New molecular target for disrupting development of venous malformations

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Video Byte

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Abstract

Venous malformations are the most common vascular anomaly around the world. Extensive malformations cause pain, bleeding, anatomic distortion, and organ dysfunction. Current therapies are invasive and rarely curative, making it necessary to develop new treatment options. To that end, researchers examined one molecular mechanism known to lead to venous malformations, the interaction between endothelial and smooth muscle cells. The researchers screened 5 patients with the mutation known to cause venous malformations. Malformations with that mutation showed lower expression of platelet-derived growth factor beta and alpha-smooth muscle actin. In addition, mutant endothelial cells showed enhanced cell viability and motility, and decreased tube formation. These conditions could be reversed in vitro by rapamycin, which, experiments showed, worked by inhibiting the AKT-mTOR pathway. This pathway could therefore be an important target for disrupting the formation of venous malformations.