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Clinical Updates on Hereditary Hemorrhagic Telangiectasia (HHT) and Other Vascular Diseases

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Message from the Guest Editor

Hereditary hemorrhagic telangiectasia (HHT) is listed in both the capillary (telangiectasia subgroup) and the arteriovenous malformation (AVM) groups in the 2018 classification from the International Society for the Study of Vascular Anomalies (ISSVA). The diagnosis of HHT remains a challenge, as it is a rare disease (with a prevalence of 1:5000–1:10,000) with age-related penetrance of its multisystemic symptoms. The average diagnostic time lag (the interval between the first symptoms and the diagnosis of HHT) is still almost three decades for probands and only somewhat shorter for at-risk family members. Other vascular diseases resembling more or less HHT, especially the capillary malformation-arteriovenous malformation (CM-AVM) syndromes 1 and 2 (caused by RASA1 and EPHB4 mutations, respectively), are even less prevalent (approx. 1:100,000). Considering the clinical and genetic peculiarities of the above conditions, their diagnosis and therapy are the subject of extensive research. Groups from all clinical specialities are encouraged to submit their original research and review manuscripts to this Special Issue of the Journal of Clinical Medicine.

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