Pediatric AVMs: A Case Report of Wyburn-Mason Syndrome in a 7 yr. old

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Introduction
Wyburn-Mason syndrome is characterized as a neuro-oculocutaneous syndrome in which the development of arteriovenous malformations (AVM) are identified not only in the retina, but in the brain as well. Also known as Bonnet-Dechaume-Blanc syndrome and retinoencephalofacial angiomatosis, it is a rare vascular disorder that is congenital in origin, often creating multiple AVMs in various locations in the brain and body throughout the lifespan.

Clinical Manifestations
Average age at presentation is 2nd or 3rd decade with symptoms of headaches, seizures, visual disturbances, and/or subarachnoid or intraventricular hemorrhage with a ruptured AVM. Additionally, 8% of patients with intracranial AVMs also present with ipsilateral retinal AVMs. They may be accompanied by AVMs in the facial skin, oronasopharynx, orbits, lung, and bone.

Presentation
AC presented in 2008 at 17 months old s/p tonic-clonic seizure requiring intubation. CT scan showed enlarged ventricles, intraventricular hemorrhage, and small right basal ganglia hemorrhage. Diagnostic angiogram demonstrated right thalamic AVM and an associated deep venous aneurysm/pseudoaneurysm. This was treated with 0.3ml NCBA glue (Onyx™) with occlusion of a substantial portion of the AVM as well as the venous aneurysm. During this hospitalization, she received a VP shunt and feeding tube for poor nutrition. She was discharged after 6 weeks to an inpatient rehab center for an additional 6 weeks.

Treatment Plan
Five months after initial treatment with NCBA glue, her angiogram continued to show two dominant feeding vessels, but no venous aneurysm/pseudoaneurysm. At age 3, she was treated with stereotactic radiosurgery (Gamma Knife™) using 32 shots (4mm and 8mm) using a dose of 16Gy to the 50% isodose line.

Follow up angiogram at age 7 showed an incomplete obliteration of the existing AVM and an additional development of vascular growth along the right subfrontal region with a small nidus. A second Gamma Knife™ treatment was performed using 16 shots (4mm, 8mm and 16mm) using a dose of 16Gy to the 50% isodose line. Due to the size of the remaining portion of the AVM, it was elected to treat this AVM in two separate sittings, approximately 9 months apart. The third procedure was terminated due to an increase in contrast enhancement on the MRI, suggestive of radiation effect from the prior treatment. Additionally, two small pial cerebellar AVMs were noted close to the midbrain and thalamus that were not previously seen on angiogram or MRI.

Long-Term Care
Consideration should be given to serial MRI and angiographic evaluation of the AVM as needed. The development of angiomatosis outside of the intracranial cavity should be monitored, particularly when inability to provide hemostasis may provide a significant issue (after trauma, abnormal bleeding episodes, etc.). Continuous cognitive therapy should be provided to ensure age-appropriate developmental growth. Physical therapies and educational support may also be provided based on the individual needs of the child. As a life-long congenital illness, support should also be provided to the parents/caregivers for needed services, financial support and emotional distress.

References

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