

## Abstract

### **The Role of Alterations in the *SLC2A10*, *FBLN5*, and *ELN* Genes in the Likelihood of Developing Tortuous Arteries**

Tortuosity is defined as the bending and twisting of blood vessels, hindering the efficiency of blood flow to their respective organs and tissues. Researchers frequently report this condition in elderly populations and label it as a risk factor for transient ischemic attacks. Aging, atherosclerosis, and hypertension, among other conditions, have been linked to the development of tortuous arteries. Multiple studies have focused on surgical interventions aimed to eliminate or alleviate the symptoms. However, its etiology is still not widely understood, especially from a genetic perspective. The current study aimed to determine the genetic predisposition of developing tortuous arteries based on occurrences of SNPs in the *SLC2A10*, *FBLN5*, and *ELN* genes, as determined by next generation sequencing. The study examined eight human cadavers for the presence of tortuosity. A synonymous SNP in *FBLN5* [g.92347680A>G (p.Ile315=)] was detected in six patients and a nonsynonymous, missense SNP in *ELN* [g.73470714G>A (p.Gly422Ser)] was detected in four patients. Further analysis revealed these variations are not associated with minimal to severe tortuosity of arteries. Future studies should include whole-genome sequencing for identification of additional candidate genes and variants for tortuosity.