SUPPLEMENTARY MATERIAL

Table S1: Postzygotic *CCM1* and *CCM2* missense and in-frame variants that had previously been reported in the literature.

Reference	Gene	Constitutional or high-frequency variant identified in blood and/or CCM tissue[(a)]	Late postzygotic variant[(a)]	Frequency of the late postzygotic variant	Familial / sporadic CCM
[1]	*CCM2*	none	c.355_369del, p.(Asn119_Ala123del)[(b)]	1.1 %	sporadic
[1]	*CCM2*	none	c.611_622del, p.(Val204_Glu207del)[(b)]	2.8 - 5.1 %	sporadic
[2]	*CCM1*	none	K569E and F97S	-	sporadic

[(a)] Reference sequences: *CCM1*: LRG_650t1, *CCM2*: LRG_664t2; [(b)] in-frame deletion in the phosphotyrosine binding (PTB) domain of CCM2.

References:

1. McDonald DA, Shi C, Shenkar R, Gallione CJ, Akers AL, Li S, De Castro N, Berg MJ, Corcoran DL, Awad IA, Marchuk DA. Lesions from patients with sporadic cerebral cavernous malformations harbor somatic mutations in the *CCM* genes: evidence for a common biochemical pathway for CCM pathogenesis. Hum Mol Genet 2014;**23**(16):4357-70 doi: 10.1093/hmg/ddu153.
2. Kehrer-Sawatzki H, Wilda M, Braun VM, Richter HP, Hameister H. Mutation and expression analysis of the *KRIT1* gene associated with cerebral cavernous malformations (CCM1). Acta Neuropathol 2002;**104**(3):231-40 doi: 10.1007/s00401-002-0552-6.