

# Genes involved in Cerebral arteriovenous malformation rupture risk

A [single nucleotide polymorphism](#) (SNP) in ALK-1 is associated with AVM susceptibility, and SNPs in IL-6, TNF- $\alpha$  and APOE are associated with AVM rupture.

A study aimed to gain insights into the potential [mechanism](#) involved in [ruptured cerebral arteriovenous malformation](#).

Sixty-five [cerebral arteriovenous malformation nidus samples](#) were collected, among which 28 were ruptured and 37 were un-ruptured. Then, next-generation RNA sequencing was performed on all of them to obtain [differential gene expression](#) (DEGs) between the two groups. In addition, [bioinformatics analysis](#) was performed to evaluate the involved biological processes and pathways by [Gene Ontology](#) and [Kyoto Encyclopedia of Genes and Genomes](#) analysis. Finally, they performed a univariate [Cox regression](#) analysis to obtain the early rupture-prone DEGs.

A total of 951 genes were differentially expressed between the ruptured and un-ruptured BAVM groups, of which 740 genes were upregulated and 211 genes were downregulated in ruptured BAVMs. Then, bioinformatics analysis showed the biological processes and pathways related to the inflammatory processes and extracellular matrix organization were significantly enriched. Meanwhile, some downregulated genes are involved in cell adhesion and genes participating in response to muscle activity and the terms of nervous system development. Finally, one hundred twenty-five genes, many were involved in inflammation, were correlated with the early rupture of BAVMs.

The upregulated genes in the [ruptured cerebral arteriovenous malformation](#) group were involved in inflammatory processes and [extracellular matrix organization](#). Some of the downregulated genes participated in [cell adhesion](#) and [myofibril](#) assembly, indicating the role of enhanced inflammation and reduced inflammation vessel strength in [ruptured cerebral arteriovenous malformation](#) <sup>1)</sup>.

<sup>1)</sup>

Li H, Yan Z, Huo R, Ya X, Xu H, Liu Z, Jiao Y, Weng J, Wang J, Wang S, Cao Y. RNA sequencing analysis between ruptured and un-ruptured brain AVM. Chin Neurosurg J. 2022 Jun 2;8(1):13. doi: 10.1186/s41016-022-00282-4. PMID: 35655323.

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