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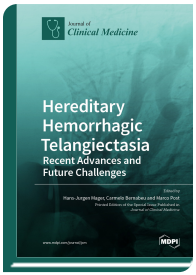
Special Issue Reprint

Hereditary Hemorrhagic Telangiectasia



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Hereditary hemorrhagic telangiectasia (HHT) is an inherited disease that affects the blood vessels, and is characterized by direct connections between arteries and veins with no intervening capillaries. These abnormal vessels may appear in the skin as tiny red dilated blood vessels in the mouth, lips, fingers and toes. The presence of these vascular lesions in the mucosa can lead to spontaneous and recurrent nose bleeding, typically beginning in mid-childhood, and this is the most common clinical manifestation of HHT, occurring in over 90% of patients. Gastrointestinal bleeding, derived from mucocutaneous vascular lesions, affects approximately 25% of patients, almost always presenting after the age of 50. Chronic nasal and gastrointestinal bleeding can cause iron-deficiency anemia, and current therapeutic strategies are trying to minimize iron and blood transfusions. HHT patients also present large vascular lesions, known as arteriovenous malformations, that occur in internal organs like lungs, liver, and brain, and may result in life-threatening complications often related to the shunting of blood. This book not only highlights the current knowledge regarding diagnosis and treatment of HHT, but also the newest insights in the molecular basis of HHT, the understanding of which is essential for the development of new medicines or therapeutic strategies.

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