

Table S1. Quantitative real-time PCR primers and probes for characterization of the *KRIT1* genomic deletion in case 7.

Gene	Ref Seq	Exon / Intron	UPL Probe	Primer F	Primer R	Amplicon (nucleotide)
<i>AKAP9</i>	NM_005751.5	Int 16	#18 (cat. no. 04686918001)	tcttttgttagaacctgagg	aaagaaataaaagcaaacaataaagga	(125 nt) tcttttgttagaacctgaggccatgctttttctgtttatacaattttg agtactaatttacttttaaaftcctgctgatattctaaatctaactcttta ttgttgctttatttcttt
		Int 38	#7 (cat. no. 04685059001)	gaatatagccagtatccaacagc	agaaaaatcagaacaatgattacctct	(68 nt) gaatatagccagtatccaacagccccttctccaagcacatgctc ctctgagtaacttagaggtaatcattgttctgatttttct
<i>CYP51A1</i>	NM_000786.3	Ex 1	#52 (cat. no. 04688490001)	accatctgccagcttctctc	ttcactccgtcggaacact	(110 nt) cccaatgtcatgaatgtcctttaaagattcttacatgtttatatttattat tgacatgttttactttgtaggctggctctcgtggtaaaactgtaatgaa gctaaatgg
<i>KRIT1</i>	NM_194456.1	Int 19	#54 (cat. no. 04688511001)	cccaatgtcatgaatgtccttta	ccatttagcttcattaacagttttacc	(110 nt) cccaatgtcatgaatgtcctttaaagattcttacatgtttatatttattat tgacatgttttactttgtaggctggctctcgtggtaaaactgtaatgaa gctaaatgg
		Ex 13	#84 (cat. no. 04689089001)	tctgtgaattgaagcatgga	ttcttgagagagacgcattcc	(72 nt) tctgttgaattgaagcatggaaataataccacagtgcagcagataat ggaaggaatgcgtctctctcaagaa
		Int 4	#72 (cat. no. 04688953001)	tgaacagtaaagatgcagtcaaa gt	ttgaaaacttttattccacataatca	(127 nt) tgaacagtaaagatgcagtcaaaagtcattcctggcttgccttttagg cactgtatattctagatagagaacatttttttttaatttttcgac atgattatgtggaataaaagtttcaa
<i>ANKIB1</i>	NM_019004	Ex 6	#22 (cat. no. 04686969001)	ccaacgatcaggtgttcaaa	gcgtgtcccaggcattatac	(62 nt) ccaacgatcaggtgttcaaatgccaactccaccaccaagtgggtat aatgcctgggacacgc

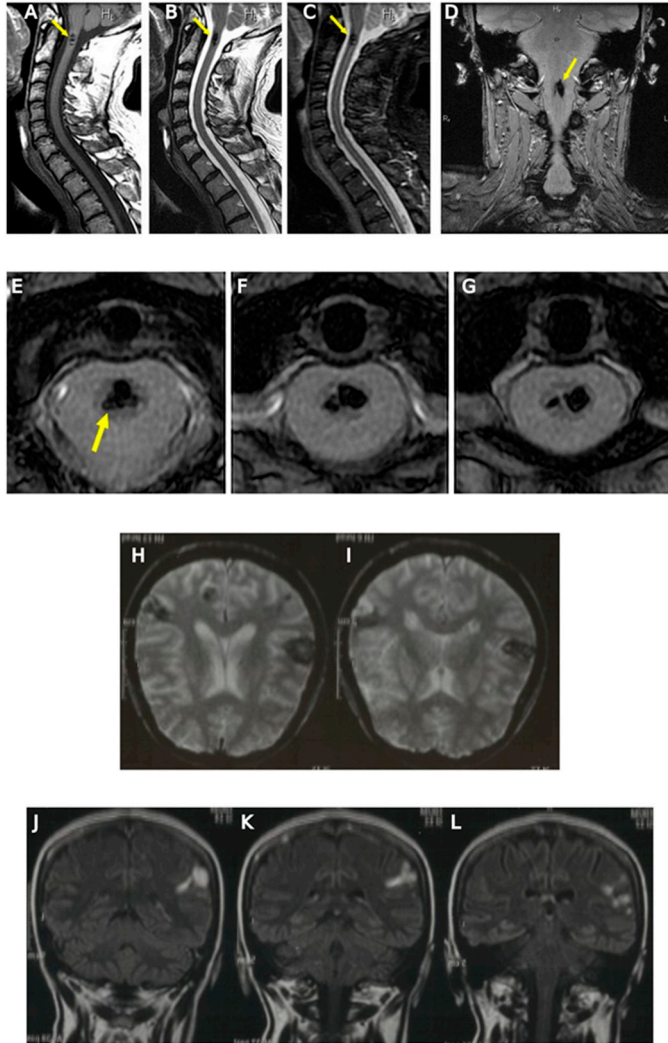


Figure S1. MRI of patient II.3 of family 12. Sequential sagittal (panels A-C) and coronal (panel D) MRI sections showing a cavernoma at the foramen magnum, posterior to the odontoid process, characterized by an elongated shape. Sequential axial MRI sections (panels E-G) showing two additional cavernomas in correspondence of the anterolateral horns of the medulla in D2 and the cauda equina in L3. Sequential transversal (panel H, I) and coronal (panels J-L) MRI section showing lesions in the left superficial white matter of the frontal lobe.