Liquid biopsy may avoid complications, improve treatment for children with arteriovenous malformation gene mutations

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A study presented today at the Society of NeuroInterventional Surgery's (SNIS) 20th Annual Meeting indicated that it may be possible to use a less invasive technique to identify the gene mutations responsible for some arteriovenous malformations (AVMs) in pediatric patients.

AVMs, including vein of Galen malformations, which are usually diagnosed soon after birth, are tangled blood vessels that disrupt blood flow and oxygen circulation, and are most often found in the brain and spinal cord. These vessels can rupture and cause brain hemorrhage, stroke, and brain damage for children and adults. Many complex AVMs in children are caused by a few specific gene mutations, but it's difficult to find out which mutation is responsible (and therefore, which treatment to use) without taking a surgical biopsy of the AVM.

A surgical biopsy comes with a risk of brain bleeding. As a result, researchers in Australia opted to try using "liquid biopsy"—taking blood samples from veins near or within the AVM—to identify the gene mutations and move forward with treatment plans for the affected children. They were particularly interested in the local mutations in vein of Galen malformations, which have not been previously identified because the risk of a surgical biopsy was too high.

In their study, "Liquid Biopsy Identifies Somatic KRAS Mutations in Paediatric Cranio-Spinal Arterio-Venous Malformations: Preliminary Results," the researchers were cleared to take liquid biopsies from 11 patients with documented brain or spine AVMs. So far, seven patients have received genetic testing, and three of those patients were found to have gene mutations related to AVM. Two patients have commenced treatment with gene-directed targeted medications, with improved health or reduced symptoms since their treatment.

"These early results are promising and show that liquid biopsy is a potential option for identifying gene targets for pharmacotherapy in

these complex AVM cases in children that cannot be safely biopsied by open surgery," said Dr. Kartik Bhatia, a pediatric interventional neuroradiologist Sydney Children's Hospitals Network in Australia. "Being able to more easily and safely identify these gene mutations means that more children with these disorders may be able to get access to treatment that improves their quality of life."

Provided by Society of NeuroInterventional Surgery

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