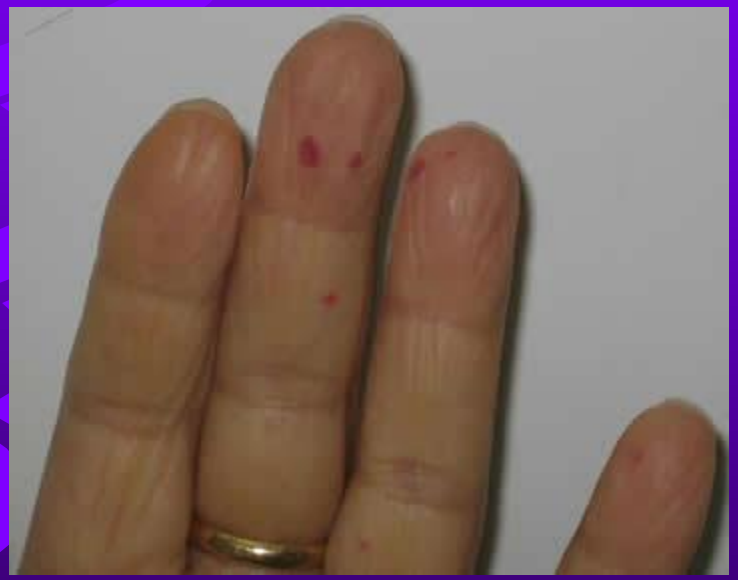
Signs and Symptoms

- Telangiectases



Signs and Symptoms





Signs and Symptoms

- Nosebleeds

Signs and Symptoms

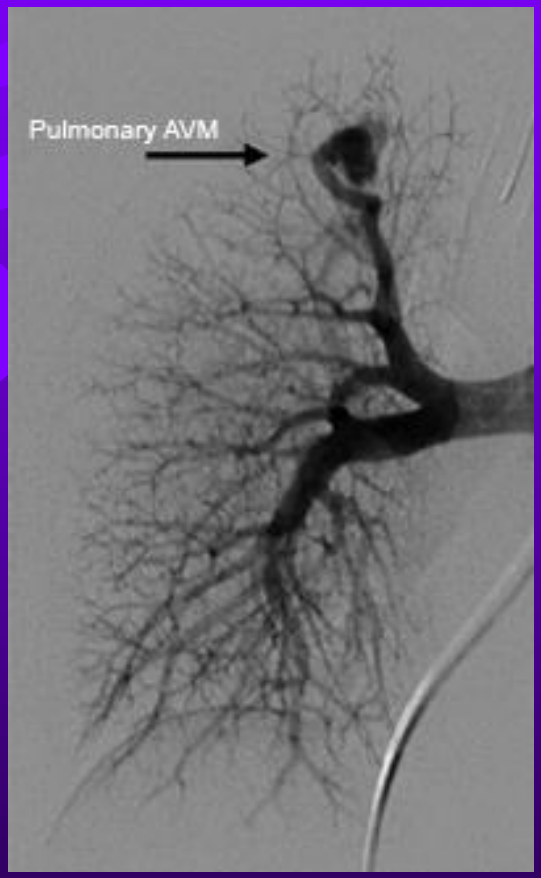
- GI Bleeding

Signs and Symptoms

- Arteriovenous malformation (AVMs)

Signs and Symptoms

- Pulmonary AVMs



Signs and Symptoms

- Brain AVMs

Signs and Symptoms

- Liver AVMs

HHT Manifestations

- 90-95% of individuals with HHT will have nosebleeds by adulthood, but they vary from infrequent and minor to daily and severe.
- 90-95% develop at least a few telangiectasia (small red or purple spots) on the skin of the face and/or hands by middle age.

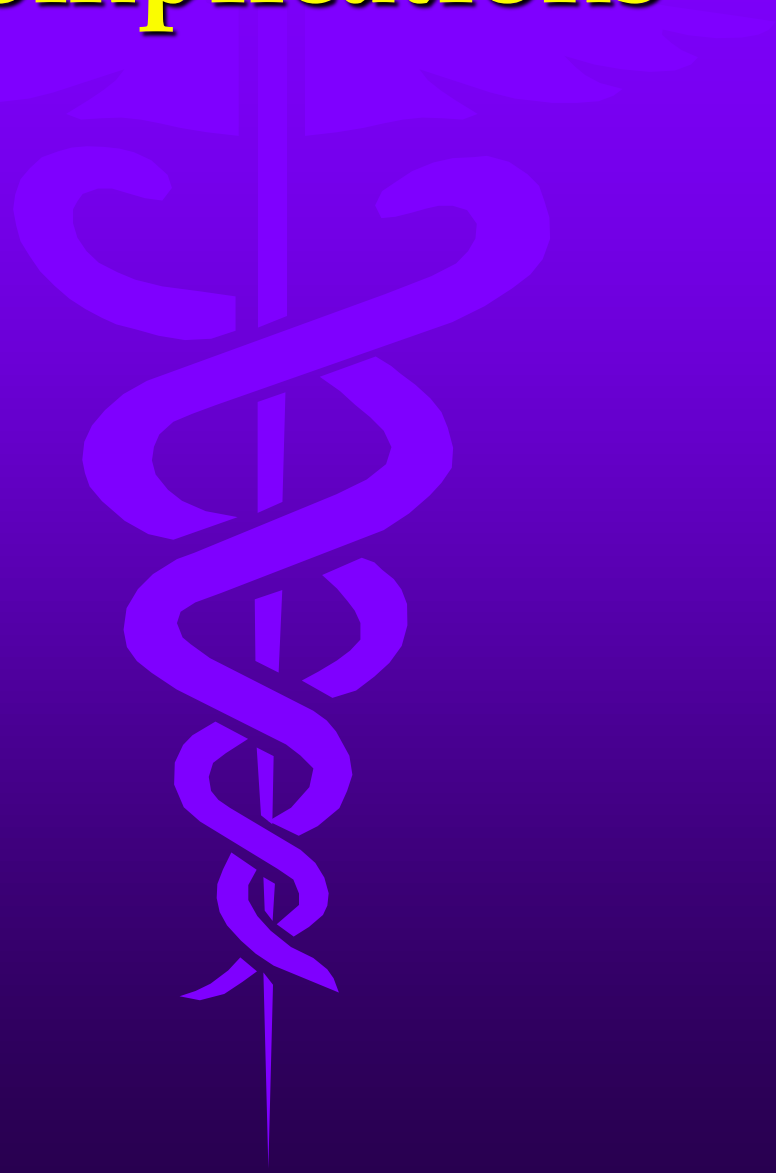
HHT Manifestations

- 20-25% develop bleeding in the stomach or intestines, but rarely before 50 unless affected with juvenile polyposis in conjunction with HHT.
- 30-50% have an arteriovenous malformation (AVM) in their lungs. These are usually present at birth and pose significant risk if not treated properly.

HHT Manifestations

- 5-20% have at least one AVM in their brain. These are present at birth and pose significant risk if not treated properly.
- Hepatic AVMs are relatively common, approximately 5% are symptomatic.
- Abnormal blood vessels in the liver are relatively common, but most cause no symptoms. Biopsy or treatment should rarely be done and specifically, embolization should not be done.

Complications



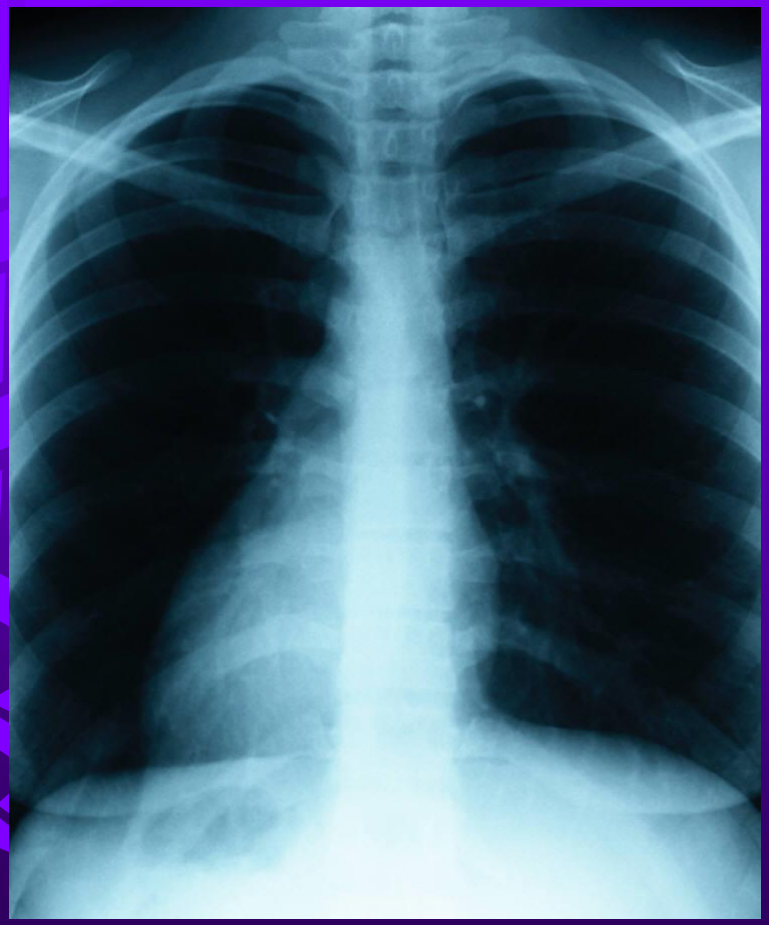
- Anemia
- Stroke

Treatment

- Nosebleeds

Treatment

- AVMs



Genetic Testing

- Endoglin and ALK-1 genes
- Few labs in United States

Genetic Testing

- Family Members

Genetic Testing

- Results
  - Positive for a mutation
  - Negative for a significant DNA sequence variation
  - Uncertain

HHT Case Study

- 34 year old female arrived in the ER by ambulance
- Presents with mental confusion, and facial drooping

History

- Diagnosis of HHT at age 9
- Para 5/gravida 3

Initial Testing

- Laboratory Testing
- CT
- MRI

Further Testing

- Echocardiogram

Testing Results

Initial Diagnosis

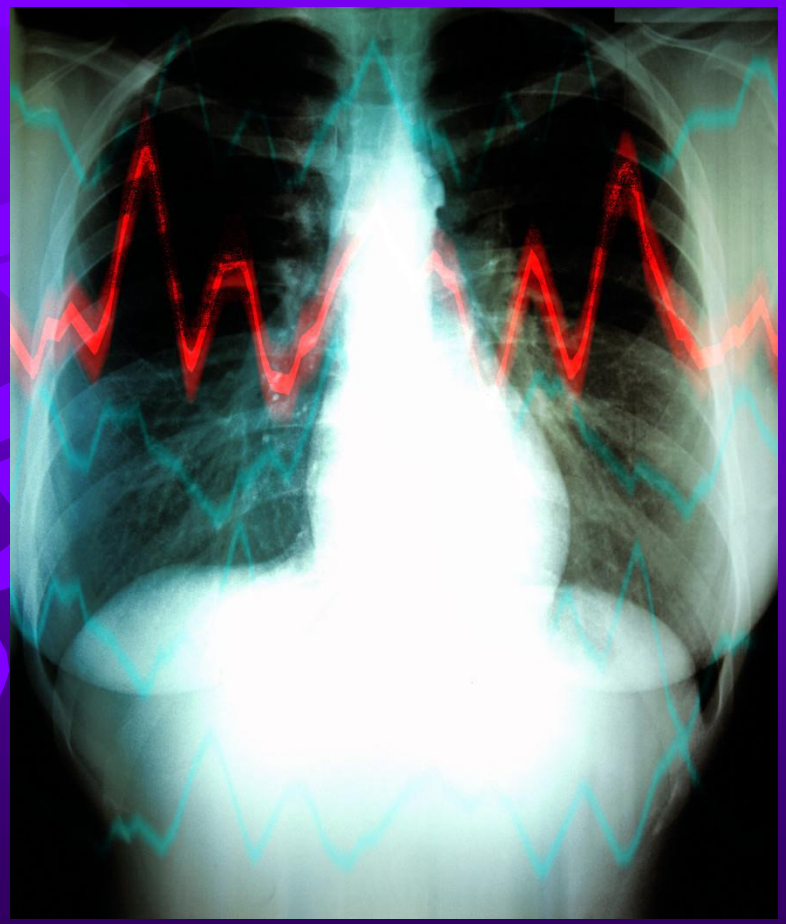
- Stroke because of hole in the heart??

Repeat Testing

- Echocardiogram

CT of Lungs

- Normal??
- Abnormal ??



New Diagnosis

- Stroke due to presence of Pulmonary AVMs

Treatment and Prevention

HHT Centers of Excellence

- GEORGIA HEALTH SCIENCE UNIVERSITY HHT CENTER
- JOHNS HOPKINS HHT CENTER
- MAYO CLINIC HHT CENTER
- OREGON HEALTH & SCIENCE UNIVERSITY HHT CENTER
- UNIVERSITY OF ARKANSAS for MEDICAL SCIENCES
- UNIVERSITY OF CALIFORNIA at LOS ANGELES HHT CENTER
- UNIVERSITY OF CALIFORNIA at SAN DIEGO HHT CENTER
- UNIVERSITY OF NORTH CAROLINA at CHAPEL HILL HHT CENTER
- UNIVERSITY OF PENNSYLVANIA
- UNIVERSITY OF TEXAS – SOUTHWESTERN
- UNIVERSITY OF UTAH MEDICAL CENTER
- WASHINGTON UNIVERSITY SCHOOL OF MEDICINE
- YALE UNIVERSITY SCHOOL OF MEDICINE

Embolisation of Pulmonary AVMs

Genetic Testing

- This patient is a carrier of the **c.263delA mutation in the *ENG gene*. This result is consistent with a diagnosis of hereditary** hemorrhagic telangiectasia (HHT), however, the severity of the symptoms cannot be predicted.

Genetic Testing of Family

- Children
- Other Family members

Further Testing of Family Members

- CT of Brain
- Echocardiogram

HHT Awareness Video More than a Nosebleed:HHT

- https://vimeo.com/242973923


BENTON