Sturge-Weber syndrome associated with arteriovenous malformation in a patient presenting with progressive brain edema and cyst formation

Case report

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Sturge-Weber syndrome (SWS) is a neurocutaneous disorder presenting with a facial port-wine stain, along with an occipital leptomeningeal angiomatosis that is typically located ipsilateral to the stain. In this paper, the authors present a rare case of SWS associated with an arteriovenous malformation (AVM) instead of an angiomatosis in the ipsilateral occipital lobe. While the patient was in the care of the authors, the AVM progressively enlarged, and was accompanied by progressive stenoocclusive changes of the venous system. The resulting brain edema finally brought about a serious neurological condition 13 years after the initial diagnosis. Transarterial embolization and medical treatments decreased the edema. Subsequently, however, a large intraparenchymal cyst appeared, aggravating the patient’s motor weakness. Aspiration of the cyst ameliorated these symptoms. The analysis of the fluid from the cyst revealed that it contained a very high concentration of protein. Although there is no proven pathogenic mechanism to explain these protein concentrations and the enlargement of the AVM, the authors hypothesize that the progressive edema resulted from a synergic augmentation of the inflow from the AVM and the progressive obstruction of venous drainage that is a hallmark of SWS. The formation of the cyst probably resulted from the blood vessel hyperpermeability that is inherent to SWS. (DOI: 10.3171/2010.1.PEDS09140)

**KEY WORDS** • Sturge-Weber syndrome • arteriovenous malformation • brain edema • cyst • embolization

**Abbreviations used in this paper:** AVM = arteriovenous malformation; MCA = middle cerebral artery; MMA = middle meningeal artery; SSS = superior sagittal sinus; SWS = Sturge-Weber syndrome.
had a posterior third occlusion causing sluggish blood flow in the region. We determined that the subcutaneous mass resulted from an engorged scalp vein, which was connected to the SSS by a diploic vein (Figs. 2C and D), and the lesion was consistent with sinus pericranii. No treatment was considered because the patient did not exhibit any neurological deficits.

When MR imaging was used for evaluation at 9 years of age, the occipital A VM appeared as a large cluster of flow voids, accompanied by localized white matter edema (Fig. 3A). At this point, the patient presented with homonymous hemianopia on the right side. Despite the fact that follow-up MR imaging analysis revealed a gradual expansion of the edema, the patient showed no remarkable change in her neurological condition other than infrequent epileptic seizures. When the patient became 16 years old, she showed a limitation of ocular movement (one-and-a-half syndrome) and right hemiparesis, followed by a disturbance of consciousness. Magnetic resonance imaging analysis revealed further expansion of the edema, producing midline shift as well as the development of a small cyst in the basal ganglia (Fig. 3B), and CT showed inherent calcifications as well as the cyst (Fig. 3C).

Angiograms revealed a marked enlargement of the AVM within the temporoorcippital region (Fig. 4A). These images indicated that the lesion was now also fed by the MMA, a feature not observed on any previous angiograms (Fig. 4D). On the venous side, the SSS and the left transverse sinus were now completely occluded. It was difficult to find evidence of the ascending cortical veins in the left hemisphere. An engorged vein of Galen and a vein running parallel to the SSS apparently served to drain the AVM (Fig. 4B). Ascending cortical veins in the right hemisphere showed evidence of flow stagnation, and the drainage of this region was dependent upon the superficial middle cerebral vein (Fig. 4C).

**Operation and Postoperative Course.** An osmotic agent and a glucocorticoid were administered to the patient upon her admission to our hospital, and transarterial embolization was performed to reduce the volume of blood entering the AVM. To avoid an abrupt alteration of blood flow, the embolization procedure was divided into 3 sessions that took place at intervals of 7 days. Each procedure was performed under local anesthesia. A microcatheter was positioned into a feeding artery, and a provocation test was performed in advance of the embolization. In the provocation test, 20 mg of lidocaine was injected into the feeding artery. If the patient did not show any neurological aggravation after the injection, the test was judged to be “negative,” and N-butyl cyanoacrylate (diluted by 20–30%) was injected into the artery. During the first session, the embolic agents were injected using 3 branches of the MMA. For the second procedure, 2 branches of the MCA and a branch of the MMA were embolized. The embolic agents were injected using a branch of the MMA and a branch of the MCA during the third session (Fig. 4E). Angiograms obtained after the third procedure indicated that the flow from the MMA was markedly reduced (Fig. 4F), and the embolization slightly decreased the inflow from the MCA.

Following the embolization treatments, the patient’s condition improved. The disturbance of consciousness resolved, her ocular movement soon normalized, and the

**Fig. 1.** Photograph showing the port-wine stain noted on the ophthalmic distribution of the left trigeminal nerve.

**Fig. 2.** Left (A and B) and right (C and D) internal carotid angiograms of the patients obtained at 3 years of age. The anteroposterior (A) and lateral views (B) of the arterial phase reveal AVMs in the temporoorcippital region and basal ganglia. The left sigmoid sinus is occluded in these images. The anteroposterior (C) and lateral views (D) of the venous phase document the occlusion of the distal SSS. Note an engorged scalp vein in conjunction with the SSS (arrow).
right hemiparesis also improved. The patient was able to resume a normal life and return to school. An MR image obtained 6 months later revealed a decrease of the edema but a slight enlargement of the cyst (Fig. 5A). The patient presented with a worsening right hemiparesis 17 months after the embolization regimen, and MR imaging showed a marked enlargement of the intraparenchymal cyst (Fig. 5B). Although Gd-enhanced images demonstrated an oval enhanced area adjacent to the cyst, it was considered a dilated vein instead of a tumor (Fig. 5C).

The patient’s neurological deterioration was ascribed to a mass effect due to the cyst. We punctured the lesion using a stereotaxic apparatus, aspirated the fluid, and placed an Ommaya reservoir into the cavity. The fluid obtained from the cyst contained a very high concentration of protein (2713 mg/dl), but red and white blood cells were not detected. The procedure resulted in collapse of the cyst (Fig. 6), and the patient’s right hemiparesis improved soon after the procedure. A follow-up CT scan, however, indicated that fluid was once again accumulating in the lesion. The fluid was aspirated from the reservoir 3 months after the initial drainage session. Aspiration is now used to treat the patient, depending on the size of the cyst, as revealed by regular follow-up CT analysis. The patient’s condition has been stable for 30 months after the first aspiration, and she has mild right hemiparesis, hemianopia, and infrequent seizures.

**Discussion**

Leptomeningeal angiomatosis in SWS is characterized by an increase in the number of capillaries and of a few small veins throughout the affected pia mater and underlying surface of the cerebral cortex. The angiomatosis has been ascribed to the failure of the primitive cephalic venous plexus to regress and properly mature. It is presumed that the differentiation failure occurs at approximately 5 to 8 weeks of gestation.

Our patient presented with abnormal vessels in the occipital lobe that were ipsilateral to the facial angiomata. However, the arteriovenous shunts formed by the abnor-
mal vessels prompted us to give a diagnosis of an AVM rather than a more typical angiomatosis. Most theories definitively state that an AVM occurs during embryogenesis and that the lesion is present at birth, but the origin of the condition remains undetermined. Mullan et al. speculated that AVMs form during a stage of absorption of the multiple pial-dural subarachnoid veins at 9 to 12 weeks of gestation (fetal body length 40–80 mm), allowing for further growth of the lesion. Several reports have described cases of SWS that were associated with AVMs. Laufer and Cohen reported a case of SWS associated with large hemispheric AVMs, although the venous structure was not mentioned. Two other case reports demonstrated SWS variants that were associated with multiple AVMs as well as venous sinus occlusion, but neither case showed progressive brain edema as noted in our case. All reported cases of SWS associated with AVM occurred in infants, but the underlying mechanism for such an association remains unknown. Perhaps the failed brain vessel differentiation connected with cases of SWS that are associated with an AVM occurs at a different stage than what is accepted for classic cases of SWS.

It is presumed that the main factor responsible for neurological complications of SWS is the chronic ischemia of cortical tissue that occurs because of impairment of venous drainage, resulting in cortical atrophy accompanied by neuronal loss and astrogliosis in the affected region. Our patient presented with progressive brain edema instead of atrophy, but the venous system showed the typical stenoocclusive changes that are the hallmark of SWS. It is likely that the edema resulted from a synergistic effect provided by the progressive obstruction of venous drainage and the increasing shunt flow due to the growth of the AVM. Several authors have suggested that AVMs tend to enlarge in young patients, but a proven pathogenic mechanism to explain the growth of AVMs in these patients is still lacking. In our case, the AVM not only grew but also developed a new blood supply from the external carotid artery. These observations indicate that AVMs might possess a self-proliferative or innate potential for expansion.

Our patient developed an intraparenchymal cyst that emerged in the basal ganglia. The cyst markedly enlarged after the edema decreased. The growth of the cyst led

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**Fig. 4.** Angiograms and a fluorogram obtained in the patient before (A–D) and after (E and F) embolization. Arterial phase (A) and venous phase (B) left internal carotid angiograms show an enlarged AVM and the disappearance of almost all of the ascending cortical veins. A venous phase (C) right internal carotid angiogram demonstrates the occlusion of the whole length of the SSS. A left external carotid angiogram (D) reveals newly developed feeders from the MMA. A fluorogram obtained after final embolization (E) shows glue casts in the MMA and MCA. A left external carotid angiogram obtained immediately after final embolization (F) demonstrates a marked reduction of flow via the MMA.
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to further neurological deterioration in our patient. Several authors reported cases of AVMs that were associated with a cyst, and in most cases, it was suggested that the cyst developed as a result of hemorrhage.\textsuperscript{2,8} Our case, however, experienced no obvious hemorrhagic episodes. Rather, we observed that the cyst fluid contained high concentrations of protein and no blood cells, indicating that the cyst might have been caused by plasma extravasation into the area. Studies examining the ultrastructure of brain vessels affected by SWS have provided evidence of increased capillary permeability, allowing for the abnormal passage of proteins and calcium.\textsuperscript{15} Therefore, we

![Fig. 5. Magnetic resonance images of the patient obtained after the embolization procedure. A: Axial T2-weighted images obtained 6 months after embolization show decreased edema. B: Axial T2-weighted images obtained 17 months after embolization show marked enlargement of the cyst. C: Axial T1-weighted images with Gd enhancement demonstrate a dilated vein (arrow) adjacent to the cyst.](image1)

![Fig. 6. Plain axial CT scans obtained after placement of the Ommaya reservoir reveal the collapse of the cyst. The reservoir tube is observed in the cyst (arrow).](image2)
hypothesize that the affected cerebral blood vessels in our patient may have ultrastructural characteristics of SWS allowing for the steady accumulation of plasma in the cyst. In addition, the fragility of the white matter exposed to the long-standing edema and the decrease in intracranial pressure following the series of treatments performed to treat her condition may have all contributed to the expansion of the cyst.

We have presented a rare case of SWS associated with an AVM. The progressive brain edema observed in our patient might be due to macroscopic vascular abnormality, including an apparent arteriovenous shunt and stenoocclusive change in the venous system. Furthermore, the inherent hyperpermeability of the capillaries affected by SWS might have contributed to the growth of an intraparenchymal cyst containing a very high concentration of protein.

Disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author contributions to the study and manuscript preparation include the following. Reviewed final version of the manuscript and approved it for submission: K Nishino, Y Ito, T Sorimachi, J Shimbo, Y Fujii.

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