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# Hereditary Hemorrhagic Telangiectasia

National Cancer Institute

## Source

National Cancer Institute. *Hereditary Hemorrhagic Telangiectasia*. NCI Thesaurus. Code C35064.

An autosomal dominant hereditary disorder caused by mutations in the ACVRL1, ENG, and SMAD4 genes. It is characterized by the presence of telangiectasias in the skin, mucous membranes, lungs, brain, liver, and gastrointestinal tract. It results in hemorrhages from the affected areas.