Novel Vascular Anastomoses and Moyamoya Disease in a Woman with Down Syndrome

Miles Timothy Byworth, James Ian Moffatt, Kanjana S Perera

Keywords: Moyamoya, Ascending pharyngeal artery, Carotid-vertebrobasilar anastomoses, Occipital artery, Down syndrome, Stroke, Stroke imaging, Vascular neurology, Vascular, Brain development

doi:10.1017/cjn.2020.195 Can J Neurol Sci. 2021; 48: 417–418

A 56-year-old woman with Down syndrome presented with right-sided weakness and dysarthria, and was found on CT/CTA to have a left middle cerebral artery infarct secondary to moyamoya disease. Her left posterior inferior cerebellar artery (PICA) was fed both by the left vertebral artery, and the left ascending pharyngeal artery (APA), with a variant origin from the internal carotid artery (ICA), then passing through the jugular foramen (Figure 1). Her right PICA originated exclusively from her right occipital artery, also via the jugular foramen (Figure 2). The left vertebral artery originated directly from the aortic arch, whereas the right vertebral artery originated from the brachiocephalic trunk. In addition, she had a trifurcated anterior cerebral artery (ACA), and just prior to this trifurcation, her left ACA was partially supplied by the left ICA, via a superior hypophyseal artery. This case is noteworthy for several reasons. First, though it is exceedingly rare to have the PICA supplied by the jugular branch of the APA, this is the first reported case with an ICA origin of that APA.[1,2] The fact that both PICAs in this patient originate from the anterior circulation should remind clinicians that in unexplained posterior circulation infarctions, vascular anatomy should be explored, as carotid-vertebrobasilar anastomoses such as these are rare, but possible. Lastly, the conjunction of moyamoya disease and anomalies of the vertebrobasilar system in a patient with Down syndrome raises interesting questions about the influence of trisomy 21 on the developing vasculature. Connections from the APA to the vertebrobasilar system are hypothesized to result from a lack of regression of an embryological anastomosis, in line with the more common persistent trigeminal and persistent hypoglossal arteries.[1] Patients with moyamoya disease have a significantly higher rate of persistent carotid-vertebrobasilar anastomoses than the general population,[3] and are also 26 times more likely to have Down syndrome.[4] Correspondingly, patients with Down syndrome have significantly higher levels of moyamoya disease, and are more than 10 times as likely as the general population to have abnormalities of the Circle of Willis[5] and vertebral arteries.[6]

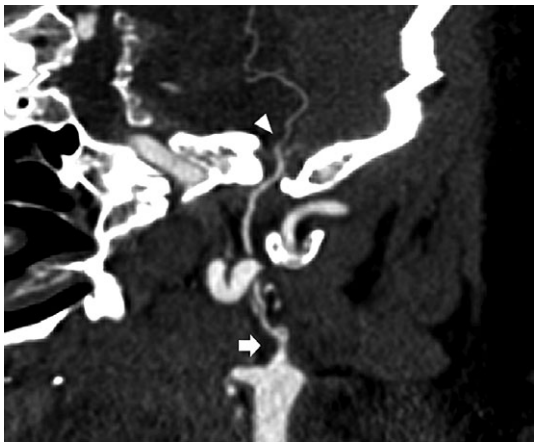

Figure 1: *Left ICA to PICA via the APA.*
Origin of the left APA from the left ICA (arrow) with a branch passing through the jugular foramen to supply the left PICA (arrowhead).

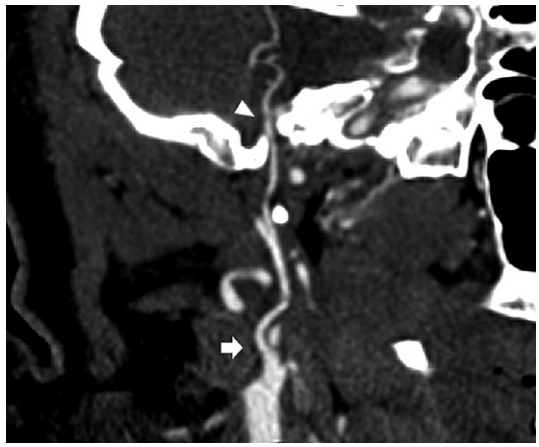

Figure 2: *Right ECA to PICA via the occipital artery.*
Origin of the right occipital artery from the right external carotid artery (arrow) with a branch passing through the jugular foramen to become the right PICA (arrowhead).

From McMaster University, Department of Neurology, Hamilton, ON, Canada (MTB, KSP); and McMaster University, Department of Radiology, Hamilton, ON, Canada (JIM)

Received April 7, 2020. Final Revisions Submitted August 6, 2020. Date of Acceptance August 27, 2020.

Correspondence to: Miles Timothy Byworth, Department of Neurology, Hamilton General Hospital, 237 Barton St. E, Hamilton, ON L8L 2X2, Canada. Email: miles.byworth@medportal.ca

Several genes on chromosome 21 are known to affect angiogenesis, namely collagen XIII/endostatin (COL18A1), DYRK1A, and Down syndrome candidate region 1 (DSCR1), possibly through inhibition of VEGF activity.[7] Whether additional copies of these genes are responsible for the anomalous vascular development seen in Down syndrome, in turn predisposing to the development of moyamoya disease, could benefit from further exploration.

Disclosures

Dr. Perera reports grants and non-financial support from Bayer AG, personal fees from Abbott, outside the submitted work. The remaining authors have nothing to disclose.

Statement of Authorship

MTB: Conceptualized study; interpreted the data; drafted the manuscript for intellectual content. JIM: Major role in the acquisition of data; interpreted the data. KSP: Conceptualized study; revised the manuscript for intellectual content.

References

1. Fujihara F, Takahara M, Katsuta T, Takemoto K, Higashi T, Inoue T. Posterior inferior cerebellar artery originating from the jugular branch of the ascending pharyngeal artery. NMC Case Rep J. 2019;6:21–24.
2. Uchino A, Suzuki C. Posterior inferior cerebellar artery supplied by the jugular branch of the ascending pharyngeal artery diagnosed by MR angiography: report of two cases. Cerebellum. 2011; 10:204–07.
3. Uchino A, Saito N, Takahashi M, Kurita H, Ishihara S. Cerebral arterial variations associated with moyamoya disease diagnosed by MR angiography. Neuroradiol J. 2014;27:697–701.
4. Kainth D, Chaudhry S, Kainth H, Suri F, Qureshi A. Prevalence and characteristics of concurrent Down syndrome in patients with moyamoya disease. Neurosurgery. 2013;72:210–15.
5. Mito T, Becker L. Vascular dysplasia in Down syndrome: a possible relationship to moyamoya disease. Brain & Dev. 1992;14:248–51.
6. Rathore M, Sreenivasan V. Vertebral and right subclavian artery abnormalities in the Down syndrome. Am J Cardiol. 1989;63: 1528–29.
7. Greene A, Kim S, Rogers G, Fishman S, Olsen B, Mulliken J. Risk of vascular anomalies with Down syndrome. Pediatrics. 2008;121(1): e135–40.

https://doi.org/10.1017/cjn.2020.195 Published online by Cambridge University Press