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Perspective

Medical Translation in Hereditary Hemorrhagic Telangiectasia and Mouse Models Translational Medicine

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DESCRIPTION

A multidisciplinary branch of biomedicine called translational medicine (TM) promotes a continuous flow of information transfer between the laboratory and the clinic, always keeping the needs of the patient in heart. It plays a critical role in improving the bench-to-bedside transition. Here, we offer various TM techniques and a summary of current research using the disease model of hereditary haemorrhagic telangiectasia (HHT). The emphasis of our work is on how the intersection of basic and clinical research affects the on-going clinical management of HHT patients today and in the future. The outcomes of this bench to bedside shift should be evaluated in additional randomised clinical trials with HHT patients. The advantages of this combination of basic and clinical research may not only be significant for people with HHT, but also for those with other vascular disorders that share antigenic abnormalities.

Systemic telangiectasia and bigger vascular abnormalities are characteristics of the rare autosomal dominant vascular illness known as hereditary haemorrhagic telangiectasia (HHT), also known as Rend-Osler-Weber syndrome. Telangiectasia, which are dilated post capillary venules directly associated with dilated arterioles losing the capillary bed, are the distinguishing feature of HHT. HHT can be identified using either a molecular genetic test or the curacao clinical criteria (recurrent epistaxis, cutaneous/mucosal telangiectasia, visceral VMs, and a first-degree relative with HHT). About 90% of cases submitted for molecular diagnosis had mutations in the endoglin (ENG) and active receptor type II-like kinase 1 (ACVRL1) genes, which produce HHT1 and HHT2, respectively. The endothelial cell (EC) surface co-receptor Enderlin (encoded by ENG) facilitates BMP9 signalling by way of active in receptor-like kinase 1. So, rather than being a disease of the TGFB pathway, HHT is now thought to be a disease of the centre created by BMP9-Endoglin-ALK1-Smad.

There are still some uncertainties around the many HHT features that have been clarified. The most frequent clinical manifestation of telangiectasia is recurrent epistaxis, which is caused by them in the nasal mucosa and fingertips of the hands. It is still uncertain why pulmonary and hepatic arteriovenous malformations (AVMs) are more prevalent in HHT1 individuals than HHT2 patients. Furthermore, the genetic makeup of HHT is unknown because 10-15% of individuals with the HHT phenotype are genetic orphans. Additionally, there is significant inter- and intra-familial variation in vascular involvement and clinical symptoms despite equal pathogenic mutations. Identifying HHT is very difficult, particularly in unusual circumstances, due to the low prevalence and great clinical variability. Together, these open issues have complicated efforts to create a treatment services for HHT patients. In order to gain knowledge about the molecular control of vascular growth and to enhance patient treatment, it is essential to further our understanding of HHT.

Mouse models in translational medicine preclinical mouse models have evolved into an effective tool for researching new treatments and comprehending the pathogenic mechanisms behind HHT. However, it is difficult to simulate the HHT-related AVMs in mouse models. This is partly because there aren't any underlying theories explaining how these mutations cause vascular malformations. On the other hand, homozygous deletion of the HHT genes after, specifically in the mural cells, did not cause AVMs. Instead, loss of ACRL1 in these cells rewires the phenotypic of vascular smooth muscle cells from contractile to synthetic, having implications for pulmonary and cardiac hypertension.

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